Trinity College Dublin
Coláiste na Tríonóide, Baile Átha Cliath
The University of Dublin

"I have the child who has something else."

Entering the Worlds of How Mothers Make Sense of 'Diagnosis' within the context of their Child with a Rare or Undiagnosed Neurodevelopmental Condition in Ireland: An Interpretative Phenomenological Analysis

Thesis submitted in fulfilment for Doctor in Philosophy in Clinical Speech and Language Studies.

February 2024

by Beth Milofsky

Dr. Irene P. Walsh (Primary Supervisor), Dr. Caroline Jagoe (Co-Supervisor)

Department of Speech and Language,
School of Linguistic, Speech and Communication Sciences Therapy, Trinity College Dublin

Declaration

I declare that this thesis has not been submitted as an exercise for a degree at this or any other university and it is entirely my own work.

I agree to deposit this thesis in the University's open access institutional repository or allow the Library to do so on my behalf, subject to Irish Copyright Legislation and Trinity College Library conditions of use and acknowledgement.

I consent to the examiner retaining a copy of the thesis beyond the examining period, should they so wish (EU GDPR May 2018).

Signature:  Date: 26/02/2024

Summary

Information on the experiences of parents whose children have rare or undiagnosed conditions is lacking in Ireland. It can be hypothesised that these parents experience a unique set of challenges, in comparison to parents of children with conditions which are more widely understood. To the researcher's knowledge, this is the first qualitative study to exclusively explore how parents make sense of 'diagnosis' in relation to their child who has a rare or undiagnosed condition, living in Ireland. It also affords the opportunity to interrogate 'diagnosis', as a phenomenon, more broadly. Five mothers participated in semi-structured interviews with the researcher online, via the video-conferencing platform Zoom (Zoom Video Communications Inc., 2016). One participant volunteered a subsequent written update, which was included in the data set. The interviews were transcribed, and the data was pseudonymised. Information which was considered potentially identifying for the participants or their families was redacted. Interpretative phenomenological analysis (IPA) was the primary analytic method and methodology, with some borrowed insights from narrative analysis (NA).

IPA's main theoretical underpinnings include: (1) phenomenology; (2) hermeneutics; and (3) ideography. Phenomenology accounts for IPA's focus on lived experience. 'Daisein' (Heidegger, 1927/2008) relates to the idea that one cannot be disentangled from pre-existing cultures and comes to the world with existing views. This position recognises the researcher as an inextricable part of the research process, in line with IPA's hermeneutic phenomenological stance. The 'fusion of horizons' (Gadamar, 2004), refers to the belief that one's interpretation of the present is a product of the interaction between our current existing horizons fusing with meaning attributed to our continuing life experiences. IPA is said to involve a 'double hermeneutic' in that the researcher is making sense-of the participant, who is making sense of their experiences. The 'hermeneutic circle' refers to the idea that to understand the whole, one needs to examine its parts, and to understand its parts, one needs to consider the whole. Idiography pertains to IPA's commitment to how a *particular* experience is made sense-of, by a *particular* individual, at a *particular* point in time. The involvement of the researcher is considered a laudable element of IPA. As recommended within quality IPA, I engaged in continuous reflection and reflexivity in order to understand my own horizons of understanding and the potential influences on the research. I attended to recommended quality markers and domains for qualitative research, and specifically IPA.

Analysis was consistent with IPA methods and involved attending to each individual case on its own in full, before considering cross-case analysis to identify group level themes. I identified six general experiential themes (GETs) from my iterative analysis of the date, with several subthemes, as follows: (1*) Entering the world of diagnosis: "There's nothing wrong but everything is wrong";* (2) *The world of rare disease: What's in a name?;* (3) *The world of mothers in healthcare systems: "You adjust to it because you have to, because your child needs you to";* (4) *Etching of the mothers' inner worlds: "I'll never forget"*; (5) *Living within a sociocultural world: Constructions and perceptions of 'disability' - "Why do I have to use the word disorder to describe my child?"*, (6) *The world of the sibling: "Second fiddle".* The findings suggest it is important that healthcare professionals attend to the common and unique needs and preferences, of these parents (i.e., hearing parents' concerns, 'boundary-ing' parent roles to prevent pressure, transparency, and better co-ordination of care). In terms of healthcare communications, the mothers expressed a desire for candid communication, clear information regarding the diagnostic processes (i.e., being prepared for an appointment, a private space, opportunity for follow-up care) and other medical information relating to their child, and greater attention to the words and non-verbal communication used when speaking with parents and families. This study provides consensus on parents' wish for a specifically dedicated support group for parents of children with rare or undiagnosed conditions, and sibling supports.

The findings may suggest an extension to the definition of diagnosis, as defined by Blaxter (1978) in terms of 'category' and 'process', with the inclusion of diagnosis as '*relative phenomenon'.* In this way, this study argues for a third dimension to be added to that original concept of diagnosis by Blaxter (1978). Diagnosis as additionally a 'relative phenomenon' speaks to it as *relative* to experience, space, time, and telling. Whilst the findings suggest there is a common trajectory for mothers of children who have rare or undiagnosed conditions, there are unique elements for every individual family. This individual variation may be heard as the participant's narrative 'sense', which may be defined in this study as the emotive force and underlying tension within the participant's account, regarding diagnosis. The findings suggest this position and a participant's overall vantage point on living with diagnosis is not static. Rather, it is in a constant state of evolution through their pre-existing histories and cultures with their present. This study also provides an example of the use of IPA with elements of NA in yielding rich insights into lifeworlds of the participants on the phenomenon being studied.

Implications of the study are discussed in terms of improved healthcare communication, services and procedures and policy, which considers the experience of the family as central to the 'diagnosis' discussion. Implications are also considered in terms of inclusion of information on rare diseases and undiagnosed conditions in undergraduate healthcare education and continuing professional development. Limitations might include the fact that the phenomenon was explored from the position of mothers only. Further research exploring the phenomenon being studied in this research from a multiperspectival dimension (such as fathers, siblings, and healthcare professionals) is recommended to support further advancements in the field.

For my Papa

Acknowledgements

I would like to express my deep gratitude for the five mothers who participated in this study, for their *whole* participation in sharing and recounting their personal experiences with me. I hope that I have been able to interpret your stories in ways that resonate as meaningful for you and that together, we have produced findings that will provide a window into the experience of what 'diagnosis' means for mothers of children with rare and undiagnosed neurodevelopmental conditions, in Ireland. In doing so, I hope the findings will support improvements in undergraduate and clinical education, healthcare communications, services, procedures, and policies to enhance the overall experiences of children and families and stimulate further research in the field. I would also like to recognise the families whom I worked with who provided inspiration for this research.

Words cannot describe my appreciation and admiration for my primary supervisor, Dr. Irene Walsh. Irene has been a role model and teacher to me since my undergraduate degree. She has been a constant force of support and encouragement, calming influence and wisdom for many, many years. Irene has been endlessly generous with her time. Irene has taught me how to be a speech and language therapist, and more than that, a clinician, a researcher, and also how to live, to know and listen to our own bodies. Her presence, ways of thinking and guidance have had a profound influence on my life so far, and in ways that I know will be eternal. I would also like to express sincere thanks to my co-supervisor, Dr. Caroline Jagoe, who has too known and fostered my growth since my undergraduate degree. Caroline has been a source of unbounded knowledge in discussions and giving of her time in providing considered feedback. It has been a privilege and a joy to be able to embark on the PhD journey with you both paving and illuminating the way.

To my incredible Mum and Dad who have always supported me unconditionally. Thank you to my brother, Josh, for having the perfect inspirational quote for every occasion. To my boyfriend Ronan who has been there with loving encouragement since my final month of writing. To my Mama for her constant love, my Great Uncle David for his thoughtful discussions and academic insight and my extended family. To my best friends and work colleagues for whom I am fortunate enough to also call my friends. I am so thankful to you all for your continued confidence in me, good times, joy, and laughter, of which I couldn't have gone without over this period. I look forward to spending even more time with you.

Table of Contents

List of Tables

List of Figures

List of Acronyms

The participants use certain abbreviations in the interviews, which relate to disability and school services in Ireland, as well as health terms or diagnoses. Please see the table of below to support understanding of these terms.

Table A

Acronyms with accompanying Definition and Description

Acronym	Definition	Description
AIM	Access and Inclusion Model	Aims to create a more inclusive environment in pre-schools by providing a graded level of supports to preschool settings.
ASD	Autism Spectrum Disorder.	A diagnostic term as defined in the Diagnostic Statistic Manual-5th Edition (DSM-5, American Psychiatric Association [APA], 2013). In practice, this diagnosis is preferably referred to as 'autism', in line with neurodiversity-affirmative[1] language. Where I refer to the definition in this thesis as defined in the DSM-5 or by other researchers, I will refer to it as ASD. Where I am speaking of autism myself, I will refer to autism or autistic person.
CDNT	Children's Disability Network Team	An interdisciplinary team of health and social care professionals who provide services for children with complex needs within a specific geographical area as per the Progressing Disability Services for Children and Young People Programme (PDS).
DCA	Domiciliary Care Allowance	A monthly payment to the carer of a child with a severe disability who lives at home.

[1] (Reframing Autism, 2023)

EIT	Early Intervention Team	A term previously used to denote a team of health and social care professionals who supported children and families in disability services between birth and five years of age. EITs have been replaced by the CDNT under the PDS reconfiguration.
HSCP	Health and Social Care Professional	A group of professionals in the health service who work across therapeutic, social care and diagnostic domains who may practice in a variety of settings including but not limited to acute, community and disability services.
NG	Nasogastric tube feeding	A form of eternal feeding (i.e. non-oral, mouth, feeding). A tube that enters through the nose and delivers food through the oesophagus to the stomach.
OT	Occupational Therapist	Healthcare Professional.
PDS	Progressing Disability Services	The new structure for children's disability services in Ireland.
PLWRD	Persons Living with a Rare Disease	N/A
SAT	School Intervention Team	A term previously used to denote a team of health and social care professionals who supported children and families in disability services between six and eighteen years of age. SATs have been replaced by the CDNT under the PDS reconfiguration.
SLT	Speech and Language Therapist	Healthcare Professional.
SENO	Special Educational Needs Organiser	Support and advice parents regarding the availability of special classes, special schools and other education supports, within a geographical area.
RDD	Rare disease diagnosis	Will be used by the researcher to denote a specifically named rare disease. I have not named the specific condition in order to protect the anonymity of the participants, the child in reference, their family members and healthcare and social professionals

	involved in supporting their care, for whom this information may be identifying or sensitive if it relates to their living experience of care / teaching, in the case of the professional or educator. See *5.2 Researcher Reflection* for further discussion.

"I have the child who has something else."

Entering the Worlds of How Mothers Make Sense of 'Diagnosis' within the context of their Child with a Rare or Undiagnosed Neurodevelopmental Condition in Ireland: An Interpretative Phenomenological Analysis

1.1 Inspiration for the Research

This research set out to explore the meaning and 'living'[2] experience of 'diagnosis' for parents of children who have a rare or undiagnosed ('SWAN')[3] neurodevelopmental[4] condition in Ireland today. This question came about through my own practice as a clinical Speech and Language Therapist working within an Early Intervention Team (EIT) at a service for children with intellectual disabilities and their families in Ireland. Parents spoke of their unique experience in

[2]I will use the term 'living' as opposed to 'lived' to refer to the participants' experiences as ongoing, to encapsulate that their meaning-making is not fixed and is in constant evolution, resonating with Gadamer's (2004) concept of 'horizon of understanding', whereby one's perspective is a fusion of their previous and current influences, as shaped by continued experience. Gadamar (2004) wrote "The historical movement of human life consists in the fact that it is never absolutely bound to any one standpoint, and hence can never have a truly closed horizon" (p. 303).

[3] Where 'undiagnosed' or 'syndromes without a name' ('SWAN') refers to "a term used when a person is believed to have a genetic condition and testing has failed to identify its genetic cause" (Fletcher-Dallas, 2007, para. 1). These children are often referred to as having 'global developmental delay' or 'failure to thrive' and may present with a range of symptoms and medical needs such as learning and / or physical disabilities, feeding difficulties, epilepsy, respiratory issues, unusual physical features (Fletcher-Dallas, 2007).

[4] The term "neurodevelopmental disorders" (NDDs) was first introduced as an umbrella diagnostic category in the Diagnostic and Statistical Manual, fifth edition (DSM-5; APA, 2013). The DSM-5-TR (APA, 2022) defines neurodevelopmental disorders as "a group of conditions with onset in the developmental period. The disorders typically manifest early in development, often before the child enters school, and are characterized by developmental deficits or differences in brain processes that produce impairments of personal, social, academic, or occupational functioning" (p.35). Neurodevelopmental disorders include intellectual developmental disorders such as global developmental delay, communication disorders, ASD, ADHD, specific learning disorder, and stereotypic movement disorder. Specifiers can also be applied to acknowledge the etiology of the condition such as "associated with a known genetic or other medical condition or environmental factor" (DSM-5-TR; APA, 2022, p. 37).

relation to having no overarching[5] diagnosis for their child. They spoke of isolation, fear and the lack of parent support groups, particularly in comparison to other more commonly known or recognised diagnoses such as autism and Down syndrome.

From my interactions with children and families in the clinical context, parents express a desire to be connected with other families who are experiencing a similar situation. Reflection on these parents' insights also inspired my interest to better understand the construct, culture, and phenomenon of 'diagnosis' in Ireland at large, that is, what it means to both the individual and society. To better understand the meaning of 'diagnosis' seemed pertinent in order to enhance professional knowledge and to identify gaps within policy and services within the Irish healthcare system. The ultimate aim of this increased understanding is to improve the quality of experiences for children and their families as they navigate their journey through the Irish healthcare system. The importance and prominence of this research question continues to be reinforced for me through my everyday interactions and ongoing clinical relationships while currently working within a children's disability network team (CDNT) in Ireland.

1.2 Research Questions

1.2.1 *Main Research Question*

How is the phenomenon of 'diagnosis' experienced in Ireland today by mothers within the context of their child with a rare or undiagnosed neurodevelopmental condition?

1.2.2 *Sub Questions*

- How is the construct of 'diagnosis' perceived in the contemporary context in Ireland?
- How does 'diagnosis' interact within daily life for the individual and society today, in the context of childhood rare or undiagnosed neurodevelopmental conditions?
- What importance does 'diagnosis' hold for parents of children with diagnosed and undiagnosed conditions, in Ireland?
- What are the unique challenges concerned with the experience of parenting children with diagnosed and undiagnosed conditions?

[5] The child may have a collation of medical symptoms such as epilepsy, feeding difficulties, low tone, learning disability but no overarching name which can explain for the collection of symptoms. The definition of 'SWAN' will be further discussed in *3.2 Current Landscape: Rare Conditions and Undiagnosed 'Syndromes Without A Name ('SWAN') Policy and Service Advancement.*

- How is disability and parenthood lived and perceived in Ireland today?
- What can health professionals and others learn from parents' experiences?
- How can these parental accounts inform service and policy improvement within the Irish healthcare system?

1.3 Research Design

This is an exploratory, qualitative study using interpretative phenomenological analysis (IPA) as the primary analytic methodology, with borrowed insights from narrative analysis. Participants were recruited using purposive sampling. Five individual in-depth semi-structured interviews were carried out with mothers, via an online video conferencing platform (i.e. Zoom, Zoom Video Communications Inc., 2016). Video recordings with audio were saved using this platform. To avoid the risk of any researcher bias, any participant who had previously or was currently working directly with the researcher in providing clinical interventions with their child were excluded from participating. There was no age restriction for the child whom the participant was referring to (i.e. the child may have been over 18 years of age), as it was anticipated that the mothers' experiential accounts could reference both retrospective and current experiences relating to their child.

1.4 Rationale for the Current Research

1.4.1 *Novel Accounts*

To the researcher's knowledge, this is the first study to exclusively explore parents' sense-making surrounding the concept of 'diagnosis' where their child has a rare or undiagnosed conditions, living in Ireland, using IPA, an in-depth qualitative analytic method. In accordance with the research design, the study aims to provide novel insights into the lifeworlds[6] of the participants on the phenomenon being studied. This research therefore aspired to contribute novel accounts to the Irish healthcare literature base and to 'illness narrative'[7] as a whole.

[6] Mishler (1984) introduced the idea of the 'voice of medicine' and the 'voice of the lifeworld' as representing two different ways of conceptualising and understanding patients' problems. Mishler (1984) posited that humane medical care requires increased attentiveness to the voice of the lifeworld (VoL) as defined as "patient's contextually-grounded experiences of events and problems in life, timing of events and significance are dependent on biographic situation and position in social world" (p.104).

[7] The term 'illness narrative' will be elaborated on in *1.4.3 Illness Narrative* below and further in *3.3 Illness Narrative: Definitions and Considerations.*

Information on the experiences of parents whose children have rare or undiagnosed conditions is particularly lacking in Ireland. The Health Service Executive (HSE) has acknowledged that, "In Ireland, there is limited information available for those who have an undiagnosed condition" (*Information for the undiagnosed,* n.d., para 1). Exploring the parents' 'living' (i.e., to suggest or emphasise ongoing experience as explained in footnote 2) experience may help to illuminate gaps within the Irish Healthcare system. Implications of this research could therefore lead to the creation of new systems, which could support parents in navigating and procuring relevant and meaningful services.

The notion of 'Dasein' is integral to the phenomenological aspect of IPA. '*Dasein*', a term coined by philosopher Martin Heidegger (1927), acknowledges that one is born into a pre-existing world and cannot be detached from pre-existing cultures and people and objects. This idea further validates the need for the current and contemporary research, which sets out to collect data pertaining to parents' experience of the phenomenon of diagnosis for their child *in Ireland.* The findings as they relate to these parents' experiences are therefore novel in comparison to any similar studies carried out in other parts of the world, as parents' experiences of diagnosis will undoubtedly be influenced uniquely by the Irish context, that is, the historical and current social and cultural constructs of health, family, disability and diagnosis in Ireland.

1.4.2 *Unique Set of Challenges*

It can be hypothesised that parents of children with rare or undiagnosed neurodevelopmental conditions experience a unique set of challenges, in comparison to parents of children with other conditions which are more widely understood. These challenges may include lack of professional and public knowledge, uncertainty, delays in genetic testing, lack of a clear diagnosis or disease trajectory, identifying appropriate management and services. The journey for people with rare or undiagnosed conditions is widely referred to as the '*diagnostic odyssey'* (Basel & McCarrier, 2017; Bauskis et al., 2022; Bouwman, et al., 2010; Kole & Hedley, 2021). Although some parental experiences will be shared amongst families raising a child with a diagnosed condition, raising a child whose condition remains undiagnosed contributes a layer of complexity (Lewis et al., 2010). Through my own clinical experience, working alongside parents whose children have no known diagnosis, the experience for these parents is reportedly fraught with fear, isolation and a sense of 'not belonging'. Parents' accounts have indicated that they face an isolating journey that is often pre-occupied with a persistent search for a diagnosis. Parents have also expressed a lack of collective identity, identifying that they do not 'fit' into 'main

diagnosis' groups or services, in comparison to other families whose children present with diagnosed conditions accessing the same service.

1.4.3 *Illness Narrative*

In recent decades, there has been growing respect for the need to hear directly from patients to gain a better understanding of their experiences, hopes, and expectations for interactions in healthcare. Illness narratives were once considered an "orphan genre" (Frank, 1994, p. 2). With the shift from medical to social models of healthcare came growing acceptance and recognition of the importance of patient experience in understanding illness and providing holistic treatment. A recent study carried out in Northern Ireland by McMullan et al. (2020) concluded that care and support for those living with rare disease is inadequate and that further education and training is needed for health care professionals. A pertinent finding was that future research within the realm of rare disease should include carer perspectives. Similarly, Somanadhan et al. (2021) explored the impact of caregiving on families and healthcare professionals and adults living with mucopolysaccharidoses (a group of one of the many rare inherited metabolic disorders) in Ireland. The authors indicated in their implications for policy and practice that further education and training around communicating sensitive information and supporting parents' needs at the diagnosis stage of the condition studied and other rare diseases is needed. Somanadhan et al. (2021) also qualify that there is limited understanding of the family experience of children with rare disorders and that further qualitative research into rare, paediatric, life-limiting illness is required to inform interventions and support both the families and health and social care professionals. Implications from these studies and an emerging body of research on rare disease internationally provide further justification for the current research.

1.4.4 *Contribution to National and International Government Health Policy Agenda and Service Reform*

> People living with rare conditions are psychologically, socially, culturally and economically vulnerable, facing discrimination and challenges in healthcare, education, housing, employment and leisure. (Rare Diseases Ireland, 2021, para 3.)

The need to address challenges for people living with rare diseases and their families amongst government policy and healthcare has received growing recognition in recent times, following many years of campaign from national alliances, advocacy groups, charities and individuals. Many

advancements have been made in directing government agendas for action in this regard with the development of documents such as the 'National Strategy for Accelerating Genetic and Genomic Medicine in Ireland' (Health Service Executive, 2022), 'Recommendations from the Rare 30 Foresight Study: The future of rare diseases starts today' (Kole et al., 2021), 'An Easyguide to Rare Diseases in Ireland and Consensus for Action' (Rare Disease Taskforce, 2020), 'National Plan for Rare Diseases 2014-2018' (Department of Health, 2014), 'Recommendations for the development of National Plans on Rare Diseases: Guidance Document (Europlan, 2010) and the 'Position paper: Patient's priorities and needs for rare disease research, 2014-2020' (Eurordis, 2011). These papers ultimately outline that the needs of those living with rare disease have been unintentionally marginalised, given the inherent nature of some condition being less common, affecting less of the population, and being less understood in terms of a clear diagnosis, disease trajectory and treatment.

The aims of this research project align with, and may fulfil, elements of national and European government agendas and pursuit, as identified through earlier plans and research. A summary review of the current landscape of existing supports, services and advancements, within the field of rare disease is included in the literature review (see *2.6 Current Landscape: Rare and 'Syndromes Without a Name' (SWAN) - Policy and Service Advancement*).

1.5 Summary

In summary, this research projects aimed to provide entry into the living experiences of the participants to understand how the phenomenon of 'diagnosis' is made sense of by mothers of children with rare or undiagnosed neurodevelopmental conditions *in Ireland*. This research is specific to illuminating new understanding on the experience in the Irish context specifically, at the present time the research was being conducted. Caregiver experience of rare disease is an under-researched area, particularly within the Irish cultural context and therefore, this study hopes to add new findings to the healthcare literature base. The study aims to fulfil research needs as identified by local and government political agenda, and previous research. By offering novel insights into the experience being studied, the research aims to contribute to elevated levels of professional understanding and in doing so, improve the quality of care, experiences and interactions for children and families within Irish healthcare systems and society. In the subsequent chapters, Chapters 2 and 3 I will provide background literature relative to the study, Chapters 4 and 5 will focus on Methods and Methodology, in Chapters 6 through 12 I will present

the study's findings and in Chapters 13 to 15 I will conclude with the Discussion. I have also included researcher reflections in boxes throughout the thesis.

Chapter 2: Literature Review: Historical and Theoretical Underpinnings of 'Diagnosis' Across Fields

2.1 Introduction

This chapter introduces the cultural context of 'diagnosis', that is, the concept and socio-cultural construction of diagnosis, while examining its origins and trajectory in psychology, criminology and sociology. Throughout the chapter, I will highlight some of the perceived values of diagnosis against the contested negatives outcomes. This 'diagnosis debate'[8] thread will continue in the second literature review chapter.

2.2 Concept and Construction of 'Diagnosis': What is 'Diagnosis'?

2.2.1 Definition and Practice of 'Diagnosis': A Versatile Concept in both its Linguistic Form and Pragmatic Function

What is 'diagnosis'? 'Diagnosis' can be viewed as both a noun (i.e., a/the diagnosis) and a verb (i.e., to diagnose). Blaxter (1978) terms this characteristic duality as both "category" and "process". The Collins Dictionary (2007) definition of diagnosis aligns with this duality, citing diagnosis as both "an opinion or conclusion so reached" and "the identification of diseases by the examination of symptoms and signs and by other investigations" (p. 456). The origin of the word diagnosis stems from the Greek word 'dia' (apart) and 'gignoskein' to know (Collins Dictionary, 2007). The meaning of the word is highly malleable, subject to alteration in its linguistic form such as by the addition of, (i) an affix, in the case of 'misdiagnosis' and 'undiagnosed', (ii) adjectives such as 'lay', 'medical', 'disputed', 'predictive' and 'differential' or (iii) the suffix to denote occurrences pertaining to diagnosis as 'diagnostic' in the event of 'the diagnostic moment' (Heritage & McArthur, 2019; Jutel, 2019) or the 'diagnostic announcement' in disclosing the news to others (Jutel, 2019).

[8]The 'diagnosis debate' may be considered as referring to the critical analysis of 'diagnosis' as a construct and medical practice, in relation to it being medically, socially, and individually formulated. Many authors including Blaxter (1978), Brown (1995) and Jutel (2009, 2019a, 2019b) have explored the 'existence' (i.e., ontology) of diagnosis with regards to its social creation. Considerations to stigma, legitimacy (in terms of social acceptance and/or medical origin), validation, visibility, experience of 'illness' versus diagnosis, and possible pathologizing effects, are amongst often considered topics within the ongoing 'diagnosis debate'. Some of these issues will be explored in more detail in Chapters 2 and 3, with reference to the literature base.

In the first instance, 'diagnosis-as-category' (Blaxter, 1978), refers to a label, a superordinate name assigned to a collection of signs and symptoms. Diagnosis can be considered both a conclusion and a starting point. Its predecessor is often the emergence of symptoms, which trigger the diagnostic procedure. In the second instance, 'diagnosis-as-process' (Blaxter, 1978) refers to the active work, the process of information gathering, hypothesis formation and testing, culminating in a conclusive finding which sets in motion a sequela of actions such as the exploration of treatment options, communications with other professionals, and family members. In the case of inconclusive findings, this may cause challenges for identifying subsequent steps as previously discussed in the introduction in the case of undiagnosed or rare conditions.

2.2.2 Diagnosis-as-Category

Suzanne Fleishman (1999), a linguist, documents her own experience of illness through a linguistic lens. Fleishman was diagnosed with myelodysplasia or myelodysplastic syndrome(s) (MDS) which is a rare disease. Fleishman (1999) speaks of diagnosis as marking a boundary between life "before" and "after diagnosis". Jutel (2019a) refers to the 'diagnostic moment' as the occasion which brings about this separation. Fleishman (1999) also makes a case for a distinction between "illness" and "disease". She delineates these terms as 'illness' representing the individual's "experience" whereas 'disease' denotes a "category of clinical taxonomy" (p. 7). Her description that *illness* is not experienced in the same way by two people with the same term *diagnosable disease* helping to illuminate the distinction between these terms. Fleischman (1999) examines the language we use to name disease and what this means for how we view disease in position to ourselves. She discusses how some diseases are more acceptingly integrated into the self with 'I am' definitions whereby others are more appropriately positioned externally, outside of the individual, with 'I have' or 'I suffer from'[9]. Fleishman (1999) examines the responsibility on the patient inferred by the different verbs used to ascribe how the disease was attained, for example "fall" as if by accident, and "get" which are passive, in contrast to "catch" which prescribes some level of responsibility. Fleischman (1999) discusses how she refers to her illness as 'diagnosed with' which allows for interpretation that the illness may not be there. She explores additional challenges involved in the case of rare disease which she recounts are "absent from most people's lexicon of disease" (1999, p. 4).

[9] The idea of how language effects how we position a condition in respect of our self and our identity is revisited in *2.5.3 Contributing to the Demise of Previously Recognised Disorders*, in reference to what is currently regarded as preferred terminology surrounding autism diagnosis.

Shanahan (2020) in his book, entitled '*The Language of Illness',* explores what we can learn from a greater understanding of the words we use to describe illness. Shanahan discusses the application of 'labels' to denote normal variance of being human as harmful amongst other negative impacts of labels. Shanahan proports that "normal should tolerate imperfection" (2020, p. 131). This seems to suggest that the definition of normality does not preclude flaw and that flaws are subsumed within what is to be considered normal. He balances his argument of the "dark side" (p. 129) of labels with potential benefits of diagnosis, such as directing treatment and de-mystifying symptoms, as echoed in other research.

2.2.3 Diagnosis-as-Process

The process of diagnosis is not always strictly linear, the diagnostic procedure can involve cyclic events of testing and re-testing varying hypotheses and utilising the evaluation of the effect of treatment to assist in transforming a diagnosis from probable to definitive (where possible). In medicine, it is likely that diagnosis precipitates the treatment. Conversely, in psychiatry, prescription of treatment medication is often used as a tool to aid diagnosis, in that a patient's response to treatment may help to reach a diagnosis (Cooksey and Brown, 1998). Some diagnoses remain 'probable' until the point of medical post-mortem by a trained pathologist, such as to confirm the existence of Alzheimer's disease. Others again, are contained in a constant state of revolving flux, with the diagnostic process not culminating in diagnosis, as is often the case with undiscovered and rare illnesses.

Blaxter (2009), chronicles her experience as a patient, over 141 days between the first diagnosis of a lung tumour on X-ray and the commencement of intervention. She does this explore the 'vanishing'[10] effect of new technologies on the patient. In her work, Blaxter (2009) refers to herself as patient P. She notes potential methodological limitations of the fact the patient, is also the author, and limitations of the single case-study. Blaxter (2009) acknowledges her attempt to bracket emotions but that the potential of emotion colouring the narrative cannot be ignored. The author also discusses the potential influence of her position as a medical sociologist. Blaxter (2009) attributes the day the tumour was identified on X-ray as *Day 1*. The identification of the tumour on visual imagery marking the start of the illness calls to question the authority of

[10] Blaxter (2009) uses the term 'vanish' (p. 762) in this context to describe the process of the patient as person being dissolved or disembodied by medical imaging procedures.

diagnosis: was 'illness' experienced by the patient before confirmation by medical testing or is it only confirmation using medical testing that demarcates the start of the illness journey? Blaxter later attributes the events prior to Day 1 as the "prologue", the "lengthy and confused period" which preceded diagnosis and which she terms the "initial 'silent' disease" (2009, p. 766).

2.2.4 Diagnosis in Medical Discourse

Duchan and Kovarsky (2005), in their edited volume, bring together an array of notable authors to discuss the pervasive nature of diagnosis in Western society, examining how diagnoses are formulated, and experienced by individuals and their families, including the impact of diagnosis. Western culture is very much hinged on procuring a 'diagnosis' to explain presentations. Jones and Beach (2005) analyse the discourse between general practitioners and patients from video recordings of naturally-occurring medical visits, using conversation analysis methods. The authors found that patients use various mechanisms to solicit diagnosis from physicians in early interactions. Jones and Beach's (2005) research illuminates the significance of diagnosis for patients in understanding their presentation and their dissatisfaction when requests to solicit diagnosis are not met by practitioners.

Gill et al. (2010) analyse data from medical consultations in the USA using conversational analysis. The authors delineate the occurrence of 'pre-emptive resistance' (Gill et al., 2010, p. 1) whereby patients describe the problem and offer counter evidence that precludes a possible explanation for the origin of that problem which the patient has already considered and ruled out. They suggest that patients contribute to the sense-making process of symptoms within medical discourses in that patients do more than convey information about their signs and symptoms, but also offer interpretations that may aim to solicit a particular interpretation or course of action on behalf of the medical professional. Conversation on the role of the patient (or patient as carer) in diagnosis, will be continued in *2.5.4 Reaching Diagnosis: A Co-constructed Practice?*.

2.2.5 The Subtle Art of Diagnosis in Everyday Non-medical Transactions

In her book '*Diagnosis: Truths and Tales'*, Jutel (2019a) addresses how diagnosis is conveyed in the arts and the media and what these representations can tell us about the construct and concept of diagnosis. Jutel (2019a) asserts that to truly understand the entirety of diagnosis we must examine its situation within multiple disciplines. Diagnosis exists far beyond the boundaries of the medical profession. Jutel (2019a) talks about diagnosis as a "tool in the scriptwriter's bag" (p. 98). She describes how diagnosis may be used in 'foreshadowing', an

ancient literary device common in Greek tragedy, which creates a sense of 'dramatic irony', whereby the audience know something ahead of the characters (Johnson, 1928; Muecke, 1983). Jutel (2019a) maintains diagnosis of course features in medical dramas such as *Greys Anatomy* (Rimes, 2005-present), *The Good Doctor* (Highmore et al., 2017-present), as to be expected, but that diagnosis is central to the plot of many popular films and TV shows that are not housed in the clinical context, such as *Breaking Bad* (Gilligan et al., 2008-2013) and *Still Alice* (Glatzer & Westmoreland, & Lutzuz, Brown, & Koffler, 2014).

Trix (2015) moves the concept of diagnosis into a more abstract realm entirely, likening diagnosis to the process of reviewing applications from prospective medical faculty members applying to teach at a large university in the United States. Trix (2015) analysed three years of letters of recommendation of successful applicants using methods from corpus and discourse analysis. The prospective faculty members' subjective qualities and objective achievements are viewed as the signs and symptoms from which the eventual label is determined – 'hire' or 'reject'. This example illustrates how the practice of diagnosis permeates our daily activities, without us being necessarily cognisant that we are engaging in the art of diagnosis.

As illustrated and as will be further depicted in *2.5 Diagnosis and the law* below, diagnosis has many functions within and outside the field of medicine. These functions often fall into arguments within the 'diagnosis-is-good' versus 'diagnosis-is-bad' debate. The role of diagnosis and its perceived benefits and criticisms within socioemotional individual and family contexts, legal, health and mental health fields, will be explored in the subsequent subsections. I will not explore in detail in this thesis but would like to briefly mention other examples of how diagnosis is utilised in our everyday interactions, including providing the legal basis for social welfare benefits, a determinant in the procurement of health or life insurance, precluding the pursuit of certain employment and career opportunities (e.g., a history of epilepsy typically prevents someone from being able to become an airline pilot; Epilepsy Ireland, n.d.). These examples further illustrate the authoritative and pervasive cultural practice of diagnosis (as alluded to by Trix, 2015).

2.3 Origin of the Concept of Diagnosis in Psychology

The Diagnostic Statistic Manual of Mental Disorders (DSM), published by the American Psychological Association (APA), is the universally recognised tool used by researchers and clinicians to classify and diagnose mental health disorders. Now in its fifth edition (DSM-5-TR; APA, 2022), the manual has seen many changes, brought about by changing societal attitudes, cultural context, and shifts from a medical to a biopsychosocial conceptualisation of human health and

disease. Revisions of the text have seen further classification of disease and outdated diagnoses discarded. Tracking these changes highlights the impact of context on diagnosis.

The DSM was first introduced as a "common language" (Blashfield et al., 2014) for those involved in the delivery of medical care. Diagnosis functions to facilitate communication about disease amongst clinicians, scientists, and researchers, in health care professions (Surís et al., 2016). The genesis of nosology and nomenclature are regarded as critical precursors and precipitators in the advancement of medicine (Surís et al., 2016). Accurate diagnosis and differential diagnosis can inform patient prognosis and treatment. In 1944, psychiatry was first recognised as a medical specialty in the United States by the APA. The DSM served to legitimise the field of psychiatry.

The initiative to develop standardised diagnostic criteria was prompted by the U.S. Census Bureau to estimate the prevalence of mental disorders for the 1920 census (Surís et al., 2016) and to develop mental health policies (Grob, 1991). By the 1950s, five separate diagnostic classification systems were in use amongst different contexts in the United States (Fischer, 2012). The inaugural edition of the DSM, published in 1952 by the APA, saw the creation of a unified diagnostic system. The International Statistical Classification of Diseases and Related Health Problems (ICD), published by the World Health Organisation (WHO), now, in its eleventh edition (ICD-11; WHO, 2022), was designed to classify data relating to diagnosis or other abnormal findings for storage, retrieval and analysis.

2.3.1 Critical Analysis of the DSM

The establishment of a diagnostic manual was considered to be an objective tool. Cooksey and Brown (1998) reference that diagnosis through DSM provides detachment by separating the person from their social world. Over thirty years ago, Loring and Powell (1988) set out to question the claim that the advent of the DSM minimises the influence of race and gender on the reliability and validity of diagnosis. In their study, the authors provided two case vignettes to 290 psychiatrists. Their findings showed that when information regarding the gender and race of the client was absent, there was a reasonable degree of consensus regarding diagnosis. Their study suggests that the diagnostic process is not exempt from being influenced by gender and race of the client and the clinician. Loring and Powell (1988) recognise that other characteristics may influence diagnostic decision-making such as years of practice or workplace setting. There is a sense in the medical profession that the practitioner must be able to execute their expertise in responding objectively to patient concerns whilst still seeking to attend to the social and

emotional experience of illness for the patient (Kovarsky et al., 2005). The diagnostic process however has been shown to be vulnerable to personal, professional and cultural biases. For example, the diagnosis of 'depression' necessitates a distinction must be made between a depressive episode and what can be considered 'normal' sadness and grief. This example showcases the margin for subjectivity in this interpretation. The reliance on a diagnostic manual has also been said to perpetuate the disparity of power in patient-professional interactions.

2.4 Diagnosis: Shaped by the Individual and Society

> *"Stories echo other stories, with those echoes adding force to the present story. Stories are also told to be echoed in future stories. Stories summon up whole cultures." - Arthur Frank (2010, p. 37)*

The above quote by Arthur Frank (2010) eloquently encapsulates the idea that stories do not exist in isolation, they are shaped by those who came before them, and will have irrefutable influence on those that follow. Here, Frank's (2010) assertion is reminiscent of the concept of 'Dasein', as previously introduced, that people cannot be detached from the world they are born into, and therefore their stories reflect the prevailing cultures of the time. In this subsection, I will discuss the literature surrounding the interrelationship and connectivity between diagnosis, the individual, and society.

2.4.1 What's in a Name? Diagnosis as Malleable in Response to Social and Political Reform

Does the removal of a "diagnosis" mean to say that the presentation no longer exists or that the presentation is no longer considered outside the realm of "normalcy". The diagnostic procedure using the DSM is hinged on the distinction between normalcy and pathology. At what point do symptoms become pathology? Diagnoses thrive or falter in response to current thinking, the parameters between "normal and problematic" (Jutel, 2019a, p. 281) as set by individuals, professions and society.

Walsh et al. (2018) put forward a narrative account of a therapeutic journey experienced by two of the authors. Delmar is an adult with a diagnosis of autism spectrum disorder (ASD) and Walsh is speech-language pathologist/researcher. In this paper, Delmar's reaction to the revision of Asperger's syndrome from a separate diagnostic category to within ASD can be identified as a challenge to her identity. Her critical remark *"Do I no longer have 'Asperger's'? I'm cured"* (Walsh

et al., 2018 p. 118) astutely provokes the reader to question the relationship between naming a 'thing' and the existence of a 'thing' – does the existence of a 'thing' necessitate its naming or does the naming call into existence the emergence of the 'thing'? And if so, does the removal of the name, cease the existence of the thing previously named? Delmar poses another provoking question *"Whose decision was that?"* (Walsh et al., 2018 p. 118). This question opens the readers' eyes to the cultural influences on diagnosis and what bodies facilitate the renaming of disorder. (This question will be further addressed in *2.5.3 Contributing to the Demise of Previously Recognised Disorders*).

2.4.2 Thrive: Assisting the Emergence of 'New' Disease

Lay people are often responsible for the 'discovery' of new named illnesses (Brown, 1995; Jutel, 2009). The patient recounts their experience of a collection of symptoms as a problem to a physician. Brown (1995) expresses that lay-led discoveries may not lead to the legitimisation of illness in their own right and often require the support of social movements to advocate for the existence, and acceptance, of the person's experience. In the case of war veterans, veteran organisations were pivotal in the acceptance of the addition of Post-Traumatic Stress Disorder (PTSD) to the DSM-III (Brown 1995; Jutel, 2009; Scott, 1990). Jutel (2009) highlights that in the case of Alzheimer's disease, it was the advancement of medical technology that facilitated the establishment of the diagnosis, with lay participation serving to later promote the diagnosis, as opposed to deliver its creation. Early patient reports of dementia were previously linked with normal aging. Other sources are cited as responsible for raising public awareness, acceptance and authorisation of illness within society such as through marketing campaigns where "the patient becomes 'consumer'" (Jutel, 2009, p. 292).

Conversely, a later study examined and attributed "lay" diagnosis talk about menopause for medicalising a natural life event as something in need of treatment. Suopis and Carbaugh (2005) discuss how 'lay' diagnosis of menopause, a natural stage in the female reproductive cycle, pathologises a natural phenomenon. In their analysis, they maintain however that menopause is viewed differently to medical diagnosis in that it is spoken about as something one is "in" as opposed to something one "has" like a medical diagnosis.

2.4.3 Contributing to the Demise of Previously Recognised Disorders

The DSM-III-R (APA, 1987) saw the removal of "sexual assault disorder", "masochistic personality disorder" and "premenstrual syndrome" (Cooksey & Brown, 1998). Removal of these

terms as identifiable conditions exemplifies the influence of women's rights' movements on diagnostic practices and psychiatry. It was viewed that the first two of these diagnoses may assist in the defence in rape trials by placing blame on the victims and the third, for pathologising the female reproductive cycle. It is interesting to note that diagnostic labels were often first altered in response to societal responses before being completely done away with. "Masochistic personality disorder" was replaced by the term "self-defeating personality disorder" (SDPD) before the concept of this diagnostic label was removed entirely in the DSM-IV (APA, 1994). The abolishment of "homosexuality" as a diagnosable disorder saw many revisions to its terminology before the concept was completely removed from being branded a mental disorder. The DSM-II (APA, 1968) first saw the chameleonic term replaced by a new label "sexual orientation disturbance" in the 1974 version and retained as "ego-dystonic homosexuality" in 1980, "sexual disorder not otherwise specified" in 1987, and finally dropped in the DSM-II-R, 1987. This shift saw a triumph in the power of political and societal views to re-shape the lens of psychiatry.

Brown (1995) and Jutel (2009) discuss how a diagnosis may as equally be rejected by the individual as by society, in relation to illnesses which carry stigma, such as AIDS, and obesity, respectively. The above exemplars show how diagnosis is not immune to cultural, social, environmental and political factors, thus reinforcing the notion that context is grounded at the centre of our discourse (Spekman & Roth, 1988).

Autism, as a diagnostic term, has seen many changes across revised editions of the DSM. How autism is viewed in society and in clinical practice continues to evolve today. The word 'autism', first featured to denote certain behaviours in the inaugural edition of the DSM (1952) under the diagnosis of *schizophrenic reaction,* childhood type. In the subsequent DSM II (1986), autism still featured within the description of childhood *schizophrenia*. The DSM III (1980) saw the advent of *infantile autism* as a new diagnostic category within a new class of conditions termed the *Pervasive Developmental Disorders (PDDs)*. In the revised DSM-III-R (1987), the term was adapted to "autistic disorder" with a list of criteria arranged as pertaining to impairment in three major domains. The DSM-IV (1994) saw the introduction of *Asperger's syndrome* as a diagnostic subcategory within a spectrum of disorders including *Autistic Disorder, Pervasive Developmental Disorder Not Otherwise Specified, Childhood Disintegrative Disorder* and *Rett Syndrome.* The DSM-V (2013) saw previously identified separate disorders consolidate under the diagnosis of *autism spectrum disorder*, which is maintained in the most recent version of the DSM, DSM-V-TR (2022). This change saw the extinction of *Asperger's syndrome* (AS) as a distinct sub-category.

The deconstruction of previously recognised conditions is not always driven by social movements, as was the case for the aforementioned examples. The unitary diagnosis of *autism spectrum disorder* was brought about by researchers and clinicians in order to eradicate misdiagnosis and inconsistencies in the application of the previous diagnostic subcategories. The impetus for this change answers the question of *"Whose decision was that?"* posed by one of the authors in Walsh et al. (2018) paper, as discussed above. Huynh et al. (2020) investigated the subsequent effects of changing diagnosis on the people to whom these labels are attributed to. The authors conducted semi-structured interviews with adults with AS, analysing their accounts using thematic analysis. This study found that many participants socially identified with the label of AS, as their symptoms matched those described in the DSM-IV (1994). Doing away with the existence of this diagnosis was perceived as a threat to identity, social status and access to supports. In this example, the eradication of a diagnostic category shook people's sense of belonging within their social world. This finding links to arguments surrounding the functions of diagnosis for validating experiences both internally for the individual and externally with regards to how they interact with and are perceived by the world around them. These arguments will be explored further in *'2.8 Individual, family and societal sense-making in response to diagnosis',* below.

The current neurodiversity movement may influence changes to the diagnosis of ASD within future revisions of the DSM and may alter medical practices surrounding diagnosis and autism (Cooper, 2005; Dyck & Russell, 2020). Within the neurodiversity movement, autism is viewed as a normal human difference (Kapp, et al., 2013; Jaarsma & Welin, 2012). Autism is celebrated as an identity and rejects the medical model's propensity towards 'cure' or eradication of autistic behaviours. The neurodiveristy movement favours identity-first terminology (i.e., "autistic person") over previously advised 'person-first' terminology (i.e., person *with* autism). This preference is founded in the assertion that autism is a positive part of the person, integral to their construction as a person and a part which cannot be detached, as some 'less preferred' element. A certain dichotomy is also captured within the neurodiversity movement which both rejects labels in terms of their disabling and stigmatising effects whilst also embracing labels as an inherent part of identity and shaping of communities (Kapp et al., 2013; Dyck & Russell, 2020). This example showcases the complexity entrenched within the practice of diagnosis and how the actual value of that diagnosis, or otherwise, rests on the individual and society's perceptions of it.

Diagnosis has also been said to afford the power of legitimising illness. By applying a formalised label to a reported set of symptoms, the person's "complaint" can be transformed from a

subjective experience to an objective illness. Jutel (2009), in her review, references this phenomenon as "permission to be ill" (p. 278), "sorting out the real from the imagined, the valid from the feigned" (p. 279). If diagnosis can serve to validate a person's experiences, then it can be understood that where no diagnosis emerges, the acceptability of the person's accounts may be called into question in both the medical and social world. Swoboda (2008) examined how physicians navigate the ambiguity of known contested illnesses such as chronic fatigue syndrome (CFS), multiple chemical sensitivities (MCS) and gulf war syndrome (GWS). The existence of these conditions is often questioned due to the lack of an identified biological or pathogenic etiology. Swoboda (2008) drew the conclusion that physician diagnosis can serve to provide public acceptance of these illnesses within society. In a much earlier paper, Brown (1995) too regards diagnosis as the determiner of the existence and legitimacy of a condition.

2.4.4 Reaching Diagnosis: A Co-constructed Practice?

Diagnosis has often been studied as an institutional frame among patient and practitioner interactions. As discussed in *2.2.4 Diagnosis in Medical Discourse*, patients may initiate diagnostic-solicitation in medical interactions. Beach (2001) views patients' narratives as the fulcrum in patient-centred practices. Diagnostic certainty has been said to be improved when patients are asked to be 'co-producers' of diagnostic practices (Swoboda, 2008). In the paediatric context, Stivers (2002) explored the implications of two practices employed by parents when presenting their child's problems – namely the "symptoms only" presentations versus "candidate diagnosis" problem presentations whereby the parent presents a lay diagnosis which they are seeking the medical professional to confirm and provide treatment for. This research too highlights the patient or caregiver's role in influencing the trajectory of the diagnostic journey.

Patient involvement in co-constructing diagnosis represents a levelling of power in practitioner-patient interactions, which comes as a reflection of a changing social and medical world. In modern society, the widespread accessibility of the internet and the invention of a multitude of online medical databases and phone applications (i.e., APPs) see access to medical information brought to the fingertips of the lay person. With the advent of these technical innovations, patients now often enter interactions with a diagnostic hypothesis to be verified, as opposed to listing a set of signs and symptoms. The wide-spread access to the internet has been a large propellant in the pseudo-equalling of power in institutional talk about diagnosis.

The rise in the recognition of the value of illness narratives and the shift from a medical to a biopsychosocial model (Engel, 1977) has also contributed to the lessening of the power imbalance within patient-practitioner interactions. With this shift, the patient is bestowed the title of 'expert' in his or her own condition. Walsh et al. (2018) paper cites 'borderland' as a figurative term used to articulate the person-centred therapy approach adapted, whereby the professional works alongside the person in a truly collaborative manner. Within this paper, the authors cite consideration to the physical space for clinical discussions in echoing the dynamic of the balanced relationship (e.g., with reference to some meetings taking place in more informal locations such as cafés rather than more traditional clinical spaces).

2.4.5 Labeling Theory

'Labeling Theory' is considered a sociological and criminological theory, in that negative reactions from society in response to a person's ill-doing can lead to the person becoming more deviant (Vance, 2021). The premise of this theory dictates that the action of naming a criminal a "criminal" may contribute to the likelihood of re-offending and can be understood as a self-fulfilling prophecy. The theory posits that the perpetuating effects of labels is incurred by way of two distinct processes, firstly by negatively impacting a person's self-concept, and secondly, by creating societal barriers that block conventional opportunities (Vance, 2021). There is a distinction to be made between primary and secondary deviance (Lemert, 1951). Primary deviance relates to the multitude of factors that lead to a person committing an offence in the first instance. Secondary deviance refers to where a person inherits the deviant status into his or identity.

Labeling theory can also be applied in understanding how social status causes and / or exacerbates mental illness (Cooksey & Brown, 1998). How does being diagnosed with a mental health disorder influence a person's recovery? Cooksey and Brown (1998) question why so much effort is placed on the diagnostic process where effort could be diverted elsewhere such as in the provision of chronic care. Timimi (2014) further argues for the 'doing away with' of psychiatric labels. He contends that for a diagnostic system to carry value, it should demonstrate that the application of such labels enhances treatments and outcomes for the individuals to whom they are applied. Timimi (2014) maintains that such labels increase societal stigma and campaigns for the abolishment of diagnostic systems. In her edited work 'Drop the Disorder' (Watson, 2016), Watson challenges the practice of psychiatric diagnosis. Berkovits et al. (2020) explored adolescents' perceptions of their autism diagnosis. Thirty-eight teenagers participated in semi-structured

interviews. The findings showed the participants viewed their diagnosis as having mainly negative effects with roughly half the participants referenced perceived experiences or predicted stigma in association with their diagnosis. More negative associations with diagnosis, in comparison to the more positive findings of diagnosis as validation in studies with adult participants (e.g., Leedham et al., 2020; Portway & Johnson, 2005), as will be discussed in *3.4.4 Validating Experiences for the Individual ('Inner') and Society ('Outer'),* may be influenced by the developmental time of adolescence which is marked by a strong desire for social assimilation (Berkovits et al., 2020). The 'inner' response of the teenagers may also be influenced by the context of the 'outer' society and predominant cultures along with prevailing narratives surrounding autism and being autistic in the teenagers' environment, as considered by the authors.

2.5 Diagnosis and the Law

Within criminology and the legal system, diagnosis has been presented as a mitigating factor to reduce a person's accountability for their actions, altering their subsequent sentencing in line with their diagnosis. In critical analyses and judicial proceedings, this defence has been accepted as justifiable and also been rejected as a manipulative device in an attempt to evade responsibility for one's actions (Sparks, 1964). The Criminal Law (Insanity) Act 2006 defines that:

> Where a person is tried for murder and the jury finds that the person…did the act alleged, was at the time suffering from a mental disorder, and the mental disorder was not such as to justify finding him or her not guilty by reason of insanity but was such as to diminish substantially his or her responsibility for the act, the jury or court ... shall find the person not guilty of that offence but guilty of manslaughter on the ground of diminished responsibility. (Section 6, p. 9)

Additionally, a person may be found to be "*not guilty by reason of insanity"* (section 23, p. 23), where the person did the act or made the omission if he or she is found to have had a mental disorder at the time of committing the offence, such to the point that he or she cannot be held responsible for the alleged act or omission by the reason that he *"did not know the nature and quality of the act he was doing, or he did not know what he was doing was wrong, or he was unable to refrain from committing the act or making the omission".* Both verdicts of "diminished responsibility" and "not guilty by reason of insanity" have powerful implications on the sentence delivered. They also have an effect on the public perception of how responsible a person is for an offence, thus influencing his or her re-integration back into society and contributing to his or her

sense of well-being, self-construct and identify which may have cascading effects on their future actions and course of life.

2.5.1 History and Critical Analysis of the Defence of Diminished Responsibility

By the 19th Century, the defence of diminished responsibility was established in Scottish Law (Kennefick, 2011). England followed with inclusion of the partial defence in Section 2 of the Homicide Act 1957 a century later. By this time, some feared it was becoming a "loophole for murders" (Kennefick, 2011, p. 271). It was criticised as a "win-win" as opposed to the insanity plea, which usually resulted in the accused being sent to a detention centre for an indefinite time period. The doctrine was accepted into Northern Ireland Law under the Criminal Justice Act (Northern Ireland) 1966. In Ireland, the first introduction of the defence appeared in the Infanticide Act 1949, which states the jury could return a verdict of "infanticide", in place of murder, the punishment for which was aligned with manslaughter. It was not until the enactment of the Criminal Act (Insanity) 2006 that the partial defence of diminished responsibility was adopted into Irish Law, one century after the concept was established into English Law, and two centuries after the concept was adopted in Scottish Law. There are strong arguments in favour and against the partial mitigation. The mitigation has been viewed as both an unjust 'get-out-of-jail-free-card' and a valid mitigation of responsibility by virtue of a diagnosis, as delivered by a consultant psychiatrist. The rationale for the partial defence stems from the ethical and moral acceptance of the frailty of the human condition. Sparks (1964) discusses early applications of and reactions to use of this mitigation. Sparks (1964) protests that it is never appropriate to base mitigations of punishment on mental disorder. It is clear to see considerable controversy surrounds the instatement of this defence. The following case illustrates the potential influence of sociocultural beliefs surrounding the legitimacy of a diagnosis or diagnostic label on legal proceedings.

Case Example. The power to determine whether the truth of the facts on which a diagnosis is based, as brought before the court by a consultant psychiatrist, is a valid defence, lies with the jury. In the case of *The People (DPP) v Alchimionek* [2019] IECA 49, the jury returned a majority guilty verdict, despite a consultant psychiatrist for both the defence, and the prosecution, stating that the accused was suffering from a mental condition. The appellant was convicted of manslaughter of one named man and of assault causing harm to another. On appeal, this verdict was overruled and a new verdict of "not guilty by reason of insanity" was assigned. This case calls into question the notion of a "valid" diagnosis. Here, 11 out of 12 Jurors, chose to deny the accused

the plea of insanity, despite strong medical evidence that the accused was suffering from a mental disorder. The delivery of a guilty verdict, in the presence of an established mental health disorder, implies the jury did not view the diagnosis as a valid reason to evade or diminish responsibility for the man's actions. Within this case, there appears to be a disparity between the perception of the layman versus judiciaries with regards to the credit and weight of diagnosis that can be attributed to the crime .

2.6 Summary

As shown here, diagnosis had origins in supporting language and communication across professionals. The appropriateness of certain diagnosis have evolved over time, through political movements and shifts in societal understanding of difference. Diagnosis is not something that is only couched with the field of medicine but also widespread in the arts, being the theme of poetry, television and film, and has relevance in legal proceedings and societal justice systems.

Chapter 3: Literature Review: Application of 'Diagnosis' in the Current Context of Living and Experience

3.1 Introduction

In this second literature review chapter, I will examine diagnosis as it relates to people's experiences. I will review research findings surrounding individual sense-making in response to receiving a diagnosis, caregivers' experiences of diagnosis for their children in several contexts, including the case of rare or undiagnosed conditions, autism, Down syndrome, and life-limiting illness. The discussion will be extended further to look at 'predictive diagnosis' for the person and the family. I will discuss other fields such as those within *critical disability studies* and the notion of *illness narrative* and their relevance to the current research. I will finish with a look towards the global climate of the pandemic -Covid-19- the onset of which occurred and endured while this research was being conducted in turn affecting the research design.

3.2 Current Landscape: Rare Conditions and Undiagnosed 'Syndromes Without A Name' ('SWAN') Policy and Service Advancement

In the European Union, a disease is considered 'rare' when it affects less than 1 in 2,000 people (EURORDIS, n.d.). Most rare diseases have a genetic origin (80%, as according to NGO Committee for Rare Diseases, 2018). Wakap et al. (2020) investigation on the prevalence of rare diseases concluded that, at the time of the study, rare diseases affected 3.5-5.9% of the population worldwide, translating to 263-446 million people worldwide at any one time. This figure presents an estimate of those people living with rare disease and does not include those *affected* by rare disease in terms of recognising illness as an 'ill unit' which extends to include family and siblings. Viewing this projected cumulative global figure through such a family-focused lens would approximately triple the number of people affected by rare disease.

The need to address challenges for people living with rare diseases and their families amongst government policy and healthcare has received growing recognition in recent years, following many years of campaign. In September 2021, representative organisations supporting the rare community in Ireland submitted a letter to An Taoiseach, Micheál Martin at the time, to call for Ireland to support adoption of UN Resolution for Rare (Rare Diseases Ireland, 2021). In December 2021, the UN General Assembly adopted the resolution on addressing the challenges of persons living with a rare disease and their families. This resolution was led by the Non-Government Organisation Committee for Rare Diseases, Rare Diseases International and EURORDIS-Rare. The

document centres on non-discrimination and inclusion in the UN Sustainable Development Goals such as supporting access to education and employment, reducing poverty and supporting participation in society. The most recent four-year National Rare Disease Plan for Ireland was published in 2014 (Department of Health, 2014). The Minister for Health, Stephen Donnelly, announced on 28th February 2023, plans to develop a new National Rare Disease Plan.

> Improving and expanding the care and treatment of patients with rare diseases is a priority for the government, with work spearheaded by the HSE National Clinical Programme for Rare Diseases. (Department of Health, 2023, "Minister for Health announces development of new National Rare Disease Plan", para 3)

In 2017, European Reference Networks (ERNs) for rare diseases were established to share information amongst healthcare providers to enable discussion on complex and / or rare diseases and conditions that need highly specialised knowledge, resources and treatment (European Commission, n.d.). In 2018, The Rare Disease Research Partnership (RAinDRoP) was launched as a research partnership amongst a variety of stakeholders in the rare community in Ireland (Somanadhan et al., 2020). 'Care Pathways for Rare Diseases' (HSE, 2023) have been developed by the National Rare Diseases Office (NRDO) to describe the processes involved in managing a clinical condition. Rare Diseases Ireland, patient experts, medical and health and social care professionals contributed to the development of these integrated pathways. On 28th June 2023, rare disease national alliance patient groups and patient representatives met in the Dáil (home of the Irish Government) to present on the challenges as the closing event for the 'Get Rare Aware' campaign (https://rdi.ie/gra/). This campaign focused on highlighting the challenges faced by parents and patients along the diagnostic odyssey, particularly in relation to accessing genetic services and wait times for genetic services in Ireland. Solutions were put forward to government to resource more genetic assistants to carry out triaging of referrals in order to increase clinical time of genetic counsellors. The most current campaign, run by Rare Diseases Ireland, *Rare Ireland* and *Takeda Ireland 'I am Number 17'* launched on the 7th February 2024. This is a public awareness campaign that aims to raise insight into what it is like to live with a rare disease in Ireland. The aim of these campaigns aligns with one of the main objectives of my own research, to increase public and professional understanding of the mothers' experience of 'diagnosis' for their child with a rare of undiagnosed neurodevelopmental condition.

In the UK and Australia, specifically dedicated support networks exist for parents and families living with a child or young adult with a syndrome without a name. These groups go by the acronym 'SWAN' to denote '**S**yndrome **W**ithout **A N**ame'. SWAN UK defines 'SWAN' as "a term used when a person is believed to have a genetic condition and testing has failed to identify its genetic cause" (Fletcher-Dallas, 2007, para. 1). These children are often referred to as having 'global developmental delay' or 'failure to thrive' and may present with a range of symptoms and medical needs such as learning and / or physical disabilities, feeding difficulties, epilepsy, respiratory issues, unusual physical features. There are approximately 6,000 children born annually in the UK with a condition that is likely to remain undiagnosed (Fletcher-Dallas, 2007). In Ireland, a similar group is currently being pioneered but is not yet established as a registered charity, like its SWAN international predecessors.

In 2022, *Rare Ireland Family Support Network Ireland* achieved their status as a registered charity. This organisation aims to support families living with rare conditions in Ireland. Rare Ireland and SWAN Ireland are members of Rare Diseases Ireland, which is the national alliance for voluntary group representing people affected by rare disease in Ireland. Understanding the current landscape of rare disease in Ireland, and internationally, has relevance for the reader in situating the participants' accounts within this terrain and applying the implications from the research in context. My findings may present a step forward in addressing this issue by contributing insights on the mothers' experience of 'diagnosis' for their child with a rare or undiagnosed condition.

3.3 Illness Narrative: Definitions and Considerations

There are many ways of hearing, engaging and classifying participants' accounts or stories. One way is to consider the work of Arthur Frank (1995, 2010, 2013). Frank (1995) provided a classification system of narrative typologies within which there are a variety of narrative types, which are intended to act as gateways to help readers understand the main underlying plot and tensions of a story. He defines a narrative type as "the most general storyline that can be recognised underlying the plot and tensions of particular stories" (2013, p. 101) which can be regarded as "ways of entering" (2007, p. 25) a person's story or as "listening devices" (Frank 2013, p. 103) to help the listener understand where storytellers are in their experience of illness. Frank (2010) describes three narrative types: *the restitution, quest and chaos narratives.* The *restitution* voice reflects the desire to return to health (Frank 1995, 2013). The basic storyline can be summarised as "yesterday I was healthy today I'm sick, but tomorrow I'll be healthy again" (Frank, 2013, p. 104). The *chaos* narrative is the most difficult to hear as it does not foresee life getting

better (Frank, 1995, 2013). The ill person's belief that there is something to be gained through the experience of illness is exemplified by the *quest* narrative, which can be further specified by the memoir, manifesto or automythology style (Frank, 1995, 2013).

Frank (2010), however, cautions there is a danger that story types can be overly adhered to and declares that his intention was not for these three types to be exhaustive. Harrington (2008) developed six narrative templates from her study of mind-body science (including 'the power of positive thinking', 'broken by modern life', 'the body that speaks'). Harrington explored the ways in which these narratives have power to shape people's experience of illness. Benbenisye et al. (2020) introduced a new narrative type, namely the '*Sisyphean Narrative',* to support interpretation of parents' experiences of chronic childhood illness for their child. Benbenisye and colleagues (Benbenisye et al., 2020) carried out semi-structured interviews with twelve parents of children with end-stage renal disease and analysed the data using IPA, coming up with a new narrative type to understand the experience for caregivers of the recurrent cycle of end-stage renal disease for their children. The Sisyphean Narrative is based on the myth of Sisyphus, who was condemned by the gods to the burden of endlessly rolling a rock to the top of the mountain, which inevitably rolls back down again, in an infinite loop. The authors conclude that producing this typology may denote a collective experience for this group of parents, from which professionals may gain understanding of the shared experience. The authors advise that future research examines the application of this narrative type in the case of other chronic diseases. The branding of the 'Sisyphean narrative' typology may present a constricted representation of the parents' experiences as an exact repetitive loop. One may argue that there may not have been complete circularity in the situations described and use of such a comparison fails to promote positive differences, physical or emotional, and however small, for the children and parents as achieved through parent and / or professional endeavours along the journey of chronic illness (D. Abrahamson, personal communication, December 3, 2019). The use of the comparison to 'Sisyphus', who is being punished for his crimes in the original myth, may for some parents also risk presenting the idea that their child's chronic illness is in some way a prescribed punishment (D. Abrahamson, personal communication, December 3, 2019).

Stories have been described as performative acts in that they actually 'do' something (Eldershaw et al. 2007). Storytelling, as an act, has been said to have an organising effect on the listener in making sense of their story. This notion of 'doing things with words' was first introduced by Austin (1975) in his Speech Act Theory, which dictates that all utterances are a form of action. It is

through storytelling that the narrator experiences distance from the stories that they tell, enabling the person to reflect upon and move through their situation (Frank, 2007). Narration not only allows a person to order and make sense of events, but also constitutes the way in which people witness who they are (Kilty, 2000), and how they define and express themselves (Murray, 2008). Spillman et al. (2017) reference that the process and opportunity to share their story appeared to be appreciated by the parents in their study. Providing people with a stage to share their stories awards 'tellability' which has been said to reaffirm for a person that their experiences are worth living and reclaiming (Frank, 2015; Smith & Sparkes, 2008). The research on the power of storytelling is relevant in considering what benefits may have come about for the participants in this study as an outcome of participating.

Previously, Gray (2001) proposed three narratives of autism, that of *accommodation, resistance* and *transcendence* following interviews with parents of children with the condition living in Australia. The researcher used narrative analysis of "a thematic nature" (Gray, 2001, p. 1249) as the chosen method of analysis. The narrative of *accommodation* was identified as the most common narrative among the parents in the study, described as being aligned with acceptance and belief of a biological origin for autism with the remaining *resistance* and *transcendence* representing alternative narratives where the parent either redefines themselves in terms of political activism or looks to religious beliefs as a way to understand autism. These narrative constructions are said to align respectively with higher cultural narratives of science, politics, and faith.

Fisher and Goodley (2007) interviewed mothers of children with special care needs in the UK to identify "empowering understandings and discourses around disability" (p. 69). The authors present three narratives within the mother's stories: *the linear narrative, the narrative of challenge and the philosophy of the present and becoming.* The *linear narrative* is aligned with the medical framework of disability which conceptualises diagnosis as an important factor in determining future development and profession with hopes for 'recovery' and return to 'normality'. Within the *narrative of challenge,* the mothers offer perspectives that are more aligned with the social model of disability which positions the child's 'disability' as originating in social regulations, with a rejection of 'normality'. Emergence of a 'fighter' against social norms is often heard within the narrative of challenge. This viewpoint can be seen as more enabling and empowering (Fisher & Goodley, 2007). The *philosophy of the present and becoming* interpret "uncertainty as an opportunity to focus on the quality of life in the present" (Fisher & Goodley,

2007, p. 74) with recognition that the future is not always known or controllable. Fisher and Goodley (2007) note a polysemous quality to the mothers' narratives in that within many stories, all three narratives were identified as interwoven. The authors conclude "*interwoven and multi-layered narratives reflect complex lifeworlds and suggest that mothers' understandings of their child's disability are constantly open to renegotiation and flux"* (p. 76). Consideration to previously defined narrative types, whilst remaining open to the potential of new narratives, may serve as useful in entering the stories of the participants in this research project. Narrative inquiry will be further elaborated on in *4.3 Consideration to Alternative Methodologies*.

Telling stories of illness and disability have the power to change not only the storytellers but the story listeners (Kilty, 2000). The illness narrative can therefore be thought of as an act of "dual reaffirmation" (Frank, 2013, p. 83). For families experiencing similar situations, the power of reading a story which resonates with the listener's own experience can foster a sense of togetherness, ease isolation and offer alternative ways of perceiving the experience (Easton & Atlkin, 2014) and by doing so, enhance their abilities to cope (Madeo et al., 2012).

My research explores parents' experience of diagnosis for their children with neurodevelopmental conditions which can be more astutely considered to be a caregiver illness narrative. Knepper and Arrington (2018) in their analysis of messages from an online support group for parents of children with persistent hyperplastic primary vitreous, a rare vision disorder, introduce the idea of "witness stories" as distinct from existing illness narrative typologies. Witness stories distinguish that the parents' experience represents a caregiver illness narrative typology and is not based on first-hand experience of illness, in comparison to illness narrative in the more traditional sense. The is the intended meaning where the term *witness stories* is used elsewhere within this thesis.

3.4 Individual and Family Sense-Making in Response to Diagnosis

3.4.1 Access Key or Padlock?

The value of receiving a diagnosis for accessing services and validating experiences has long been juxtaposed with the potential stigma and preconceptions associated with being 'labelled'. Additional, unique challenges and considerations are cited in the growing research for those with rare and undiagnosed syndromes in terms of, what is widely considered to be, the

'diagnostic odyssey'[11]. These varying perspectives in relation to family and individual sense-making in response to diagnosis have been documented in the literature and will be explored in this section.

3.4.2 In the Case of Rare and Undiagnosed Conditions

Although some experiences of parents raising a child with no known diagnosis, are shared with the reported experiences of parents whose child *has* a diagnosis, parenting a child with no diagnosis is said to "adds a layer of complexity" (Lewis et al., 2010, p. 807). Lewis et al. (2010) explored parents' experiences of raising a child with no diagnosis in the United Kingdom. They gathered data from fourteen parents through conducting semi-structured interviews, which they subsequently analysed using Grounded Theory methods (Charmaz, 2009). The researchers comment that they selected this method of analysis for the generalisability it affords in being able to develop explanatory theories in connected with the topic being studies. Lewis et al. (2010) found that the challenges faced by these parents could be divided into two distinct components: (1) the inner emotional experience (i.e. realisation there is a problem, experience of testing, reasons for wanting a diagnosis, emotional impact and active coping mechanisms) and (2) the outer sociological experience (i.e., experience with professionals, various support networks issues with education and housing) experiences. Frustration was evident as being common across the journey. The Genetic Alliance (2018) identified that one of the biggest challenges families face is the lack of understanding from non-specialist health professionals regarding having no diagnosis. A host of unique, specific, issues associated with having no diagnosis have been identified in the existing research base including difficulty accessing services (Genetic Alliance, 2018; Lewis et al., 2010; Rare Disease UK, 2016), feelings of isolation (Rare Disease UK, 2016), emotional distress (Lewis et al., 2010; Spillmann et al., 2017), lack of communication amongst professionals (Genetic Alliance, 2018; Lewis et al., 2010), complexity of care (Genetic Alliance UK, 2018; Spillman et al., 2017), the number of professionals involved and appointments, and frustration (Lewis et al., 2010).

According to Madeo and colleagues' (Madeo et al., 2012) study, parents who experience greater uncertainty feel less control over their child's condition, which may lead to the use of less effective

[11] 'Diagnostic odyssey' is a term used in genetics and academic research to refer to the, often lengthy, journey for people living with rare disease to procure a diagnosis (Basel & McCarrier, 2017; Bauskis et al., 2022; Bouwman et al., 2010; Kole & Hedley, 2021).

coping strategies and poor adaptation. Madeo et al. (2012) carried out quantitative analyses to identify the relationships of key variables to perceived uncertainty, based on data gathered through mixed-methods survey administered electronically. Bourke et al. (2014) explored mothers' and fathers' experiences of having a child with Klinefelter syndrome (KS; a genetic condition which is often undiagnosed) by carrying out in-depth interviews, analysed using thematic analysis, in Australia. The findings identified that parents' experiences were influenced by the timing of diagnosis, who provided the diagnosis, what information was given from health-care professionals and found online. Individual support networks and specific support groups were identified as essential by the participants in the study to support positive outcomes regarding parents' adjustment to the situation.

Rosell et al. (2016) looked at parents' perceptions of genetic testing where their child has an undiagnosed condition and identified what parents value. Rosell et al. (2016) conducted semi-structured interviews with parents who had undergone whole exome (i.e., regions of a genome) sequencing and had the results communicated to them with the outcomes ranging from 'definite' through 'likely', 'possible diagnosis' to 'no diagnosis'. For those who obtained a definite or likely diagnosis for their child, parents felt medical care was more specified and there was a reduction in worry despite their being no treatment. For some families in this group, there was a reported continued sense of isolation and frustration given the limited information available on the rare condition and the inability to connect with other families.

Spillman et al. (2018) analysed written illness narratives of people affected by undiagnosed conditions for narrative content and type, finding the narrative type, of 'chaos', as defined by Frank (1995, 2013), coexists with being undiagnosed. Gimenez-Lozano et al. (2022) carried out a study in Spain which included adults and children who had a confirmed or suspected rare disease diagnosis. For the paediatric group, families faced higher financial burden in comparison to the adults with rare disease. A majority of the households reported a negative impact on their life and emotional state as a result of the rare disease diagnosis. The authors concluded that further education is needed for healthcare professionals to understand the risks to socio-health associated with rare disease to support the child and family.

Simon et al. (2022) carried out interviews with parents surrounding their diagnostic journey in relation to their child with a neurodevelopmental condition of genetic origin in Los Angeles. The authors interviewed a large number of participants (n=37), accessing a range of perspectives. Their

findings from a qualitative, primarily descriptive approach to analysis, identified key themes including delays in accessing a timely diagnosis from first parental observations of symptoms, obstacles accessing clinical interventions (including long wait times for an appointment, lack of insurance coverage, availability of local evaluations, transportation difficulties, and native language differences), the importance of being part of a patient advocacy and unique challenges for those aged eighteen years and over.

In the Australian context, Baukis et al. (2022) conducted interviews of parents whose children are living with undiagnosed conditions to understand parents' experiences and perspectives relating to this experience. The authors used thematic analysis to identify three main themes from the findings, including (1) responding to significant care needs, (2) the diagnostic journey, and (3) the value of diagnosis. The final theme was subdivided into two juxtaposing subthemes of 'lowered expectation of diagnosis and limitations of diagnosis' and 'hopes of what a diagnosis might provide'. The authors found that while all participants felt there were benefits to diagnosis, some parents identified that a diagnosis would not alter the child's health (e.g., "It doesn't matter-and the answer…it won't make difference to our journey or [child's] life because it's unlikely to change", Baukis et al., 2022, p. 9). Benefits cited include diagnosis as helping to know '*what to look out for'* in terms of future problems, supporting knowledge around risk of reoccurrence for siblings when they have children, connecting with other families with a child with the same diagnosis, and improved access to education.

3.4.3 Parents of Children with Chronic or Life-limiting Conditions

Jacoby et al. (2020) conducted semi-structured interviews with 12 parents of children who had various stages of renal failure. They analysed the data using IPA, identifying eight themes which they divided in terms of the 'intraspective' (i.e., loneliness, responsibility and guilt, fear and suffocation, inhibition of subjectivity) and 'intersubjective' experience (i.e., lack of understanding, tension in the family, the health-care system: between satisfaction and disappointment, objectification and depersonalisation). Similar to the division of themes into 'intraspective' and 'intersubjective' dimensions, Lewis et al. (2010) found that raising a child with no known diagnosis can be understood in terms of the 'inner emotional' and the 'outer sociological' experience. The identification of a binary experience, relating to the inner and outer worlds of parents, is a common phenomenon identified by Jacoby et al. (2018) for parents' experiences of raising a child with chronic-recurring illness and in earlier work by Lewis et al. (2010) relating to parents' experience raising a child with no diagnosis.

Cote-Arsenault and Denney-Koelsh's (2011) qualitative study on the experience of a lethal fetal diagnosis (LFD) in pregnancy for mothers also identified themes across two dimensions, namely the 'Personal Pregnancy Experience' (i.e., *grieving multiple losses; arrested parenting; my baby is a person*) and 'Interactions of Others' (i.e., *fragmented health care, disconnected family and friends*), with the theme of "utterly alone" spanning both dimensions. These findings may collectively allude to the duality of experiences for parents as a more universal phenomenon, in terms of the inner experience for the individual and in interacting within society.

3.4.4 Validating Experiences for the Individual ('Inner') and Society ('Outer')

In this section, I will present a review of the literature as it relates to (a) first-person experience of illness, and (b) the parental experience of illness for their child. For both groups, I will consider the impact of diagnosis across two dimensions, namely the 'inner emotional' and the 'outer sociological' experience, as previously established.

3.4.4.1 First-Person Experience of Illness: 'Illness Narrative'. Arantzamendi et al. (2020) conducted a secondary analysis of qualitative data gathered in a phenomenological study designed to gain understanding of the lived experience of having cancer. Their research concluded in a refined theory which identifies a five-phase iterative process for living well with advanced cancer, which added to the previously defined Theory of Living with Chronic Illness (Robinson, 2017). Arantzamendi et al. (2020) paper centres around the notion of understanding one's experiences of living while dying with life expectancy is both uncertain and short. This concept and adapting to how to live with the impeding expectation of death called to mind a two-fold visualisation of lack of diagnosis as both a 'shield' and a 'blindfold'. On the one hand, lack of knowledge of diagnosis can act as a protective barrier from the potential dangers and at the same time, lack of knowledge denies the opportunity to prepare emotionally and logistically for the individual and their family members.

In the case of Autistic Spectrum Disorders (ASD), much of the research and empirical evidence alludes to the belief that diagnosis can be critical for understanding how one's own self navigates the world. In some cases, diagnosis is viewed as an antidote which nurtures self-compassion and awareness for not only the parents but also for the autistic person. Portway and Johnson (2005) explore the risks associated with having undiagnosed Asperger's syndrome (AS) in childhood. In this study, the authors analysed data using constant comparative analysis, gathered through carrying out unstructured interviews with adults and parents of adults with AS. The authors

identified various every day and long-term risks for those who remain undiagnosed including being misunderstood, social isolation, receipt of inadequate services, prolonged dependency on parents, and unhappiness.

Leedham et al. (2020) explored the lived experience of women with autism who received the diagnosis of an autistic spectrum condition over the age of forty years. The data was analysed using Interpretative Phenomenological Analysis. Leedham et al. (2020) findings provide strong support for the function of diagnosis as a critical facilitator which enabled these women to understand and make sense of their own behaviour and life experiences. Some of the participants speak about how failed social interactions led to internalised beliefs that they were 'wrong', 'bad' or 'broken' (p. 138). Diagnosis provided the panacea for these self-deprecating constructions by offering an alternative explanation. In their study, the participants' response to diagnosis is largely positive, with diagnosis cited as enabling transition from being self-critical to self-compassionate. The underlying tension in the women's narratives in Leedham et al.'s (2020) study may be viewed, as a re-birth through the reconstruction of identity on the autism spectrum. Some participants spoke of diagnosis with gratefulness for finally being accepted for who they are. For others, diagnosis was said to absolve feelings of blame and instigate a positive shift in mental health and birth a sense of power, pride, strength, and freedom. Understanding of the participants' experience may be enhanced by viewing their recounted experiences through the lens of Frank's *automythology* narrative (Frank, 1995, 2013) which is a subtype of the 'quest' narrative, which speaks to the total reinvention of self. Sadness and anger are also referenced in relation to the delay in diagnosis. The participants express frustration and sympathy for their younger selves in that diagnosis, and that the resultant understanding of their own difficulties, did not come sooner. This study particularly emphasises the function of diagnosis in being able to support the process of self-awareness and re-construction of identity. The findings in Leedham et al.'s (2020) poignantly speaks to the argument of diagnosis as a validating experience, in the context of adulthood diagnosis of autism.

3.4.4.2 Parental Experiences of Diagnosis. The receipt of an ASD diagnosis has also been said to provide relief from blame amongst the identities of the primary caregivers, as well as assisting greater empathetic understanding in onlookers when it comes to explaining a person's behaviour. Foster-Galasso (2005) discusses the efforts of her family to cope with their son's diagnosis of a developmental disorder. She states that the diagnosis gave her relief from the diagnosis of "Bad Mom" by providing a reason for her son's behaviour, which she could verbalise

both for herself and public perception. Denman et al. (2016) also reference the usefulness of diagnosis in providing explanation for non-typical behaviours that assist parents in the sense-making process, providing counter evidence to the internal and external beliefs of the 'bad' parent. Foster-Galasso (2005), with reference to the developmental disorder, makes the statement "names have power" (p. 27). This perhaps provides some rationale for patients' eager solicitation for diagnosis. Foster-Galasso (2005) goes on to question that without a name, how does one source information? Although she lauds diagnosis for its ability to provide a gateway to service provision and navigating social interactions, she remains open to the controversies of labelling. In defence of diagnosis, she makes the point that, denoting that diagnosis does not create the problem, but merely names a problem which already exists and has prompted the person to present to a healthcare professional for investigation.

Avdi et al. (2000) explored parents' perspectives of the "problem" during this diagnostic inquiry of autism spectrum disorder for their child. Diagnosis was largely viewed as a positive description and process for the families in the current study. Avdi et al. (2000) carried out semi-structured interviews with sets of parents whose sons were undergoing assessment for "communication difficulties" in the United Kingdom. The researchers state they chose a form of this methodology, in contrast to 'micro-level' analysis, to identify discourses in the parents' talk and how they are connected to broader representations of development, disability and medicine. In their analysis, Avdi et al. (2000) identified parents repeatedly construed diagnosis as an "antidote to uncertainty" (p. 248). It was praised as a "label which would transform the 'problem' from something vague and poorly understood to a knowable, defined 'thing' with a predictable future, known causes and treatment" (p. 248). Diagnosis was said to validate parents' anxieties, provide explanation and understanding of their child for themselves and in explaining their child's behaviours to others, being said that diagnosis carried an inherent 'built-in' explanation. In one parent's discourse, they liken diagnosis to solving a mathematical equation. Another function of diagnosis identified was as an explanation for the disease origin which served to mitigate parental guilt and externalise the problem as distinct from the person.

Avdi et al.'s (2000) paper is of particular relevance to the current research as it explores parents' journey from undiagnosed to diagnosed and the meanings and functions they bestow upon diagnosis. Avdi and colleagues (Avdi et al., 2000) concluded that diagnosis was viewed as both a "relief and terribly distressing" yet no diagnosis was viewed as creating an "almost untenable position of uncertainty" (p. 251). The parents' conceptualisations pre-diagnosis may provide some

paralleled insights into the thoughts and cognitive process of parents in the current study of children who present with undiagnosed conditions. The participants in Avdi et al. (2000) may also carry some similar thought processes to the second sample group, parents of children who received a diagnosis of Down syndrome peri- or postnatally. Avdi et al. (2000) offers a hypothetical bridge between the two proposed sample groups for the current research, where parents move from one defined group to the other, namely from a place of where their child has no diagnosis to having procured a diagnosis. Avdi et al.'s (2000) paper references that the analysis may be generalisable to broader applications to understand how medical diagnosis is constructed by parents and families. However, it must be acknowledged that considerable differences between the sample groups in the current research project remain, such as the cited reduced visibility of autism ("not an obvious disability", p. 243), the age at which diagnosis is typically reached and nature of the diagnostic label itself with regards to its origin, symptoms, and construction within society.

Werkhoven et al. (2022) cite this 'sense-making' effect of diagnosis as helping children and parents see why a person experiences certain challenges. The authors question do we need labels to provide this internal freedom and liberation from guilt and blame? They posit: does the origin of the problem lie in societies? This may call to mind propositions of critical disability studies (see *3.7 Critical Disabilities Studies*). Werkhoven et al. (2022) reference the use of labels in the neurodiversity movement, which is working for society to recognise ASD and ADHD as acceptable forms of human diversity (Aftab, 2022).

3.4.5 Stigma

Conversely, the following study includes positive participant reflections where there is a delayed or no diagnosis. Godley and Tregaskis (2006) analyse parental accounts of their experience of professional health and social care support since the birth of their child with a disability in Britain, as obtained through conducting parent interviews, using multi-methods of analysis including narrative inquiry, discourse analysis and Grounded Theory. Of most relevance to the current study, the researchers identified "diagnostic stories" (pp. 636-638) and "conceptualising impairment" (pp. 638-640) as key super-ordinate themes. The researchers' analysis provides insights into parent perceptions, where an early certain diagnosis was provided and where no certain diagnosis was identifiable. The analyses revealed some benefits of delayed definitive diagnosis including forming a strong sense of one's child's worth. One mother also alludes to the "liberation" associated with no diagnosis referencing "when there's no label…that's

fortunate because you make your own way then" (p. 639). This participant's comment was striking, in that it contrasts greatly with my personal experience of parents' experiences of no known diagnosis. Another cited benefit to receiving a delayed or uncertain diagnosis is the provision of a longer time for parents to accept and come to terms with their child's impairment. One participant criticised diagnosis for negatively influencing their child's care, referencing that the health care providers were helping with the 'Down syndrome', as opposed to offering guidance around child-care queries such as how much sleep or milk should the baby be having.

Linton (2014) explored the experiences of diagnosis for adults with Autistic Disorder (AD) and Asperger's syndrome, from a variety of countries, using a phenomenological inductive content analysis. Linton's findings also support the argument that clinical diagnosis exacerbated stigma. Approximately in the past two to three decades, there has been a shift in the way healthcare services are provided from 'diagnosis-led' to 'needs-led' service provision (Parry-Jones & Soulsby, 2001). Along with this shift, came the increased awareness of 'person-first' thinking (Mead & Bower, 2000). Person-first thinking calls for professionals and lay people to recognise the person-as-person. The person comes before the diagnosis, acknowledging that the diagnosis does not define the person. Gillman et al. (2010) explored the implications of diagnosis for people with learning difficulties and their family carers. They conclude with proposing an alternate way of viewing diagnosis from a social constructionist perspective:

> If diagnosis is regarded as a hypothesis that is neither true nor false, but more or less useful, then consideration could be given to the efficacy of specific diagnoses in terms of the opportunities they create or the possibilities they limit. Furthermore, viewing diagnosis as tentative or one of many possibilities affords those who are the recipients of diagnosis the choice to accept or reject it. (Gillman et al., 2010, p. 405)

Gillman et al. (2010) make a plea to professionals to part with the 'certainty' they attach to diagnosis and instead view diagnosis in terms of the value it offers for the person involved. More recently, Werkhoven, Anderson and Robeyns (2022) present the benefits and caveats of using diagnostic labels for developmental disorders, namely attention-deficit/hyperactivity disorder (ADHD) and ASD, across four contexts of scientific, therapeutic, social and administrative. The authors conclude that critique and defence of labels needs to consider the interests of all stakeholders across these four contexts. Further criticism of diagnosis and its impact for the

individual within their social world will be further explored in '*1.5.5. Diagnosis: Shaped by the individual and society'.*

3.5 Looking Beyond the 'Diagnostic Moment': What Comes Next?

Several studies have investigated what supports the emotions and events that ensue following a receipt of a diagnosis. Clark et al. (2020) put forward a model of family sense-making after a child's diagnosis of Down syndrome. The origin point of this model is 'hearing the diagnosis', which acts as the lever that sets in motion a cogwheel of feelings and actions that help bring the family into 'rescuing hope'. This model called me to question - what happens for those who have received no diagnosis? What is their origin point to start the necessary sense-making journey in order to re-shape their narrative and re-claim their story? A model of sense making needs to be identified for these families. The current research could help (i) to establish this theoretical model which health care professionals could use to recognise where a family is on their sense-making journey, and (ii) how to support them in navigating the continued path to reach Frank's (1995) *quest* narrative, where one learns to live a life re-imagined, with illness.

Rabbitte et al. (2017) and Braiden et al. (2010), explored parents' experiences of the diagnostic process of autism (once again the area where many studies have been conducted). In Rabbitte et al. (2017), a number of themes were developed from parental accounts, obtained through semi-structured interviews, pertaining to the benefits which accompanied receipt of diagnosis, including access to support and help, better understanding of their child and child's needs, and helpful for their child in making sense of their own experiences. The researchers identified the emotional impact in terms of "fear of the unknown" (p. 59) and "optimism and hope" (p. 59) as themes that followed receipt of diagnosis.

Braiden et al. (2010) conducted semi-structured interviews with parents on their experiences of autism diagnostic procedures. Braiden et al. (2010) analysed these parental accounts using thematic analysis and identified factors which contributed to parents' overall experience of the diagnostic process for autism, including having their initial concerns listened to and receiving written information. The provision of adequate written information appears to be a recurring finding that contributes to parents' positive experiences of diagnosis, regardless of the specific diagnosis given. Waxler et al. (2013) and Skotko and Bedfia (2005) found that receiving adequate written information and referrals to appropriate services (e.g., genetic counsellors, parent support groups) influenced parents' perceptions of the experience of diagnosis, of William's syndrome,

and Down syndrome, respectively, for their child. Skotko and Bedfia (2005) identified further factors which influenced mothers' experiences of receiving a post-natal diagnosis of Down syndrome for their child, contingent on the professionals' delivery of the diagnosis, including timing, location, sympathy, language used, and content (i.e., whether positive or negative aspects of the diagnosis were emphasised).

3.6 Predictive Diagnosis

One of the founding principles of medical ethics is 'do no harm'. Genetic susceptibility testing calls to question this guiding principle. The ethical dilemma in genetic testing for hereditary diseases such as the 'cancer gene', Huntington's disease, Alzheimer's is widely known. This ethical debate owes to the power associated with 'knowledge' of a probable or definitive future diagnosis. It can be postulated that the sheer process of knowing one's likelihood of developing chronic, and potentially life-threatening illness in the future, would likely be hugely influential on the way one views and interacts with the world. This type of knowledge is irrevocable. Once it is known, it cannot be taken back.

Hamilton and Robson (2019) reviewed the literature on psychosocial effects of testing for cancer susceptibility genes. They reported that generally negative emotional effects of predictive genetic testing have been limited. The research in this field is however reported to be both limited and flawed. Studies involved small samples and neglected to explore long term effects. The effects of certain gene variants are uncertain, in that a presence of such genes is not sufficient to predict a diagnosis of cancer with certainty.

A study by Timman et al. (2014) explored the cascade of psychological responses to a positive genetic susceptibility test for Huntington's disease, over time. They concluded that carriers and their partners experienced more distress immediately following test results, while their outlooks improved over successive 2-3 years. Emergence of psychological effects, including hopelessness, were identified as the carrier approached the age of onset. I wish to further explore the concept of 'hope'. How does hope function within patient coping and recovery in response to diagnosis? Do healthcare professionals recognise the potential utility of this emotion in their interventions and provision of care?

Roberts et al. (2011) assessed the impact of genetic susceptibility testing on asymptomatic individuals at risk for Alzheimer's. Presence of the searched gene variant again presents a

significantly higher risk of developing the disease, but alone is not sufficient to offer a certain predictive diagnosis. The aforementioned studies concur that further research is needed to elevate understanding of the psychological effects of probable or possible future diagnosis to support practice in this field and safeguards for the individuals undergoing testing.

Predictive diagnoses have also been studied in the case of neurodevelopmental disorders, such as Down syndrome. Yau and Zayts (2014) examine the 'risk of knowing' talk in medical consultations with parents around prenatal screening for Down syndrome in Hong Kong. The authors refer to the 'risk of knowing' talk as the consequences that follow pre-natal testing. Yau and Zayts (2014) concluded that medical agenda favoured performing tests over attending to 'risk of knowing' talk. The authors advise that in order to maximise informed-decision making, the 'risk of knowing' talk should be included in discussions around pre-natal screening. Stefansdottir (2020) examined two views on prenatal testing for Down syndrome from the perspectives of mainstream views in medicine and the perspective of Down syndrome activities. The medical view is criticised for disregarding the potential and value of the person living with Down syndrome. As of 1 January 2019, the Health (Regulation of Termination of Pregnancy) Act 2018 commenced in Ireland. The act legalises abortion to be carried out if the pregnancy is no more than 12 weeks. After such time, abortion is permitted under special circumstances only. Huge controversies and variations exist within individual and societal views, medical practices, laws and political reforms fields, within and across countries, exist as to whether pre-screening for neurodevelopmental conditions is ethically or morally valid. Stefansdottir (2020) concludes that non-invasive prenatal testing is not an isolated example which calls for ethical consideration in modern medicine and cites the use of cochlear implants as another such example. One school of thought on the use of cochlear implants may be seen to infer that the life of a Deaf person is lesser than that of a hearing person (see discussion in Ladd, 2002; Blume, 1994; Wrigley, 1996). This leads me onto a brief reference to critical disability studies which looks to examine society's role in defining what constitutes disability.

3.7 Critical Disability Studies

Goodley (2016) defines critical disability studies as a "location populated by people who advocate building upon the foundational perspectives of disability studies whilst integrating new and transformative agendas associated with postcolonial, queer and feminist theories" (p. 190-191). In a 2013 paper, Goodley explores the emergence of the field of 'critical disability studies' (CDS). The field of *critical* disability studies appears to be markedly different from a more generic

disability studies' approach in its more universal application of 'disability' to represent a form of social oppression, resulting from any societally perceived difference from 'ablest' norms. Campbell's (2009) work in critical disability studies forefronts the ableist body, as the cast against which *the Other* is forecast, where *Other* refers to any self which is outside the expected normal being such in terms of ethnicity, class gender, sexuality. Critical disability studies include examination of language that is ableist or dis-ableist[12]. Titchoksky (2015) cautions of the dangers when disability is used as a metaphor for deficit. Eilers (2020) looked at the *'Disneyfication'* of the well-known fairytale, *Beauty and the Beast*. The author highlights how the plot rests on two paradoxical principles – (1) the notion that beauty lies within and (2) the binary notion of good/beautiful and bad/ugly pairings. Eilier (2020) maintains the film promotes the thinking that a 'disabled body' is a problem that must be fixed with the help of the heroic able-body. Considering the foundation beliefs within CDS may be of relevance when hearing the participants' accounts and making sense of their sense-making.

3.8 The Global Pandemic: Covid-19

During the time period in which this research was conducted, the World Health Organisation (WHO) declared the Covid-19 outbreak as a pandemic on 11 March 2020 (*WHO Director-General's Opening Remarks at the Media Briefing on COVID-19 - 11 March 2020*, 2020). The pandemic has had a direct influence on the nature of this study in terms of methodology (see *5.3 Data Collection*) and how the participants' experiences were lived through this time, at the level of the individual and society. From a whole culture perspective, the term 'diagnosis' has gained global recognition and diagnosis talk has permeated through society. The practice of diagnosis, historically reserved for the medical professional, has been entrusted to the public through self-identification of symptoms and home testing procedures. Ultimately, the Covid-19

[12] 'Disablism' has been defined by Thomas (2007, p. 73) as "a form of social oppression involving the social imposition of restrictions of activity on people with impairments and the socially engendered undermining of their psycho-emotional well-being". 'Ableism' can be understood as the societal preference and prescription for non-disability (Campbell, 2009; Goodley, 2014). Campbell (2009) described ableism results in a "network of beliefs, processes and practices that produces a particular kind of self and body (the corporeal standard) that is projected as the perfect, species-typical and therefore essential and fully human" (p. 44).

pandemic has established terms surrounding diagnosis such as 'symptoms', 'testing positive / negative' 'contact tracing' into our common everyday vernacular.

The authority of diagnosis has also been globally spotlighted. The diagnostic process has played a key role in managing the crisis. Kocak et al. (2020) in their paper on "crushing the curve…" cite the grave importance of diagnosis in the case of managing a pandemic in that "patients may be lost without a proper diagnosis". In these times, the evolving incidence of Covid-19, is determining how our economy and society functions. The example of a global pandemic also shines light on another property of 'diagnosis', its perishability and fragility. The emergence of Covid-19 catapulted the world's leading scientists in motion to search for suitable treatments and a vaccine that would prevent and eradicate the existence of Covid-19. 'Diagnosis' is therefore not a finite entity but something that can be altered, challenged or cured through the course of a treatment or passing of time.

3.9 Summary

From the discussion across Chapters 2 and 3 on previous research in the field, it is evident that diagnosis is a concept which permeates everyday life, within and outside of the practice of medicine and psychology. The impact of diagnosis on the 'inner' and 'outer' experiences of the individual and from the perspective of the caregiver has been cited in the case of a variety of chronic illnesses or differences[13]. Benefits of diagnosis have been associated with own and other identity construction, validation of internal and external feelings, and aiding family sensemaking. Caveats of diagnosis have been referenced in terms of pre-conceptions, labelling and stigma. I have attempted to present the pendular nature of diagnosis, which seems to swing in accordance with the perceived values and stigmatisations associated with a particular diagnosis, at the level of the individual and the society, which may be considered fluid and changing with social and political movements. My research aims to add to understanding as to the meaning of 'diagnosis' as it is experienced by mothers of children with a rare or undiagnosed neurodevelopmental condition in Ireland today.

In Chapters 4 and 5 respectively I will now discuss the methodology and methods for the current research, including a section on reflexivity.

[13] In keeping with preferred terminology used within the neurodiversity movement as it relates to autistic individuals.

Chapter 4: Methodology

4.1 Introduction

In this chapter, I will discuss my rationale for choosing IPA as a suitable primary methodology to address the research question at hand. I will also justify my decision for use of IPA informed by Narrative Analysis and briefly explore other considered methodologies. I will conclude with a detail of my own positionality as a researcher, and individual 'kaleidoscope' lens, through which I viewed each stage of the research process.

4.2 Justification of the Analytic Methodology

IPA is an especially appropriate research method for studying topics which are "complex, ambiguous and emotionally laden" (Smith & Osborn, 2008, p. 41). Core to IPA is the commitment to understanding the lived experience of a particular phenomenon for the participant in the context of which they live, as described in the extract below:

> With IPA, the objective is to get as close as possible to the lived experience of participants so that it can be examined in detail. Accordingly, IPA researchers aim for insight into what it is like to have an experience from the point of view of the person who has had it to elicit rich descriptions, trying to capture the emotions surrounding the experience and how people understand it and make sense of it. The personal meanings associated with the lived experience are considered particularly important in IPA, as it is how the lived experience related to people's views of their world and their relationships. (Smith & Nizza, 2021, p. 4)

IPA research is committed to understanding how people make sense of their experience of a particular phenomenon, which in this case refers to parents' experience of the phenomenon of 'diagnosis' as pertaining to their child with a neurodevelopmental condition. Smith (2011) speaks about the potential of finding *'gems'* in IPA research, with the gem as described as "the relatively rare utterance that is especially resonant and offers potent analytic leverage to study" (p. 6). Smith (2011) advises the gem is not guaranteed to be uncovered within every data account but that where it is, it offers windows of insight into the phenomenon being studied. The different types of gems, '*the spectrum of gems'* as described by Smith (2011, p. 6), will be elaborated on in *5.6 Reliability and Validity Considerations.*

IPA has been applied as a qualitative methodology to understand how individuals make sense of their experiences in a variety of health, disability, economy, societal and psychological contexts, such as but not limited to, the experience of living with chronic fatigue syndrome (Dickson et al., 2008), foster placement breakdown (Rostill-Brookes et al., 2011), the first episode of depression (Mith & Rhodes, 2015), following brain injury (Dwyer et al., 2019), bereavement following suicide (Gordon & McElvaney, 2021), chronic pain (Kirkham & Smith, 2015), and professional identity (Huff et al., 2019; Oakland et al., 2013). IPA has three philosophical underpinnings, which inform its approach. These are discussed by Smith et al. (2022), and as summarised below.

4.2.1 Phenomenology

Phenomenology accounts for IPA's interest in exploring the human experience, and on reflection (Smith et al., 2022). Heidegger (1927/2008), a phenomenological philosopher, described that a phenomenological method sets out to explore how we consciously interpret our experiences. Heidegger (1927/2008) speaks of intentionality in our attendance to something, whether that be real or perception of remembered reality. Husserl (1952/1980) recommended that the researcher needs to 'bracket' their own pre-conceived notions on the experience, to attend to the experience being studied. Heidegger and Gadamar are philosophers credited for *hermeneutic* phenomenology. *'Dasein'* (already referred to earlier in Section 1.4.1) translated as *'there-being'* is a concept coined by Heidegger, as discussed in his major work *Being and Time* (1927), which refers to the idea that one is born into a pre-existing world and cannot detach from pre-existing cultures and people and objects. In *Being and Time,* Heidegger (1927/2008) explains the derivatives of the term 'phenomenology' as being rooted in two Greek words: *phainomenon* and *logos.* Heidegger discusses the interpretation of these terms at length, cumulating the total definition of phenomenology as "to let what shows itself be seen from itself, just as it shows itself from itself" (Heidegger, 1927/2008, p. 81)

Van Manen (2017) regards phenomenology as the study of lived experiences, as detailed in the quotation below:

> Phenomenology, if practiced well, enthrals us with insights into the enigma of life as we experience it—the world as it gives and reveals itself to the wondering gaze— thus asking us to be forever attentive to the fascinating varieties and subtleties of primal lived experience and consciousness in all its remarkable complexities, fathomless depths, rich details, startling disturbances, and luring charms. Genuine phenomenological inquiry is challenging and

> satisfying precisely because its meaningful revelations must be originary and existentially compelling to the soul. (p. 779)

Van Manen (2017) criticises IPA's inclusion of 'phenomenology' in its title and proports that interpretative *'psychological'* analysis would be a more fitting term given within this methodology, given the researcher is said to be making sense of the participant who is making sense of their experiences, described as the 'double hermeneutic' in IPA (as detailed in *4.2.2 Hermeneutics* below). For van Manen (2017) it is this attention to how a person *understands* their experience, which moves the methodological aims from phenomenological, which pertains to the experience itself, to the more psychological realm. One might argue that in speaking about the experience (i.e., the phenomenological aspect), the participant is invited to both recount and meaning-make, at the same time (i.e., accounting for the more hermeneutic roots) and represents the constellation of methodological concerns underpinning IPA.

4.2.2 Hermeneutics

Hermeneutics contributes to IPA's focus on interpretation (Smith et al., 2022). IPA involves a 'double hermeneutic' (Smith & Osborn, 2003), in that the researcher is making sense of the participant's narrative, who is making sense of their experiences. The "hermeneutic circle" (Heidegger, 1927/2008) refers to the idea that to understand the whole, one needs to examine its parts, and to understand its parts, one needs to consider the whole. The notion of the hermeneutic circle has particular relevance during in-depth analysis whereby the researcher must analyse iteratively. Smith et al. (2022) detail that what constitutes 'the part' and 'the whole', exists across varying levels, for example *the part* may be the word to which *the whole* is the sentence containing that word, an extract as a part pertaining to the complete interview as *the whole,* and the interview *as part* relative to the entire research project.

Gadamer (2004, p. 370) introduced the notion of 'horizons of understanding'. He posits that researchers come to understand text through their own existing horizons and that new understanding of others' experiences brings with it a shift in horizons, denoted 'fusion of horizons'. Much of Gadamer's (2004) writings are concerned with the influence of the past on experience. He maintains that "every experience has implicit horizons of before and after, and finally fuses with the continuum of the experiences present in the before and alter to form a unified flow of experience" (p. 237).

4.2.3 Idiography

Idiography is concerned with the particular, namely how a particular phenomenon is experienced by a particular individual, within a particular context (Smith et al., 2022). Idiography also refers to IPA's ability to utilise rich analyses of individual cases to assist the formulation of more general claims.

4.3 Consideration to Alternative Methodologies

In the following chapters, I will show how IPA presents a good fit to address the research question at hand. However, in the process of designing this study I considered several alternatives. In this section, I will detail some of the qualitative methodologies which I considered and, where referenced, have borrowed from in order to enhance the use of IPA as applied to this topic.

4.3.1 Narrative Research: Borrowed Insights

Narrative research as a methodology lends itself well to the aims of my research study in terms of how I interpreted the data, and how I considered the implications of the findings. Chataila (2005) described narrative inquiry as "the process of gathering information for the purpose of research through storytelling" (p. 2). Stories heard through narrative research offer insights into cohorts which have been underrepresented and less heard (Goodley, 1998; Lincoln & Guba, 1985), which is relevant to the topic of my research (i.e., rare and undiagnosed neurodevelopmental conditions). In narrative research, similarly to IPA, the researcher occupies an active role in the process of hearing stories and making those stories 'hearable' and has been described in the literature in terms of various artforms such as comparisons to craftspeople who paint or sculpt their impressions (Hollway and Jefferson, 2000), cake makers (Chataila, 2005) or tailors (Cotterill & Letherby, 1993). Narrative research is concerned with the legitimacy of truth by experience as opposed to objective factuality (Chataila, 2005). This assertion aligns with the view of IPA, that the objective truth is not under speculation of the researcher. The researcher is instead interested in pursuing truth as *perceived, remembered* and *made sense of* by the participant.

Narrative research has variants, adopted by different researchers. The type of narrative analysis (NA) I will now refer to is that described by Josselson and Hammack (2021). I feel NA has shared epistemological underpinnings, research objectives and methods, to IPA. NA can be considered a hermeneutic endeavour in its intent to shine new understanding on a living experience through meaning-making. Similar to IPA, NA is concerned with understanding the experience of *'how'* something is lived and is less interested in generalisation. In NA and IPA, in-depth semi-structured interviews are often used as a vehicle to gather rich data. NA also recognises the role of social

context on how people perceive reality. A commitment to analysing the data for stated and hidden meanings is also common to both approaches. In NA, researchers look for "explicit" and "implicit" meaning. This is similar to what researchers denote "descriptive" or "conceptual" notes in IPA, respectively. What NA terms 'hermeneutics of demystification' (Josselson, 2004) could be considered similar to the process Smith (2011) describes in his titled article "'We could be diving for pearls': The value of the gem in experiential qualitative psychology". Here, Smith (2011) talks about the process of 'peering' and 'unpeering' in identifying a 'gem' (p. 1) from a participant's whole account. Smith (2011) elaborates that there is a spectrum of gems, from 'shining' through 'suggestive' to 'secret', and they are differentiated based on the amount of detective work required to surface their meaning. It is evident that NA and IPA share some theoretical underpinnings and overall determinations. Given this similarity, I consider it methodologically aligned and congruent to borrow from NA in order to address the research question at hand. I have named each narrative account as a named participant's 'story'. This denotation, which was reached upon through close readings of the transcripts, applies a narrative analytic lens which was employed in order to best make sense of, and present the data.

Additionally, I consider the work of Arthur Frank (1995, 2005) on illness narrative and narrative typologies as relevant to illuminating the experience of the participants in this study. Theories and methods from Frank's (2010) dialogical narrative analysis were useful in assisting interpretation of the data, specially what he refers to as "*'getting it'"* (p. 87). Frank (2010) speaks about interpretation being inherent in storytelling. He qualifies that both tellers and listeners are constantly interpreting each other. This notion can be viewed similarly to IPA's focus on 'double hermeneutics', in that there are multi-levels of analysis occurring. I have previously introduced the work of Frank (1995, 2010) (see *1.4.3 Illness Narrative; 2.4 Diagnosis: Shaped by the Individual and Society; 3.3 Illness Narrative: Definitions and Considerations)* and will elaborate further on the relevance and application of his work to the current study as relevant. Within *14.3.1 Exemplar in IPA: Borrowed insights from Narrative Inquiry* I will present a narrative 'sense' for each of the participant.

Criticism of narrative research questions the balance between representing the storyteller's voice and the researcher's interpretation (Goodley, 1998; Chataila, 2005). Indeed, the right of the researcher to use 'disability' as an object of intellectual inquiry has been questioned by Goodley et al. (2019). A potential answer for this might be in looking to the 'so what?' of narrative research. Chataila (2005) reconciles this concern by asserting:

> The goal of narrative research is to provoke commentary on what researchers, readers and activists might do next. In other words, narrative research can be justified if it involves using stories that are worth telling; that have moral and political purpose to effect positive change in policy and practice, and that progress understanding. The question then that lies to narrative researchers is not to undermine disability activism through the stories. The issue is not of reducing lives of disabled people to mere stand-alone stories. (p. 8)

In conclusion, ultimately, I favoured IPA as my primary methodology. IPA's main objective is more closely aligned to my exact research question, to understand how the participants' make sense of a *particular* phenomenon, in a *particular* context. The participants in my research were asked to share how they experience the phenomenon of study, as opposed to tell a story of their journey (or more astutely, '*witness'* illness experience). Narrative Analysis, although similar in its methodological intent, is first and foremost 'story work' so to speak, that is, understanding through *storytelling*. I was also attracted to IPA's method of analysis, notably its explicit reference to 'linguistic' commentary as part of exploratory note making. As a speech and language therapist, this specific pursuit to examine language in order to understand meaning for the participants in their experience, matched my natural analytic lens; it supports me to gain entry into clients' or others' worlds through attending to *how* they speak of their experiences. The multi-layered analytic process (i.e., to make descriptive, linguistic, and conceptual commentary on the data) allowed me to excavate the accounts to reveal the living experiences of the participants.

4.3.2 Discourse Analysis

Other analytic methods, such as discourse analysis (DA) is concerned with examining the co-construction of meaning through the study of interactions amongst particularly institutional and other discourses (Gale, 2010). It draws parallels with IPA with regards to its attribution of the influence of context on any interaction. The notion of dual hermeneutics in IPA odes to this same viewpoint in that the researcher's role in the data collection episode is not regarded as a passive, uninfluential entity but as a key component in the double sense-making process which has influence on the analyses and conclusions drawn from the data. For example, DA may provide a useful analytic tool for a healthcare clinician to shed light on how one's talk influences the

interaction in clinical practice, for example in the area of dysphagia[14] practice (Walsh & Leahy, 2009) and working with individuals with communication disorders associated with stroke and schizophrenia (Walsh, 2007). Although the context of the phenomenon being studied (i.e., the sphere of 'diagnosis' in childhood rare or undiagnosed conditions) will likely include interactions in clinical discourse, the current research question is primarily concerned with *how* a particular human phenomenon is experienced as opposed to how the experience is navigated through talk. IPA is therefore a more appropriate methodology to address the current research.

4.3.3 Grounded Theory

Grounded theory (GT) was developed by Glaser and Straus (1967). GT is a method for researchers aiming to study processes (Charmaz, 2012). Charmaz (2012) discusses how GT address the "why" questions, as opposed to the "what" or "how" questions. IPA as a method and methodology seeks to understand *how* a particular experience is made sense of. GT sets out to make more generalisable claims about specific groups and often achieves this by employing a larger sample size (Smith et al., 2022). One of the key differences that separates GT from most other qualitative approaches is the focus on simultaneous data collection and analysis. In grounded theory, data analysis and data collection occur together, in that analysis of data guides where to source further data (Foley et al., 2021). Theoretical sampling is employed in search for an abstracted theory that is grounded in the data (Glaser & Strauss, 1967/2012; Foley et al., 2021). Theoretical saturation is said to be achieved when no new categories emerge from analysis of new data (Charmaz, 2012, 2020; Charmaz & Thornberg, 2021). The aim of grounded theory is to arrive at theoretical saturation of the data where all concepts and categories to arrive at a theory. Grounded theory is a commonly employed method in healthcare research to understand processes from patient perspectives (e.g., the healthcare experience of people with amyotrophic lateral sclerosis as detailed by Foley and Timonen (2015). While GT is concerned with identifying processes, IPA is looking for rich, thick descriptive accounts pertaining to the particular. IPA lends itself to exploring experiences from individual accounts in great detail, which can often provide the basis for macro analysis of the same topic as carried out using grounded theory. I chose to employ IPA's more detailed lens for the current research due to the scarcity of parental accounts of diagnosis in the Irish context so that subtle, deep insights can be brought into the professional,

[14]Dysphagia refers to eating, drinking and swallowing difficulties (Royal College of Speech and Language Therapists [RCSLT], 2022).

medical, research, political and legal fields. Future research may seek to understand healthcare processes for this cohort, using GT.

4.3.4 Thematic Analysis

Braun and Clarke (2021, 2022) discuss the main differences between IPA and reflexive thematic analysis (TA), a sub-type of TA described by Braun and Clarke (2021, 2022), which involves coding and researcher generated themes through subjective interpretation. The key differences include IPA's "*dual analytic focus"* (Braun & Clarke, 2021, p. 41) in attending to the generation of themes across individuals and at the level of the individual, and IPA's approach to analysing individual cases before looking to identify themes across cases. TA instead identifies themes across participants from codes assigned to all accounts. Braun and Clarke (2022) distinguish TA from other qualitative methods in asserting it is more of a method than a methodology. Braun and Clarke (2022) describe three variants of TA. Reflexive TA recognises the role of the researcher as a valued element. Similarly to IPA, themes are considered produced by the process of the researcher actively engaging with the process and data, as opposed to being passively uncovered. Reflective TA's interpretative commitment assimilates with the fundamental pursuits of IPA. IPA also includes a linguistic focus at initial coding, which is not included in TA. This addition is welcome to me as a researcher, given my linguistics' orientation as a Speech and Language Therapist, as previously referenced within my justification for choosing IPA as my primary methodology. As such, I am interested in the discrete and nuanced capabilities of language and discourse.

IPA is ultimately recommended when addressing research questions that aim to understand human experiences and meaning-making in relation to a particular phenomenon with a small homogenous sample (Braun & Clarke, 2021). IPA allows for a deeper engagement with the data and preservation of the participant's *unique* and idiosyncratic experience within their lifeworld, which aligns with my research aims within the current study.

Therefore, having remained open to considering other qualitative methodologies throughout the research process, I ultimately found that IPA offered the most fitting approach to answer the research question at hand, along with borrowed insights from narrative research. The theoretical origins of IPA and the proposed methods, including the attention to descriptive, linguistic, and conceptual elements within the data (as will be explored in *5.5 Procedure for Analysis*) also align with my own positionality as a researcher (as detailed in *4.4 Researcher Positionality: Influence of*

the Researcher below). Smith (2019) positions the "centre of gravity" (p. 167) of IPA at the level of "what does it mean?" (p. 168), which - along with how they make that meaning- is ultimately what I want to know for the mothers in relation to the phenomenon under investigation.

4.4 Researcher Positionality: Influence of the Researcher

The researcher arrives to the participant with their own pre-existing knowledge and preconceived notions of the world, which they have acquired through their own social experiences and the existing historical world they were born into. What the researcher brings to interpretation contributes to what makes this, and by extension other IPA studies, inherently unique. Another researcher will undoubtedly be looking through a different lens in terms of their prior entry into the social world. As already referred to previously, the researcher within IPA is regarded as an active contributor to the research design and process. This active contribution is aligned with hermeneutic phenomenology (Laverty, 2003). In contrast, purely phenomenological approaches unlike IPA, require the researcher to engage in a period of self-reflection and reflexivity for the purpose of bracketing so that one's own biases do not influence the interpretation (Laverty, 2003).

Reflexivity has been defined as a process that helps researchers to consider their position and influence during their study and helps them know how they have constructed and sometimes imposed meanings on the research process (Crotty, 1998). Reflexivity is "self-critical sympathetic introspection and the self-conscious analytical scrutiny of the self as researcher" (England, 1994, p. 244). Using Gadamer's (2004) terminology a researcher's viewpoint can be understood as our '*horizon of understanding'*. Transferring my thoughts onto the page, in this section, for me embodied what Smith et al. (2008, p. 1395) described that "writing brings the unsaid into the open space where ideas are exposed to interpretative gaze, to wonder, and to ask still more questions". As referred to in *1.5 Summary*, from this point on I have included some samples of my own reflections, which I maintained in a reflective log throughout the research process. These will be highlighted as numbered 'Researcher Reflections' (in line with IPA methodology, as exemplified by Walton, 2018).

4.1 Researcher Reflection

Within personal communications with Michael Larkin (July 7, 2022), a founder of IPA, he outlined IPA's commitment to understanding the person-in-context "is very specifically consonant with a hermeneutic phenomenology, and probably doesn't need the broader landscape of contextualism". From his point of view further detail on ontological position is not

required, as he attests when collecting data within any research, ontology as methods which collect data assume there is a real world. Larkin (July 7, 2022) advises in explaining which components of phenomenological and hermeneutic ideals are relevant to the researcher and how the researcher intends to analyse and make sense of the data, the researcher has sufficiently described their epistemological position.

4.4.1 Researcher Stance: My 'Kaleidoscope' Through which I View the World - The Shape-Thrower which Crafts my Research Framework

Savin-Baden and Major (2013) define 'personal stance' as the position taken towards an issue derived from one's own beliefs and views of the world. This stance offers a projection of one's core moral code. It can be more wholly defined using the German term '*Weltanshcauung',* coined by Immanuel Kant (1970) and later used in the workings of Wilhelm Dilthey (1976), which translates as '*worldview'.* I perceive '*Weltanshcauung*' as not a static entity. I believe it is co-constructed through acculturation and is in constant evolution as we encounter new exchanges, experiences, and environments. I perceive the human mind and body as frail and vulnerable to influences from external and internal sources which can enact transient or lasting changes to one's world view. I would like to introduce the notion of a kaleidoscope which I envision as symbolic of my *Weltanschauung*. The combination of the whirling swirls, constitution and distribution of shapes, the explosion and amalgamation of colours is representative of my world view. As I see it, this kaleidoscope reflection is constantly in flux and sensitive to time. Each time I peer through the viewpoint, the image generated is specific to that moment in time, and will be different, either discretely and markedly, on the next turn. I would like to acknowledge the scope of our oscillating landscape may be somewhat bounded by less consciously visible constraints. Such constraints may be ascribed to our sociocultural milieu and broader worldview, as determined by our own individual contextual upbringing within Western society. In saying this, I would like to now invite the reader to look through my 'kaleidoscope', which I have attempted to bring as consciously as possible to the forefront, in recognition of the potential limitations of my lens. The purpose of doing so is to be transparent about the potential influences on my research process and findings.

As a healthcare professional, I am bound by ethical, legal, local and professional standards. In my training, my modules, lectures, peers, practice educators and clients all had influence in shaping my identity as a Speech and Language Therapist. My specific place of work, the clinical caseload, colleagues, and structural organisation all contribute to shaping my evolving practice, perspective and philosophies as a clinician. It was my chosen profession and career path that led me to

cultivate my research question in the first instance. My personal experience of working with families whose children had diagnoses of complex neurodevelopmental disorders, and notably, those who had no named diagnosis, propelled me to wonder how these parents experienced diagnosis as a phenomenon. I acknowledge that I have some pre-conceived notions of what this experience might be given my consultation with families within my daily practice, my interest and active engagement in the field through literature reviews and attending symposiums on the subject. It is important that I acknowledge any pre-existing ideas I may have about the parental experiences so that I can consciously put this information aside when engaging in interviews with participants to actively seek the truth and not the potential truths that I have pre-determined.

In addition, I would like to acknowledge my specialised skillset as acquired through my professional experience that may act as enabling factors in my data collection. Inherent to my professional skills and practice is the practice of discrete observational skills that is, monitoring, noting and responding to participants verbal and non-verbal behaviour in real time whilst simultaneously being engaged in other tasks. Building relationships with clients and colleagues is another core requirement of my role. These skills supported me in facilitating the participant to be at ease and facilitate more in-depth, raw reflections which will in turn provide more complete data for analysis and ultimately enhance the validity and richness of later findings.

As a healthcare professional, I am hardwired, likely through my own inherent personality traits which drew me to the profession in the first instance, and through my professional training, to possess a sense of compassion for the patients who I work alongside. One definition put forward by Gilbert (2009) defines compassion as "a deep awareness of the suffering of another coupled with the wish to relieve it" (p. 13). On reflection of my own emotional state directly following participant interviews I have noted a concoction of conflicting emotions that ensue. My immediate feelings post interview appear to drop like coins, each with two sides, a sense of pride and a pull of despair.

4.2 Researcher Reflection

At times during the participant interviews, I noted feelings of intimidation or fear in interactions with the participant. I was so astutely aware of the parents' disappointment and strong references to the impact of the words of healthcare professionals looks, lasting words that there was a heightened sense of responsibility I felt to not be another source of disappointment or annoyance for the participant. At times, it was hard for me to hear the participants dismay for their experiences

in settings that emulate where the I work, as a representative of one of the healthcare professionals referenced. I was cognisant of so desperately not wanting to add to any burnings on the participants memories – 'pyrography' - words and looks of healthcare professionals leaving permanent marks on the participants' memories like pyrographic markings on a wooden surface.

Outside of the workplace, family, friends, and travel experiences will undoubtedly influence the way I think and approach situations and how much I can relate with the participants' experiences. Hellawell (2006) speaks to the "inside-out" (p.483) phenomenon in relation to students' relationships with their informants. She describes how researchers can relate to their participants along some dimensions of "insiderness" (p. 490) and simultaneously hold "outsiderness" (p. 490) along other dimensions. She attributes self-awareness of this position as something which can aid doctoral students in achieving higher quality levels of reflexivity. Perhaps my above reflections and sense of emotional conflict can be explained by this "inside-out" paradigm. On the one hand I am very much "inside" the informants' experiences, 'I' am referenced in many of their accounts, not me personally per se, but as a representative of my profession 'speech and language therapist' and by extension, the wider title of 'health care professional' or indeed any medical personnel. Participants recount their experiences in the very setting which I work (i.e., acute paediatric hospital) and settings which I have previously worked (i.e., paediatric disability service). This brings me in, as very much part of the participant's experience, a symbolic character in their narratives. On the other end of the spectrum, I am *not* the speech and language therapist or HSCP in question. I was not, or am not, responsible for the participant's experience in healthcare or determining management and interventions. I also differ from my participants in that I am *not* a parent. I am *not* married. I also often differ on the age continuum to my participants. In this way, I am "outside" their experiences. My level of "inside-out" may vary amongst participants. For example, one participant is also a healthcare professional which gives further "insiderness" and assimilation with this participant's experiences. Understanding and naming this parallel has aided me in 'making-sense' of my reactions to 'making-sense' of the participants 'making-sense' of their experiences. This layer of reflexivity might be termed a *'triple'* hermeneutics.

My professional role as working in the paediatric hospital at the time of conducting the parent interviews afforded me another layer of "insiderness" being able to share the visions of the physical space, mental processes and inner workings of a paediatric hospital setting. This experience helped me to be 'on' the journey with parents as they detailed their experiences and surroundings. This ability to mentally transport, with the participants storytelling, the place

setting of their experiences, served to enhance my ability to see and understand their situation and ultimately the influence of these structures on their individual experiences.

As a researcher, in keeping with Larkin's (July 7, 2022) guidance, as referenced in *4.1 Researcher Reflection*, and in terms of hermeneutic phenomenology, I strongly support the notion of 'Dasein' and lean on it heavily in terms of how I interpret my data. I find the following quote from Arthur Frank (2010) nicely illuminates the philosophy of 'Dasein' that is "Stories echo other stories, with those echoes adding force to the present story. Stories are also told to be echoed in future stories. Stories summon up whole cultures" (p. 37). Goldspink and Engward (2019) re-purpose the use of the term "echoes" to refer to the interlacing of the participant's and researcher's words and experiences during the research process. Goldspink and Engward (2019) refer to the 'professional-self' and 'researcher-self'. I would like to acknowledge a third self, the 'personal-self'. This conscious separation of the selves and multiplicity of identities foregrounds the intricacies of achieving the balance between faithful description of the participant's accounts in combination with suspicious and curious interpretation of the analyst, both of whom are people-in-context, and cannot be separated from their life-worlds.

In sum, my professional, research and personal selves all contribute to how I conducted this research, made sense of the participants making sense of their experiences, interpreted the data and presented implications for IPA, diagnosis, policy, education, and practice. This individual influence is to be lauded within IPA, as described by Braun and Clarke "researcher subjectivity is a fundamental resource for IPA" (p. 41).

4.5 Summary

In this chapter, I introduced interpretative phenomenological analysis (IPA), as the most suitable choice of methodology to explore the research question at hand, discussing its theoretical underpinnings and overall pursuits. I introduced how and why I will use borrowed insights from narrative analysis to complement my chosen methodology and to best illuminate the findings. I also offered an exploration of other qualitative research methods and their potential strengths and weakness in studying the phenomenon of focus in this research. I concluded this chapter with an introduction to my position as a researcher, in keeping with IPA methodological considerations. The next chapter, Chapter 5, will focus on methods used.

Chapter 5: Methods

5.1 Research Design

This is a qualitative study exploring how five mothers make sense of 'diagnosis' in relation to their child with a rare or undiagnosed neurodevelopmental condition using Interpretative Phenomenological Analysis (IPA) with borrowed insights from Narrative Analysis (NA). Accounts were gathered through individual in-depth semi-structured interviews carried out, via an online video conferencing platform (i.e. Zoom, Zoom Video Communications Inc., 2016). The video recordings were later transcribed, pseudonymised and analysed using a method consistent with IPA methodology.

5.1.1 Change to Research Design

Originally, in this research, I intended to study parents' experience of diagnosis where their child had a rare or undiagnosed disorder, exclusively. During the early stages of forming the research question and in consultation with my research supervisors, I altered my original research design to include parents from two groups, namely (i) parents of children who have a child with a rare or undiagnosed neurodevelopmental condition, and (ii) parents of children who have Down syndrome. I had planned to conduct semi-structured interviews and then to invite participants to be involved in an optional subsequent, multi-perspectival focus group, with a maximum of 10 participants. The logic was that parents of children with Down syndrome had a specifically different diagnostic journey in that diagnosis is typically antenatal or soon after birth. Diagnosis can also be strictly confirmed on genetic testing and there is wider public and professional knowledge, support services and clinical pathways established for children with Down syndrome, in comparison to those with rare or undiagnosed neurodevelopmental disorders in Ireland. The thinking was that consideration of the parents' varied experiences of 'diagnosis' may help to illuminate the unique experiences of each group's living experience (i.e., parenting a child with a known genetic diagnosis or parenting a child with a rare or unknown diagnosis).

No parents of children who have a diagnosis of Down syndrome expressed interest in participating in the study. Therefore, I proceeded to conduct the interviews with the cohort of parents of children with rare or undiagnosed neurodevelopmental conditions. Having completed these interviews, I believed the data I obtained could contribute unique and novel findings on an under researched area (i.e., how is the meaning of 'diagnosis' for parents of children with rare or undiagnosed neurodevelopmental conditions experienced and perceived, in the Irish context). On

reflection at the time (and since), and in consultation with my supervisors, I also determined that inclusion of a second parent group would have ultimately dilute the breadth of findings that could be analysed and presented relating to the original topic inspiring the study. In terms of a resultant smaller sample size, Smith and colleagues (2021) explicitly state "there is no right answer to the question of sample size" (p. 46). The authors advise that the richness of the individual case and in-depth of analysis and write-up is more important than quantity. The authors also note that given the complexity of human experience, which IPA aims to understand, such studies often in fact *benefit* from smaller sample sizes. Smith et al.'s (2021) advice provides understanding and justification for my decision to include five participants only in this study.

5.1 Researcher Reflection

"Stories are powerful research tools. They provide us with a picture of real people in real situations, struggling with real problems" (Witherall & Noddings, 1991, p. 280).

The above quotation for me provides further support for the decision to include one group amongst the research sample (i.e., parents of children with rare or undiagnosed conditions). As well as considering including a different cohort of parents, I considered gathering information from healthcare professionals on the same phenomenon. Larkin et al. (2019) explored how multi-perspectival designs can permit attention to further dimensions of a chosen phenomenon. Larkin et al. (2019) proposed multi-perspectival designs can increase the inferential leverage of experiential studies. However, given that the group being studied are inherently a marginalised cohort and there is limited research on parental experience of rare or undiagnosed conditions in Ireland, I decided privileging the experience of these five participants seemed an important first step to add new insights to the literature. I also had concerns that including additional sample groups would by virtue, dilute the depth of focus that could be afforded to each of the five mothers.

5.2 Participant Recruitment

I disseminated the Participant Information Leaflet (PIL) (see Appendix B) to potential participants via two gatekeepers: Rare Diseases Ireland (RDI) and Down Syndrome Ireland (DSI). Other relevant interested parties (e.g., 'SWAN Ireland - Syndromes Without A Name' parent support network), re-shared the link to RDI and DSI's invitation to be involved in research across social media platforms.

5.7.2 Inclusion Criteria

Any participant who I had previously or was currently working with in providing direct clinical interventions with their child was excluded from taking part in this research. There were no age restrictions pertaining to the definition of 'child' at the time of the interview (i.e., parents could participate if their child was over 18 years of age).

5.7.2 Demographic Information

Table 5.1 summarises the profiles of the participants. Table 5.1 Introduces the pseudonyms assigned to the mothers, their child, and other named family members referenced by the participants during their interviews. I also included information surrounding the child's diagnosis, and age at the time of the interview, (as collated from the participants' accounts) to support the reader in contextualising the findings.

Table 5.1

Participant Descriptive Information

Participant (Mother)	Child	Diagnostic context / child presentation	Child age at time of research interview (years)
Robyn	Nathaniel	Antenatal diagnosis of brain malformation. Rare disease diagnosis at around one year of life.	7
Claire	Rose	Public health nurse referral to physiotherapy and speech and language therapy following 12-week developmental check. Identification of seizures at around 5 months of age. RDDX at around 10 months of age, which was later revised to RDDY approximately 6 months later.	3
Mary	Alexander	No unifying diagnosis (i.e., syndrome without a name).	7
Judy	Declan	Recurrent chest infections and GP visits from 7 months of age. Other issues including a skin condition, speech difficulties, movement condition. RDD at six years of age.	7
Olivia	Anna	No unifying diagnosis (i.e., syndrome without a name) and unknown disability.	3

Note. The mothers are listed above in the order in which they attended for interview; mothers and children are referred to via pseudonyms; RDD refers to rare disease diagnosis.

5.3 Data Collection

The semi-structured interview has been described as "…the means by which the researcher can gain access to, and subsequently understand, the private interpretations of social

reality that individuals hold" (Minichiello et al., 1990, p. 87). It allows for increased relationship building with the participant and collation of rich data (Smith & Osborn, 2008). The aim of the semi-structured interview is to facilitate the participant in telling you what it is like "to live in their personal world" (Smith et al., 2009, p. 61). The researcher's role in the semi-structured interview, is as a facilitator to guide, rather than direct the interview (Smith & Osborn, 2008). The individual interviews were carried out online between May-October 2021, via the video conferencing platform, *Zoom* (Zoom Video Communications Inc., 2016). Face-to-face interviews were not permitted at the time of data collection, due to public health guidelines to prevent the spread of Covid-19. I consulted examples in the literature illustrating the robustness of qualitative interviews remotely to justify use of this method of data collection (e.g., Archibald et al., 2019; Dodds & Hess, 2020; Eigege et al., 2022). Each interview was 1-1.5 hours in duration.

The practice of preparing an interview guide is advised to support the researcher in enquiring about potentially sensitive questions (Snow et al., 2009). I prepared an interview template with guiding probes (see Appendix C) based on a critical review of the existing literature on the subject being explored, and my own clinical experiences working with families of children with rare and undiagnosed conditions. I also used several techniques, typical to interviewing, such as 'funnelling' to gradually approach more sensitive issues and prompts to use, as needed, to support the participant in accessing the question, particularly for more abstract questions.

5.3.1 Subsequent Data: Update to Olivia's Story

The main body of the data, therefore, consisted of 5 transcribed interviews, and subsequently formatted to allow for IPA analysis. An update was received from Olivia by email in April 2022 in response to my request to all participants to review the transcript for any identifying details. Olivia provided written consent for the inclusion of this update within the dataset. I have chosen to include Olivia's update as it captures the notion the mothers' narratives must be considered 'living' (i.e., a term previously introduced in 1.1. Inspiration for the Research, see footnote 2) experiences. Life is not static and is in constant evolution. How we perceive our continued realities is subject to new experiences and evolving social, cultural movements, which overlays our existing histories and worldviews. This data should however be considered distinct in comparison to that gathered 'live', as part of an unfolding dialogue, within the semi-structured interview. Written entries also afford the time and space for participants to reflect on their lifeworlds and to narrate from a point of retrospection (Cudjoe, 2022). Cudjoe (2022) illustrates the use of written diary entries as a method of data collection in line with the underlying methodology

of IPA. Where data from Olivia's written account is included in the analysis, the line number will be marked with 'w', so as to distinguish the line numbers from her spoken narrative.

5.4 Ethical Issues

This research project received full ethical approval from the Research Ethics Committee (REC), School of Linguistic, Speech and Communication Sciences, TCD on 30/04/2020 (see Appendix A). Further to review of the requested documents, the Data Protection Officer confirmed he was satisfied that the intended processing of personal data for the purposes of the study were compliant were data protection legislation and commended the quality of the document as prioritising the importance of the participants' privacy rights throughout the data processing lifecycle. I will now detail how I obtained informed consent and how I considered the need to safeguard participant anonymity, with reference to the benefits and costs of these measures in terms of contextualising the sociocultural identity of the participants.

5.4.1 Informed Consent

Participant Information Leaflets (PIL) were disseminated by gatekeepers to potential participants (see Appendix B). Informed consent was ensured by providing potential participants with relevant information about the study, such as the aims and objectives, and an opportunity to contact the researcher or research supervisors should they have any further queries regarding the research. At the outset of the interview, I reviewed an abridged summary page of the PIL (see Appendix E) with the participant via sharing the document on *Zoom (Zoom Video Communications Inc*., 2016). I emailed the participant a link to access to the online informed consent form, created using Microsoft Forms. I provided support for the participant to complete this form, as needed.

5.4.2 Anonymity and Confidentiality

To protect the anonymity of all those involved within the child and parent illness experience, participants were asked at the outset of the individual interviews, to avoid using real names or providing other identifying information. Raw electronic data (i.e., saved interview files from *Zoom Video Communications Inc*., 2016) were stored on the researcher's individual password protected computer within encrypted external hard drives. Participants were allocated pseudonyms for use on subsequent stored data records, including transcriptions and hard copies of transcripts. The code key that linked personal data to other data was stored in a secured location in the research supervisor's office in the host institution, in a separate location to the stored raw data. Only the researcher and supervisor had access to the data in its original form. If there were any incidents relating to breaches (or suspected breaches of personal data) the researcher and supervisor pre-agreed to inform the Data Protection Officer at Trinity College Dublin. No such

incidents occurred. Participants were given the opportunity to review (within a specific time period) their pseudonymised transcripts and to suggest any changes to further redact any information which they believe may have been identifying.

In addition to the pseudonymisation of participants' names, I chose to gather limited information on the sociocultural identity of the participants (such as geographical location, marital status, highest level of education, parent occupation, ethnicity, socioeconomic status, family structure, etc.). Such information is typically sought in qualitative studies to assist interpretating the generalisability of the findings to other similar groups, and to support understanding of what factors may influence how a phenomenon is experienced. However, I decided to specifically not seek detailed demographic information, to further protect the confidentiality of the participants, given the rare nature of their children's' diagnoses and the exceptionally small community of such parents in Ireland (which would make it easier for them to be identified). For this same reason, I redacted information on the child's specific rare disease diagnosis and replaced the term with the abbreviation 'RDD' (i.e., to denote 'rare disease diagnosis'). The cost of omitting detailed demographic information may render less insights on how sociocultural factors may have influenced the participants' experiences. Such information may be sought in future studies with less qualitative focus where less sensitive and personal data is presented. Given the participants in my study also share information on other persons in their child's life, such as siblings and husbands (who were not required to consent to have information about them included in this study), I considered it important to take extra precautions to protect the participants' *and* their families' identities lest referencing others in the data could result in upset, on identification.

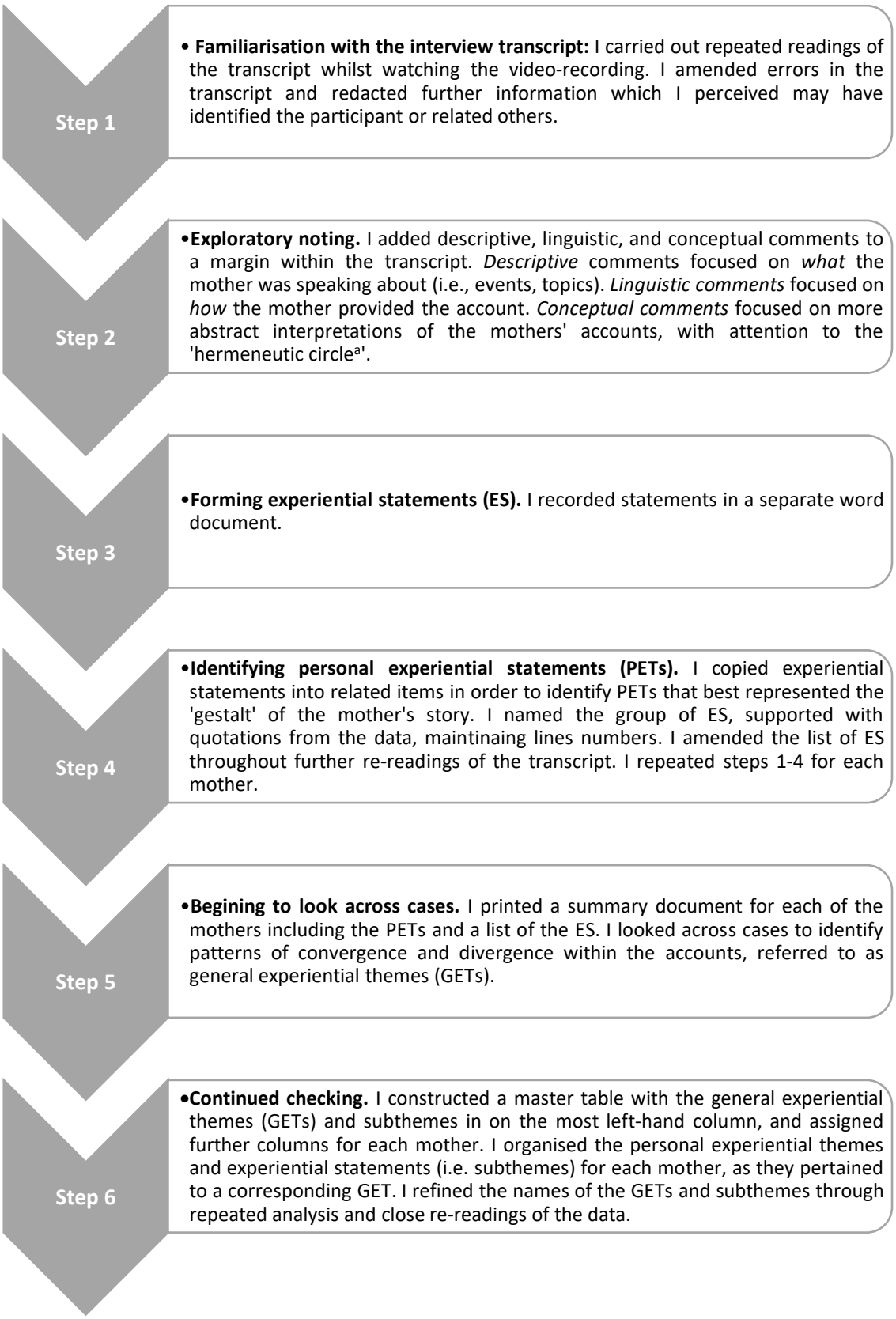
5.5 Procedure for Analysis

IPA provides a flexible set of guidelines for analysis which are in line with the interpretative, idiographic, and hermeneutic nature of the approach (Smith & Osborn, 2003; Smith & Nizza, 2022). I have followed guidelines for the novice IPA researcher (Smith et al., 2021; Smith & Nizza, 2022), in combination with some accommodations to suit my personal analytic style as a researcher. I have used the new terminology proposed for IPA research, whereby *emergent themes* were replaced by *experiential statements* (ES). A collection of related experiential statements now forms *personal experiential themes* (PETs), which were previously denoted *subordinate themes.* These new titles are considered to be more reflective of the analytic process (Smith et al., 2021). Each individual's interview transcript was analysed on its own (i.e., Steps 1-4) before looking to cross-case analysis (i.e., Steps 5-6) to identify *group experiential themes* (GETs), as is consistent with IPA approach (Smith & Nizza, 2022; Smith, Flowers & Larkin, 2021). Please refer to Figure 5.1 below for a visual

presentation of the procedure I used for analysis based on guidelines from Smith et al. (2021) and Smith and Nizza (2022).

Figure 5.1

Procedure for Analysis – Adapted from Smith et al. (2021) and Smith & Nizza (2002)



[a]The 'hermeneutic circle' (Heidegger, 1927/2008) refers to the idea that to understand the whole, one needs to examine its parts, and to understand its parts, one needs to consider the whole.

5.6 Reliability and Validity

Yardley (2000, p. 219) presented four domains to be considered when assessing validity in qualitative research, namely: (a) sensitivity to context; (b) commitment and rigour; (c) transparency and coherence; and (d) impact and importance. More recently, Nizza et al. (2021, p. 3) identified four quality indicators specific to IPA, as defined as: (a) Constructing a compelling, unfolding narrative; (b) Developing a vigorous experiential and/or existential account; (c) Close analytic reading of participants' words; and (d) attending to convergence and divergence. I will now detail how I attempted to fulfil each of these markers in my current research.

5.6.1 Sensitivity to Context

Goldspink and Engward (2019, p. 298) stated "Data analysis is not a single, detached activity, but one that is intrinsically connected to the complex and dynamic life world of the researcher". In recognition of this, Yardley (2000) advises the researcher attends to reflexivity. My researcher stance has been introduced in 4.4.1. As previously introduced, I also maintained a research log throughout the research process which I used to record, and bring to conscious awareness, my own reactions. I have included these throughout the write-up in boxes labelled '*Researcher Reflection'.* I maintained sensitivity to context during the analytic process (as defined in *5.5 Procedure for Analysis* above), through iterative readings and re-reading of the transcript. Additionally, I included for each participant, my invitation to begin their narrative (as shown in *6.4. Entry Point to the Mother's Accounts)* to attempt to acknowledge the co-constructive nature of the dataset.

5.6.2 Commitment and Rigour

Commitment and rigour pertain to the sufficient sampling of the data. Hennink and Kaiser (2022) concluded that saturation can be achieved with small sample sizes, particularly with homogonous groups and where the study has discrete aims. I have clearly outlined my rationale for the data sample within *5.1 Researcher Reflection.* O'Reilly and Parker (2012) contend that relevance of saturation needs to be considered within the context of the research aims, claiming "The adequacy of the sample is, therefore, not determined solely only the basis of the number of participants but the appropriateness of the data" (p. 195). For this research, I considered deep, rich, and thick descriptions illuminating both similar and different experiences on the parents' meaning making of diagnosis in relation to their child with a rare or undiagnosed neurodevelopmental conditions, as sufficient (i.e., as discussed in *5.1.1 Change to Research Design*, where I explored my rationale for not including other groups amongst the researcher participants, such as parents of children with Down Syndrome or healthcare professionals).

5.6.3 Transparency and Coherence

Coherence is achieved through the application of the research methodology, IPA, to gain understanding into the participants' lifeworlds. I have attempted to be transparent in detailing how I completed the analysis, including a sample of the clustering of experiential statements in identifying personal experiential themes for one participant, namely Olivia (see Appendix G). Including individual story summaries (*see 6.4 Participant Story Summaries*) was also included in effort to fulfil this quality marker. I have included any identified limitations of the research in Chapter 15, Section *15.6 Study Limitations and Direction for Further Research*.

5.6.4 Impact and Importance

The rationale for the current research, as outlined in *1.4 Novel Accounts*, outlines the impact and importance of the study in that the population included are part of a marginalised group. Little in-depth qualitative research exists on the experience of mothers of children with rare or undiagnosed conditions in Ireland. I have outlined what importance the research findings may hold in *1.2.2 Sub Questions*. The importance of the research will be discussed more fully in Chapters 14 and 15 as part of the discussion.

5.6.5 Constructing a Compelling, Unfolding Narrative

Nizza et al. (2021) describe constructing a compelling, unfolding narrative, involves carefully chosen extracts from the data to present the narrative of the overall findings. I have attempted to support the reader in accessing each of the mother's stories, through producing individual participant story summaries, as represented in *6.2 Participant Story Summaries.* I have also aimed to fulfil this validity marker through careful organisation of the data within each GET, including extracts from a variety of mothers with my interpretation on the mothers' meaning-making, relative to the GET at hand. As advised by Smith et al. (2022), I include the mother and child's pseudonym for all extracts to support the reader in being able to "follow the story of each individual through the analysis" (p. 113). The inclusion of transcript line numbers may also help orient the reader to at what point in the account discussions occurred.

5.6.6 Developing a Vigorous Experiential and/or Existential Account

This quality marker is concerned with showcasing what is important to the participants. To assist me in understanding what was significant for the participant, I considered the frequency with which they referred to certain events or interactions and attended to linguistic and conceptual comments in the data. In my commentary, I have also made explicit reference to why I have interpreted certain moments as significant for the participant.

5.6.7 Close Analytic Reading of Participants' Words

This criterion refers to the researcher's interaction with, and interpretation, of the participants' words. I have attempted to achieve this by considering the participants' words in individual parts whilst maintaining perspective as to how these fit within the context of the whole of participant's whole account (i.e., attending to the 'hermeneutic circle', Heidegger, 1927/2008). I have included line numbers with the extracts so that the readers can re-contextualise quotes and extracts within the context of the surrounding dialogue to support the transparency of my interpretations. I have also included detailed explanation for why I reached certain interpretations (i.e., based on evidence within the transcript).

5.6.8 Attending to Convergence and Divergence

This validity marker involves retaining idiographic focus on the participants whilst highlighting similarities and difference across their accounts. I considered presenting five single-case studies followed by a briefer overview of group experiential themes. I found precedence for this type of presentation, in Riggs and Coyle's (2002) study of young people's accounts of homelessness. Riggs and Coyle (2002) presented the analysis by identifying personal experiential themes for each of the four individual participants, in turn, followed by a conclusion about the group of cases. However, based on advice from Smith (personal communication, 19 March, 2022) that "Constructing GETS is not just a matter of accumulating all the PETS you have for each participant. Another phase of interpretative analysis occurs here which involves selection, synthesis, transformation, distillation", I considered presenting only a brief cross-case analysis would less successfully engage in IPA analysis. In IPA, cross-case analysis is concerned with identifying shared and individual features across accounts. Given, IPA's inherent commitment to maintain an idiographic focus, I concluded that presenting the mothers' accounts as individual case studies was not needed to preserve the individuality of their stories. Within my analysis, I have also signposted which themes or ideas are most evident within a named participant account. I have also explicitly highlighted where one mother's perspective is similar or different from another's.

5.7 Analytic Interpretation

5.7.1 Multiple Meanings and Oscillating Horizons

Spence (1984) makes a distinction between 'narrative' and 'historical' truth. Narrative truth involves a constructed account of experience, not a factual record of what "really" happened. The question of factual accuracy is not the focus of IPA or narrative analysis (Frank, 2010; Josselson & Hammack, 2022), which are concerned with hermeneutics and meaning. Understanding this belief is foundational to my research purpose and analysis. Validity and truth lie in the memory and interpretation of experience for the individual. Validity is not being compared against an exact

historical recording of events. This belief can best be understood by Heidegger's 'minimal hermeneutic realism', as termed by Dreyfus (1995), that reality exists is an interpretation which is in opposition to the position of objective reality. Smith (2011, p. 38) similarly maintains "in this area of social science there can never be a one-and-only- true account or perspective". From a sociological narrative perspective, Frank (2010) concurs "no one's meaning is final, and no one meaning is final" (p. 99). The first half of this statement can be interpreted to offer an extension on this idea, in that participants' interpretations of their experience are also not absolute. This shifting vantage point can be exemplified through a written update received from Olivia (see Section 5.3.1). A change in the narrative 'sense' underlying Olivia's data, pertaining to each time point, can be seen in attending to Table 14.1 *Overall Narrative 'sense' Felt Within each of the Mothers' Accounts.* Similarly, as Clark et al. (2020) found that 'rescuing hope' was a way which enabled families to adopt more productive narratives towards making sense of the diagnosis of Down syndrome for their child.

5.7.2 More than Five Voices

According to Frank (2010), stories incorporate multiple points of view, that is their polyphony. This terminology and concept from Narrative Analysis, which is relevant to considering my data. There are many other voices woven throughout the participant's narratives, included through recounts of dialogues with recollected reported speech, perceived thoughts and looks of others, overheard interactions and supposed interpretations of other's experiences. Through the filter of the mothers, we also gain entrance into the thoughts, feelings and actions of healthcare professionals, grandparents, siblings, colleagues, onlookers, thereby hearing from the voices of many others. This polyphony alone signifies lives are not lived in isolation but influenced and constructed in the context of society and the communities in which a person lives.

5.8 Summary

This chapter has provided a summary of the practical considerations regarding how the research was carried out including the research design, participant recruitment, data collection, ethical considerations, procedure for analysis, and attending to reliability and validity. I also presented reflections on the careful considerations I made during the research process in attending to sensitive issues. References to how the methods align with the methodological underpinnings of both IPA and NA are also included within this chapter. In the following chapter I will present and introduction to the findings and analysis.

Chapter 6: Introduction to Findings and Analysis

6.1 Introduction

In *Chapter 6 Introduction to Findings and Analysis*, I have included individual 'story summaries' for each participant, following by an overview of the general experiential themes (GETs) with sub-themes. I also present the participants' entry points into their narratives within the context of my opening comment at the outset of each interview which invites each participant to begin their narrative. A detailed analysis of each GET in turn will follow in Chapters 7-12. In the context of this thesis, I will use the terms 'account' and 'narrative' synonymously to refer to the participant's data in the semi-structured interview, as recorded in the lines attributed to the participant within their individual transcripts.

6.2 Participant Story Summaries

Within the text boxes below, I have summarised elements of each participant's account to present five individual stories which aim to represent each participant's story. I am taking 'story' to mean the predominant elements of the participant's journey in response to the research question being posed. I interpreted topics, events, or experiences to be significant "markers"[15] within each participant's journey if they were explicitly named by the participant as such, or as interpreted by me as significant through attending to the frequency of that topic recurring within the accounts or by the wording used (i.e., way in which it was spoken about including choice of wording and paralinguistic features). I have chosen to include these summaries to preserve individual *people* journeys, and to assist the reader in getting to know the *mothers*, as their story elements become interwoven within the subsequent GETs as a product of in-depth cross-case analysis[16].

In staying close to the data, as required with an IPA approach, I chose to name the participant summaries as each of the mother's stories (i.e., Robyn's story; Mary's story etc.) based on insights from Claire who stated in the summary of her interview "my story, you know, does come back to…"

---

[15] Weiss (1994) defined 'marker', within the context of qualitative interviewing as "a passing reference made [in a field interview] by a respondent to an important event or feeling state" (p.77) which the interviewer should note to return to for further exploration if not volunteered by the interviewee.

[16] Cross-case analysis is considered an intrinsic step in the IPA process (Smith, Flowers & Larkin, 2021; Smith & Nizza, 2022) and pertains to Nizza et. al's (2021) fourth quality marker of IPA research "attending to convergence and divergence" (p.3). In presenting individual story summaries, I also aim to attend to the idiographic commitment of IPA (Smith, 2021) and quality markers of "transparency and coherence" (p.222) for qualitative research (Yardley, 2000).

(lines 1158-1159). The participant's use of the phrase "my story" called me to explicitly question 'whose story, is it?'. Examination of Claire's use of the phrase helped me to interpret that each of these stories can be attributed to the mothers, above the child, as it is the experience *'as told by'* each of the mothers, and how that experience was lived by them. This naming aligns with the distinction made by Knepper and Arrington (2016) in their analysis of messages from an online support group for parents of children with persistent hyperplastic primary vitreous, a rare vision disorder. They introduce the idea of "witness narratives" (Knepper & Arrington, 2018, p. 326) as distinct from existing illness narrative typologies, in that the parent's experience represents a caregiver illness narrative typology and is not based on first-hand experience of illness, in comparison to illness narrative in the more traditional sense. My research explores mothers' experiences of diagnosis for their children with neurodevelopmental conditions which can be more astutely considered to be a caregiver illness narrative.

6.2.1 Robyn's Story

Robyn is mother to Nathaniel, seven years of age at the time of interview. Robyn received prenatal knowledge at the 20 week-scan that Nathaniel had a brain malformation. Nathaniel later received a genetic diagnosis of a rare disease at around one year old. Robyn's story includes positive reflections on the benefits of the early intervention service and advice to other parents to engage with what is offered and to embrace thinking that one-to-one intervention in not the "silver bullet" (line 657). In relation to diagnosis, Robyn discusses considering "what's the actual purpose for it" (line 441) at a particular moment. Robyn asks questions which allude to the potential longevity of diagnosis, in considering what impact public knowledge of her son's rare diagnosis could have on Nathaniel in the future, in the context of living in a small town. She wonders if people know about her son's diagnosis, would it stay with him the rest of his life and damage him if he's doing great and people are "still like he's that kid with that weird diagnosis" (line 560-561). Robyn expands on this point of view when discussing considerations for testing for autism. She identifies that this is not something they need now but if when her son is older and "if he stopped improving then maybe that diagnosis is for *him* and maybe it gives him clarity with regards to maybe why he's a little bit weird" (lines 418-419), maybe then an autism diagnosis might be needed.

Robyn's story offers insights into perceptions on the value of diagnosis in the cultural context. Robyn references others are "freaking obsessed" (line 770) with diagnosis and "diagnosis-centric" (line 458) and that this is to access services. Robyn views the fact that diagnosis is the most important thing in getting services as "really crap" (line 714). Robyn talks about the challenges associated with a rare genetic condition in terms of accessing support groups, lack of understanding in education and when/how to "embrace it when the kids are older" (line 729).

Robyn talks about the impact of living in a small town in relation to concerns with a rare diagnostic label. Robyn discusses antenatal testing and heritability in terms of informing siblings, which she concludes "we'll cross that bridge when we come to it" (line 547). Robyn's story includes an awareness and explicit recognition that her experience is different to a lot of other parents and repeated expressions of gratitude, feeling "lucky" (line 78, 245, 314, 359, 667). Robyn finds fortune in that her child is progressing well, they had early access to services and a positive experience of early services and supports within their family network.

6.2.2 Claire's Story

Claire is mother to Rose, three years of age at the time of interview. Claire describes her own story around having or not having a diagnosis as about "the whole 'and there was something' or 'getting the diagnosis'" (line 1159), "getting a diagnosis and getting a label" (line 1163) and then "how do you tell people" (line 1164). Rose was initially diagnosed with one named rare disease, which I will refer to as RDD X, it was later confirmed that Rose does not have RDD X, she has RDD Y. On giving birth to Rose, Claire recalls she was told "you have a perfect baby girl" (line 41). Claire describes how everything was fine and Rose "was doing all the things that a baby should do" (lines 49-50) however there was something "niggling" (line 52) at her. Claire describes an all-consuming anxiety around there was something about Rose and the change to her identity as a new mother. For Claire, "getting an answer mattered. Getting a, getting a reason, *mattered"* (lines 574-575).

Claire provides a day-by-day account of one significant week in the hospital. Claire describes, in explicit detail, the events of one week that is "burned" (line 100) into her memory when Rose was five months old, beginning with the moment Claire identified Rose was having a seizure. Claire describes some "relief" (line 784), for want of a better word, in this discovery in that she had somewhere to channel her concerns. She includes anecdotes demonstrating becoming institutionalised living in hospital such as "you create these narratives about people and like 'you selfish bastard, what are you doing spraying your hair' (laughing) like get out, I need to get in the shower (laughing)" (lines 394-396) and recalls interactions and dialogues with healthcare professionals.

As a health care professional herself, Claire discusses the experience of "switching out of mothery mode" (line 782) into healthcare professional mode and dissolving back into parent mode. Claire's story includes a description of how she navigated the diagnostic disclosure to her husband. Claire describes their search for answers and references the main reason for wanting a diagnosis as to access a community group of other parents. Claire explores the challenges in "how do you tell

people" (line 1164) in terms of being aware of how the parents talk about Rose will make others feel and project how the parents view their own daughter. Claire discusses factors which influence the conversation around Rose such as the other person's experience and the context of old or familiar colleagues at work. Throughout Claire's story, she expresses strong rejection of 'pity' from others, "there's no poor Rose here, cut out that shit, there's no poor Rose" (lines 1091-1092) and a conscious representation of Rose as a person, "all the wonderful personality traits that she has and interests" (line 524).

6.2.3 Mary's Story

Mary is mother to Alexander, seven years of age at the time of interview. Mary has been searching for and would love a diagnosis for her son. Mary believes a diagnosis could help give Alexander the best care and quality of life and help other people to understand Alexander. Mary describes the birth of her son Alexander as "very traumatic for [her]" (line 33) and the opposite of what she had wanted to happen and a scenario which she had not been prepared for. Mary had a normal pregnancy and expectations for a natural birth, with no indication that anything was wrong. Upon labour, Mary describes she was rushed to theatre for an emergency section and when Alexander was born, he was taken away from her. She recalls lying on the operating table exclaiming "'where's my baby, what's wrong, what's happening'" (line 62). Mary talks about her fears of her son dying, symptom monitoring and fear about the future that accompanies having no diagnosis for her son. There is a sense of 'impending doom' and fear that transcends Mary's story in the emergence of a new symptom, wondering "is this ultimately going to be the thing that takes him away?" (line 863).

Mary's story also includes many recollections of conversations with healthcare professionals and Alexander's experience in interactions with healthcare professionals and undergoing medical procedures. Through examples of professionals she has encountered, Mary identifies qualities of a good doctor, including someone who "just takes the time to be kind" (line 1282), "spoke to us just about the facts" (line 361), "a man that you know cares about this child" (line 375), "doctors that do listen" (line 594), and "believe me" (line 637). She explains because you can trust these doctors, and "take him seriously" (line 637). Mary identifies that the doctor doesn't need to know everything and that she would prefer honesty, saying "a doctor that says I don't know if you don't know because then at least, you know, you trust these doctors more" (lines 601-602). Mary contrasts this with examples of what was not helpful, "he just didn't know how to, play with him" (line 1293), "harshness of the other doctor's delivery if things" (line 353), "talks around an issue

like a politician" (line 600), "don't look at me as [if] I'm a crazy person" (618) which leads to maternal doubt and questioning.

Mary discusses her own journey in learning to advocate for her son and how she sees her role in supporting his management and obtaining best care. Mary's sister "nearly died" (line 386). Mary attributes her past experience of her sister's illness as likely influential on her experience of trauma and adopted role relating to Alexander's health. Mary provides insight into her experience of disability and reactions in the community. She talks about how family and other people don't understand and in relation to Alexander's invisible disability "some people just kind of look at you, like you know, like oh my god what's wrong with that child and they have no concept of em of his disability at all because he looks perfectly normal" (line 1069-1073). Mary also references that her family did not live nearby, there was no history of special needs in the family, and how these things made the experience very isolating and lonely.

6.2.4 Judy's Story

Judy is the mother to Declan, seven years of age at the time of interview. Declan was born full term as a "perfectly healthy" (line 11) baby. At six months he got his first chest infection, which set the trend of being regularly in and out of the GP for the first three to four years of his life. Judy describes her priority concern was Declan's speech difficulties. Judy praises the special language class Declan attended which she views as an "*absolute,* like salvation" (line 543) for supporting the progress Declan has made which she presents as a silver-lining to receiving a late diagnosis. Judy attributes the chair of the support group for her son's rare diagnosis as an "absolute angel" (lines 251-252) for the information and reassurance she provided. These reflections suggested to me a sense of spirituality and gratitude to Judy's' account.

In relation to diagnosis, Judy describes how she was 'blind-sided' when her son received the named rare disease diagnosis at around six years of age and described the diagnosis as a "*complete* left-of fielder" (lines 480-481). Judy describes issues around preparing parents for the moment the diagnosis was delivered, how the diagnosis was given and the lack of follow-up care. At the end of the initial diagnosis meeting, Judy recalls being given a "a very badly photocopied piece of paper" (line 485), "here's a badly photocopied piece of paper about your child, go home and read it and come back to us in six months?" (line 1103-1104). Judy discusses issues in healthcare and the lack of transparency, including not being told the purpose of blood tests, appointments, and not being given notice before appointments. Judy expresses frustrations that nobody linked anything together sooner and the delays in genetic testing. Judy provides advice for healthcare

professionals that there should be 'an actual protocol' to follow if you get a rare disease diagnosis. Judy talks about lack of knowledge amongst professionals regarding her son's rare diagnosis.

Judy discusses her own role within the family before Declan's diagnosis and her position at the time. Judy describes she was not searching for a diagnosis as such, "I felt I was firefighting you know, my husband's kind of health issues, my son's health issues, and I was kind of working full time to keep the whole on the road" (lines 903-906). Speaking in retrospect, Judy exclaims "it's great in hindsight to be able to say this was linking it altogether" in contrast with at the time "you just plod by day by day, .h, hour by hour probably sometimes and you know, the day gone, you know that chest infection clears up am, and you just roll onto the next one" (lines 909-911). Judy comes back to evaluations on healthcare practice here saying "That's the way it was. Am but nobody ever stopped and said, this child has so much going on, why don't we send him for paediatric (.) review?" (lines 911-913). Judy speaks about lessons learned through her experiences within healthcare systems, how she has learned to be that parent with the "little notebook" (line 797) to get information on her child's medical care. Judy expresses strong dismay at the inclusion criteria for language class, that Declan would not have been able to access this class if he had received his genetic diagnosis earlier.

Judy talks about the negative impact the Covid-19 pandemic had on medical appointments, school closures and loss of speech and language therapy services within language class. Judy's brother has special needs. In relation to her son Declan's siblings, Judy speaks about some of the perceived challenges for them and what she considers to be their role. Judy also describes how she explains Declan's diagnosis to his siblings as well as to their family and others in the community.

6.2.5 Olivia's Story

Olivia is mother to Anna, three years of age at the time of interview. Olivia expresses strong abhorrence for public health services describing "the public sector was so poor and so shite and it lets you down at *every single turn*" (line 442). Olivia questions who to trust, describes conflicting findings and recommendations from healthcare professionals. Anna had her first seizure at seven months old. Anna had some eye bulging and reports of choking on saliva when she was younger which Olivia describes she hadn't made a connection between those earlier medical things and the seizures. Olivia described being given contradicting advice from medical professionals regarding the relevance of the EEG results (i.e., an electroencephalogram which is a test that measures electrical activity in the brain). Olivia questions "who do you trust?" (lines 1098-1099), "who are you actually supposed to listen to?" (line 1101-1102). Olivia recalls conversations with healthcare

professionals and discusses how her concerns about seizures were being dismissed by medical professional in the hospital and GP service, as "she's just having seizures" (lines 39-40), "no, no, look she's just had the flu" (line 130). Olivia describes this in contrast to her concerns and that she was "*freaking* out" (line 141). Olivia relates her concerns regarding Anna's development, and how her insistence that something was wrong was completely dismissed, which lead Olivia to question herself "I' must be going crazy" (line 254).

Anna has a "just has, syndrome without a name and unknown disability from the assessment of need process". Olivia describes how she would love a diagnosis to allow her to find that group of other parents who could relate to similar experiences, to access information, supporting Anna, and access to education. Olivia describes her hopes for an autism diagnosis because then she would be able to find out more information on it, ways to support her, access to education, and support groups for her and Anna's brother. Olivia describes within the context of her family, a diagnosis of autism would not be a big deal, "sure we're a family of oddballs, wouldn't be that big of a deal" (lines 112-113). There is a family history of epilepsy, intellectual disability, and autism. Olivia describes feeling that healthcare professionals expect parents to figure it out and her fight to get an appointment for early intervention services. Olivia disagrees with "negativity around [diagnosis]" (line 670) and expresses frustration in professionals' 'gatekeeping' diagnosis for fear of 'labelling', "<the labelling thing drive me fucking spare>" (lines 1071-1072). Olivia discusses how she feels withholding diagnosis causes more suffering and denies access to appropriate services for the child and family, "stop framing it as negative, like, whatever the diagnosis is, like by gatekeeping it (.) you're not just harming the child and their prospects of accessing education, of accessing supports within their community but you're also affecting the whole family because they can't access the support they need because they don't know where to go" (lines 1111-1115).

Olivia references "the lack of sleep and challenging behaviours" (examples given include headbutting, biting as the "biggest difficulty", line 454, and lack of support from public services. Olivia discusses the impacts on the challenges on Anna's brother and the whole family in day-to-day situations.

6.2.6 Summary

Now that I have presented each mother's story, I will move to introduce the themes that I have identified as existing *across* the mothers' accounts (i.e., GETs). As outlined by Smith et al. (2022), each GET serves to showcase the divergence and convergence of the participants' experiences and perceptions pertaining to a common theme. I will maintain an idiographic lens, as

is inherent within IPA methods and methodology, to preserve the individuality of the mothers' reflections whilst attending to these overall shared entities (i.e., GETs).

6.3 Overview of General Experiential Themes (GETs)

I have conceptualised the GETs which are representative of the journey of the participants in relation to the phenomenon being studied, as understandable a series of 'worlds'. The themes and subthemes have been presented in the table below and illustrated within the Figure 6.1. Each theme will be discussed in detail in its own subsequent chapter (*Chapters 7 through 12).*

Table 6.1

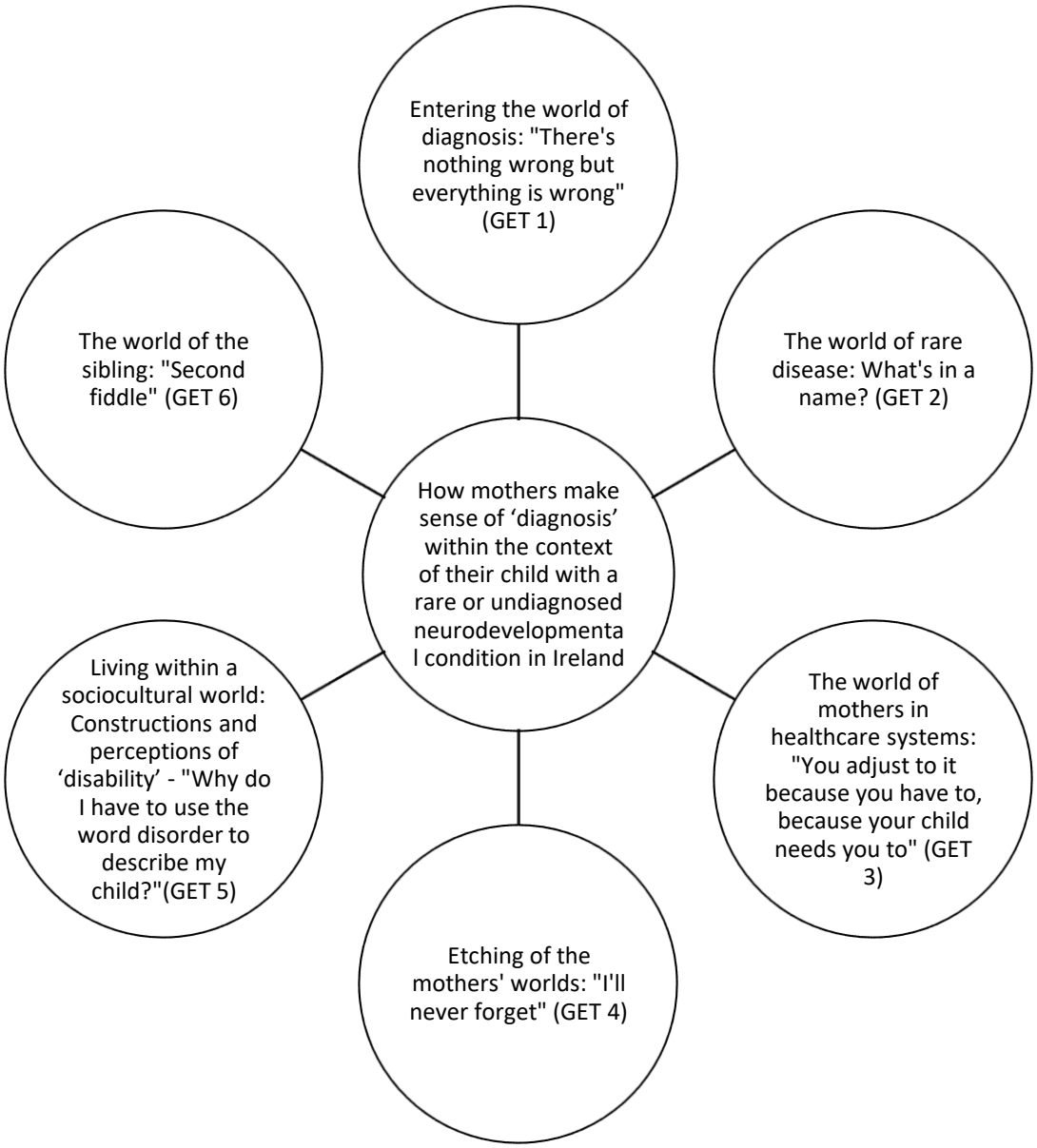
Overview of GET with Subthemes

General Experiential Themes (GETs)	Subthemes
Entering the world of diagnosis: "There's nothing wrong but everything is wrong"	*Searching for an answer or "firefighting" moment by moment?* *Dismissal and maternal questioning: Being "pooh-poohed" - "I must be going crazy"* *A fragmented "jigsaw" puzzle: Lack of transparency and co-ordinated care*
The world of rare disease: What's in a name?	*Rare disease: "A series of letters and numbers that doesn't mean anything to anyone"* *Not fitting in with the "ASD Moms… or Down syndrome Moms"* *Learning to live: "We just need to live our life now"*
The world of mothers in healthcare systems: "You adjust to it because you have to, because your child needs you to"	*Maternal role and responsibility: Balancing trust and onus* *Finding your voice, lessons learned.*
Etching of the mothers' inner worlds: "I'll never forget"	*Looks that speak volumes and words I'll never forget* *Chronology: Moments remembered in timelines*
Living within a sociocultural world: Constructions and perceptions of 'disability' – "Why do I have to use the word disorder to describe my child?"	*Labelling* *How do you tell people?* *Onlookers: "To me he's a beautiful child inside and out, no matter, disability or not, you know but other people don't understand it"*

The world of the sibling: "Second fiddle"	*Experience of the sibling: 'Sibling voice'* *Role of the sibling* *Telling the sibling about their brother or sister's diagnosis or difference*

Figure 6.1

Graphical Depiction of General Experiential Themes in response to the Research Question 'How is the phenomenon of 'diagnosis' experienced in Ireland today by mothers within the context of their child with a rare or undiagnosed neurodevelopmental condition in Ireland?'



6.4 Entry to the Mothers' Accounts

In this section, I will present the opening exchange of each participant's interview that ensued further to my initial remark, which invited the participant to begin their narrative. I have chosen to highlight this 'entry point' to allow consideration for any potential shaping of my interview question on the subsequent accounts provided by the participants. This conscious and explicit reflection is in keeping with the recognition of the role of the researcher across all stages of the research process within IPA and attending to quality markers for IPA and qualitative research, (notably '*sensitivity to context')* as previously discussed in *4.4. Researcher Positionality: Influence of the Researcher,* and *5.8 Reliability and Validity*, respectively. In the table below, 'R' is used to refer to researcher and 'P' for participant. Please refer to Appendix F for Table B which describes the meaning of any transcription symbols used.

6.4.1 Entry Point for Robyn, Mother to Nathaniel:

R ***So it was really just today, like I was saying to hear about your experiences so even if you want to start and tell me a bit about^***

P Cool

R *^your child [or your family]*

P [so day one] we'll go (laughing)

R *Okay, day one, start at the very beginning (laughing)*

P (laughing) so, em, I suppose, we were a little bit different to most in that, um, I found out at my, my 20 week scan that there was an issue so the first issue was that Nathaniel had [brain malformation] so one of the ventricles in his brain was enlarged so that kind of brought us down the route of (.) more scans, and more observations and everything like that. Em, we did have an amnio-he's my third child

Robyn seems to identify the beginning of their story in pregnancy with in-utero detection of a medication symptom. Robyn continues to describe her child's birth and ensuing days, followed by referrals and healthcare professionals including services and further genetic testing.

6.4.2 Entry Point for Claire, Mother to Rose:

R ***Yeah, so do you want to tell me a bit about eh your daughter then?***

P Yeah, em, so Rose is eh three, she's three since January and em: Rose is (..) Rose is (closed eyes) the most wonderful girl in the world (laugh)

R *Aw*

P So, em

R *[xxx]*

P Pardon?

R Sad I don't get to meet her now

P Yeah, actually, you may, we might because she's here

R Ok (laugh)

P So, she:, eh, Rose is three, she loves music, she loves Reggae music in particular, em, which we discovered, eh she loves very, she's a real foodie for a child who's tube fed, she's a really foodie, em so what she does eat, it has to be tasty and she likes her kinda em, yeah curries and Bolognese and things like that, em she loves trampoline and swing and em, she likes, she's got kind of a few of her favourite toys, she goes to a special preschool, which she also loves, em and she gets em you know, a lot of her therapies in school, physio, SLT, OT and all the rest and loves her school and she also has, em outside of that then, we have em nurses and carers that come in and help her as well and she seems to love all them and she has loads of cousins and em, yeah and a little brother, of course, she's a little brother, em and eh

Claire begins with a description of Rose's interests, education settings, therapies, and family network. In the subsequent lines, she describes this description as "how we see her in our day-to-day life" (line 29) which she distinguishes from "her medical side of things" (line 30), which begins with her birth. Considering the above segment within the context of Claire's whole account may help us to understand Claire's thinking on why she begins with this description and on her views of disability. Within her account, Claire expresses views on how she wants Rose to be viewed and her discomfort and apprehensions in doing Rose a disservice if she describes Rose in terms of what she *cannot* do. Claire later references the description she provided in lines 522 "I wanted to give the kind of description I gave you at the start".

6.4.3 Entry Point for Mary, Mother to Alexander:

R Like I was saying it's really to hear from you, so I'll be talking a more listener role and just hearing abou- so if you want you start even just talking about Alexander

P Yeah, em, (laughing), where do I start? How much history do you want, do you want from birth or (laughing) do you want like current^

R ^Wherever's kind of relevant to you that you feel plays into factor on your day to day and your experience of diagnosis for Alexander

P Well maybe, I'll just give you a brief overview around his birth and kind of what happened there because that is where it all started and you know

R Where it began

P Yeah, yeah. So I had a very normal pregnancy, em (.) very wanted little boy

Mary's response to the researcher's invitation to begin her account, appears to reflect the dyadic nature of the semi-structured interview and the influence of context on the information shared given the participant is demonstrating engaging in some level of selection in terms based on what she interprets the researcher would like to know. This opening dialogue brings into more explicit focus where is the start of the experience for Mary, which she consciously identifies as her expectations during pregnancy and her experiences at birth.

6.4.4 Entry Point for Judy, Mother to Declan:

R ***So yeah, really, I don't know if you know but I'm a speech and language therapist during the day, normally, but em, today I'm just going to be here to listen and to learn about your personal experience, em (.) around (.) you know, your search for a diagnosis for your little boy***

P Yeah, yeah, for Declan

R For Declan, yeah

P Yeah, perfect

R Lovely. So if you want to tell me kind of where the journey begins for you

P Yeah, em, ok, I suppose like Declan was born at what we would say full term, you know, perfectly healthy, good healthy weight (.) you know, didn't- wasn't sick at birth or anything like that and actually he was almost bang on six months when he got his first (.) chest infection, so (.), .hh you know, little did we know, when I look back now that was the start of a very, very long road, em so Declan was a June baby so he's just had his 7th birthday actually

For Judy, I 'honed' the research question more specifically to include reference to 'diagnosis', similar to the follow-up clarification I provided to Mary, further to her request for clarification, as was illustrated above. Similar to Mary, Judy attributes the beginning of her experiences on diagnosis with Declan's birth.

6.4.5 Entry Point for Oliva, Mother to Anna:

R ***Ok, so yeah, em I think I said to you on the phone call before that I am a speech and language therapist but I'm doing this kind of from- as a researcher, and want to hear from you about your experiences, em and then the ultimate aim is to help others understand what it's like, you know, in your experience around diagnosis and to help inform services. So if you want to start off wherever, you know wherever is the beginning for you***

P (facial grimace) ok, so, em, so em, when Anna was like seven months she had her first seizure, she was taken by ambulance into the hospital and they thought it was like a febrile

> convulsion because it turned out she had a temperature that only arrived after the seizure, so they were like, ah that's a febrile convulsion, that's fairly normal, and they were actually about to send us home and she started having more so they admitted her and em (.) she was in there then for a week (throat clear) and they said she had flu so they were still kind of like these are unusual for febrile convulsions because they're not really stopping when her fever is down and they never actually did stop after that so em the, they actually did em, we have a hu:ge family history, em

Olivia begins her narrative at the point when something was first medically noticed and the events regarding that initial hospitalisation.

6.4.6 Concluding Remarks

The order of the segments presented above are aligned with the order in which the interviews were conducted. As evidenced above, my opening question initially asks to hear about the child, and family on one occasion for Robyn, and becomes more specific to include reference to the experience of 'diagnosis', further to Mary's clarification request on where to begin. Most participants (Robyn, Mary, Judy) start by describing the events around pregnancy and birth. In contrast, Olivia begins the story at the point of where something was first noticed. This mostly relates to theme of '*Entering the world of diagnosis: "There's nothing wrong but everything is wrong"* in terms of expectations and comparison to 'normality' at birth versus foreshadowing[17] of what we know as researcher and reader to expect deviance from this 'normal' trajectory at some point in the narrative given the phenomenon being studied. Claire is the only participant who begins with a person-centric description of Rose as opposed to information surrounding pregnancy or birth or first medical symptom.

6.1 Researcher Reflection

During the time of writing up, I wondered whether I should have asked a consistent opening question to all participants to reduce the variability in data collection methods. However, on reflection I feel that flexibility is something which is acknowledged and appreciated within IPA (Smith et al., 2022). Having an undefined opening question allowed for more natural communication and I don't think would have changed the participant accounts provided or

[17] An ancient literary device common in Greek tragedy, which creates a sense of 'dramatic irony', whereby the audience know something ahead of the characters (Johnson, 1928; Muecke, 1983), as referenced in *2.2. Concept and Construction of 'diagnosis': What is 'diagnosis'?* Chatman (1978) refers to 'foreshadowing' as the two timelines in fictional writing and film, one which is being lived by the characters in the story and one which is available to the audience.

resulting analysis. All participants were also primed before the interview to know the researcher topic of interest based on the information provided in the Participant Information Leaflet.

6.5 Direction for Next Chapters

I will now move to a detailed description of each GET within Chapters 7-12. Quotations and extracts from the data have been included as part of fulfilling quality criteria for qualitative research (Nizza et al., 2021; Yardley 2000). I have included some longer extracts so that readers can resituate quotations referenced in the commentary (marked with line numbers) within the body of the wider dialogue, to honour IPA's commitment to the 'hermeneutic circle'[18] (Heidegger, 1927/2008). Providing such context attempts to fulfil Nizza et al.'s (2021) referenced quality marker of IPA ('*Close Analytic Reading of Participants' Words'*, see discussion in *5.6 Reliability and Validity*). In some instances, I have shortened some segments, where the content is not directly required to support the analysis being made, by inserting ellipsis markers '…' to indicate where lines have been removed. The line numbers in the left-hand column are also adjusted accordingly. This is to support ease of readability of the extracts. Finally, for most extracts I have added what I interpreted to be a key phrase (or phrases) to the extract title and bolded some identified key phrases *within* extracts, to enable easier identification and highlight relevant emphasis. However, this is not to exclude other phrases of potential importance in the extracts, which may be the focus of another researcher or reader's analysis[19]. This formatting is merely intended to assist the reader in attending to and retaining the participant's message in the extract under discussion. I have also maintained some of the markings from the transcript, in the extracts featured in the analysis in order to convey paralinguistic features within the participant's speech, where I interpreted these had relevance to understanding the participant's experience.

6.2 Researcher Reflection

Additionally, as a healthcare professional, I think retaining longer extracts pertaining to the mothers' detailed recollections of their internal dialogues and evaluations on their experience, contributes a 'palpable' quality to the accounts which permits greater entry into the mothers' lifeworlds. Such deep insights may better contribute to effecting change within healthcare communication and procedures. Suggestions for clinical practice will be detailed in Chapter 15.

[18] The 'hermeneutic circle' (Heidegger, 1927/2008), as previously defined, is an iterative process, which involves circular examination of the data moving from the parts to the parts within the whole, to understand the data.

[19] IPA as a methodology recognises the integral role of the researcher in interpreting the participant's sense-making of the phenomenon being studied (Smith & Nizza, 2021; Starr & Smith, 2011).

Chapter 7: Entering the World of Diagnosis: "There's nothing wrong but everything is wrong" (GET 1)

7.1 Introduction

The theme of *'Entering the world of diagnosis: "There's nothing wrong but everything is wrong"'* focuses particularly on the mothers' varied experiences, as one participant put it, before they entered into "the world of understanding what diagnosis meant" (Robyn, lines 791-792). This theme includes participant reflections looking back on their children's early years from a position of hindsight, with what they know at the time of the research interviews. There is a sense that the participants are narrators of their own stories. It seems they are using the literary device of 'foreshadowing' (see footnote 16), which creates a sense of dramatic irony for the readers, in that we anticipate certain elements are going to change, of which the mothers, who might be considered akin to 'protagonists' in their own story at the time of *living* the experiences being described, would have been unaware. The use of 'foreshadowing' is particularly prominent within this theme but is undoubtedly present within the style of the whole narratives as captured within other GETs.

'Entering the world of diagnosis: "There's nothing wrong but everything is wrong"' includes participants' expectations from pregnancy, experiences of giving birth, from when something was first noticed, and either confirmed in medical symptoms or reportedly felt by the participants as a sense that something was not right. This theme also includes responses from healthcare professionals to mothers' concerns and the impact of these responses on the mother, namely self-questioning and self-doubt. The idea of a fragmented 'jigsaw' puzzle comes directly from a metaphor[20] used by Olivia, in relation to the lack of co-ordinated care and consensus across professionals. This analogy is extended in Judy's experience, where she describes "nobody linked anything together" (line 43) regarding the disappointment with follow-up care to receiving a rare diagnosis.

[20] Neefjes (2022) offered an accessible definition of a metaphor as "the linguistic expression of a leap of thought that finds similarities between two dissimilar circumstances" (2022, p. 427). Further metaphors used by the mothers will be discussed, as they are identified, within the Findings chapters (Chapter 7-12). The evaluation of metaphors will be revisited within the Discussion chapters (see Chapter 14, Section *14.3 IPA as Permitting Entrance into the Participants' Lifeworlds).*

Table 7.1

Summary of Subthemes within GET 1

General Experiential Themes (GETs)	Subthemes
Entering the world of diagnosis: "There's nothing wrong but everything is wrong"	*Searching for an answer or "firefighting" moment by moment?*
	Dismissal and maternal questioning: Being "pooh-poohed" - "I must be going crazy"
	A fragmented "jigsaw" puzzle: Lack of transparency and co-ordinated care

7.2 Searching for an answer or "firefighting" moment by moment?

The subtheme '*Searching for an answer or "firefighting" moment by moment?*' deals with whether the participants were actively searching for a 'unifying'[21] diagnosis for their child and what projected value 'diagnosis' held for each of the mothers. Claire, Mary, and Olivia all spoke about actively searching for a diagnosis for their child (e.g., "getting an answer mattered", Claire, line 574, later Extract 7.12) and detail what importance they perceived in finding a diagnosis. In contrast, Judy talked about how she was responding to challenges for her son day-by-day (e.g., "you just plod day by day…that chest infection rolls up and you just roll onto the next one", lines 908-910, Extract 7.2).

'Searching or "firefighting" moment by moment?' also addresses the 'entry' point into most of the participants' narratives in response to my opening invitation to begin their account (as detailed in *6.4 Entry to the Mothers'* Accounts), representing the point at which their journey was perceived to begin each of the mothers. For example, Robyn had pre-natal knowledge of something not being right and received an early genetic diagnosis when Nathaniel was one year old. For Claire and Judy, Rose and Declan were described as 'perfect' ("'you have a perfect baby girl'", Claire, lines 40-41, Extract 7.9) and 'healthy' ("born at what we would say full term, perfectly healthy", Judy, lines 10-11, Extract 7.1) respectively, at birth. For Olivia, health concerns for Anna also did not become apparent until later in infancy. For Mary, she had expectations for a natural delivery and instead there was unexpected trauma at birth which was the start of her continued journey in hospitals and

[21] In this context, I am using 'unifying' to refer to an overall diagnosis which may account for a multitude of medical symptoms or other diagnoses (such as epilepsy, developmental co-ordination disorder), delayed or disturbed developmental milestones (i.e. 'global developmental delay'), behaviours of concern.

healthcare services with Alexander. Experiences of the child during early infancy and childhood are also presented within this subtheme.

Robyn is the only participant who was informed of something wrong during pregnancy. At the 20-week scan, she was informed Nathaniel, had a brain malformation. At one year old, Nathaniel was given a diagnosis of a specific rare syndrome. When asked if getting a name or a diagnosis was something that was important to Robyn, she stated "not at all (shaking head)" (line 786). Robyn reflects "he has this [brain malformation], okay, that's fine, we have this medical thing, we understand what that is, and we wouldn't really have, I suppose I just wasn't in the world of understanding what diagnosis meant" (lines 789-792). Robyn described her mindset as "that was my questioning rather than, em, than oh well can we get a diagnosis for something for this you know, it was really just from a medical perspective, what do we need to do" (lines 807-809).

Robyn's experience is similar to Judy's, in that she was not searching for a diagnosis. For Judy, the diagnosis of a rare genetic condition came as a shock, referenced as "a *complete* left-of-fielder" (line 481). The unexpected shock of a genetic diagnosis is in comparison to the experiences of Mary, Claire and Olivia whose accounts feature a sense a strong desire and elicitation for an underlying answer to explain their child's development and medical symptoms. Judy describes her experience of Declan's birth below:

Extract 7.1

Judy/ Declan: *"little did we know"*

10	P	Yeah, em, ok, I suppose like Declan was born at what we would say full
11		term, you know, perfectly healthy, good healthy weight (.) you know,
12		didn't- wasn't sick at birth or anything like that and actually he was almost
13		bang on six months when he got his first (.) chest infection, so (.), .hh you
14		know, **little did we know**, when I look back now that was the start of a
15		very, very long road, em so Declan was a June baby ....
~~16~~		~~7th birthday actually~~

In Extract 7.1 above, the phrase "little did we know" (line 14) appears to contribute to the effect of 'foreshadowing' (as referenced in *7.1. Introduction* above). The phrase "when I look back now" (line 14), is seen repeated in line 907 in the next extract, Extract 7.2, which may imply a level of in real-time sense-making and reflection taking place through the action of storytelling.

For Judy, a specific search for diagnosis was not part of her journey. Declan's early years were characterised by repeated illnesses (e.g., "really just in and out of the GP, chest infection after chest

infection" lines 30-31). Judy described the time she and her husband were due to have their first night away with a small baby and had to return as Declan was sick. She describes this as "the trend for the next, eh, what'll I say, kind of three four years probably, em, he was chronically in and out of the GP" (lines 26-28).

When asked directly if diagnosis was something Judy had been looking for, she described she was living "day by day to be honest with you, day by day you know, incident by incident" (lines 861-862) and that Declan's speech took over her priority of concerns. In Extract 7.2 below, Judy discussed how they were living, before diagnosis:

Extract 7.2

Judy / Declan: "I felt I was firefighting you know"

896	P	A:m but I think .hh yeah, so like, it's great in hindsight to be able to say this
897		was linking it altogether but in the time, in the manner of it, now there was
898		other stuff going on here at home as well probably in the background that
899		just made our life was a little bit more chaotic, am you know, my husband's
900		mental health wouldn't have been great and things at the time as well, am,
901	P	so we had a lot going on here as well, you know and **I felt I was**
902		**firefighting you know,** my husband's kind of health issues, my son's health
903		issues and I was kind of working full time to keep the whole show on the
904		road as well with the, you know, with the family and all that am
905	R	[xxx] tackling every day
906	P	When you look back, yeah, you know those kind of times when you look
907		back and you're like how did you do that, how did you get through that
908		but you just do, **you just plod day by day, .h, hour by hour** probably
909		sometimes and you know, the day gone, you know that **chest infection**
910		**clears up am, and you just roll onto the next one**. That's the way it was. Am

The metaphor which is included in the title of this subtheme (i.e., "firefighting") can be seen in line 902 in Extract 7.2 above. The phrase "day by day" (as seen earlier in lines 861-862, in the quotation included above) can be seen again in line 908 in Extract 7.2 above. The inclusion of two phrases with the same repeated structure (e.g., "day *by* day, hour *by* hour", line 908) may serve to echo and add a certain repetitive rhythm the cyclical manner in which Judy perceived her (and her family's) daily life.

7.1 Researcher Reflection

In the first half of the extract above, Judy gave further details to their family context at the time (lines 897-904). As highlighted already many times during this study, a person's experience is influenced by the

'context' in which they are living and is reflective of past and present prevailing cultures (i.e., 'Dasein'), and ongoing interactions between the individual and society. The implications for practice must be considered with recognition to context as a non-constant entity, for example, considering the onset and impact of Covid-19 on people's experiences as was referenced within the current study. The implication for healthcare professionals is to remain open to the potentiality of new levels of sense-making and changing narratives for the people whom we are working with, over the course of an intervention. This flexibility and adaptability are needed to best understand where families are in their journey, changing needs and priorities in order to identify appropriately placed supports.

This subtheme of *'searching or "firefighting" moment by moment?'* also includes a referenced lack of transparency between professionals and families (as will be attended more specifically in the third subtheme of this GET, *7.4 A fragmented "jigsaw" puzzle: lack of transparency and co-ordinated care*). In terms of the events preceding diagnosis, Judy outlined "I went up completely unprepared for this diagnosis" (lines 476-477) and "I wasn't aware that they were even testing for this [RDD]" (line 458). Judy, in describing her then-upcoming appointment with the consultant, explained she was not expecting to be informed of a unifying diagnosis.

In Extract 7.3 below, Judy described how she thought the meeting would be around the confirmation of the diagnosis proposed by the OT (i.e., "he'd had an OT assessment and the OT had said…I would diagnose him with [movement condition]…a medical person has to sign off on it…so I thought, this is what we were going up for", lines 111-116):

Extract 7.3

Judy / Declan: "so I landed up to this appointment...on my own"

98	P	the paediatric consultant for a sec- for six months' time again and we got
99		a call about, maybe about a week beforehand, you know, make sure we
100		were going up for the appointment and I said, yeah, I'll be up and at the
101		time I just assumed it was Covid related that they were checking that we
102		were going up, I don't know in hindsight, was it because of Covid or was
103		it because they were, anticipating giving us a diagnosis, I suspect the
104		later. Am, **so I landed up to this appointment**, with Declan, **on my own**, in
105		the *absolutely* horrible day back in March, it was probably the worst day
106		for travelling, I mean the roa- there was ferocious flooding around here,
107		…
110		school, get up to the appointment, am, absolutely awful weather, **on my**
111		**own**, into the appointment, now, he'd had an OT assessment and the OT
112		had said look, yeah, I suspect or I would diagnose him with [movement
113		condition], but she said the pae- you know the

doctor, a medical person has to sign off on it so she said because he was
under the care of the paediatrician, she just referred to the paediatrician
to sign off on it, so **I thought, this is what we were going up for**. A:m, he'd

In the Extract 7.3 above, Judy used the phrase "landed up to this appointment" (line 104). The choice of verb "landed" may be seen to reflect the sense of unpreparedness and abruptness in which Judy feels she presented for the appointment in hindsight.

As seen in Extract 7.4 below, discussion around being unprepared for diagnosis features readily and recurs throughout Judy's account (e.g., "so I went up *completely* unprepared", line 476, in Extract &.4 below), which appears to show the significance of this experience in relation to her whole journey to date, at the point of the research interview:

Extract 7.4

Judy / Declan: "it was a complete left-of-fielder"

P about those, **I wasn't *aware* that they were even testing for this [RDD].**
Like, they just told me they were, I don't know how they phrased it
to be honest Beth but the impression I came away - what I was able to
relay as a lay parent to my husband was, they just ran tests, like they were
just running bloods. Am, you know, I wasn't expe-now I, may be they
told me and maybe I signed the forms but he was seen in person, just before
…
R He was seen, yeah, sorry February 2020, yeah yeah yeah. So he was seen
February 2020, yeah (laugh) that's where the date is wrong, just before
Covid 19 hit and we didn't get a diagnosis until March '21
R Yeah
P So kinda like 12 months later. And like I wasn't told, so we-and we had one
teletherapy appointment in between and I wasn't told at that stage whether
there were bloods still outstanding, **I wasn't even aware that there were still**
bloods outstanding you see, so I went up *completely* unprepared (.)
R Yeah
P for this diagnosis. It it, I mean, and it did I think (.) if I *had* been prepared
for it and I knew they were looking for it, now of course me being me I
would have looked it up online (laughing), am, *but*(.) **it was a *complete***
left-of-fielder and (.) that in itself is traumatic, and (.) requires processing,

In Extract 7.4 above, Judy emphasises how she did not know there were testing for rare disease or that there were outstanding blood results (i.e., "I wasn't *aware* that they were testing for the [RDD],

line 459; “I wasn’t even aware that there were still bloods outstanding”, lines 475-476). Judy discusses the trauma that being unprepared for the diagnosis (i.e., *“but*, it was a *complete* left-of-fielder and, that in itself is traumatic, and, requires processing”, lines 480-481).

In the Extract 7.5 below, Judy described a perilous journey that ensured as a result of not being properly informed on what to expect from the appointment so that she could have brought somebody with her:

Extract 7.5

Judy / Declan: “how I got home I do not know”

187	P	Aw, completely, yeah, am, I don't remember getting the the hand-, coming
188		away to read at home, we did the bloods, again Declan was crying because
189		I'd promised him there'd be no bloods, I wasn't expecting bloods, and
190		again, I went up thinking like [XXX] and this [muscle weakness] and really it
191		wasn’t mentioned at all, am, during the appointment, so I came away, back
192		down through those awful floods, actually went through one flood that in
193		hindsight, did about 500 or 600 euros lots of damage to the car that I, I,
194		I, you know, if the car had stalled in that flood, I actually would have
195		written off the car in the flood. I was completely (.) not (.) with it, you know
196		in terms of like, just .hh, you know, **how I got home I do not know**

We may interpret further trauma for Declan, who was six at the time, in the experience (e.g., “Declan was crying because I’d promised him there’d be no bloods”, lines 188-189) and witnessing upset for his mother in car home (e.g., “I was completely, not, with it”, line 195). Use of “in hindsight” (line 192-193) appears to show Judy’s is evaluating the experience in her narrative re-telling of the events.

The following extract, Extract 7.6, provides Judy’s reflection on her thoughts during the moment of diagnosis:

Extract 7.6

Judy / Declan: “I was just kind of thinking”

149	P	And, so I mean, you know, .hh (.) she, she- I just remember Beth from that
150		appointment, am, I mean I was **on my own**, Declan at this stage had had
151		so many hospital appointments, he doesn't like hospitals, he *hates*
152		needles, doesn't like getting bloods done, so **I was on my own with him,**
153		**she told me she was just diagnosing him with this,** am, I kinda s-, I don't
154		even think I got the leaflet into my hand to be honest, am, she just, I,

you know, **I have two memories from it**, am, she mentioned heart defects and she mentioned scoliosis, now **she *may* have said other things** but they're my two recollections, am, you know and I'm there with my six-year-old, **on my own**, and I'm thinking, heart defects, cripes almighty, and he has a diagnosis of a cold induced urticaria and we would have seen an allergy specialist a number of years ago and you know I remember thinking, he told us at the time, kind of again a bit, almost, kinda, in a you know blasé, kinda funny way, em, it's it's just cold induced urticaria so kind of don't ever go from extreme temperature into the other, am, kinda don't ever let him jump off a cliff into the cold water, send him into cardiac arrest so I was like in my head when she mentioned cardiac, am, you know, review or cardiac issues, I was like, and heart defects, I was like oh that could be linked to that, so that was, my brain was, twigging away on that, I was pinging off my, you know in the back of my head, and then she mentioned scoliosis, and I was just, I I have a recollection you know of sitting there, Declan was pulling at me for something, he wanted to know something, and **I was distracted with him and I was just thinking in my head** scoliosis, I was like oh my god there's such long waiting lists for that, you know, they're the kind of kids you see in pain on you know, Claire Byrne Live, Primetime Special, you know about how bad our services are, and that's what was going through my head up in that appointment, em and I was just kinda thinking, oh my god like, am, am, so, you know, she did various (.) activities with Declan, she, I know she checked his spine on the day, am (.) she got him to do a few exercises on the floor and things like that, am, and she did say that myself and my husband would need to be tested as well, so I was sent for bloods and she was sending Declan for bloods as well that day, and again, sending Declan for bloods but when I came home I couldn't tell my husband what the bloods were for, you know am, so I was probably a bit, now in hindsight, **I think you know that kind of, you're there, you're responding, but you're not actually processing the information,** I was at that stage you know (laughing) am

In the Extract 7.6 above, the interjections and hesitations in Judy's speech may appear to echo the bafflement she felt at the time resurfacing as she actively recalled her thinking in the moment (e.g., "I was just kind of thinking" lines 176). Judy repeats "on my own" three times within this extract (lines 150, 152, 158) which seems to show the gravity for her in not being informed and prepared to bring another person for the diagnostic moment (as seen elsewhere in her account, such as "I think one thing that irks me is, obviously I was on my own at that diagnostic appointment in March, I think we should have been called in together", lines 390-392; "you should have somebody with

you, even if we couldn't be there together", line 298; "I shouldn't have been driving home on my own afterwards", lines 298-299). In line 156, Judy referenced "she *may* have said other things". Viewing the use of "may" together with Judy's earlier comment "maybe they told me and maybe I signed the forms" (lines 462-463) might suggest Judy is owning to the possibility that she was informed but could not *hear* the information in that moment due to the way the diagnosis telling was carried out.

Near the close of Judy's interview, when I prompted her to consider if there was anything further that she wanted to share, Judy advised of two procedures surrounding the 'diagnostic moment'. As shown in the Extract 7.7 below, firstly she references "the importance of following protocol, you know two parents should be there, or two people" (lines 1088-1089) and "a heads up that something big is coming down the line" (lines 1096-1097):

Extract 7.7

Judy / Declan: "a heads up that something big is coming"

1089	P	parents should be there, or **two people should be at that meeting** and things
1090		like that and Covid aside, and I *know* Covid changed things, but I mean .hh,
1091		especially for the diagnostic meeting and especially when it was coming left
1092		of field for us and we weren't expecting it, am, I really needed somebody
1093		else there, you know and somebody rang the week before and said are you
1094		coming to the appointment, even to be told look, have somebody in the car
1095		with you, you know the doctor needs to talk to you on your own, even if
1096		they'd only allowed one of us in and you know **a heads up that something**
1097		**big is coming** down the line, have somebody with you, that would have been
1098		important to me and I think you know, I did kind of say at the time I'll

Secondly, she referenced poor information (e.g., "badly photocopied piece of paper, line 1103) provided at the 'moment of diagnosis' and a lack of follow-up care (e.g., "that whole disconnect afterwards", line 1102-1103; "*huge* disconnect", lines 1105), as seen in Extract 7.8 below:

Extract 7.8

Judy / Declan: "here's a badly photocopied piece of paper"

1101	P	so you know, I think if they have protocols in place on paper, they're good
1102		protocols, they should to be followed and then that, **that whole disconnect**
1103		**afterwards**, I just felt really- **here's a badly photocopied piece of paper** about
1104		your child, go home and read it and come back to us in six months? .h (.)
1105		that's not good enough. You know, there's **a *huge* disconnect there**
1106		when you're given a rare diagnosis and something needs to be, you know,

1107 here's a contact number, here's a support group, you know am

Suggestions surrounding how parents should be supported throughout various stages of the diagnostic process, feature frequently throughout Judy's narrative and are re-iterated in her closing remarks, which may signify the importance of these experiences for Judy in relation to her experience of an 'unexpected' diagnosis for her son. In relation to this subtheme of *'Searching or "firefighting" moment by moment?'*, Judy's description of a diagnosis that came unexpectedly represents a very different experience to that of Mary, Olivia and Claire, who were actively searching for a diagnosis.

It is Claire's words that form part of the title of this GET "there's nothing wrong but everything is wrong but there's nothing wrong feeling" (lines 796-797). Within her account, she described an "all permeating" (line 796) anxiety in feeling something was wrong with Rose before any symptoms of medical significance were, or could be, identified by a healthcare professional. As seen in Extract 7.9 below, Claire recalled the words spoken by the Paediatrician on Rose's birth, "handed her back to me into my arms and used the sentence 'you have a perfect baby girl', that is still true (laugh), but that was where we, we started life" (lines 40-42). Claire's description of "that is still true" might be an early indicator within her narrative regarding how she views Rose as a person. Claire does not preclude the term "perfect" from being applicable to her daughter. Claire describes how she wants to present Rose to the world and at times in contrast to how family or others may view her (this desire will be discussed in more detail in the later firth GET *"Living within a sociocultural world: Constructions and perceptions of 'disability' - "Why do I have to use the word disorder to describe my child?"*).

A sense of foreboding or 'dramatic irony'[22] can be felt in Claire's account of Rose's birth ("you have a perfect baby girl'", line 40-42, as referenced in the paragraph above) and early development (e.g., Rose described as "fine", line 42; repeated as *"fine",* line 46; "nothing medical of any significance" (line 44); "doing all the things that a baby should do", lines 49-50), as evidenced in the extract below:

Extract 7.9

Claire / Rose: "you have a perfect baby girl"

40	P	And handed her back to me into my arms and used the sentence **"you**
41		**have a perfect baby girl",** that is still true (laugh), but that was where
42		we, we started life. Em, and eh the first eh couple of days, she was **fine**

[22] The effect of 'foreshadowing' as previously detailed in footnote 1.

43 but we had em, just challenges establishing breastfeeding but em
44 **nothing medical of any significance**, she was very sleepy for the first
45 twenty-four hours after she was born and they tested all her blood
46 sugars and everything but it was **fine** and we eventually got established
47 on breastfeeding and got home from hospital a couple of days later
48 and she came home and everything was great, so she was feeding, she
49 was growing, she was thriving, she was **doing all the things that a baby**
50 **should do,** in terms of wet and dirty nappies and blah blah blah. But I

In the Extract 7.9 above, we as the reader, get a sense that things are not likely to stay *fine*. In the following extract, Extract 7.10, Claire's described "there was something niggling" (lines 51-52) at her and begins self-questioning any influence of her as a new mother (which will be explored in the subsequent subtheme 7.3 *'Dismissal and maternal questioning: Being "pooh-poohed" - "I must be going crazy"'.*

Extract no.7.10

Claire / Rose: "there was something niggling"

51 P suppose kind of somewhere along the way, **there was something**
52 **niggling** definitely at me, em, so after the birth I, I just the combination
53 of em, I think probably something niggling about Rose but just the whole
54 new mother, big change of identity, all of a sudden, you know, all of that
55 just hit me like a ton of bricks, you know all the hormones, everything

In terms of the 'search' for Claire, initially it was about seeking verification that something was not right, as opposed to a diagnosis, *per se*. As evidenced in Extract 7.11 below, when Claire identified Rose as having a seizure, at around five to six months of age, the search appeared to the move to *'why?*'. Claire used the term "hounding" (line 363) to describe her interactions with doctors in soliciting a diagnosis, which may serve to convey the intensity for which she was seeking a diagnosis. Claire recalled:

Extract 7.11

Claire / Rose: "tell me anyway"

363 P just kind of given up on it. And I remember hounding the doctors saying,
364 if you are looking at my child and thinking "oh my god, classic", just a
365 classic (.) *bluh^*
366 R Mmm
367 P ^but we can't tell the parents that until we get the genes back, I said will
368 you **tell me anyway** and I understand that when the genes come back

you might have to retract that diagnosis but *is* there anything, are you looking at her thinking **something weird, wild and wonderful** that we don't know about^

Extract 7.11 above may signify Claire's wish to know any potential diagnosis for Rose, regardless of with what certainty the diagnosis could be confirmed. Similarly, Mary and Olivia appear to echo this preference for honesty and transparency, regardless of whether there can be certainty in the information. For Mary, she said "if they're honest with you and tell you, I really don't know what's happening here that's fine" (lines 616-617). Olivia expressed her views on this in relation to dismay for 'withholding' of diagnostic labels (as will be discussed later in the fifth GET*, 'Living within a sociocultural world: Constructions and perceptions of 'disability' - "Why do I have to use the word disorder to describe my child?"* within the subtheme of *'Labelling').*

Of note also in Extract 7.11, is Claire's reference to "weird, wild and wonderful" (line 370). She uses this whole phrase elsewhere in her account (as seen in lines 589-590, extract Extract 7.12). The alliteration in the phrase is striking and the semantic meaning of each of the adjectives chosen may be considered suggestive of Claire's views on rare disease and disability more broadly (views on disability will be considered further in the fifth GET '*Living within a sociocultural world: Constructions and perceptions of 'disability' – "Why do I have to use the word disorder to describe my child?"').*

Returning to considering on the importance of diagnosis for Claire, she described for her, initially, "getting an answer mattered. Getting a, getting a reason, mattered" (lines 574-575, see Extract 7.12). This statement shows that a label or a diagnosis was something of importance for Claire. Claire described that in their search for diagnosis "we had of course googled a gazillion things, and then, we stopped" (lines 376-377). At the end of a recollection of Claire's experiences of becoming institutionalised in the hospital (e.g., "you're, you're you know, you're, you just become institutionalised", lines 387-388), we can interpret a shift in the family's priorities in terms of seeking diagnosis as seen elsewhere in her narrative, "we just parked the diagnosis…we just went look, we just need to get out of here, we just need to get through this, we just need to survive" (lines 404-406). The repetition of 'just', and again in line 406 "we just need to survive" (line 406) may suggest a sense of pragmatism and resignation regarding hopes for receiving a diagnosis.

Later in Claire's narrative, she explicitly stated "we had kind of made our peace with that and we said look, if we don't get an answer, fine, and if we do get an answer, <*fine>,* it's not going to make a difference and we didn't really care" (lines 593-596). This shift away from expectation of receiving

a diagnosis may continue to be felt in the lines, “whereas, I was *really* anxious to get that epilepsy panel back from [hospital], mehh (gesturing), I didn’t mind so much about this one, it will come back when it comes back. Until then, we’re just going to live life. So diagnosis at that point, didn’t matter” (lines 595-599).

Claire’s meaning-making around this shift in attitude in pursuing diagnosis, may offer insights that the intensity of the search for diagnosis can be fluctuating. We might consider what factors may have influenced this shift for Claire. In Extract 7.12 below, Claire recalled a conversation between herself, and her husband being told that one third of participants in a research study will not find anything:

Extract 7.12

Claire / Rose: “chances are it’s not gonna mean anything anyway”

574	P	enrolled in the [X] study. And, yeah, getting an answer mattered.
575		Getting a, getting a reason (.) *mattered.* Em, an::d, I::, yeah we got into
576		that study and then I think (eyes closed) at the, they kind of meeting
577		with the researcher for that we were told, there is a, a large p-, I think
578		it's like two thirds of people that go through that, will find out (.)
579		*something.* One third *won't.* They won't find anything in a third. And I
580		remember, kind of going, oh. Okay, well now we're kind of back
581		(gesturing) to that whole thing before were we really wanted to be in
582		that 10% (gesturing) (laughing). Em, and you're kind of back looking at
583		statistics and I kind of said to Tom, I said**, look we're not going to get**
584		**an answer, that's fine. And even if we *do* get an answer**, given that she
585		has, they've tested her for so many genes already, we know it's not this,
586		it's not this, it's not this, it's not this (gesturing on digits), no matter
587		what they find, **chances are it's not gonna *mean* anything anyway**
588		because there won't be the parent support group, there won't be a
589		whole lot of research behind it, it's going to be weird, wild and
590		wonderful**, it's not going to give us (.) an answer for the future, it's not**
591		**going to give us, a community**

The recalled dialogue, presented in Extract 7.12 above, might help us to understand the context of Claire’s changing hopes or expectations for receiving a diagnosis for Rose (e.g., “look we’re not going to get an answer, that’s fine. And even if we *do* get an answer…chances are it’s not gonna *mean* anything anyway…it’s not going to give us, an answer for the future, it’s not going to give us, a community”, lines 583-591).

Claire and Olivia name a main reason for searching for a diagnosis as wanting to access a group of parents whose children had the same diagnosis. For Claire, accessing a group of other parents was named as the primary reason, as described in the extract below (i.e., "*MORE* than anything…", line 529):

Extract 7.13

Claire / Rose: "there would be other parents"

525	P	Yeah, instead of people saying things like oh well, Rose, has
526		a, eh, well she can't walk, she can't talk, she can't eat, you know. I just, I,
527		I didn't know so I wanted to have something that I could then tell people
528		she has this and this means blah, blah, blah. And the other thing that I,
529		***MORE* than anything** was, there's a, kind of a, vibrant parents [RDD X]
530		you know Facebook group, there's a [RDD X] charity in
531		Ireland, **there would be other parents** that I could (.) just make a
532		connection with and say, you know, we have a daughter with [RDD X],
533		**thank God there's someone else who knows what this is like**? And what
534		have you learned and what can you teach us (.) So, as it transpired Rose

As discussed earlier in relation to Extract 7.12, similar to Claire, Mary described a change in her pursuit for diagnosis as seen in the extract below (i.e., "there does come a point where you have to kind of say…we need to just live our life now", lines 1083-1085):

Extract 7.14

Mary / Alexander: "we need to just live our life now"

1082	R	And is it something you feel that you're actively still looking for
1083	P	.h up until about a year ago, I would say I was, em, and **there does come**
1084		**a point where you have to kind of say, ok, you know, em, we need to just**
1085		**live our life now** and get on with things, and I think I've reached that
1086		point, I **still hold out some hope, em, and I'm always kind of keeping an**
1087		**eye on articles and you know, and new research**, things like that, em,
1088		you know, even after chatting on that [support network] call, em, I
1089		contacted some people after that call, after the advice I was given by the

On examining the extract above, it appears Mary may be saying she felt a need to stop searching for diagnosis to live, she had not completely let go of searching for one ("still hold out some hope, em, and I'm always kind of keeping an eye on articles and you know, and new research", lines 1086-1089). In this sense, and similar to Claire, the pause in the search for diagnosis may have been viewed by Mary as out of necessity.

The following extract, Extract 7.15 below, shows Mary's continued solicitation for diagnosis (i.e., "every appointment I ask him, you know is there anything else we can be doing to try and get a diagnosis", lines 1107-1109):

Extract 7.15

Mary / Alexander: "is there anything else we can be doing to try and get a diagnosis"

1105	P	Dr. [X] for at our next appointment so you know, I still, Dr.[X] I'd say is sick
1106		of me at the point, because, you know he's like (laughing) just let it go
1107		because you know we've done everything like and **every appointment**
1108		**I ask him, you know is there anything else we can be doing to try and get a**
1109		**diagnosis** and he kind of said to me at the last appointment, you know,
1110		no matter whatever, city (.) country, you lived in, Alexander has had all
1111		the testing, you know, **and I do have to at some point just accept that**
1112		**and you know, leave it** be because you drive yourself mad, eh to be
1113		honest
1114	R	Mmm
1115	P	Em (.) chasing it, and you put a lot of energy into it, em

In lines 1111-1112 in the extract above, Mary said "I do have to at some point just accept that and you know, leave it". The present tense used by Mary here in "I do" may imply that she is cognisant that she needs to leave it but has not reached this point at the time of the interviews. In the extract, Extract 7.16 below, Mary describes she is "trying" (line 1124), again showing that stopping the search for diagnosis appears to be a thoughtful and ongoing process for her:

Extract 7.16

Mary / Alexander: "accepting that we don't have answers"

1117	P	So, I've started to, instead, just put my energy into our life and you know,
1118		just living again because we feel like we've been on hold for so long,
1119		really since he's been born, you know, and you stop living because
1120		you're always waiting for what's coming next, what's happening next,
1121		you know, em (.) so we've started to kind of, you know, just, just try,
1122		try and live again, you know, and try just kinda get back to, em, just
1123		taking it day to day and that's a really hard and difficult thing to do
1124		for me (laughing) but you know, I am **trying** to kind of em, step out of
1125		that *fighting* zone that I've been in for the last you know six years or
1126		whatever it's been and, and just **accepting, accepting** Alexander as he is,
1127		**accepting that we don't have answers**, em, but, but also not accepting
1128		substandard care for him, you know, so^

In the Extract above, the repetition of "accepting" (lines 1126-1128) may signify the active work Mary is doing to try and adopt this stance (similarly, she uses "trying" in line 1124). This may suggest stopping the search for diagnosis appears to be a thoughtful and ongoing process for Mary. Later in her narrative, Mary provides further insights surrounding her search for diagnosis (e.g., "to have that overall diagnosis would just be (.) I still (laughing), I still dream about getting it, you know, but (laughing) I am more realistic", lines 1224-1226).

It appears Mary believes that answers can provide elevated care for her son Alexander. Within her account, she referenced previous anecdotal evidence where discovery that Alexander had a named congenital condition led to Alexander being monitored by the cardiology department (lines 1185-1198). Mary talked about how answers may come with "massive impact" (line 1201) on parents but the benefit for their son far outweighed that psychological weight for herself and Alexander's Dad (i.e., "that's just something that we have to deal with, you know, it's about Alexander's health and it's about helping him", lines 1202-1203; "for me, it's about his, his health and his care, em and not so much maybe about the emotional impact it has on us, that's just something that we, as his parents, just need to manage ourselves", lines 1240-1242). Mary's reflections here touch on parental roles and responsibilities (which will be explored in the third GET, '*The world of mothers in healthcare systems: "You adjust to it because you have to, because your child needs you to"*').

Elsewhere in Mary's account, she talks about the upset of being told bad news against benefit of knowing what's coming ("at least you know, and you know what to look out for", lines 892-893). Mary discussed what this means for parents in terms of symptom monitoring and potential benefits for the child's care (i.e., "then as soon as I see a symptom, I can alert them and we can do our best to help him or you know, even just give him a better quality of life", lines 895-897). It seems from what Mary is saying that knowing the trajectory of her sons' condition would give her more control in being able to best help her son ("answers *can* only help the child", line 1200; "he can only get better if we have a full proper answer and that's a good example of it", lines 1203-1204). She returns to consideration to her earlier idea that it depends on the diagnosis with the acknowledgement that a rare diagnosis that there will be no treatment. She expresses that even in this case she feels knowing "can surely help him a little bit better" (lines 913-914).

Mary speaks to the experience of being undiagnosed (i.e., "Nobody had any answers so nobody could tell us why this child was born sick", lines 219-220; "you're just kinda desperate for somebody to tell you something, em. And nobody had any answers…that's been the story of our life you know, people just don't have answers (laughs) for us, like", lines 232-236). Looking for answers that healthcare professionals cannot give her Mary's may be understood to encapsulate the main tenet

of her narrative. The search for '*why*' (line 220 in the extract above) was previously referenced above in relation to Claire's journey.

In Extract 17.7 below, it seems Mary is giving an example where her pursuit for a diagnosis may have resulted in possible harm for Alexander (e.g., "one of the nurses said "I just…can't do that to him. It's not right and it's not fair", lines 431-432):

Extract 7.17

Mary / Alexander

427	P	^and really, you know, this needs to be done, this needs to be done now
428		and what else can we do, and they were like he needs a broviac, that's
429		what he needs but we can't get a slot in theatre and em when it came to
430		a day where one of the nurses said "I just, I can't actually get a line, you
431		know there's nowhere to get a line on this child, **I can't do that to him. It's**
432		**not right and it's not fair".** Em, I said, ok, I and I gave out that day, like I

7.2 Researcher Reflection

Mary's response to my direct question as to whether she thought diagnosis would be helpful, "depends on what it is" (line 884) may highlight the idea that not all diagnoses are created equal[23].

Mary verbalised the potential benefits and caveats to diagnosis in what appeared to me as a live to-and-fro debate, unfolding during the process of the interview (i.e., "so I support to have a clear path would be helpful, even though when it's very rare, maybe they can't be so clear anyway, maybe the diagnosis wouldn't ultimately help, well I don't know, it feels, it feels (laughing) like it would" (lines 987-991). I find Mary's contemplation may be represented as a balance scales:

"is there a need to tell me then if you don't actually know when because I'm going to sit here now and worry" (lines 922-924)

"Ultimately I absolutely want to know what's going to happen to Alexander and you know, how long is he going to live for, will he have a *full* life albeit in his own

[23] This brings to mind a remark from Jan Grue's (2021) in his memoir. The author has a diagnosis of spinal muscular atrophy from infancy. He quotes Han Solo "*Never tell me the odds*" (p.9) in relation to how the author surpassed his forecast limitations. This brings to mind questions about the future, the worth of knowing. I am reminded of what Yau & Zayts (2014) referred to as the 'risk of knowing' talk in their article in relation to decision-making in prenatal screening for Down syndrome in Hong Kong, as discussed in *3.4. Individual and Family Sense-making in Response to Diagnosis*.

"but at the same time then you know, there's no real answers" (line 916)	way, you know, em, or will he die young" (lines928-931)

Mary addresses the question of the value of diagnosis as a difficult one. After discussion, giving weight to the positives and negatives, Mary appeared decisive that for, ultimately, the scales are tipped in favour of diagnosis, even if brings challenges for her as a parent.

In the Extract below, Mary's appears to summarise her thoughts on the perceived value of diagnosis (i.e., "would love a diagnosis…you'd know, *roughly,* maybe, what to expect", lines 959-960):

Extract 7.18

Mary / Alexander: "ultimately I would love a diagnosis"

955	P	Yeah (.) yeah: yeah, yeah but (.) yeah, **ultimately I would love a diagnosis**
956		for Alexander so that you do know, and these things that come up
957		aren't causing you, they're still going to, obviously they are still going
958		to stress you out, when your child is getting new things and having new
959		symptoms, even if you had a diagnosis, but at least with a diagnosis you'd
960		**know, *roughly,* maybe, what to expect**, what's coming, em. Alexander's

In summary, the mothers occupy different circumstances and pursuits regarding a search for diagnosis. For Robyn, there was pre-natal knowledge of something wrong and an early genetic diagnosis. For Judy, a rare genetic diagnosis was something that took her by surprise and was something she described she was completely unprepared for. Claire, Mary and Oliva describe a journey of searching for answers. Claire describes most of all wanting access to a community. Mary, describes that in her view, information surrounding diagnosis had led to improvements for Alexander's care in the past and that more answers, even if causing worry for parents, would support the provision of optimum care for Alexander and allow for future planning. Claire and Mary also recognised and named the search for diagnosis as a barrier to living, always waiting and being in fighting mode. For Olivia, she too is searching for a diagnosis for access to health care, services, and the community group (the mothers' expectations, hopes and caveats, attached to diagnosis will be further explored in *Chapter 8 'The world of rare disease: What's in a name?').*

7.3 Dismissal and maternal questioning: Being "pooh-poohed" - "I must be going crazy"

The next subtheme of '*Dismissal and maternal questioning: Being "pooh-poohed" - "I must be going crazy"'* mostly features the mothers' experiences of having their concerns dampened by

healthcare professionals. This subtheme is particularly prominent in Olivia, Mary and Claire's accounts. Claire's experiences appear to express more feelings of self-doubt and maternal questioning whereas Olivia and Mary named feeling dismissed by professionals, leading to feeling *mad* or *crazy.*

Olivia referenced her concerns surrounding Anna's seizures and difficulties sleeping being downplayed as 'normal' illnesses by the medical professionals (e.g., "she's just having seizures", lines 39-40; "she had the flu", line 66; "no, no look she's just had the flu", line 130; "I have no concerns here", line 132; "no, no, there's no concern here", line 139). In the Extract 7.19 below, Olivia describes a mismatch between her own concerns (i.e., "I was *freaking* out like", line 143):

Extract 7.19

Olivia / Anna: "there's nothing there…I must be going crazy"

245	P	And it was affec- it was starting to affect her development, and they were
246		like she's grand, look at her, she's pulling to stand, she's doing this, she's
247		doing that, and I was like, you're here for 30 seconds, **I am telling you**
248		**something's wrong**
249	R	Mmmm
250	P	And they **just completely dismissed it** and (.) so they just sent us out of
251		[hospital] and they were like yeah, it just doesn't exist, don't know what
252		she was looking at, don't know what all those people down in [county] were
253		looking at, **there's nothing there** and we were like, I was like, okay, so I
254		literally went to my GP and I was like**, I must be going crazy**, can you, em, like
255		send me to a psychiatrist or something and she was like, Olivia, you're not
256		crazy, like there's something going on her to be fair like. Em

In Extract 7.19 above, Olivia describes the dismissal of parental concern led her to attend her GP questioning her own health (i.e., "I must be going crazy, can you, em, like send me to a psychiatrist or something", lines 254-255). It is possible to interpret Olivia may have been using some hyperbole in her request to see a psychiatrist to highlight her feelings of self-questioning in response to the medical team's dismissal. Maternal questioning is similarly evident in Mary and Claire's accounts.

Mary referred to a doctor who "talks around an issue like a politician and then will basically (.) brush it off" (lines 600-601) as "as the ones that you know, basically make you feel like you're a bit mad (laughter) you know because they don't actually have the expertise to deal with your child" (lines 605-608). In the Extract 7.20 below, Mary talked about how this attitude leads you to question yourself (i.e., "I start to doubt myself and wonder am I imagining these things", lines 620-621):

Extract 7.20

Mary / Alexander: "I start to doubt myself and wonder am I imagining these things"

615	P	These are the people, you know, that are supposed to have the know
616		how and if they're honest with you and tell you I, I really don't know
617		what's happening here that's fine but **don't look at me as if I'm a**
618		**crazy person** (laughing) you know, and that's your only eh, your only
619		kinda attitude and that's just not helpful you know to Alexander or me
620		because then **I start to doubt myself and wonder am I**
621		**imagining these things**, and you're not, you're absolutely not

In discussing what are helpful qualities in healthcare professionals, Mary professed doctors that are going to listen and believe her are important qualities ("that's the big, that's the big thing, are they going to listen to me and are they going to believe me", lines 635-636). Mary explained "the doctors that do listen are the actually really good doctors, em, you know that trust your instincts and trust what you're saying to them, you don't have to prove it" (lines 593-595).

Claire described questioning herself in relation to the unshakeable anxiety she described as "there was something" (see also "something niggling", line 53, as introduced earlier in Extract 7.10). Claire recalled "if I couldn't look at her without bursting into tears, what was she going to smile *at,* to be fair*"* (lines 115-116) and "what if it's me, it's post-natal depression. I'm not bonding with her, that's why she's not engaging with me" (lines 118-119). In the Extract 7.21 below, Claire gave descriptions of the thoughts that went through her head the moment she confirmed Rose was having seizures (e.g., "'oh thank god'", line 858; "like a little bit of justification", line 860):

Extract 7.21

Claire / Rose: "thank god I'm not crazy…like a little bit of justification"

856	P	fell out of that then again but no in that moment of knowing that there
857		was somewhere to channel all this (gesturing) (..) *anxiety* energy,
858		brought with it, a sense of **"oh thank god",** you know, **thank god I'm not**
859		**crazy**, (facial expression) even though that wasn't really my bother but
860		you know and, and then, **like a little bit of justification** "I knew it! Oh my
861		god I knew it! I knew it, I knew it". Em, you know and the, the the
862		(scrunching facial expression) non-descript is she, "YES!", "ok, she's
863		tracking and following now *BUT SHE DOESN'T"* and "yeah, no, she's not
864		sick and she hasn't got a temperature *but* there's something wrong"
865		and I think I felt justified maybe in that

In the Extract below, Claire continued to describe a sense of relief, of sorts, that came from the realisation that Rose was having a seizure (e.g., “at least I have something to worry about now”, lines 785-786):

Extract 7.22

Claire / Rose: “at least I have something to worry about now”

783	P	into work, and I just went, it was kind an I *knew* it, kind of, and I don't
784		want to use the word relief but I can't think of a better word of just that
785		whole (inhale, hand on heart) oh, well **at least I have something to worry**
786		**about** now whereas this *general* (gesturing), you know, over*whelming*
787		anxiety, I mean over*whelming* anxiety was just, I couldn't put a finger
788		on it, I, I, it was just like I was (..) *DISSOLVING* into this anxiety, that I
789		had nowhere to channel it, I had nothing to hang it on, and I, when I
790		saw the seizure, I think I could nearly feel like this, (large in breath)
791		.hh my god, ok *now* I have something to *worry* about (laugh) ok, it's
792		not like I'm not anxious or it's not like I'm not concerned or it's not like
793		but [it's just

For Claire, her experiences seem slightly differentiated from that of Olivia and Mary, in that Claire doesn’t bring up feelings of being pooh-poohed. She reflected positively on her encounter with the public health nurse during the twelve-week check-up. Claire recalled “I have to say our public health nurse was great, em and she probably handled me pretty well. Em, I suspect she *probably* thought I was over-reacting a little but. Em: (.) but having said that, she took everything I said on board and took it seriously” (lines 869-871), “she didn’t blanket over my concerns and she, em (.) she based everything she said to me on evidence” (line 886-887).

Reference to being “pooh-pooh-ed” appears to occupy only a small component of Claire’s account. Claire volunteered the use of the term “pooh-pooh” when I specifically asked about whether she had experienced this sensation (i.e., “he just goes "<enjoy it. Enjoy it while it lasts> because it's all going to change and you'll be run off your feet"…and I remember saying, well I can't seem to relax around her…am I okay?.. "look, it's baby blues" and he did pooh-pooh me at that six week, developmental check (lines 910-915).

Claire further reflects on this experience, in the extract below. It may be that, in hindsight, Claire appreciates maybe there was nothing that could have been known until seizures were visible for Rose (i.e., “it probably would have just had to have gone to that point”, lines 974-975).

Extract 7.23

Claire / Rose: "it probably would have just had to have gone to that point"

965	P	I dealt with was the public health nurse. I hadn't actually brought her to
966		the GP, *POSSIBLY* (facial expression) (.) because of that six week
967		experience but like that, she wasn't *sick,* em (..) and maybe it's, it's,
968		actually afterwards, you know months later when I talked about it with,
969		with my *own* GP, who's *WONDERFUL*, she said to me, she cou- she said,
970		Claire I probably wouldn't have done anything more, you know if you
971		came to me and told me she wasn't smiling, she wasn't engaging, I would
972		have been looking at the Mum and post-natal, she goes I'll be honest,
973		I wouldn't have been worried about Rose, there was nothing to be
974		worried about, em (.) and you know, that **it probably would have just had**
975		**to have gone to the point** where she was having visible seizures for
976		anybody to do anything because there was nothing (.) to be done.

Returning to Olivia's experience, she described frustration in having been given conflicting information (as will also be discussed within the next subtheme *'A fragmented "jigsaw" puzzle: lack of transparency and co-ordinated care')*. Olivia reflected on a conversation she had with a member of the early intervention team where she recalled she was told her child did not have complex needs and would send her to primary care (i.e., "she was like, oh look, 'I mean we're a very busy service, we have children with complex needs and your child doesn't have complex needs, so I'll send them to primary care", lines 310-313).

Olivia described how the professional's comments were in contrast with the assessment of need report which she received three weeks prior to the appointment. Olivia referenced "the public sector was so poor and so shite and it lets you down at every single turn" (lines 421-422). In contrast, Olivia lauded the support she got from the poison control service (i.e., "oh my god they're the best, you can call then, you can call them pretty much anytime", line 985-986; "they are actually the best resource ever to be honest with ya (laughing)", line 983). This example may seem to serve to further illuminate the unhelpfulness of the public health centre teams.

Olivia's abhorrence for public health service is highly palpable throughout her account. This may be indicated by her use of repeated strong language, "dark humour" (i.e., "I always find the more exhausted I am the more dark my humour", line 2w) and comparison between the efficacy of the public system in contrast with pest control referring to them as). Olivia's subsequent written update offers an entirely different narrative 'sense' (this term will be further defined in *14.3.1 Exemplar in IPA: Borrowed insights from Narrative Inquiry*). Further comparisons will be drawn between Olivia's

narrative account provided during the interview and her subsequent written update, in relation to relevant parts of the data.

In the Extract 7.24 below, Olivia recounted response from healthcare professions when she sought advice desperately to manage Anna's behaviours (e.g., "you just need to be more positive", line 748):

Extract 7.24

Olivia / Anna: "you just need to be more positive"

745	P	when they were here, we were like oh can you please give us any advice of
746		what we can do or where we can go to get any kind of help or support like
747		we don't know what else to do anymore and they were like em **you just**
748		**need to be more positive** because there are children out there who can't
749		walk and your child can walk so you just need to look on the, you know, at
750		the positives? Em (.) so (.) we put in a formal complaint about that because
751		obviously it was wildly fucking unhelpful

In Extract 7.25 below, Olivia relays a conversation between herself and a health and social care professional during a home visit where again Olivia was seeking advice on managing behaviours (e.g., "have you ever thought about watching [television series on parenting], lines 472-474):

Extract 7.25

Olivia / Anna: "have you ever thought about watching…"

463	P	Em, so they were like **have you thought about eh watching [television series**
464		**on parenting],** and I was like I feel like that won't really stop her head butting
465		and (.) you know smearing an:d all those kind of- like I mean
…		
470	P	You know [names lead cast member]? They suggested I do that
471	R	Okay (.) and .hh
472	P	To deal with like, challenging behaviours, you know like. I was like (.) I mean,
473		she- she bu-like, I mean I was asking them for help because her like, she'd
474		had a particular period where like things were really difficult for her

Later in her narrative, Olivia refers to this health and social care professional as the one who "the one who said em that we should watch [television series on parenting]" (lines 741-742) which may suggest how strongly this ill advice stayed with Olivia. It seems laughing, in line 462, may be used to highlight the incompetency of the early intervention team.

The guidance to "be more positive" (line 748, within the earlier Extract 7.24) appears similar to dialogue Robyn recalled from her initial meetings with the paediatrician (i.e., "look, look around you, your baby's not so bad (.) and be be, happy with what you have 'cause there's other kinds in here who are, so I was, but look, we didn't know, so I was like, maybe he's not, like I don't know anyway", lines 40-43). Robyn later referred back to this conversation citing the paediatrician's words "be grateful for what you have, you don't have a child with you know, *rea:lly* bad special needs, and I don't", lines 289-291). In contrast to Olivia, Robyn is not recalling this dialogue in relation to citing feelings of dismissal (I will return to the idea of lasting dialogues in *the fourth GET, 'Etching of the mothers' inner worlds: "I'll never forget").*

7.3 Researcher Reflection

Hearing Olivia's disappointment for public health care services ("the public sector was so poor and so shite and it lets you down at every single turn", lines 421-422), was particularly difficult for me, as a health care professional working with families in public service, to hear at the time of the interview and to review on continued analysis and write up of this thesis. Olivia's words, and the words of all the mothers in my study, have continued to stay with me in my own practice and contribute an added emotional weight and conscious awareness to the mother's experience in healthcare systems. Olivia's description of being told her child does not have complex needs and that her needs would be best met by primary care services, is a conversation that can be considered common place in clinical practice. The learning from carrying out this research, as a clinician, to be so astutely aware of the mother's experience, may be considered a huge benefit in terms of having possibly heightened understanding of the mother's experience. At the same time, I feel this knowledge and foresight adds to weight of compassion and despair as a clinician in times where having this conversation is needed. The thought of possibly being responsible for contributing to a feeling of disappointment in services, for any mother, feels crushing to me as a clinician. Perhaps in this way, mothers and families whom we work with have the potential to leave etchings on my memories (as will be discussed in Chapter 10, *'Etching of the mothers' inner worlds: "I'll never forget",* within the subtheme '*Looks that speak volumes and words I'll never forget').*

7.4 A fragmented "jigsaw" puzzle: Lack of transparency and co-ordinated care

This final subtheme of '*A fragmented "jigsaw" puzzle: Lack of transparency and co-ordinated care'* within the GET *'Entering the world of diagnosis: "There's nothing wrong but everything is wrong"* deals with the mothers' reflections on the lack of transparency and co-ordination in healthcare systems.

Judy focused mostly on her experience of being uninformed and unprepared for receiving a genetic diagnosis for her son and how there was “nobody linked anything together” (line 43) and “they could never pinpoint anything” (lines 45-46). In the extract 7.26, Olivia discusses the challenge surrounding the lack of co-ordinated care (i.e., “sending you from post to pillar”, line 1025):

Extract 7.26

Olivia / Anna: “nobody who knits it all together”

1024	P	Em, I suppose, the biggest, the biggest issue as well I suppose is that when
1025		these medical professionals are **sending you from post to pillar** or whatever
1026		and when this person is saying that she needs occupational therapy or
1027		this person is saying that she needs that, em, **there's nobody who knits it all**
1028		**together**. All these different professionals **give you a jigsaw puzzle piece** but
1029		(.) they don't read each other's notes

In line 1028 in Extract 7.26 above, Olivia used the metaphor of “a jigsaw puzzle piece” to refer to segments of information provided by varying professionals. This metaphor forms part of the title of this subtheme. Elsewhere in her narrative, Olivia stated “they kept bouncing her off to other people, nobody was taking responsibility for her”, lines 579-580). Olivia offered advice to parents that it’s other parents who are going to provide information (“anything we’ve gotten so far we’ve gotten from other parents on the internet”, lines 805-806) and to get a copy of every single report on their child’s health so that they don’t miss crucial information (i.e., “they won’t give you the notes, unless you get them under the Freedom of Information Act”, lines 1031-1032).

In her account, Olivia offers some possible explanations for lack of co-ordinated care and not giving feedback to parents (i.e., in reference to the acute paediatric A&E services in Ireland, “it’s so understaffed”, line 628; “they’re just putting out fires” (line 629); “they don’t have the time to even deal with the patient in front of them, never mind the patient’s paper trail” (lines 1045-1058).

Similar to Olivia, Judy spoke about having to look for follow-up herself following receiving the diagnosis for Declan (“I had to chase and almost fight to get the answers to those questions” line 370). Judy referenced how the consultant did later return her call, without notice. Judy uses multiple phrases to describe how she “wasn’t expecting a call” (line 379) which appears to emphasise how unfavourable this was for Judy (i.e., repetition that the call was “out of the blue”, lines 371-372, 378; “threw me for a loop”, line 380).

In terms of inadequacy of services, Judy also spoke about delays in genetic testing, as seen in Extract 7.27 below (i.e., “that’s twelve months after Declan has had a diagnosis before we find out if myself and my husband are carriers”, lines 404-405):

Extract 7.27

Judy / Declan: “we’re waiting twelve months”

403	P	They're probably going to take, if they take six months to come back again,
404		we're back into March, **that's twelve months after Declan has had a diagnosis**
405		**before we find out if myself and my husband are carriers** and like, you know
406		you compare it to like, there's a lot of the online support groups are like
407		American based and it's like they get a diagnosis and got both straight in
408		and they had the results within a couple of weeks like and they're giving
409		out that they had to wait a couple of weeks for these results, I'm like
410		**we're waiting twelve months** like am (laughing) you know it's, it's
411		*complete* complete, you know when you compare it to what *can* be done
412		it's because we just accept it because it's what *we* know, but when you see
413		how other countries deal with it, you know, **they all have their team in place**
414		almost**- it seems to be in an instant**, you know it seems to be that whole
415		battery of referrals are done straight away. I only know that Declan was

In the above extract Judy compares the efficiency of the genetic testing in Ireland to those services in other areas (i.e., “they have their team in place…it seems to be in an instant”, lines 413-414). In her account, Claire similarly references the issue of delays in receiving feedback from genetic testing (i.e., “And we were told that was going to be six weeks, then it was twelve weeks, then it was sixteen weeks, then it was six months and we just kind of given up on it” (lines 360-363).

Judy offered an anecdote which showcased a consequence of the lack of transparency in healthcare services. She described how she arranged a dental appointment to check Declan’s teeth, given that low calcium levels are associated with the specific rare diagnosis Declan has. Judy advised she was later informed that his calcium levels had been previously checked in one of blood tests drawn on the day of the diagnostic meeting (i.e., “do I need to get his, his nutrient levels checked...And she said to me, well no, we did all of that. No, his calcium levels are within normal range, so I didn't even know that's what they were checking”, lines 775-778). Judy described the resultant trauma for Declan in this situation, “I had traumatised the poor child trying to get bloods” (line 788). Judy’s use of the first-person here (i.e., “I had traumatised…”, line 788) may suggest she assumed some responsibility for the trauma endure by Declan. Judy described “they were poking and prodding him” (line 784). The reference to “tried three times” (line 783) and alliteration in the phrase “poking and prodding” may echo the repetitive action with the needle.

Judy discussed another insult caused by the lack of transparency from medical professionals in that she "had taken time off work to do that appointment". This is in reference to the fact that the appointment was in vain on two accounts. They "never got the bloods done" (lines 788-789) and the bloods were not required as Declan's levels had already been checked and were fine. Judy also spoke about the lack of transparency in terms of the inaccessibility of blood test results. Judy described the report as "just a whole list of medical terms and figures, and stats" (lines 85-86). Judy concluded "I basically had to get that report translated into layman's terms for me" (lines 94-95).

In relation to receiving a diagnosis of a rare genetic condition Judy spoke about the lack of follow-up care (as discussed earlier in relation to Extract 7.6). The badly photocopied piece of paper is mentioned many times throughout Judy's account (i.e., "got handed a leaflet, that was *very* badly photocopied", lines 135-136; "It took me, it actually took me three or four readings before I actually realised, it was a skipping in a pattern, but it took me three or four readings to figure out what the pattern was" lines 145-147), which may attest to the grave significance of this incidence for Judy.

7.5 Summary

In summary, the GET of '*Entering the world of diagnosis: "There's nothing wrong but everything is wrong"* represents the varied early experiences of the mothers, from the pregnancy or birth of their child through to infancy and in some cases, the experiences of receiving a rare diagnosis. In the case of no known diagnosis, Claire, Olivia, and Mary speak about searching for answers. Claire and Mary spoke about weighting this search against the need to live and accept their situation. For Mary, she exclaimed she would ultimately love a diagnosis for Alexander and believes this would only be helpful in directing Alexander's care and anticipating what's coming. Experiences of the child's needs being unidentified, the mothers being *pooh-poohed* by healthcare professionals and a lack of co-ordination and transparency in healthcare services are also explored within this theme. The next GET *'The world of rare disease: What's in a name?'* will offer insights into the mothers' reflections on living within the realm of rare diagnosis, having entered the world of diagnosis (whether diagnosis had been explicitly sought, given or continues to be unknown).

Chapter 8: The World of Rare Disease: What's in a Name? (GET 2)

8.1 Introduction

The GET of *'The world of rare disease: What's in a name?'* describes the mothers' experiences of having a diagnosis which is 'rare' and not at all as understood by professionals and people in the community as more mainstream diagnoses, comparably Down syndrome and autism, as referenced specifically by the participants. All participants reference wanting a diagnosis to gain entry to a group of parents who can resonate with their experiences, possibly provide guidance for the future and be a source of support for themselves as parents and for the child's siblings. This theme highlights the various specific challenges which the participants describe accompany a *rare*, less known, genetic diagnosis in terms of professional knowledge and community understanding as well implications for family planning and the future.

Table 8.1

Overview of Subthemes within GET 2

General Experiential Themes (GETs)	Subthemes
The world of rare disease: What's in a name?	*Rare disease: "A series of letters and numbers that doesn't mean anything to anyone"*
	Not fitting in with the "ASD Moms… or Down syndrome Moms"
	Learning to live: "We just need to live our life now"

8.2 Rare disease: "A series of letters and numbers that doesn't mean anything to anyone"

'Rare disease: "A series of letters and numbers that doesn't mean anything to anyone" examines what 'rare' or 'undiagnosed' means for the participants. Robyn discussed a rare genetic diagnosis as it pertains to accessing supports in education and services. Judy offered insights into her experience of professional understanding of Declan's specific rare genetic diagnosis. Mary and Olivia spoke about what *not knowing*, in association with their child being undiagnosed, means for them and their hopes for what a diagnosis could bring. Claire questioned what a *rare* diagnosis tells you. It is her words that comprise part of the title of his subtheme, "a series of letters and numbers that doesn't mean anything to anyone" (lines 693-694, see Extract 8.1 below for surrounding context).

8.1 Researcher Reflection

Claire's words, "a series of letters and numbers that doesn't mean anything to anyone" (lines 693-694), prompted me to consider all words. Are all words just a series of letters until understanding is built upon them? With further research in the field, in the future, will a named rare disease diagnosis perhaps be as comprehensible to the lay person as now more commonly known diagnoses? Will there by a new host of diseases labelled with letter and number combinations that will remain arbitrary until they have more understanding attached to them? Covid-19 was something that held no meaning for the common person prior to the emergence of the global pandemic in 2020. Depending on the cohort and geographical location, perhaps to some scientists this word was known amongst their vernacular, but not to the everyday person. Now, having lived through a global pandemic, including the emergence of variants, the discovery of vaccines, our schema for what this word means has grown considerably and continues to grow with new knowledge acquired. The variant types of Covid-19 were named and associated with particular symptoms, prior to any meaning being imposed upon these variants, their names may have been considered merely be a combination of letters and / or numbers.

Claire's daughter, Rose, was initially diagnosed with one named rare disease, which will be referred to as RDD X (as discussed in *5.2 Researcher Reflection*) at around 10 months of age. This diagnosis was later revised to another named rare disease, RDD Y. In the Extract 8.1 below, Claire describes the rarity of the specific rare disease RDD X (e.g., "I think forty or fifty kids on record that have her kind of clinical presentation", lines 695-696):

Extract 8.1

Claire / Rose: "a series of letters and numbers…it doesn't even have a name"

691	P	and I am exactly the same where Tom was, so I know that Rose has
692		the one and only, I think we've been told there might be one other kid,
693		with her specific mutation of [RDD Y], **a series of letters and numbers**
694		**that doesn't mean anything to anyone** so she has a genetic mutation of
695		her [RDD Y] gene, there's may:be I think forty or fifty kids on record that
696		have her kind of clinical presentation and like that, if you were to go
697		into it, you're like there's the cohort that inherited it, there's the cohort
698		that are de novo, there's the, and it's kind of related to [RDD Y] and
699		[RDD Y] and I know all this because I've done all the reading, but (..) **it**
700		**doesn't even have a *na:me*** and like it, you know, [symptom diagnosis] still

For me, Claire's reflection "it doesn't even have a *na:me*", in lines 699-700 in the Extract 8.1 above, raises the question on the importance of nomenclature and may provide an argument in favour of

labels. How can we come to understand something that has no referent? The emphasis and prolongation Claire puts on the word 'name' may be interpreted as emphasising the importance she attributes to a condition having a name. The term "the one and only" (line 692) seen earlier in the extract may be understood to imply isolation. This interpretation may be further supported when viewed in the context of the other mothers' experiences, as will be elaborated on in the discussions below.

8.2 Researcher Reflection

Claire's comment in lines 699-700 "it doesn't even have a *na:me"* evoked a question for me surrounding the term 'SWAN', 'syndromes without a name' – does this provide a label? Based on Claire's reflection that RDD X does not have a name, should we infer that SWAN too is a collection of letters that does not quality or afford status of a name?

For Claire, at this point in the diagnostic odyssey[24], having entered the world of diagnosis and consequently the world of rare disease, there appears to be a shift in the value Claire imposed on getting a 'label', as she had come to know from previous extensive genetic testing that the results are unlikely to yield what she was hoping a diagnosis would bring (i.e., "not going to give us an answer for the future, it's not going to give us, a community", lines 590-591; as discussed in relation to Extract 7.6 in the previous chapter). Many genetic tests had already been carried out for Rose which did not yield any conclusive findings (i.e., "we know it's not this, it's not this, it's not this…" lines 585-586, as can be seen in Extract 7.6 in the previous chapter):

In the Extract 8.2 below, Robyn was referring to the experience of 'rare' disease diagnoses providing less access to services than more commonly understood diagnoses, in relation to Nathaniel's rare diagnosis (i.e., "we *have* a diagnosis, I don't need 500 more", lines 445-446):

Extract 8.2

Robyn / Nathaniel: "we have a diagnosis, I don't need 500 more"

445	P	When we already *have* a genetic one, **we *have* a diagnosis, I don't need**
446		**500 more**
447	R	Yeah
448	P	Yeah, you know, what is it, what is it gonna, you know, other than, and you

[24] Diagnostic odyssey, as defined *3.4 Individual and Family Sense-making in Response to Diagnosis*, is a term used in genetics and academic research to refer to the, often lengthy, journey for people living with rare disease to procure a diagnosis (Basel & McCarrier, 2017; Bouwman et al., 2010; Kole & Hedley, 2021; Bauskis et al., 2022).

449 know I think it's a, it's a bad way to look at it in terms of the service wise, is
450 (.), you know, just because, I don't, it's not that I'm not against it, if we have
451 to get it, we have to get it, but **what's the purpose**? Is the purpose just so I
452 tick a box to stay in a service, you know, would be my kind of thinking

Robyn's repeated emphasis on *'have'* in line 445 in the extract above might serve to show the force she has needed to use to legitimise Nathaniel's rare diagnosis to access services. When viewing the above extract within the context of Robyn's whole account, we learn Robyn has needed to defend the sufficiency of Nathaniel's rare diagnosis particularly in the education setting (i.e., "the school is like well we're really afraid of losing his SNA because the SENO doesn't know what that is (..) and she's like, can you get him an ASD diagnosis, well actually I can't just get him one, you know (laughing)", lines 159-162; "would you get an ASD diagnosis because I've seen a couple courses that would really suit", lines 403-404).

Elsewhere in Robyn's account, she similarly referenced the school reported "we'd like to diagnosis him with hyperlexia" (lines 426-428). Robyn questioned "why are we doing that" (line 429). Robyn continued the discussion "we just need a diagnosis for the SENO, and I'm like I have a diagnosis, I have a genetic diagnosis" (lines 429-430). By way of including reported dialogue, which is a common phenomenon across the participants' interviews, we again are given insights into the cultural perceptions of diagnosis (as will be further discussed in Chapter 11, '*Living within a sociocultural world: Constructions and perceptions of 'disability' - "Why do I have to use the word disorder to describe my child?"')*.

8.3 Researcher Reflection

The idea that rare diagnoses are less understood brought to mind the notion that different diagnoses carry more weight than others. This is similar to the idea that not all diagnoses are created equal, as referenced in *7.3 Researcher Reflection*, and in relation to contested diagnoses and genetic testing in the case of inheritable and predictable diagnoses, as explored in *3.6 Predictive Diagnosis.*

Robyn's account deals a lot with the notion of *'purpose'* when considering diagnosis, (i.e., "why give another diagnosis, just for, just for the sake of it", line 412). For Robyn, she referenced diagnosis must have purpose and be timely. Robyn uses the phrase "at the moment", and other phrases linked to timing throughout her account, in relation to considerations surrounding diagnosis (e.g., "I feel at the moment all the diagnoses people are doing is to try to tick a box to get a service", lines 442-443).

In the Extract 8.3 below, Robyn reference the spectrum of needs associated with Nathaniel's rare disease (e.g., "some children can't walk or talk…some people have nothing", lines 214-216):

Extract 8.3

Robyn / Nathaniel

213	P	I think there is because a lot of the time Beth, say **our genetic diagnosis**, is
214		*HU:GE.* It is a case of **some children can't walk or talk** and other children
215		say like, it comes from my husband's side of it, so he has the gene and he is
216		*fine.* So some children, **some people have nothing^**
217	R	Mhmm
218	P	So, in terms of the spectrum, is huge, so where does he lie on that, we don't,
219		at the time, I mean he's doing really well at the moment but you know when
220		I was going to the schools or the playschool they were like what to expect and
221		we were like we actually don't really know
222	R	Yeah
223	P	I don't know like we were told he couldn't walk and he's walking now so. I
224		don't know, like he's, he's, you know so that, so that is a challenge, um (.)
225		and I don't know how a doctor will help you through it because there isn't
226		enough people to, **I suppose with the mainstream, kind of diagnoses**, at least

Robyn refers to "our genetic diagnosis" (line 213) in the line before the extract above. This reference to 'our' may be considered notable in terms of the genetic diagnosis being attributed as 'theirs', as opposed to 'his', denoting the occurrence of a genetic diagnosis as a family condition[25]. In line 226, Robyn started to compare her experience to the experience for parents of children with more mainstream diagnoses (discussion on this comparison will be the focus of the next subtheme '*Not fitting in with the "ASD Moms… or Down syndrome Moms"')*.

Also in Extract 8.3 above, Robyn repeats the phrase "I don't know" (lines 223-224) in relation to the challenges of not knowing where Nathaniel lies in terms of the spectrum of his genetic diagnosis. Robyn's reference to *not knowing* with a rare condition is echoed by Mary, in relation to Alexander having an undiagnosed condition. In Extract 8.4 below, Mary describes "the complete unknown"

[25] The reference to Nathaniel's diagnosis as "ours" (line 213) calls to question the position of disease within the family unit and is reminiscent of the term coined by Jacoby et al. (2018), as defined "Based on our analysis, we argue that the illness is experiences as a mental system extending beyond the space and boundaries of the child's body to include his or her parents which we refer to as the *ill unit".* In my discussion, chapter 6, I will extend this term to include the child's siblings within the ill unit based on my findings, and as consistent with other research findings.

(line 865) as “the biggest problem” (line 859) and later reiterated this as “the biggest issue” (line 864):

Extract 8.4

Mary / Alexander: “you never know what’s coming”

859	P	^but **you never know what’s coming and, and that’s really the biggest**
860		**problem**, em, the doctor’s don’t know, eh, they can’t tell you (.) you know
861		you, you basically are always worrying when you see any new symptom,
862		is this something new, is this something big, is this going to *turn* into
863		something big, **is this ultimately going to be the thing that takes him away**,
864		you know, em (.) and that’s **the biggest issue** for me with being
865		undiagnosed is (.) **the complete unknown**, you know, the, the worry
866		that any new thing brings and of course, seeing him getting these
867		severe pains in his head was making us think there’s something
868		really wrong internally?

In the extract above, Mary refers to “always worrying” (lines 861) and “the worry” (line 865) in relation to the emergence of new symptoms, when Alexander has an undiagnosed condition. Such references to *worry* contribute to the sense of impending doom and ominous discomfort that appears to permeate throughout her account, as can also be felt in the following Extract 8.5 (e.g., “So then you’re watching him”, line 966):

Extract 8.5

Mary / Alexander: “is he just going to fall to the ground”

966	P	**So then you’re watching him**, you know, sometimes he’ll do something
967		or I’m like, o:h, today feels like a day, you know, where maybe if he’s
968		rolling his eyes a bit cause he’s tired or you know he’s just a bit, kinda
969		not right, sometimes he just doesn’t look right, you know and like he’s
970		absent a bit, em, you’re thinking is today the day the seizure’s are going
971		to start, you know, **is he just going to fall to the ground** (.) and you know,
972		will *that* be something that could take him from us, you know, and you
973		end up thinking like that *anyway*, **so I suppose** to have a clear path
974		would be helpful you know, even though when it’s very rare, **maybe they**
975		**can’t be so clear** anyway, you know, **maybe the diagnosis wouldn’t (.)**
976		**ultimately help** you know, em, well I don’t know, it feels, **it feels**
977		**(laughing) like it would**, you know, em but that’s

In lines 973-977, in the extract above, Mary questions the capacity of a rare diagnosis to provide a clear path. The words “suppose” (line 973), “maybe” (line 975), “feel” (line 976) and her laughter

may show her hesitation in committing to the position of diagnosis as helpful (see previous discussion in 7.3 *Researcher Reflection* for Mary's considerations on the value diagnosis could bring).

In relation to this subtheme of '*Rare disease: "a series of letters and numbers that doesn't mean anything to anyone",* I will now introduce Judy's experiences. In Extract 8.9 below, Judy discusses the lack of understanding of rare diagnosis in the education and professional sectors (e.g., "they don't get…what [RDD] is" lines 741-742; "I just felt it's not at all understood", lines 745-746:

Extract 8.6

Judy / Declan: "I just felt it's not at all understood"

739	P	on the street like but am, you know, from a school point of view, I mean
740		like the SNA application was rejected so we're gone to that, that exceptional
741		review stage, and **I'm like they don't get**, in the department, **what [RDD] is**
742		**(laugh)** em, you know, they don't get that his IQ levels, his deficits might
743		increase, might become more apparent as he gets older, am, you know,
744		they don't get that his processing speed is slower, he's, you know, he'll
745		need somebody to repeat instructions, am, yeah, **I just felt it's not at all**
746		**understood**, you know, and from a child's point of view, look a child is
747		linked in with, the only person actually that I met Beth who knew about
748		[RDD], is a **funny story**, am, so the the low calcium was one of the things

(The "funny story", line 748, referenced by Judy in Extract 8.6 above will be discussed with the subsequent subtheme *'Not fitting in with the "ASD Moms… or Down syndrome Moms")'.*

In the Extract 8.7 below, Judy recalled a telephone conversation she had with the manager of the Children's Disability Network Team (CDNT). Judy described that, on summarising the conversation, the manager called back the incorrect diagnosis (i.e., "I said no, no, no, it's [RDD], line 693):

Extract no. 8.7

Judy / Declan: "I said no, no, no it's…"

682	P	^and even I did talk to the, the CDNT team manager and I, so they'd
683		gotten his OT file, his SLT file couldn't be transferred until he finished
684		language class, again, data and all that kind of sharing, and all that, so
685		that was fine and I said to her on the phone, I said to her well you know he
686		has a new diagnosis as well of [RDD] so she said she saw reference to it
687		in the OT file 'cause I had just mentioned it to the OT, just as she was
688		signing over, we got the diagnosis, so she mentioned it I think in her file

689 that he had a new diagnosis but she really hadn't seen him, since the
690 diagnosis but even on that phone call when the manager was relaying back
691 to me at the end, you know we were just kind of doing a recap or whatever
692 and she said and I've noted that he has [incorrect RDD name], [as before]
693 and **I said no, no, no it's [RDD]** and I came off the phone call and I said to my

As seen in Extract 8.8 below, Judy continued to recollect the conversation she had with her husband directly following the call with the CDNT manager (e.g., 'they can't even write down the name correctly...', line 704):

Extract 8.8

Judy / Declan: "they can't even write down the name correctly…how can I have faith…"

694 P like, **you can't even write this down correctly, what faith can I have** that
695 they understand it
696 R Yeah
697 P What **faith** do I have that they (.) rec- that they understand the
698 implications and that they recognise and (.) am, what will I say
699 recognise and (.) .h agree isn't the right word but .hh understand the (.)
700 amount of additional needs and additional therapies and services that that
701 this, you know, what's the implication of [RDD] for his, for her service
702 treatment plan
703 R Yeah
704 P They **can't even write down the name correctly** Beth, **how can I have faith**
705 **that they are actually going to understand what it means**, you know, and
706 said do you know, you're just kind of, you know now you're going to be
707 telling all the pro- like you have to educate the professionals about [RDD]
708 and I'm like .hh oh my god, what Iris said is so right (laugh) am, you
709 know, I I don't know, I mean, I get on the one hand it's a rare diagnosis but
710 if you're manager of a children's disability network team, surely^
711 R Mmmm
712 P ^you know the second most common chromosomal disorder after Down
713 syndrome (facial expression) (.) like
714 R Yeah
715 P **I would have hoped**

In Extract 8.8 above, Judy questioned how she can have "faith" (line 697) that the manager understood Declan's rare diagnosis when she couldn't even write it down correctly. Judy's repetition of this question (lines 694, 697, 704) may serve to emphasise her lack of trust in services further to the incidence. The use of the word "even" (line 694, 704) may also serve to show how

Judy viewed getting the name correct as a minimum standard, which the manager did not meet (as also seen in Judy's remark "if you're manager of a children's disability network team, surely you know…I would have hoped", lines 710-715). The repetition of "no, no, no" (line 692) and "can't even write this down correctly" (line 694) and "can't even write down the name correctly" (lines 704) may also show the intensity of Judy's disappointment and the weight of this experience for Judy. There also appears to be an increased number of hesitations, re-starts and pauses in the above segment within Judy's narrative (e.g., lines 697-699). Judy was also noted to be looking to the side and appeared thoughtful. Her hesitations and body movements may also reflect the disappointment and disbelief caused for Judy by this event. These dysfluencies may also signify that further sense-making is happening for Judy in real-time, through recollecting and re-telling the story in dialogue with the researcher as part of the interview.

In line 707 of the extract above, Judy remarked "you have to educate the professionals about [RDD]". This reflection borders into theme of 'parent responsibility', which will be explored in Chapter 9 '*The world of mothers in healthcare systems: "You adjust to it because you have to"'*.

8.3 Not fitting in with the "ASD Moms… or Down syndrome Moms"

The quotation that comprises part of the title of this subtheme '*Not fitting in with the "ASD Moms… or Down syndrome Moms"* is borrowed from Robyn's words and is a common phenomenon discussed by all the mothers in the study. The mothers all reference a comparison to the experience of those who have a "mainstream" diagnosis in terms of public and professional understanding of what the 'rare' diagnosis means for their child. The mothers describe a lack of parent or sibling supports in the case of 'rare' disease as opposed to more recognised diagnoses. This subtheme also extends to address other challenges associated with rare, less understood or undetermined diagnoses, where they are discussed in direct comparison to more widely recognised conditions. It is this explicit comparative that led me to position the mothers' insights within this subtheme as opposed to the previous subtheme *'Rare disease: "A series of letters and numbers that doesn't mean anything to anyone",* whilst still belonging couched within the overall GET *'The world of rare disease: What's in a name?'.*

Robyn reflects on her thoughts following joining a support group for the specific rare disease her son is diagnosed with. Robyn references "there was a *load* on the [RDD] there was a load on it, and there was a load of research going on at the time" (lines 107-108). When we view this remark in context of the following extract, Extract 8.9 below, we may interpret emphasis on "load" to be representative of Robyn's surprise regarding how much was actually known on the RDD, in contrast

to how little people appear to *'care'* about the diagnosis (e.g., "a little bit confused…if this is actually a thing…like why does nobody care about the genetics", lines 113-115):

Extract 8.9

Robyn / Nathaniel: "because it's not like ASD or Down syndrome…people don't really care"

113	P	^a little bit confused I was like, well if you're, if this is actually a thing, then
114		why is nobody, why does, why is no one (.) like **why does nobody care** about
115		the genetics, **should I not care about the genetic diagnosis** or is that just
116		another label that we don't want or you know, cause he literally was like,
117		yeah, that's what he has, but we actually don't care about the data because
118		it's not really something that is mainstream, it's not, what I really felt Beth,
119		and I felt this the *whole* time because it's not, **because it's not like ASD or**
120		**Down syndrome or something like that, people don't really care**.

The repetition of "why" and multiple re-phrasings in line 114 in the extract above, may also signify Robyn's puzzlement regarding the disparity between people caring and how much is known about the RDD. We see how what she perceives as people's lack of caring causes Robyn to question herself or the value of the diagnosis that should be taken with "should I not care" (line 115). Viewing this extract within the body of the transcript, the "he" (line 116) referred to above appears to be the geneticist. Robyn's conclusion that 'people don't really care' because it is not comparable to more well-known conditions such as autism or Down syndrome (lines 119-120) is the quintessence of this subtheme. We don't know who the "people" (line 120) are who Robyn refers in the extract above. However, we may interpret she is referring to health care professionals and educators, or just people in general (i.e., the public).

In the Extract 8.10 below, in relation to comparing rare with more 'mainstream' diagnoses, Robyn states "I suppose with the mainstream, kind of diagnoses, at least there's a team, there's other parents who've gone through it or you know there's that kind of support group" (lines 226-227):

Extract 8.10

Robyn / Nathaniel: "the only Mom who doesn't have an ASD or Down syndrome child"

223	P	I don't know like we were told he couldn't walk and he's walking now so. I
224		don't know, like he's, he's, you know so that, so that is a challenge, um (.)
225		and I don't know how a doctor will help you through it because there isn't
226		enough people to, **I suppose with the mainstream, kind of diagnoses, at least**
227		**there's a team, there's other parents** who've gone through it or you know
228		there's that kind of support group and not to be dramatic but it's a case of
229		like, for all the things that we've ever been and again, I've had great

230		support from the early intervention centre, we've done all the training and
231		everything but I'm usually **the only Mom who doesn't have an ASD or**
232		**Down syndrome child**
233	R	Yeah, and-
234	P	You know **I have the child who has something else**

Robyn uses the phrase "something else" (line 234) in the extract above to refer to her son's rare diagnosis in comparison to the more understood diagnoses of autism or Down syndrome. Robyn's use of this term may reflect what she has come to know in terms of how rare disease is perceived by others in the community. Perhaps implying a sense of isolation, Robyn also references herself as "usually the only Mom" (line 231). Elsewhere in her account, Robyn elaborates in relation to the groups within the early intervention service her son was accessing, "there were loads of groups in the early intervention centre but they were all diagnosis specific…I'm not part of any of these groups" (lines 348-355).

8.4 Researcher Reflection

The quote "I have the child who has something else" (line 234) in the final line of extract 8.13 above comprises the titular quotation of this study. For me, these words, as spoken by Robyn, embody the entire experience of all the mothers. The sentiment epitomises the resounding thread tying mothers' accounts together and representing their common experience. "I have the child who has something else" speaks to the unique position of having a child with a rare or undiagnosed condition, which can be further unpacked and evidenced across worlds (or GETs) for the mothers.

Similarly to Robyn, in Extract 8.11 below, Mary described the experience of being with other mothers and children as "really hard" (line 555) and "very, very isolated" (line 556):

Extract no. 8.11

Mary / Alexander: "we never belonged anywhere"

555	P	It was really hard, it was really hard and you felt very, very isolated I
556		would say, especially em when **we *never* belonged anywhere**, you know
557		and we still don't really in a lot of ways like, except for SWAN Ireland,
558		you know, em, we don't belong in any real group because even em

In the extract above, emphasis on 'never' in "we *never* belonged anywhere" (line 556) may signify the weight of this reality for Mary, of 'never' belonging. Later in her account, Mary described

additional individual factors as compounding the isolation for her ("we live in an area that I didn't grow up in, em, so I don't know anybody here either which didn't help", lines 563-568).

Claire uses emphasis and increased volume in referring to what she wanted "*MORE* than anything" (line 529; as previously seen in Extract 7.13 in the previous chapter) in looking for a diagnosis, was access to that group of parents whose children shared the same rare disease diagnosis as her daughter Rose ("there would be other parents…thank god there's someone else who knows what this is like", lines 531-534, Extract 7.13). Elsewhere in Claire's account, and in relation to not having yet made contact with the [RDD X] parent group, as Claire had been told to await confirmation from the genetic panel of the diagnosis for Rose, Claire reflected "I was waiting, *waiting*, to just give that mother a ring and say, how did you get through" (lines 550-551). Repetition of 'waiting' and emphasis on the second repetition appears to convey the strength of Claire's desire to find this group of parents.

Olivia, similarly, as shown in the extract 8.13 below, described her desire for a diagnosis as "purely for (.) because, for that reason that we go and find (.) *that* group" (line 631):

Extract 8.12

Olivia / Anna: "she ate four window decals today, did you child ever do that?"

630	P	**we would love to have a diagnosis**, **just purely for (.) because, for that reason**
631		**that we could go and find (.) *that* group** and go hey, you know (.) like, **she**
632		**ate four window decals today, did your child ever do that?** (laugh)
633	R	(nodding)
634	P	What did you do about it?
635	R	Yeah
636	P	Em or, what did, you know, **how is that affecting your child twenty years**
637		**later**, you know that kind of thing. Em, so that you can know to plan ahead, a
638		little bit, em (looks in direction of child) <monkey>

The pausing and emphasis on the word *that* (line 631) may be interpreted as representative of the importance for Olivia of finding a parent support network herself. Olivia referenced projected questions she would put to the group such as "she ate four window decals today, did your child ever do that?" (lines 631-632) and "how is that affecting your child twenty years later?" (line 636). Olivia's questions are similar to what Claire referenced she would love to ask ("what have you learned and what can you teach us", lines 533-534; as can be seen in extract 8.12 above)

Robyn, Judy and Olivia all reference that there are no support groups for the siblings of the children *who have something else*. Consideration to the mother's insights into the experience of siblings will be explored in Chapter 12,*' World of the sibling: "Second fiddle"'.* Analysis of the mother's reference to the sibling experience within this subtheme are included only as they directly relate to the comparison between siblings of children with rare diagnoses as opposed to more common diagnoses or illnesses. See, as discussed by Robyn, in the Extract 8.14 below:

Extract 8.13

Robyn / Nathaniel: "you have the children's support group for kids who have autism, or…for brothers who have Down syndrome"

521	P	Yeah, just normal, em, but em, yeah and the other thing, again this is **super**
522		**dramatic** as well, is there is no support groups for them because the, you
523		know, you have the eldest, **you have the children's support group for kids**
524		**who have *autism,* or you have the children's support group for brothers who**
525		**have *Down syndrome,*** there is no support group for them either, now I don't
526		know whether they would want to go or whether they would want to do it
527		but the option isn't there for them
528	R	Yeah
529	P	You know for them to, to, I think there's like the big brother group or
530		something like that and it's for if your brother is, em, on the spectrum, em

In the extract above, Robyn used the phrase "super dramatic" (line 521-522). This phrase, or similar, is used often by Robyn throughout her account. This may link back to what she was told in an early interaction with a paediatrician, as was shared earlier in her narrative, which may have shaped how Robyn views her situation and what she feels is 'permissible' to tell. (This idea will be revisited in Chapter 10, *'Etching of the mothers' inner worlds: "I'll never forget").*

Judy similarly spoke about the lack of support for siblings, in comparison to more mainstream diagnoses. Judy references that she had signed Declan's sister up to a sibling workshop which got cancelled due to the Covid-19 pandemic (lines 1000-1001). In the Extract 8.15 below, Judy introduces a new comparison to cancer diagnosis (i.e., childhood cancer, line 1037), as another more universally understood diagnosis:

Extract 8.14

Judy / Declan: "is there a gap for these siblings"

1033	P	know, am. It's it's, but it is, it's tough on them, am, and I did say to
1034		somebody before Beth, you know .hh, it's an awful way of saying it but it's

1035 the only way I can explain it now right, so you know, if, if Declan had gotten
1036 a diagnosis of a childhood illness like, something very traumatic, and I'm
1037 not saying it blasé but something like childhood cancer and people
1038 understand like oh my god this takes so much time and attention, focus
1039 and you know, siblings get *supported* in a different way, whereas I think
1040 because this is ongoing and you know, its so, not understood, the level of
1041 awareness is so low, it's – they are forgotten about and they're not
1042 supported by, you know I wonder sometimes like could we link them into
1043 services for siblings *with* very traumatic childhood illnesses and you know
1044 that they would get a little bit of TLC, a little bit of spoiling, am but then
1045 I don't- like, *I* don't know about those services, like are they for children
1046 that are like extremely sick and then Declan isn't, and you know, and
1047 R Yeah
1048 P Is, ah, I, I just like- **is there a *gap* for these siblings,** you know

Judy placed emphasis on the word "*gap",* as seen in the final line of the extract above (line 1048). This stress may serve to reflect the magnitude of the gap for sibling supports, from Judy's perspective. Similarly, Olivia indicated about what a diagnosis of autism might provide for Anna's brother in Extract 8.16 below (i.e., "they'd all be like 'oh yeah, my sister, my sister bites me all the time too'", lines 931-932):

Extract 8.15

Olivia / Anna: "my sister bites me all the time too"

926 P He is, in fairness. See I suppose, that's the other kind of thing, is that (..) you
927 know, that a diagnosis would mean for *him* (very long pause) because like
928 even if it was em, even if, like if it was autism then, for example, the
929 [named support group] em, **they have a sibling^**
930 R Mhmm
931 P **^*group***, and they'd all be like, they'd all be like "oh yeah, **my sister, my**
932 **sister bites me all the time too"** you know

In the Extract 8.16 above, Olivia placed emphasis on the word "group" (line 931) which again may serve to show the importance she placed on what this group could offer. In the Extract 8.17 below, Olivia introduced another comparative to children with dyslexia in relation to family supports (i.e., "if you have a child with dyslexia you can go to the dyslexia association and you can talk to other people"):

Extract 8.16

Olivia / Anna: "the only reason we ever wanted a diagnosis…you can talk to other people"

422	P	so poor and so shite and it lets you down at *every single turn* (.) so **the only**
423		**reason we ever wanted a diagnosis** was because for example, in [county],
424		there, you know, **if you have a child with dyslexia you can go to the dyslexia**
425		**association and you can talk to other people** who have had a child with
426		dyslexia and figure out what helps and if she had autism, then we could go
427		to the [named support group] which is like a parents- it's in a community
428		centre and they have a play group and **they have a sibling's play group** and
429		they have you know all this kind of thing **whereas instead what we have is**
430		**absolutely nothing**. We have (.) a child who just has, you know, goes in- now

In Olivia's written update, of note, we learn since the time of the interview Anna has gotten a diagnosis of autism and intellectual disability and a genetic mutation was identified within the family. We know from the interview that Olivia expressed she felt a diagnosis of autism would be helpful in terms of accessing information ("I was like this must be autism because (.) and to be honest with you I wish that it was because then I would be able to find out more information on it" lines 362-364), and supports for herself and Anna's brother (see earlier Extract 8.16).

Robyn is the only participant who expressed some reluctance or a potential negative consequence of joining such groups (i.e., "what I would say is that sometimes it's nice not to talk constantly about your child's difficulties and so I have found that in the groups with other parents…is you're constantly talking about what's wrong with your child and I just find that exhausting", lines 239-243).

Claire also offered a nuanced insight in relation to this subtheme of '*Not fitting in with the "ASD Moms… or Down syndrome Moms"'.* In the Extract 8.18 below, Claire spoke to the individual variation that still exists amongst people with Down Syndrome[26] (i.e., "*even* a parent that says my

[26] Claire's insights in this extract are supported by the findings of Sangster et al. (2022), who based on their study, identified that although there is a dominant narrative relating to the the experience of parents raising children with Down syndrome, alternative narratives exist. One mother's quotation which exactly reverberates what Claire is saying is "When I talk to other parents of children with DS, we're not even-we're in the same book, but we're in different parts of the book" (Sangster et al., 2022, p.10). This provocation is also supported by findings from Goodley & Tregaskis (2006), where one mother speaks to the downside of a more universally understood diagnosis "The professionals that we were talking to were helping us with the 'Down syndrome' rather than helping us with child care, you know, nothing like how much milk and sleep he

child has Down syndrome, means something completely different from *another* parent who says my child has Down syndrome", lines 729-731):

Extract 8.17

Claire / Rose: "you know what people with Down syndrome can and can not do"

727	P	Yeah, yeah, so it's just one of those ones that, you know, (interjections),
728		unless you fall into the very, the ones that people know, unless you say
729		Down syndrome, and ***even*** **a parent that says my child has Down**
730		**syndrome, means something completely different from *another* parent**
731		**who says my child has Down syndrome**, you know it's not like every child
732		that has Down syndrome but I think it just cuts off the- people go I kind
733		of think I know what that means, in terms of **you know what people with**
734		**Down syndrome can and can not do** or are or are not like you know
735		**rightly or wrongly people think that**. Am

In moving to consider the issue of '*How do you tell people?*' (which will be explicitly explored in *Chapter 11, 'Living within a sociocultural world: Constructions and perceptions of 'disability' – "Why do I have to use the word disorder to describe my child?')*, in the extract below, Judy explained how she explained Declan's diagnosis to their family, using comparison to more mainstream diagnoses (i.e., "look lads, you've heard of Down syndrome, it's an extra chromosome, Declan is short a chromosome and it's kind of that same idea", lines 729-731):

Extract 8.18

Judy / Declan: "you've heard of Down syndrome…it's kind of like that"

719	P	Outside of healthcare, aw: **it's not at all understood**, not a, nada, no, em,
720		I mean, I'm actually a post primary teacher, I'd never heard of it, am,
721		Declan's, I mean, transferring school now, they've never heard of it, you
722		know, and I'm like well I can understand from a teaching point of you, I
723		wouldn't expect them to understand- to have heard of it, if I haven't
724		heard of it^
725	R	Yeah
726	P	^but again, I mean, you know I have heard of Down syndrome, you know
727		and that's the way I've been explaining it to people, you know, if I'm
728		with family and stuff, we were you know just telling family about the
729		diagnosis and the way I was explaining it to them was, well look lads, **you've**

should be having, how to change nappies, all these type of things that you know you tend to miss" (Goodley & Tregaskis, 2006, p. 638).

730 **heard of Down syndrome**, it's an extra chromosome, Declan is short a
731 chromosome and am **it's kind of like that same idea** that there's lots of
732 different levels of severity, some people have milder cases, some people
733 have more severe cases, you know, some people have heart problems,
734 some people won't and, so that's kind of just, I've been relating it because
735 **it's the only relatable**, am, I think, awareness that people have (.) am (.)
736 but

Judy described how she framed the diagnosis around Down syndrome as it is something relatable that people would have existing awareness of (see lines 734-735).

As introduced in the previous subtheme '*Rare disease: "a series of letters and numbers that doesn't mean anything to anyone"',* Judy referenced a "funny story" (line 748) that ensued when she brought Declan to the dentist, and the secretary's grandchild had the same rare disease as Declan (i.e., "oh my grandchild has…" (line 756), as detailed in Extract no. 8.20 below:

Extract 8.19

Judy / Declan: "I know someone with that"

750 P And so I said, do you know, I'll, I'll, I'll get a dental check-up done as well,
751 because and again, as well like, he was on so many antibiotics, sure his
752 teeth could be in an awful state now, you know, looking at them, they
753 seemed to be ok, am so I got a, I rang just a, so I said I'd try the school
754 dental first, I said I didn't know would they see him, and if not, I'd get a
755 private appointment but I rang the school first anyway just to see, and the
756 secretary I think, that I spoke to, she was like, oh my grandchild has [same
757 RDD] (smile) **she was the only person that I know, who was like, oh I know**
758 **someone with that**, and she was like, definitely bring him in for a review
759 and I'll tell the girls here about it and she was like (smile, laugh) you know
760 like and I suppose, this had happened at a time that I had read so much, and
761 it was all negative and I just, **that was a little glimmer of hope, in the who:le**
762 **madness of it all (elated facial expression)** so he had his dental review and

In the Extract 8.20 above, Judy's response to meeting somebody in the community who knew someone with the same rare disease as her son, highlights the joy and value for Judy in this connection. She described "she was the only person that I know, who was like, oh I know someone with that" (lines 756-757). If we examine the notes in parenthesis we see Judy showed both a smile (line 757) and an elated facial expression (line 762) as she re-lived this moment which may be interpreted as the enduring hope that the power of connecting with someone in a similar situation

brought Judy. Judy's description "that was a little glimmer of hope, in the who:le madness of it all (elated facial expression)" (lines 761-762) seems to add to the sense of optimism that can be felt as often prevailing throughout Judy's account, alongside the voice of spirituality (as will be further detailed in *8.6 Researcher Reflection)*. The prolongation on the word "who:le" (line 761) may imply Judy slowing down in relishing this precious moment. I have featured this anecdote here amongst the analysis as I think it provides an example of what all participants say they are looking for in finding as Claire said "there would be other parents that I could (.) just make a connection with and say, you know, we have a daughter with [RDD X], thank god there's someone else who knows what this is like?" (lines 531-533).

8.5 Researcher Reflection

I think it is encouraging to consider the lasting happiness and hope that this encounter with a stranger who could relate to Judy's experience brought and continues to bring Judy. This feels to me particularly comforting in comparison with the main tenet captured within *Chapter 10, '*Etching of the mothers' inner worlds: "I'll never forget"' (GET 4). Does Judy's anecdote offer evidence to support the idea that words and looks can leave lasting smiles on the mind of another as much as they have the potential to leave painful scars?

For Judy, she found membership within that group upon Declan receiving a rare disease diagnosis. Judy referred to the chair of the support group for her child's rare disease as an "angel" ("you meet angels along the way, so Iris Wright to use was an absolute angel, am gave me, you know, a lot of reassurance, a lot of (.) and just information", lines 251-253). Judy commended "I probably got more information from her than I felt I did from the appointment" (lines 227-228). The use of "probably", along with other conditional terms and seen elsewhere in Judy's account, might suggest ambiguity in this assertion or openness that her perception is not absolute.

8.6 Researcher Reflection

Iris is a pseudonym applied to the head of the support organisation for Declan's named rare disease diagnosis. In all other participants' accounts, pseudonyms were only assigned to immediate or extended family members, as referred to as by the participants. For Judy, I interpreted that this woman required a pseudonym to echo the significance Judy attributes to this woman within her story, with multiple references to Iris (lines 225-239, 333, 1108-1111) and referring to her as an "absolute angel" (lines 251-252).

Judy makes multiple references to angels within her account (e.g., "you meet angels along the way", line 251). The along with the gratitude she expresses as the 'silver-lining' in that late

diagnosis permitted Declan's entry to language class, which she regards for Declan has been an *"absolute,* like, salvation" (line 543) may signal a spiritual voice which appears to transcend throughout Judy's account and appears as distinct from the other mothers' accounts.

8.4 Learning to live: "We just need to live our life now"

The subtheme '*Searching for an answer or "firefighting" moment by moment?'* (as discussed in Chapter 7) and the current subtheme *'Learning to live: "We just need to live our life now"* are closely linked. The former primarily relates to how the mothers came to enter the world of diagnosis and how they navigated this world, in relation to whether they were explicitly seeking a diagnosis, or whether a diagnosis was given, unsolicited. The current subtheme involves learning to live within that world, irrespective of whether there is a known rare diagnosis or in the case of an undiagnosed condition.

Towards the end of her interview, Claire spoke about life expectancy and genetic testing. Claire recounted in relation to not needing to know about life expectancy "what more does *any* parent know about their kids other than they're born and life is ahead of them and that's all we really wanted to know and the [RDD Y] part just of kind of tells us that" (lines 1186-1189).

Similarly, as presented in the Extract 8.21 below, Claire recounted a conversation between herself and her husband in relation to considering what if Rose was born with disabilities (e.g., "we'll deal with it and what will be, will be", line 1247):

Extract 8.20

Claire / Rose: "what will be, will be"

1240	P	we're kind of the family that we're like the future is what the future is,
1241		you know, like I said, I was pregnant with Rose when I was at the
1242		funeral of that girl, she was 50 when she died from [RDD Z],
1243		and it sparked a conversation between myself and Tom, what if this
1244		child has disabilities^
1245	R	<mmm>
1246	P	^and I remember saying to him, well then she does, then she or he does,
1247		you know, and then, we'll deal with it and **what will be, will be**, and
1248		that's very much our attitude and has been, look **what will be, will be**.

Claire's use and repetition of the phrase "what will be, will be" (lines 1247 and 1248) may appear to show her outlook on life and living. This may be considered translating to the well-established phrase, *'que sera sera'*.

In her account, Robyn offered insights around therapies for Nathaniel, which can be viewed in terms of learning to live. In relation to accessing private and public speech and language therapy services, Robyn concluded "I was like, we're really getting nothing out of this because (laughing), she's just making me feel better (laughing). She wasn't telling us anything different…we actually are doing everything, again, it's not, you don't need to be seeing your therapist once a week" (lines 827-830). Robyn also discusses potential dilemmas with making life "*ALL ABOUT THERAPIES"* (line 699-700). In Mary's account, she described "whatever is thrown at you, you adapt, and you adjust" (lines 185-186). Mary discussed the challenges in needing to leave behind this "fighter" role in order to live, in the absence of diagnosis (see earlier extract, Extract 7.14). Both of these examples, in relation to the mother role, relate to ideas which will be explored within Chapter 9 *"The world of mothers in healthcare systems: "You adjust to it because you have to, because your child needs you to"'* (GET 3).

Considering Olivia's subsequent written update, in comparison with her spoken narrative provided at the time of interview, may provide some useful insights on the subtheme of '*Learning to live: "we just need to live our life now"*. Olivia expressed during the interview that "after we did the autism assessment, we cried for like three days" (lines 1083-1084) in relation to the fact that Anna did not meet the criteria for autism (analysis on this reaction will be revisited in Chapter 11, under the subtheme *11.2 Labelling*). In contrast to the abhorrence for healthcare services and professionals in Olivia's narrative account (e.g., "the public sector was so poor and so shite and it lets down at *every single turn",* lines 421-422, can be seen in the previous extract, Extract 8.17), we see praise for healthcare professionals in her written update "her geneticist in [hospital name] has been fantastic" (line 22w). In the summary of Olivia's email, she appeared to present an acceptance of a new perspective on way of life.

Extract 8.21

Olivia / Anna (written account)

> Though we won't get a unifying diagnosis anytime soon or maybe ever, we know we have done everything we possibly can to find answers. We have reached a place where we are ok with not knowing because she's not going backwards anymore and that's all we need, so long as we keep taking these baby steps forward and our little girl is smiling then everything else can be figured out. (lines 22-27w)

I think this summation carries a tone of hope. If we compare the language used in Olivia's spoken account versus the update provided, the words and phrases alone reflect a discernible shift in

perspective and the living experience, as described by Olivia. There is a lot of strong language in Olivia's oral account including "just bullshit really" (line 279), "so poor and so shite" (line 422), "they're so crap" (line 573), "so shite" (line 622), "wildly fucking unhelpful" (line 751), "they were just really fucking rude" (lines 757-758), "drives me fucking spare" (lines 1071-1072), and "really pisses me off" (line 1091). All of which were used in reference to healthcare services and providers. In contrast, language reflective of 'appraisal' and 'hope' can be identified in her written update, including "grateful" (lines 3w, 14w), "really beautiful progress" (line 4w), "improved enormously" (lines 4-5w), and, "fantastic" (line 22w).

It is of note that although Olivia's attitude is different in the above accounts it is not to say that all the difficulties as referenced in the oral account had been resolved. Whilst Olivia referenced "things have improved a lot" (line 2w), she wrote "sleep and pica are still intense, but we are constantly working on new ways to help meet her sensory needs and reduce these challenges for her" (lines 7-8). Sleep and challenging behaviours were named as the greatest challenges in Olivia's oral account (line 454).

8.7 Researcher Reflection

Referring back to the narrative types, as described by Arthur Frank (1994), I think viewing this update in contrast to Olivia's spoken account at the time of the interview, represents a shift in narrative types from a voice of "dark" (as used by Olivia herself) distain to one of acceptance, and learning to live once things are taking small steps forward. This new narrative type may be more closely assimilated to the "quest" narrative type of described by Frank. However, I will not reduce Olivia's story to this more constrained description which does not fully reflect the nuances of her voice and narration (see discussion 3.3 *Illness Narrative: Definitions and Considerations*). For example, the "quest" story, as defined by Frank (2007) there no gladness about illness told through the quest story (2007), which I would not believe as applicable to the underlying storyline conveyed in Olivia's update.

In more general terms, Franks quest narrative typology is all about learning to live life re-imagined, a new life *with* illness. For Mary, Claire and Olivia, in my study, it may appear that acceptance that a unifying diagnosis may not be available, and reduced expectations or 'change' in the search for diagnosis, may have supported adoption of a new narrative type, or more colloquially, a new way of living.

8.5 Summary

In summary, the GET of '*The world of rare disease: What's in a name?*' looked at the mothers' experience, having entered the world of diagnosis, specifically in relation to 'rare' diagnosis or having no unifying diagnosis. The mothers spoke of what this meant for themselves, their child, their family and in interactions with healthcare professionals and educators in accessing services. This chapter included reference to the mothers' experience of rare or undiagnosed conditions, which they contrasted directly to the experiences of family members whose child or sibling has a more 'mainstream' or commonly understood diagnosis.

Finally, within this theme, I discussed how the mothers learned to live, within the world of rare or undiagnosed disease. As stated within this chapter, there is overlap with some of the experiences the mothers describe exemplified within earlier subthemes and further description of which will be reserved for subthemes to come. The next GET '*The world of mothers in healthcare systems: "You adjust to it because you have to, because your child needs you to",* will examine the mothers' experiences, as mothers within healthcare systems, in the context of the phenomenon being studied.

Chapter 9: The World of Mothers in Healthcare Systems: "You adjust to it because you have to, because your child needs you to" (GET 3)

9.1 Introduction

The GET of '*The world of mothers: "You adjust to it because you have to, because your child needs you to"'* is specifically about the experience of having a child with a rare or undiagnosed condition for the participants, as *mothers.* Subsumed within '*The world of mothers'* is the first subtheme *'Maternal role and responsibility: Balancing trust and onus"*, which includes the "fight" referenced by all mothers in order to obtain optimum care for their child. The focal point of this subtheme is the tension suggested in the mothers' accounts between wanting to have their concerns listened to by healthcare professionals, in opposition with the pressure felt in being responsible to monitor and report on their child's health. Claire named this phenomenon as "a bit of a catch 22" (line 1006). Within this first subtheme, what should constitute parent versus professional roles is also explored. Two distinct subsections are also presented including, gender roles or individual parent roles *(9.2.1)*, and one participant, Claire, spoke to her position as both healthcare professional and parent and mother *(9.2.2).*

The second subtheme of *'Finding your voice, lessons learned'*, addresses the insights shared by all the mothers regarding having learned to understand and navigate healthcare systems further to their experiences with their child. The experience of maternal trauma (*9.3.1)* as most heavily evidenced by Mary and becoming institutionalised living in hospital as reported on by two participants, Claire and Mary *(9.3.2),* will also be discussed within two additional subsections.

Table 9.1

Overview of Subthemes within GET 3

General Experiential Themes (GETs)	Subthemes
The world of mothers in healthcare systems: "You adjust to it because you have to, because your child needs you to"	*Maternal role and responsibility: Balancing trust and onus* *Finding your voice, lessons learned*

9.2 Maternal role and responsibility: Balancing trust and onus

The subtheme '*Maternal responsibility: Balancing trust and onus'* considers the mothers assumed roles of fighting for their child's access to services, the distinction between maternal and paternal role, and considering boundaries between professional and parent roles. The "fight" for services is something common to all the participants' experiences. The qualifier '*Balancing trust*

and onus' serves to convey the juxtaposition between a want to be listened to as the mother of the child in contrast with the responsibility healthcare providers relying on a mother's observation places on the mothers in this study. Within her narrative, Robyn described the "hardest part is, every *single* thing that you get, you have to fight for and that's the, and that's the crap bit, because nobody tells you" (lines 668-670). Robyn's repetition of the phrase "the hardest part" and the emphasis on "single" in the quotation above appear to convey the struggle of the fight.

In the Extract 9.1 below, Robyn described that she was not informed of what supports she was entitled to with reference to the domiciliary care allowance (DCA) and the access inclusion model (AIM):

Extract 9.1

Robyn / Nathaniel

245	P	So, you know, but *then,* em, I was very lucky that eh, my husband's aunt,
246		she has a playschool and eh, I was really against this to begin with, however,
247		she has a, she was fantastic, she knew how to get AIM, she knew how to get
248		all the stuff for him, and whereas other parents would have told me that
249		but we didn't really have that (.) connection, so you know, no parent ever
250		told us what we were supposed to do^

In extract 9.1 above, Robyn described her position as fortunate in having a family connection to inform her in the absence of a parent support group (i.e., "my husband's aunt…she knew how to get all the stuff for him…whereas other parents would have told me that but we didn't have that, connection" lines 245-250).

Olivia, described the maternal responsibility and role, similarly to Robyn, as a "fight" for services. In the Extract 9.2 below, Olivia recounted a time when she did not agree with the advice of service providers regarding the most appropriate service to best meet Anna's needs (e.g., "I don't' agree. Respectfully, I don't agree", line 308):

Extract 9.2

Olivia / Anna: "Respectfully, I don't agree"

300	P	So like that's on, like, a fairly hefty dose of clonidine, em, so, it does work, it's
301		definitely better than it used to be but it's still pretty shit, em, so we went then
302		to the early intervention then with the [disability service] in February and they
303		were like em (shaking head), they were like she's so smiley, she's so
304		engaging, there's noth- they were like this, I actually, it was with a, with em a

practice nurse?, the appointment, em, she came in and she played a couple of games with her and she was like (hand gesture) that's the last time now I'm gonna see ya, there's no need for her to be here, am, and I was like, what? I was like, um, **I don't agree. Respectfully, I don't agree**.

Further to this extract, Olivia recalled further dialogue from this interaction, where she explicitly stated her role "I have to just advocate as best I can" (lines 320-321).

Within her narrative, Mary referred to the pressure in needing to fight (for your child's care), "it's such a pressure and such a stress on parents who have a very, very sick child you know, *knowing* you have to fight for his basic needs?" (lines 403-404, as can be seen in the later Extract 9.4). In the Extract. 9.3 below, Mary echoed the reality that it is the parent's responsibility to 'fight' for their child that leads to actions within the healthcare systems. Mary spoke specifically from the position of a mother whose child has no known diagnosis (i.e., "when you have a child that has no diagnosis…you feel like you have to fight all of the time, to get him looked after", lines 703-705):

Extract 9.3

Mary / Alexander: "when you have a child that has no diagnosis…you feel like you have to fight all of the time"

P Yeah, it's very stressful, you know and I, I like, I put a lot of pressure on myself because you feel, **when you have a child that has no diagnosis, you know, you feel like you have to fight all of the time,** em to get him looked after because **you do actually have to fight all the time** and you know, in your day to day life, like you could have, like I have a list of- a stack of papers over there that I have to get through, **there's so many forms** that come through the door you know or different things that you have to reply to or contact people about or **chase up**, like you're **constantly chasing things up** and it feels **grossly unfair** when you have a little boy like Alexander who needs so much care and you know, there's so many things that **you're constantly looking for** and, em, you know you end up having to **just chase everybody** like even (.) even just simple things like getting him an appointment you know, now to be fair, I will

In the Extract 9.3 above, the strain and hardship in the need to fight appears reflected in the choice of wording used by Mary such as "fight all of the time" (line 704), "constantly chasing things up" (line 710), "grossly unfair" (line 710), "so many forms" (lines 707-708). Repetition of "chase" and "constantly" also appear to show the struggle. Judy similarly used the words 'fight' and the 'chase' in relation to looking for follow-up further to the diagnostic appointment ("had to chase and almost fight to get the answers to those questions", line 370).

In the extract below, Mary described the responsibility to fight as a "huge pressure" (line 454):

Extract 9.4

Mary / Alexander: "you should not have to fight for anything…I was putting huge pressure on myself"

447	P	couldn't eh- or he wasn't being made a *priority* until you, you fought
448		for him you know em, so that was kind of the main thing and I was
449		happier then, I could kind of relax a little bit, em you know and get that
450		done, em but again as a parent of a child- a very, very sick child, **you**
451		**should not have to fight for anything when your child's in ICU**, you know
452		they should be prioritised and these things should be done, em
453	R	Yeah
454	P	Because it's **huge pressure**, you know**, I was putting huge pressure on**
455		**myself, feeling very responsible** to get things done yeah you know. Yeah,
456		whereas **that's a medical thing and that shouldn't be my problem** or my
457		issue you know, em

In lines 456-457 of Extract 9.4 above, Mary expressed how she did not feel it should be her role to fight to get medical procedures done ("that's a medical thing and that shouldn't be my problem", line 456).

In the Extract 9.5 below, Mary referred to a phone line where you can speak with a doctor, as helping to alleviate some of the parent onus (e.g., "the doctor knows and you can relax a little bit…that makes a huge difference", lines 753-754):

Extract 9.5

Mary / Alexander: "he'll increase his medication over the phone…they trust you"

747	P	worried, you can ring, and they will then go to speak to the doctor and get
748		back to you and say, you know, he feels that's ok, you know, you can
749		just keep noting it or record some videos and send them into us or you
750		know, **he'll increase his medication over the phone**, you know and **they**
751		**trust you,** and you know, to do the right thing and then you're
752		reassured, and it takes away a lot of the stress because you've said it
753		now, **the doctor knows and you can relax a little bit you know and that,**
754		**that makes a huge difference**
755	R	Mmm
756	P	Yeah, yeah, yeah, **because it's too much to hold, as a parent**, you know
757		it is too much, em, but yeah Alexander started-or we were, he was sick or

In the following Extract 9.6, Mary spoke to further qualities in a doctor which help to reduce the feeling of responsibility on her (e.g., "I don't have to persuade him to do his best", line 376; "the ones that really care", lines 378):

Extract 9.6

Mary / Alexander: "the ones that really care"

376	P	know **I don't have to persuade him to do his best you know**. He is just
377		going to do it and that's a key thing in, in doctors that I found over the
378		years, is **the ones that really care you can relax a little bit because you**
379		**know you don't have to fight for every little thing**, you don't have to be
380		quite as on the ball with things, em

In contrast to Mary, who reported a release in pressure as the doctor's trust in her, (i.e., "he'll increase his medication over the phone, you know and they trust you", lines 750-751, as seen in the previous extract, Extract 9.5 above), Claire reported a pressure in knowing that the medical team will make changes to medication in response to parent reports only (e.g., "I'm like *really?* You're going to make this drastic drug change…based on, *just* what I've told you over the phone", lines 1024-1026) as seen Extract 9.7 below:

Extract 9.7

Claire / Rose: "I'm like really? You're going to make this drastic drug change…based on, just what I've told you over the phone"

1022	P	how much information do you need and everything was based on *just*
1023		what I was saying and then they'd say ok, well we might increase her
1024		Epilim then, **I'm like *really?* you're going to make this drastic drug**
1025		**change** that's going to have huge side effects for my daughter
1026		**based on (.) *just* what I've told you over the phone**, are you that
1027		trusting of my (.) history giving, you don't want to see her? Do you
1028		want to make an appointment? We could bring her up next week, we
1029		could bring her up in the morning, do you want to have a look at her?
1030		No, okay, and I remember this being, you know like, ***big* decisions were**
1031		**made,** new drugs were going to be started because I said Rose was
1032		having and and like I was ***verbally* describing seizures** over the phone,
1033		nothing visual^

In the Extract 9.7 above, Claire emphasized *"really?", "just", "big"* and *"verbally"* which seems to convey her disbelief or shock that the medical team would make changes to Rose's medication based solely on parental reports. In the Extract 9.8, below, again in direct contrast to Mary, Claire

identified the fact that the doctor may make medication suggestion based on parent reports or observations alone as “a *huge* pressure” (line 1041):

Extract 9.8

Claire / Rose: “that was a huge pressure”

1035	P	^em, then you can send in videos but you know, and I *did,* eh but yeah
1036		there was new drugs like and, and I'm glad, there was once or twice
1037		were I just said do you mind if we hold off for a couple of days and
1038		see and I'm really glad we did because there's actually, there's probably
1039		two lots of drugs in this house that we never started because^
1040	R	Okay
1041	P	^it wasn'- but **that was a *huge* pressure** on (gesturing towards self),
1042		it was nearly the opposite, it was like everything is on me, I was like
1043		I'd really love it if someone just threw an eye over her and you know
1044		in case I'm crazy and what I'm seeing is not what I'm actually seeing
1045		here so there was that, as well, em.

In the Extract 9.9 below, Claire alluded to what can be considered the primary juxtaposition encased within this subtheme of ‘*Maternal role and responsibility: balancing trust and onus’*. Claire polarised the “*great* relief” (line 987) in being listened to by doctors with “an awful lot of pressure” (line 989) felt on herself in medical professionals listening to a mother’s instinct:

Extract 9.9

Claire / Rose: “a mother knows…a bit of a catch 22”

983	P	Yeah, yeah but in [hospital], they kept, and many times they said, we
984		always take a mother's instinct, a parent's instinct, you know,
985		seriously, like a parent knows, **a mother knows** and they said we don't
986		dismiss that so if you're there going, there's something off and even if
987		we can't see it, we'll listen to you and I remember getting ***great* relief**
988		with that in [hospital] going 'thank god', **actually to such a point that (.)**
989		**that I felt it (laughing) put an awful lot of pressure on me** because
990		there'd be times, oh god I remember once she woke at about 3
991		in the morning or I looked at her in the dark or something at 3 in the
992		morning and she was asleep, at long last (closing eyes), and she did
993		something funny with her mouth (demonstrating) and the nurse was,
994		happened to be in, checking her 02 or something, and I said "go:d. does
995		her mouth look a bit funny to you?" and she looked at her and she went,
996		"no, not at all" and I went "yeah, no, maybe not". "Oh no Claire, if you
997		think (.) you see something funny, you're the mother, if that's your

998 instinct I'm just going to get the doctor to come and review her and I'm
999 like "no::: (throwing head backwards) please don't, it's 3 in the morning,
1000 she's asleep. The doctor comes in and has to wake her and check her
1001 and make sure she doesn't have a droop and nothing has happened and
1002 is she ok and is she lifting both her limbs, and I just went, oh my god, I
1003 wish I had said nothing now (laughing) because sometimes that, that
1004 ***too* much pressure**, the opposite I felt, that that they really (smiling)
1005 which is great, I wouldn't- no I wouldn't change any of that but it, it
1006 can be **a bit of a catch 22** as well so (smiling)

In lines 1003-1004 in Extract 9.9 above, Claire described how, when in hospital, she almost regretted bringing her query to the nurse's attention. She named the medical team weighting so much on maternal instinct was almost "*too"* much pressure. Claire concluded that she wouldn't change being listened to but summarised this phenomenon as "a bit of a catch 22 as well so" (line 1006). The catchphrase "a bit of a catch 22", as referenced in the introduction to this chapter, summarises the feeling embodied by this subtheme.

Mary detailed her role in monitoring and managing Alexander's symptoms. Mary gave multiple examples where she reported it was her observations and pushing for things to get done which led to doctors taking actions that were of benefit for Alexander's care (such as, discovering Alexander was in pain and 'pushing' for an MRI which led to the identification that Alexander had a congenital muscle condition[27]). In the extract below, Mary describes how she monitors and shared her observation with the medical team (e.g., "the video is a powerful thing", line 847):

Extract no. 9.10

Mary / Alexander: "the video is a powerful thing"

844 P experience, I recorded it, and I sent it to his neurologist, so he could see
845 how violent this was, you know this wasn't like I've got a bit of, there's
846 some- you know a headache, this was severe, em and I've learnt that over
847 the years that **the video is a powerful thing** and it feels wrong to be
848 recording your child as they box themselves in the head and scream in
849 pain but you have to do it because it's the only way that they can really
850 get an idea, em, and you know in that instance, they, em, you know

[27]Mary's reference to her monitoring and reported of Alexander's health symptoms to medical professionals leading to positive steps in management of these symptoms bring to mind the topic of patient led 'lay-diagnosis' and patient elicitations during discourse interactions in medicine (see *2.2 Concept and construction of 'diagnosis': what is 'diagnosis'?).*

Mary's remark lines 846-847 as indicated, "I've learnt that over the years" ("over the years" can also be found in extract no. 9.5, lines 377-378) resonates particularly with the next subtheme within this GET, '*Finding your voice, lessons learned'.*

In the following examples, I will provide discussion of where the mothers addressed their perceptions of professional or parent roles. Robyn advised her and her husband "did *everything* that the early intervention centre (.) provided for us" (lines 633). Robyn continued to describe the therapists are telling you "what you have to do on a day-to-day basis" (line 638) and that in "you're not getting that silver bullet in that one-on-one meeting" (lines 657-658). Robyn appeared to have been reflecting on her experiences in comparison to the experience of other parents she encountered who she described had a negative attitude towards group trainings. Robyn's understanding of the concept of therapist, as described using the metaphor of the "silver bullet" (line 657), may be an extension of the earlier metaphor of "magic pill" (line 89) quoted by Robyn in reported **s**peech from dialogue during the geneticist meeting ("he was like you're not going to get a magic pill for this, so it doesn't matter what kind of genetics he has, it's not a, I can't fix it for you", lines 89-90). For Robyn, she advised "the appointments aren't going to do it, you're the only one who's going to do it" (lines 687-688). In conclusion, Robyn attested to the importance of the parents role in early intervention. This may be understood as extending to paediatric therapy services more generally.

It appears Olivia shared Robyn's belief on the prime role of parents in therapy as can be evidenced in her statement, "it's definitely true like parents are the best therapists and stuff like that because obviously there's so much you can do at home" (lines 780-781). Olivia proceeded to express there is a limit to what parents can, and should, be expected to do, "but at the same time there comes a point where you're *not* a professional, you're *just* a parent" (lines 781-782). Olivia returns to continue this idea later on the division between parent and professional roles "it's not, should never be our, it, it has felt the entire time that they expect us to figure it out, like that's literally, it's kind of, yeah that's totally been the feeling like" (lines 1067-1069) and "I'm not a professional (laughing)" (lines 1089). Similarly, Mary, as introduced in extract 9.3 (lines 450-452) above, referenced "as a parent of a child- a very, very sick child, you should not have to fight for anything when your child's in ICU, you know they should be prioritised and these things should be done".

In the Extract 9.11 below, Robyn discussed caveats of when the parent taking on role of therapist can go too far (as introduced earlier in the final subtheme of Chapter 8, 8.4 'Learning to live: "We just need to live our life now"):

Extract 9.11

Robyn / Nathaniel: "just a normal little boy"

694	P	BUT then, on the other hand, he's also not a job, so you do have to maybe not
695		make it all about OT and physio as well, **he's just a normal little boy**
696	R	Yeah. It sounds like you've thought of both sides so much, and have^
697	P	We try
698	R	^the balance, yeah
699	P	Because I did fall into, wrongly, in the first year or two, that it was *ALL*
700		*ABOUT THERAPIES,* and then you forget, that he's just a little boy as well
701		who actually wants to just swim in the pool, NO, SAY THIS, THE Lámh WORD
702		for swimming pool (laughing)

Robyn's reference in the extract above, to the fact that Nathaniel is "just a normal little boy", an idea which she repeats, "he's just a little boy" (line 700) may serve to remind parents that whilst parents have a role in supporting their child's development, it is important to maintain *just being parents* to their child. This contrast appears to represent another tension pair of 'parent and therapist', within the subtheme of '*Maternal role and responsibility: balancing trust and onus'.*

9.2.1 *Gender and parent roles*

Within their accounts, Judy, Claire, and Mary offered insights into their distinguished roles as mother or parent within their situation. These differences will be explored within this subsection.

For Judy, she described her role as *"firefighting" (*as introduced in Extract 7.2 and used within the title of the subtheme '*Searching or "firefighting" moment by moment?").* In examining the surrounding dialogue in relation to how Judy was living prior to the 'diagnostic moment', she described "I felt I was firefighting you know, my husband's kind of health issues, my son's health issues and I was kind of working full time to keep the whole show on the road as well with the, you know, with the family and all that" (lines 901-904).

In the Extract 9.12 below, Judy provided reflections on her perception of different understandings between herself and her husband with regards to Declan's diagnosis (e.g., "I probably have a better grasp on the implications of it, than my husband would, because he hasn't been at the appointments so it's not real to him" lines 1140-1142):

Extract 9.12

Judy / Robyn: "the woman gets the whole pregnancy thing and then the Dad…until he sess the heartbeat…it's just a piece of stick with a line on it"

1140	P	Yeah, it is and I think, like, I would say I probably have a better grasp of the
1141		implications of it, than my husband would, am because he hasn't been at
1142		the appointments so it's not real to him, I think **it's a bit like you know in**
1143		**pregnancy when like you know the woman gets the, you know the whole**
1144		**pregnancy thing and then the Dad is there and until he sees the heartbeat**
1145		maybe on the scan, it's just **you know a piece of stick with a line on it**, you
1146		**kind of *know* your partner's pregnant but you don't kind of *get* it until you**
1147		**see the heartbeat,** and it's a bit like that for me at the moment, I think it's
1148		like I get it, and I know it, and I feel it^
1149	R	Mmm
1150	P	^but I don't think at the moment, like my husband, like **he *knows* it but he**
1151		**doesn't *really* get it,** do you know what I mean

In Extract 9.12 above, Judy extends the account of her own and husband roles to more general roles of "the woman" and the "Dad". Judy appeared to demarcate different levels of '*knowing'* across parent roles within their genders. It seems, Judy used the comparison of the experience of a mother reading a positive pregnancy as a metaphor for diagnosis (e.g., "I think it's a bit like you know in pregnancy…the Dad is there and until he sees the heartbeat maybe on the scan, it's just a piece of stick with a line on it", lines 1142-1145), in terms of varied gender reactions in *'knowing' versus 'really getting it'*. In relation to Declan's rare diagnosis, Judy discussed how she interpreted her husband "*knows* it but he doesn't *really* get it" (line 1150-1151).

For Mary, in response to my prompt as to how Alexander's Dad, Jason, felt about his role, Mary discussed the differentiated role both parents play in relation to care for their child Alexander, as shown in the extract below (e.g., "he more took on the role of supporting me and looking after me", lines 480):

Extract 9.13

Mary / Alexander: "I very much took over Alexander's care…Jason…more so supported me"

458	R	And how do you think Jason felt about his role at this time or (..) was it
459		something you'd discussed
460	P	(laughing) no, I tend to (laughing) take over everything so I do, I do tend
461		to take over these things like it's my, eh, like I've quit my career basically
462		to become Alexander's carer you know and I suppose I see it as my job

463 and I think in our relationship as well I am the more *forceful* one, I am
464 the more outspoken one and, and Jason wouldn't really be you know em
465 R Mmm
466 P You- you know, it's just not his, not in his nature? to- and It's not in my
467 nature either^
468 R ^Different personalities, yeah
469 P Yeah, it's not in my nature either to be honest but I've found, I- like I had
470 to do it em but I very much, yeah **I suppose I very much took over**
471 **Alexander's care** and maybe didn't even give him a chance to (laughing) to
472 do it you know (laughing)
473 R …
477 P Yeah, yeah. You fight, like you fight for your child and you know **Jason**
478 **was, he more so supported *me*,** I would say
479 R Ok
480 P So he more so took on the role of supporting me and looking after me and
481 R Mmm
482 P And you know I in turn looked after, **I would say** that's kind of the way
483 we, we tackled it you know

On examining the Extract 9.13 above, it appears Mary viewed she assumed the role to fight for Alexander and her husband took on the role of supporting her. Mary's use of the phrase "I suppose" (line 462) and "I would say" (line 478) may imply a sense of live, in the moment, sense-making in real-time in dialogue with the researcher. Mary provides insight that she has left her job to become the carer for her child and suggests she views it as her "job" ("I've quit my career basically to become Alexander's carer", lines 461-462).

Similarly, in her account, Robyn described herself and her husband took carer's leave to make time for supporting Nathaniel's development early on in his infancy (lines 682-692). Robyn referenced this as was a "huge sacrifice" (line 690) for them in terms of financial and career implications.

Claire also provided insights into a differentiated parent role. In relation to the earlier *GET of 'Entering the world of diagnosis: "There's nothing wrong but everything is wrong"',* Claire reflected that at the time when "there was something niggling" (lines 48-64) at her about their daughter Rose, it was *her* whom her husband was worried about. Claire continued that they did agree that there were things they were expecting Rose to be doing which she wasn't. Claire described a "complete swap" (lines 821-822) in the roles her and her husband occupied at the point when she identified Rose as having a seizure and "hospital mode kicked in" (line 805), as exemplified in the extract below:

Extract no. 9.14

Claire / Rose: "hospital mode kicked in…I know this person"

804	P	know but it's going to be okay, we'll, this will be okay." And then I saw
805		the seizures and like that kind of (.) **hospital mode kicked in**, and I just
806		went we need a referral to the GP immediately so that the GP can send
807		us to A&E. Tom, you know, we need to do this and we were in my
808		mother-in-law's house, packed her up, brought her home, I was waiting
809		for the GP to ring me back, eh, Tom, I just, you know I just went into
810		sort out the car, we're going in the car and I-he, we were gone over in
811		two different cars so did a few jobs in the farm, he was home maybe
812		twenty minutes behind me. By the time he got home, I had a bag
813		packed for Rose, I had a bag packed for me, I'd never done this before
814		but I [had all her clothes^
815	R	you were in (.) mode]
816	P	^I had all my clothes packed. I had everything I needed and then let's go.
817		Yeah and we were in the car and I remember sitting in the car, in the
818		back seat behind her in the back of the car and **Tom was, "oh my god,**
819		**oh my god, oh my god"** and I was like she's fine. Tom, she's fine. She's
820		breathing, you know, I said she's not in any distress here, she's
821		absolutely fine, **don't panic,** drive the car and we did a complete (.)
822		swap (gesturing) (laughing) I felt like **I (interjections) was like I know this**
823		**person, I know this, we have an issue, and we've gone into A&E** and I
824		remember giving a history and the doctor was, I wouldn't have been

In extract no. 9.12, Claire recounted her and her husband Tom's dialogue on the journey to the hospital. Claire suggested Tom was panicked ("Tom was, 'oh my god, oh my god, oh my god", lines 818-819; "don't panic", line 821) in comparison to herself, who was it appears was activated by the incidence to switch into health care professional mode (e.g., "I was like I know this person, I know this, we have an issue, and we've gone into A&E…", lines 822-823). Claire is a healthcare professional (as referenced previously in the introduction to this chapter and within Claire's story summary, *6.2 Participant Story Summaries).* Claire's reflections on first realising Rose was having a seizure (as previously introduced within the subtheme of 'Dismissal and maternal questioning: Being "pooh-poohed" - "I must be going crazy", see extract no 7.24) appears to have been the impetus for Claire's switch into hospital mode. Further examples of where Claire shifted between these two positions of *mothery* ("mothery mode", line 782) and hospital mode will be explored in the subsection below.

9.2.2 Switching between mother and therapist mode

Throughout her account, Claire makes numerous references to oscillating between mother and professional roles, in relation to responding to Rose and within healthcare discourse.

9.1 Researcher Reflection

For me, Claire's references to 'switching' between professional and parent roles, plotted throughout her account, conjures up a parallel to the practice of "Bian Lian" within Chinese theatre. "Bian Lian" translates to "face changing" (Jernigan et al., 2009, p. 44) and is regarded as a technique used within Chinese theatre, notably Sichuan opera, to show the characters mood (Jernigan et al., 2009). Jernigan et al. (2009) discuss the art of changing masks may be considered reflective of the capacity of human beings to house multiple personalities.

In the Extract 9.14 (previously presented above), Claire's comment "I know this person" (lines 822-823) may be interpreted to mean she knew her identity as a health care professional. Her familiarity with this role might be viewed in contrast to knowing her position as a mother for, as previously referred to by Claire as "just the whole new mother, big change of identity, all of a sudden" (lines 53-54, as seen previously in Extract 7.10).

In the extract below, Claire continues to describe the events surrounding when she first identified Rose was having a seizure, as initiated in Extract 9.14 above, from the point of arriving to hospital (e.g., "we talked very much like there was a case presentation going on…we were using acronyms", lines 833-836):

Extract 9.15

Claire / Rose: "I snapped well out of therapy mode…"

833	P	knew me alright and we'd you know, we talked very much like there was
834		a case presentation going on about the patient on the bed, and I
835		remember talking like that, until the reg who I was talking to, you know
836		and we were using acronyms like any LOC, I was like no LOC, you know
837		and I could feel myself (gesturing)
838	R	You were in a clinical mind, dissociated almost from parent mode and
839		full [X] therapist
840	P	Yeah, yeah, and then the registrar was asking and I was giving the
841		history about stopping smiling, stopping this, that and the other and you
842		know it's the classic infantile spasms presentation of eh, developmental
843		regression and then the classic seizure type and as I described everything,
844	P	I could see him, he was [xxx] he had just finished his paeds (.) you know

stint after six months and he's sitting back and he's going **"oh, interesting, interesting, interesting"** and **I *sna:pped* well out of [healthcare professional] mode** and back into, actually I think I went into **[healthcare professional] tutor? Kind of mode** (laughing)

R Practice educator hat (laughing)

P Don't you *DARE* refer to my child or any patient as *interesting.* Interesting. Don't you dare, I said this is my child. I don't want to hear it's an interesting case to you and **I *dissolved* back into tears again (laughing) and all of a sudden I was out of that mode** and I was back into "oh, I don't want my child to be *interesting.* I want my child to be really boring" (laughing) you know, am, so that (laughing) yeah, kind of

In the Extract 9.15 above, Claire described how she switched out of hospital mode, "I *sna:pped* well out of [healthcare professional] mode" (lines 846-847) in response to the doctor's response "oh, interesting, interesting, interesting: (lines 845-846). The elongation and emphasis in the word snapped might echo the manner in which she halted roles. Claire also introduced "[healthcare professional] tutor? Kind of mode" (lines 848). Claire concluded the above recollection, with a description of a shift back to "mothery mode" (line 782) with "I *dissolved* aback into tears again (laughing) and all of a sudden I was out of that mode*"* (lines 852-853).

The above extract, specifically "oh, interesting, interesting, interesting", lines 845-846, will be discussed in more detail in Chapter 10, '*Etching of the mothers' inner worlds: "I'll never forget"'* GET 4.

9.2 Researcher Reflection

I wonder if my "insiderness" (Hellawell, 2006, p. 490) with Claire as a fellow healthcare professional facilitated her in sharing and narrating her experiences of switching between healthcare professional therapist, tutor and "mothery" (line 782) modes. This consideration may provide further support for the notion that researcher subjectivity which is recognised as a "fundamental resource for IPA" (Braun & Clarke, 2020, p. 41).

On another instance within her account, Claire described a switching of roles between "the crumbling mother" and "work mode" (lines 189-191). Claire referred to commencing enteral feeding ("we had to put an NG tube in", line 225) as "one of the hardest things" (line 227). Claire described what this was like given her professional identity "and you're sitting, the [healthcare professional] mother, watching 02 dipping right down into the 70s, her coughing and spluttering, and I remember saying to the nurses, this child is not safe to feed" (lines 232-234). Claire recounted

her opening a dialogue with a nurse around the "clear risk of aspiration[28] and signs of aspiration that I can't stop seeing" (lines 262-263).

In the following Extract 9.16, Claires recounts an encounter with a nurse whose words "*really* got under my skin" (line 266). Further references to changing roles can be seen in this scenario (e.g., "switching off the mammy part (gesturing pushing to the side with both hands) and just turning on", lines 273-277):

Extract 9.16

Claire / Rose

271		adding thickener and then we'd have a videofluroscopy and I just
272		remember, about 2 or 3 in the morning, and I was like that kind of
273		(closed eyes) (laugh) switching off (eyes open) the mammy part
274		(gesturing pushing to the side with both hands) and just turning **on** and I
275		go, really? that's what the speech therapist is going to do, and how
276		exactly do you suppose that this woman is going to thicken my breast
277		milk^

In extract 9.16 above, "on" (line 274), in the context of the surrounding dialogue appears to relate to therapist mode. The extract below is a continuation of the same scenario, and depicts the point at which Claire returned "to being Claire" (line 292):

Extract 9.17

Claire / Rose: "please don't make me make that decision"

279	P	^because (laugh). Explain that to me, how is she going to thicken all
280		these medications we have to syringe into my child, just explain that one
281		to me and eh, I kind of got very (.) *irate* that the the, notion of, that the
282		(dysfluency) that this nurse wasn't looking at the problem so the shift
283		changed the following morning and one of the angels came on and I
284		was still in my (sitting up straight, demonstrating) "look it"^
285	R	[xxx] angry
286	P	^"where is", and I could feel myself doing it and and the, the nurse
287		looked at me and she just said, [Mum] do you think we need an NG tube
288		and I just (eyes closed) burst into tears and I said to her (eyes open)
289		please don't make me make that decision^

[28] Passage of food, drink, or saliva beyond the level of the vocal folds and entering the airway (Irish Association of Speech and Language Therapy [IASLT], 2021).

290	R	Mmm
291	P	**^please don't make me make the decision** to put an NG tube in my child,
292		and **I went back to being Claire**. I remember- can actually remember the
293		moment of just going, ***please*** **(shaking head), don't make me make that**
294		**decision**. And, em, she said you're right, Claire your daughter needs an

In the Extract 9.17 above, we may take Claire's comment "and I went back to being Claire" (line 292) to mean she is rejecting her professional role and re-establishing herself as mother to Rose in this instance. The repetition of "please don't make me make the decision", with emphasis on the second repetition and recalled shaking of her head (see line 293) may signify the desperation in which she wishes to relinquish the decision-making authority regarding this medical element of her child's care.

9.3 Researcher Reflection

Data and analysis relating to the experience of '*Switching between mother and therapist mode'* might be best considered as 'divergence' within the group-level subtheme of '*Maternal role and responsibility: balancing trust and onus'.* It felt significant for me to include these findings within the current GET. I made this decision in order to most faithfully illuminate the experience, and meaning, of the phenomenon for the participants. IPA, as previously explained within Chapter 4, Methodology, is devoted to maintaining attention on the idiographic experience whilst presenting group-level themes (Smith et al., 2022). Including these findings relating to a smaller number of participant's, appears congruent with fulfilling Smith et al. (2022) description of the aims of IPA research. The above reflection provides rationale for including subsection *9.3.1 "Living in hospital": maternal experience of becoming institutionalised*, in relation to Claire and Mary's experiences within the subsequent subtheme of '*Finding your voice, lessons learned'.*

9.3 *Finding your voice, lessons learned*

The subtheme of '*Finding you voice, lessons learned*' concerns the phenomenon of maternal learning acquired through the mothers' experiences within healthcare systems in terms of knowing how to best advocate for their child and their child's care. I combined two phrases used by Mary ("finding your voice") and Judy ("lessons learned") in their accounts, respectively, to form the name of this subtheme. Whilst the earlier subtheme introduced what mothers felt constituted their perceived roles and responsibilities, this subtheme covers more specific examples of knowledge gained and applied in fulfilling the assumed mother roles. Both subthemes are closely

linked and together represent the overall GET *'The world of mothers in healthcare systems: "You adjust to it because you have to, because your child needs you to"'.*

Towards the end of Mary's interview, as with all participants, I prompted Mary to consider was there anything else that hadn't been covered which she felt she would like people to know or that had been an important part of her whole journey in relation to the research question. Mary responded, "I think just understanding how (.) hard the appointments are maybe for parents, you know, especially with these complex kids, em and you know (.) just you need (.) doctors that (.) care." (line 1231-1232). The pausing may be considered significant here in highlighting Mary was thinking and reflecting live, in the moment. The pauses may also function to add power and conviction to her words. Directions and considerations to what constitutes the *'good doctor'* can be seen to feature throughout Mary's accounts and constitute a personal experiential theme for her (some of which have already been seen, e.g. Extract 9.6). When stating what she needs from those supporting Alexander's care, Mary attested "you need someone that, that treats Alexander with the value that he deserves to be treated with" (lines 1237-1238). A lot of Mary's dialogue on what she wants from the *'good'* doctor appear to be based on what she has learned through her experiences to date with Alexander.

In the following extract 9.18, Mary recollected times when she interrupted to speak up for her son (i.e., "I say just stop what you're doing", lines 1276-1277; "we're not doing that again", line 1277):

Extract 9.18

Mary / Alexander: "you really find your voice"

1273	P	idea how to do it and therefore he just brute forced a child's mouth open
1274		and that then has massive repercussions for the GP who has to do it
1275		quite regularly and we've worked really hard to on that, and you know,
1276		there's times **now** where I do speak up, and **I say just stop what you're**
1277		**doing** and, you know, **we're not doing that again** or for blood tests,
1278		we have huge issues with blood tests, em, especially in [county] and I've
1279		had to, I've had big rows with doctors (laughing) you know, where
1280		they'll let the people that are learning do blood tests and I just put a
1281		stop to that **now**, straight away, you know, **you really find your voice**
1282		**and you have to speak up for him**, but, em as well it would just be good
1283		to see more (.) training done, on a, on a^

In Extract 9.18 above, the use of "now" (line 1276, 1281) might hint at the idea of learning how to *fight* for your child. This view is also supported by the phrase "you really find your voice" (line 1281).

In the extract 9.19 below, Mary described how she witnessed her mother fighting for treatments that saved her sister's life ("my mother fought and fought for to get that treatment", lines 389-390):

Extract 9.19

Mary / Alexander: "I very quickly found my voice"

384	P	fighting with the system in there and **I very quickly found my voice**, em
385		I had been through a eh (.) medical thing with my sister so my sister
386		nearly died when I was twenty and I was one of her main carers in
387		hospital so through quite a, a difficult setting before you know em, so
388		she was flown to [hospital], she was, she was on death's
389		door you know and **my mother fought and fought em for her to get**
390		**that treatment** and that saved her life and I'm going to see her this
		…
393		Yeah, yeah, so I suppose I had witnessed it you know and I had been in
394		the depths of ICU before my sister

At the start of the above extract, we see Mary's comment "I very quickly found my voice" (line 384) which I used to denote part of the title of this subtheme. In the following extract, spoke to how her earlier experiences in healthcare systems in relation to her sister may have influenced how she managed interactions within healthcare systems with regards to her son ("I knew I need to fight here", line 402):

Extract 9.20

Mary / Alexander

396	P	Yeah, yeah, so that's probably why I did find Alexander's, eh, stay in hospital
397		hospital so traumatic because I had had PTSD from [my sister's] stay but I
398		never actually realised I had it, nobody ever told me I should seek
399		counselling or anything and then when Alexander was born it just brought
400		*everything* up for me again, em, and I have since now had a lot of, a lot
401		of help with that, em, but I think that's maybe, it- **it hindered me but it**
402		**also *helped* me** because **I knew I need to fight here** you know and I need
403		to make sure he's looked after and I did you know but it's such a pressure
404		and such a stress on parents who have a very, very sick child you know
405		*knowing* you have to fight for his basic (.) needs? you know and that
406		was the case, em

There is alliteration in Mary's description of '*hindering'* (line 401) and '*helping*' (line 402) in the Extract 9.20 above, in relation to reflecting on the potential dual influence of her past experiences with her sister on how she adjusts and manages her son's health. The polarity between '*hinder'* and *'help'* appears similar to me to the juxtaposition of '*trust'* and '*onus*' as represented within the previous subtheme of '*Maternal responsibility: balancing trust and onus'.*

9.4 Researcher Reflection

Within Mary's account, she referenced that her sister "nearly died" (line 386) and that Mary "was one of her main carers in hospital" (line 386-387). Mary volunteered this information in relation to learning how to fight for her son and the grade of trauma she endured in relation to Alexander's birth and health. Mary's sharing of this information may be seen to exemplify the notion of 'Dasein[29]' (Heidegger, 1927/2008) and "the universal process of horizon-formation and fusion" (Gadamar, 2004, p. 578). Gadamar's (2004) fusion of 'horizons of understanding' appears to maintain that historical events contribute to a person's understanding of current experiences and that the influence of prior experience on the present can only be understood in hindsight. From my perspective as a researcher, acknowledgement of the notion of 'Dasein' directs me to consider how Mary's experiences and sense-making in relation to Alexander, in the context of his medical needs and an undiagnosed condition, were influenced by her prior life experiences which comprise her horizon of understanding. Although I cannot explicitly detangle the exact influence of the past on how Mary lives or understands her present, I can recognise the past as a component which contributes to Mary's sense-making of the current phenomenon being studied.

Elsewhere in her narrative, Mary described other things she learned from her experiences navigating healthcare with Alexander, including "I keep daily diaries because I've had to at this point" (lines 682-683). The function of these diaries for Mary was for symptom monitoring. Mary also advised she learned being "firm and calm and polite" (lines 805) and "politely persistent" (line 796) works best in obtaining appointments for Alexander.

In relation to Declan having a diagnosed rare disease, I specifically asked Judy whether having a 'named' diagnosis had changed her role in healthcare appointments. Judy explained having a

[29] 'Dasein' comes from the phenomenological influence on IPA which acknowledges owing to the notion that people cannot be detached from pre-existing experiences (Heidegger, 1927), as introduced in *1.4.1 Novel Accounts.*

named diagnosis altered her part, in the sense that "you have to constantly remind people" (line 833). Judy declared "I'm going to like a foghorn on repeat going into appointments" (lines 836-837).

In the extract below, we see in context Judy's use of the phrase "lesson learnt" (line 793) which contributes to the name of this subtheme. Judy appears to be detailing how she now navigates appointments to accommodate for healthcare professionals lack of co-ordinated or transparent care. This extract follows Judy's reference to the situation of when she brought Declan for blood tests at the GP which turned out to be redundant as Declan's levels had already been checked (as previously introduced in the subtheme '*A fragmented "jigsaw" puzzle: Lack of transparency and co-ordinated care'* within the first GET, *'*Entering the world of diagnosis: "There's nothing wrong but everything is wrong"):

Extract 9.21

Judy / Declan: "I'm going to be that parent now"

793	P	So, kind of in my own head, I was like, lesson learnt Judy, ask. You know
794		just be, just be, be annoying, and just ask them what bloods are you taking,
795		you know and be, be, you know **I'm going to be that parent now** at every
796		appointment with my little notebook (laugh) you know, am, but I think
797		I have to, am, so just ask like what bloods are you actually, you know what
798		is the purpose of these, besides just ***having faith*** in what they're doing,
799		you know, **kind of blind faith kind** of

In examining Extract 9.21 above, it appears Judy has learnt her previous approach of "*having faith"* (line 798)*,* "kind of blind faith" (line 799) was not enough. She now has learnt to ask what is being done and why.

In the following extract, Judy reflected on her regrets in "not being *pushier* for answers and just accepting*"* (lines 843-844) in relation to asking questions surrounding Declan's health, compared to what she knows now, having learnt from her experiences navigating health care systems (e.g., "I just think you have to be so much more proactive in asking the question", line 851):

Extract 9.22

Judy / Declan: "I do regret not being pushier…hindsight is, is invaluable"

840	P	Yeah, like I do, like as I said, I do wish .hh you know, when he was sick that
841		you'd pushed for more answers, you know, am, you know .hh, I wasn't
842		just **that hypochondriac parent** whose child had a sniffle and I was running
843		to the hospital you know, am, more, like **I do regret not being *pushier* for**

844 **answers** and just you know, accepting, you know I mean even, the the, you
845 know, being told back that he was hospitalised for pneumonia in February or
846 …
851 I just think you have to be so much more proactive in asking the questions
852 and it's very hard to ask when you don't know what you're supposed to be
853 asking, **hindsight is, is, invaluable in knowing well I should have asked** that
854 you know, am, but I didn't know to ask, so how do you, ask something
855 that you don't know you're supposed to be asking for

In the above extract, there is further signposting to an established parent 'type', "that hypochondriac parent" (line 843) in addition to Judy's earlier reference to "that parent now at every appointment with my little notebook" (Extract no. 9.18, lines 795-796). The use of the demonstrative "that" may imply a universally understood and accepted construction. At the end of the extract, Judy evaluates that "hindsight is, is invaluable in knowing well I should have asked that" (line 853). This evaluation appears to encapsulate the essence of this subtheme in knowledge gained in looking back on experiences with your child in healthcare systems.

9.3.1 Maternal trauma

Mary is the only mother in the study for whom maternal trauma constitutes a personal experiential theme (namely '*Maternal trauma and individual coping: "you adjust to it because you have to because your child needs you to"'*). Mary particularly described the trauma for her that ensued in the event of Alexander's birth (e.g., "we're going to section you" line 32; "where's my baby, what's wrong you know, what's happening" line 62). Further analysis surrounding Alexander's birth will be presented in the following chapter, Chapter 10, *'Etching of the mothers' inner worlds: "I'll never forget"*, GET 4*).*

9.5 Researcher Reflection

There are many references to death and variations of morbidity laced throughout Mary's account, in reference to her prior trauma from her sister's illness ("my sister nearly died", lines 385-386; "she was on death's door", lines 388-389) and past events and fears for the future for Alexander. Some examples are represented in the table below.

Trauma through endurance of past events	**Fears for the future**
"yours was upstairs effectively dying" (line 136)	"is this ultimately going to be the thing that takes him away" (line 858)
"Alexander was dying at that point, there was absolutely no doubt about it" (lines 299-300)	"is he doing to die from this" (lines 854-855)

"I think this child is going to die" (line 303)	"will he die young?" (lines 930-931)
""deteriorate?"…as in, die like." (lines 320-321)	"ultimately the things that are going to put Alexander in big trouble" (lines 1210-1211)
"I was really afraid then that he was dying" (lines 1173-1174).	

There is a sense of 'impending doom' and 'watchful waiting' in Marys fears for the future, in association with no diagnosis, that transcends her account. Within concerns for the future, Mary shares how she fears are the emergence of a new symptom the signal of the start of the end for her son? The depiction of the mother whaling as she holds her dead son in the study 'Mother and Child' within Picasso's 'Guernica' (1937) for me symbolises the grave parental trauma and fear of morbidity captured by Mary in this account. This depiction is particularly palpable in the lines such as "you've just taken my baby away" (line 66), "is he going to die from this" (lines 854-855).

In following extract, Extract 9.23, Mary recounted the trauma for her, surrounding Alexander's birth:

Extract 9.23

Mary / Alexander: "the complete opposite of what I wanted"

32	P	you know "we're going to *section* you" which to be honest now for me,
33		it still affects me em (.) his birth. It was very traumatic for me, em,
34		because I really wasn't- it was like **the complete opposite of of eh what**
35		**I *wanted*** y'know *to* happen basically
36	R	Yeah
37	P	I wanted no medical intervention and this nice natural birth and it turned into
38		you know (laughter) the complete other extreme em *which* I know can
39		happen for any child really you know even if they're not unwell you know that
40		kind of thing can happen but I suppose I hadn't allowed myself to really
41		kind of think about that happening which was may be my own (laughter)
42		doing (laughter)

Early in the opening of her narrative, Mary introduced the events of his birth where she alluded to the fact that things did not go as expected (i.e., "I had a very normal pregnancy...I went into labour and I had great plans for a natural birth…all these, wonderful ideals (laughter) and then I went into the hospital…and thing turned then very quick", lines 13-20). In Extract 9.23 above, it appears Mary

attributes the fact that she was unprepared and that the situation was “the complete opposite” (line 34) of what she wanted as adding to the trauma for her. This interpretation is supported by what Mary said later in her narrative (i.e., “…it wouldn’t have impacted me if I could have prepared myself in some way”, see lines 71-75, in the later Extract 9.25, below).

In the following extract, Mary reflected back to how she responded to the unexpected events surrounding Alexander's birth (e.g., “it was a really, kind of a sobering moment”, lines 178):

Extract 9.24

Mary / Alexander

178	P	Yeah, yeah, yeah and you know **it was a really, kind of a sobering moment** of
179		you know we're in trouble here you know em but at the same time you
180		kind of eh go into some kind of another *land* in your mind because it's so
181		overwhelming you know that **it becomes NORMAL very quickly** you know
182		when you're in these settings and your new normal, **you adjust to it**
183		**because you have to em, because your child needs you to** you know

In the extract above, we see the surrounding context for Mary’s reflections which make up the name for the current GET *(‘The world of mothers in healthcare systems: “You adjust to it because you have to, because your child needs you to”’),* ‘“it becomes NORMAL very quickly…you adjust to it because you have to em, because your child needs you to” (lines 181-183). In Extract 9.23, Mary attested to how the realm of normalcy transcended quickly through the experience (“you know we’re in trouble here you know em but at the same time you kind of eh go into some kind of another *land* in your mind because it’s so overwhelming you know that it becomes NORMAL very quickly” lines 179-181).

In the following extract, Mary further recalled Alexander’s birth and the surrounding events as “really scary” and “really traumatic” (line 64). Mary expressed the “not knowing beforehand and that shock” (lines 65-66) as having compounded her trauma. She also attributed the setting as having contributed to the trauma of the event (“if you’re sitting in a doctor’s office and someone tells you something shocking, you’re not quite so vulnerable as you are, in an operating theatre”, lines 79-81):

Extract 9.25

Mary / Alexander: “not quite so vulnerable as you are, in an operating theartre”

64	P	And it was **really scary, it was real- really traumatic** and its had a-that that
65		whole element of it had a *massive* impact on me, the not knowing beforehand

and that shock of you've just taken my baby away and I don't even know, I
I haven't seen him or what's wrong, or anything and that still affects me
today in in a big way, even though I've had counselling for years and I'm
still eh I still do counselling em^

R Yeah

P ^psychotherapy like I still, I still am affected by that you know and and I always
kind of stuck to my guns about-if anything had been caught like I wouldn't
have had that same trauma (.) em you know **it wouldn't have impacted**
me, even though it still would have obviously been a very hard experience
it wouldn't have impacted me if I could have prepared myself in some way
you know

R The shock

P Yeah [xxx] **the shock** yeah yeah and in that setting as well you know it's
different **if you're sitting in a doctor's office and someone tells you something**
shocking, you're not quite so vulnerable as you are (laughing) in an operating
theatre

In terms of narratability, in the extract above, Mary appears to seamlessly link her evaluations from past and present moments within her account. As evidenced in her summing up that the events "*had* a massive impact" on her (line 65) and that she "still" is affected (line 71). The statement that Mary continues to be affected also shows the enduring trauma for her. In lines 71 -76 within the extract above, Mary attested to her belief that if she could have known to expect something was not right in advance, she could have prepared, and Alexander's birth would not have been as traumatic. It appears Mary's general belief is that knowledge, even if it is 'bad news', is helpful in preparing for dealing with the outcomes.

9.3.2 "Living in hospital": Maternal experience of becoming institutionalised

As discussed in the introduction to this chapter, both Mary and Claire offered insights into their experiences of becoming institutionalised through their time living in hospital with Alexander and Rose, respectively.

Within her account, Claire expressed "you just get into full institutional mode in [hospital], when you're in there, and it's (.) the weirdest experience, you know, you're in a, literally a *glass box*" (lines 377-379). Claire offered two detailed anecdotes which she used to showcase the effect of being institutionalised. The first example is about another parent on the ward and needing to be there (where *there* relates to being in the room with Rose), in the extract below:

Extract 9.26

Claire / Rose: "full institutional mode" (line 377)

384 P Open view, you're, you're you know, a pull down bed and **I *lived* there**
385 **with Rose**, like I was living in that room with her, because you know, we
386 were on two different wards, I think one is 24 (.) **fish bowls**
387 (gesture), one is 17, one parent toilet, for all of them and you're,
388 you know, you're, you just become institutionalised, you know and you're
389 watching and you're going like there was a **Dad on the ward** for a couple
390 of weeks, maybe two weeks, and he'd go for a shower in the morning
391 and I remember looking, going, if he goes for a shower now at nine
392 o'clock, I won't get into that bathroom for 45 minutes, I don't
393 know how a man, and he'd come out with his hair sprayed and gelled
394 and you create these narratives about people and like **"you selfish**
395 **bastard, what are you doing spraying your hair"** (laughing) like get out,
396 I need to get in the shower (laughing). And then you know, if it was
397 half nine, I wouldn't go for a shower because what if the team came
398 around, what if the speech therapist came around, what if, I can't, yeah
399 I'll miss it. And it did happen, like you'd go to the loo and you'd come
400 back and someone would be like "oh the physio was here" but you
401 weren't here so I told them to come back, and you're like **"*no:*, *I was***
402 **just gone for a pee, I would have been here, oh my god, they mightn't**
403 ***come back for days*"** and you're just in this, **constant alert mode,** em (..)

In the first few lines of the above extract, Claire set the scene for the audience in a description that reads almost as a *mise en scene*[30]*.* Claire's inclusion of reported dialogue also adds to the visibility and replicability of the scene as a screenplay. Claire states "I *lived* there with Rose, like I was living in that room with her" (lines 384-385). Claires' use of 'fishbowl' in place of 'glass box' is an echo of a referent I made in my previous comment. Just prior to the start of the above extract, Claire used the metaphor "*glass box"* (line 379) to describe the hospital setting. Use of the word 'glass' in this context, to me brings up connotations of exposure and lack of privacy whilst '*box'* conjures up a sense of entrapment and confinement.

In the Extract 9.25 above, Claire's description of "a Dad on the ward" (line 389) may be interpreted as an 'extra' character in the story. The use of reported dialogue, emphasis, and the re-telling of her thought process at the time all help to entrust a living, breathing quality to the narrative. Claire's

[30]Mise en scene is defined as "the arrangement of scenery and stage properties in a play" or "the setting of an event" (Soanes & Stevenson, 2004).

reference to “constant alert mode” (line 403) is almost palpable in her retelling. The use of recounted remarks and thoughts, paralinguistic features (i.e., emphasis, prolongation), continuous speech and strong language (“selfish bastard”, lines 394-395) all appear to function to highlight the ‘irrationality’ or ‘intensity’ of thinking for Claire while being institutionalised.

The second example which Claire used to demonstrate her experience of becoming institutionalised is presented in the Extract 9.26 below. This extract deals with a recollected scenario, when Claire was asked to move hospital rooms to allow for cleaning of the room and then was not moved back to her original room:

Extract 9.27

Claire / Rose: “you just get completely institutionalised”

411	P	^yeah, the environment, and then even stupid things like they did a big
412		deep clean of the ward when were there and it was in the summer,
413		they had a couple of days were there was lots of-not lots of, there was
414		like *five* empty rooms, you know, they were, I remember them saying
415		this doesn't happen, which I understand completely but when they had
416		a couple of empty rooms, they did a big deep clean on them and they
417		had cleaned every single room on the ward and we were the only family
418		that were still not discharged but they had to go in and clean our room
419		so they said, we're just going to move you to the room *next* (gesturing)
420		door and we're going to clean your room and we'll move you back
421		and I said that's fine, but after they moved us they said, oh well there's
422		no point on moving back because like that rooms clean now so if we
423		moved you back, we'd have to clean the room you're in. **Three days. I**
424		**cried. Three days. I cried. Because I was *next door* to the room, *WHICH***
425		***WAS IDENTICAL* to the one I was in but I'd been in that room** (signalling)
426		for four weeks, that was my bed, and I just had everything set up
427		(laughing), even though I had exactly the same next door, uh (laughing)
428		ridiculous things like that. You do, **you just get *completely***
429		**institutionalised**. Am (.) so yeah, so we parked the whole diagnosis

In lines, 423-425, Claire described her intense reaction to not being moved back to her original room, “Three days. I cried. Three days. I cried. Because I was *next door* to the room, *WHICH WAS IDENTICAL* to the one I was in” (lines 424-425). Claire’s use of stress, volume, gestures and punctuation, seem to effectively portray Claire’s view on looking back on the situation (i.e., “ridiculous things like that...you just get *completely* institutionalised”).

Turning attention to Mary's account, in the extract below, Mary provided entry into how the healthcare professionals appeared institutionalised and perhaps not cognisant of how daunting an experience may be for a parent (i.e., "they're used to it so they don't necessarily really understand how daunting that is for a new parent", lines 283-284):

Extract no. 9.28

Mary / Alexander: "don't necessarily really understand how daunting that is for a new parent"

276	P	and he had all these different medical things going on so he had like these
277		lines in through his bellybutton, like his blood sugars were only .6 em, so
278		he was very very sick in many ways so we couldn't actually hold him other
279		than we'd try and give him a bottle of expressed milk or if I didn't have
280		enough, we'd give him a bottle of formula or whatever but it was very
281		(laughing) like you're literally trying to work around all these wires and
282		you know, you get used to it very quickly but it's very daunting at the
283		start, and the nurses don't necessarily, **they're used to it so they don't**
284		**necessarily really understand how dauting that is for a new parent** and^

Although Mary does not explicitly reference becoming *'institutionalised'* in the above extract or elsewhere within her account, she reported on the experience of speedily adjusting (i.e., "you get used to it very quickly", line 282 within the Extract 9.25 above) in relation to quickly adopting "a new normal" (line 182, see earlier Extract 9.23) in the hospital environment and in relation to medical decisions. This re-framing of normal may be interpreted as a definition of sorts of institutionalised.

9.4 *Summary*

In summary, the GET, '*The world of mothers in healthcare systems: "you adjust because you have to em, because your child needs you to'* centres on maternal role and responsibility, and maternal learning and adjusting, as explored by the mothers, having been thrust into the world of healthcare. This GET appears to encompass many *tensions*, as referenced throughout the chapter, namely the pairing of 'trust' and 'onus', 'value' and 'pressure', 'help' and 'hinder', 'professional' and 'parent'. Within this GET, there is also reference to the knowledge mothers gained from having passed through the healthcare systems in terms of how to best advocate for their child and manage their care. Further topics (including switching between parent and professional roles, maternal trauma, and becoming institutionalised living in hospital) were also introduced within this GET, in relation to unique qualities of the mothers' individual experiences.

The next GET, '*Etching of the mothers' inner worlds: "I'll never forget",* will address *how* the mothers appear to organise and mark their memories within their mind, as interpreted through their re-telling of events in relation to the phenomenon being studied.

Chapter 10: Etching of the Mothers' inner worlds: "I'll never forget" (GET 4)

10.1 Introduction

The GET '*Etching of the mothers' inner worlds: "I'll never forget"'* explores the ways in which the participants appear to remember, and narrate, living in their lifeworlds[31]. This GET spotlights the richness captured within the participants' data, which enables the reader to transcend and feel 'present' to witness the specific moments that are being recollected.

Table 10.1

Overview of Subthemes within GET 4

General Experiential Themes (GETs)	Subthemes
Etching of the mothers' inner worlds: "I'll never forget"	*Looks that speak volumes and words I'll never forget*
	Chronology: Moments remembered in timelines

10.2 Looks that speak volumes and words I'll never forget

The subtheme '*Looks that speak volumes and words I'll never forget'* refers to the specific looks and words of healthcare professionals, as recounted by the participants (the mothers do include recalled dialogues with other people in their social worlds, namely colleagues, grandparents, neighbours, other parents and strangers - some of which will be explored in the following chapter, Chapter 11, *'Living within the sociocultural world: Constructs and perceptions of 'disability' - "Why do I have to use word disorder to describe my child?'",* GET 5*)*. All the mothers include reported dialogue in their accounts which adds further to the layering effect of 'impasto'[32] for the reader, as discussed in *5.7 Analytic Interpretations.* The fact that the words of healthcare professionals will never be forgotten, is explicitly named by some of the participants, and can also be inferred given the inclusion of much reported speech within their accounts.

10.1 Researcher Reflection

For me, this subtheme is reminiscent of part of the title of Reiser's (1980) paper *"Words as Scalpels"* (p. 837). In this paper, Reiser (1980) discusses changing practice in 'truth-telling', over

[31] Mishler (1984) introduced the idea of 'Voice of the Lifeworld' (VoL). Mishler argues that attention to the VoL of the patient is a necessity to deliver holistic and "humane" care.

[32] An artistic technique, credited to the Impressionist era, whereby layers of paint were added to add thickness and depth to a painting. I use this term here to refer to the inclusion of other voices adding layers and breadth to the participants' accounts.

time, within medical discourse between practitioners and patients. Historically, prior to the birth of 'medical ethics' in the 2000s, it was considered best practice to conceal bad news from patients. Reiser quoted (1935, as cited in Reiser, 1980, p. 838) advice given by L. J. Henderson, a Harvard physician, to colleagues "Try to do as little harm as possible, not only in treatment with drugs, or with the knife, but also in treatment with words". It is interesting here to think of words as *agents.*

Reiser (1980) described "words can wound as deeply as knives, that what is said can be as significant as what is done" (p. 840). Reiser (1980) appears to be examining the content of the practitioner's message, "the what", how much truth to share. For the participants in my study, it appears to have been significant the information practitioners share and how they shared it. I envision the words and looks of practitioners as instruments which burn marks on the participants' memories like the art of pyrography.

The thoughts of capability of words to effect outcomes, namely as leaving permanent, irrevocable, marks on memory, calls to mind Austin's (1962) speech act theory. I am provoked to consider whether there was conscious intent or attention to the impact of the words or looks expressed by the medical professionals, as referred to by the mothers in my study and the larger cohort they represent. This theme in particular, may provide impetus for prompting medical and healthcare and social professionals to attend more considerably to the words and expressions they use in consultations with parents, children and other family members.

For Mary, this subtheme of *'Looks that speak volumes and words I'll never forget'* also constituted a personal experiential theme (PET). The following extract refers to the first time Mary was allowed to hold Alexander, after many unexpected medical complications and a traumatic birth:

Extract 10.1

Mary / Alexander: "it's the looks because they speak volumes to parents"

289	P	Yeah, yeah, em. So then, yeah anyway, so that evening I got to hold
290		Alexander and **I do remember that moment in particular** because em the
291		nurse said to me, the matron was there, and she said to me em, "have
292		you held your baby Mary?" and I said "no, like I havn't, em, I havn't got
293		to hold him yet" and the nurse, she said to the nurse, "take Alexander out
294		now, Mary is going to hold him". Because they had taken out his line from
295		his belly button so it was safer to move him around and I remember
296		the look and it was just a look but **I remember looks** and I think that's
297		something that doctors and nurses need to be a little bit more careful

about to be honest, it's**, it's the looks because they speak *volumes* to**
parents you know em. And Alexander was dying at that point, there was

In the Extract 10.1 above, Mary urged "It's the looks because they speak *volumes* to parents" (lines 298-299). Emphasis on *"volumes"* may serve to show the magnitude of potential held within a look. In attending to the linguistic features of this line, the use of 'volume' and 'speak' may be appreciated as poetic, functioning to personify a "look", which is silent, into something with grave communicative impact. In the continued dialogue, Mary further translated this look, in saying "in other words, I think this child is going to die so let-, let her hold him you know. And it wasn't that she said anything out of turn, it was just the look" (lines 302-304). Mary's evaluation here appears to showcase the power that can be held within a look.

In the Extract 10.2 below, Mary refers to how the news of Alexander's health was delivered to her, following his birth (e.g., "he's, the sickest child in this hospital", line 252):

Extract 10.2

Mary / Alexander: "there's no real need for me to know he's the sickest child in the hospital…that's just drama"

P now Beth he was just the sickest little baby you've ever seen and he was surrounded by, obviously, all these little neonates that were the size of your hand and you know these tiny, tiny babies and, and, he was quite big in comparison to these children but actually em, that evening, we got told "**he's the sickest child in this hospital"** you know, and you're looking around you going, we were feeling quite lucky you know because of his size

R Yeah

P And we were thinking oh, you know, that we're *very* lucky that he's so, so well (air quotations, laugh) you know, that's what we thought at the time

R And was a comment like that helpful or unhelpful to you

P It's unhelpful. Like it really, **those kinda comments really stick with you** and it doesn't really help you, you know, like, it's a bit of a dramatic comment, em, tell me, yes, tell me the extent of what we are dealing with and tell me you know that he is very sick, but don't, **there's no real need for me to know he's the sickest child in the hospital**

R Mm

P **That's just drama, and it's unnecessary** you know. Em, like, and I, I really **strongly believe that parents should always be told the extent of what's happening for their child and understand the seriousness of it but leave out the dramatics** (laughing) you know

Later in her narrative, Mary referred to the doctor who is referenced in Extract 10.2 above, as "this was the guy that said to us about, you know, being the sickest child in the hospital" (lines 310-311). Mary may have used this phrase simply as a referent for the researcher or she may have referenced this man as such because the words he spoke were so significant for her that this remains the defining characteristic. Similarly, Olivia used the referent "she was the one who said em that we should watch [television series]" (lines 742-743) to refer back to a healthcare professional referenced in her narrative (the context surrounding this example was previously discussed in Extract 7.25 and discussed in *7.2, Dismissal and maternal questioning: Being "pooh-poohed" - "I must be going crazy").* Returning to the extract above, Mary referenced the "drama" (line 265) in this doctor's remark as "unhelpful" (line 259) and "unnecessary" (line 265). In the final lines of the Extract 10.2 above, Mary advised "I really strongly believe that parents should always be told the extent of what's happening for their child and understand the seriousness of it" (lines 266-267). These lines demonstrate Mary's position on the medical practice of 'truth-telling' (as referred to in *10.1 Researcher Reflection).*

In the extract below, Mary reflected on how and where they received bad news[33]:

Extract 10.3

Mary / Alexander: "in front of everybody…I will never forget"

312 P was there and they had done an ECHO, and he (.) **in front of everybody, in**
313 **the ward, told us how sick our child was.** There was parents, **I will never**
314 **forget**, there was parents standing right behind us like from me to my
315 laptop away with their child and he told us all these facts about what
316 happening to Alexander and about you know, him being the sickest child
317 in the hospital and how serious the situation was. He told us you know, he
318 only had a 33% chan- you know like, 33% of children with this deteriorate.
319 **And I'll never forget that word and still that word triggers me, em, you**
320 **know like and I said "deteriorate?"** and he was like, "yes". And I was like
321 "oh". Ok. You know, **as in, die** like. And I didn't say it out loud, because I
322 didn't need to but I suppose that was, that was the fact, you know, **that**
323 **was the truth of the matter. But, but the means in which it was delivered**
324 **was overly dramatic** with the whole statement of being the sickest child

[33] 'Breaking bad news' is not a phenomenon restricted to rare disease or the process of diagnosis. Breaking bad news has been studied in the literature in relation to many disease cohorts, some examples include but are not limited to patients with cancer (Postavaru et al., 2022), providing a diagnosis of autism (Gray, 2001; Maynard & Turowetz, 2017; Rabbitte et al., 2017), or life-threatening foetal diagnosis (Core-Arsenault & Denney-Koelsch, 2011).

325 in the hospital and given, you know, **they should have brought**
326 **us somewhere, away from other parents because I'll never forget that**,
327 you know, I was thinking those people have just heard *every* medical
328 thing that's happening to our child and that's unfair, you know, you
329 should be given **at least a small bit of privacy** em or bring us over to a
330 **corner,** you know, away from where there's other parents at least, do you
331 know if there's no room available, em, to deliver information like that,

Mary paused in line 312 in the Extract 10.3 above before exclaiming "in front of everybody, in the ward, told us how sick our child was". This pause may signify the gravity of what the doctor had done for Mary by delivering the news in a public place. Mary elaborated "there was parents standing right behind us" (line 314). Mary attested "I will never forget" (lines 313-314) and "I'll never forget" in relation to being told details about her child's sickness in front of other parents, which again may show the lasting effects of the doctor's practice. In line 319, Mary said "I'll never forget that word and still that word triggers me". This is in reference to the word "deteriorate" (line 318). Mary explained how she translated this word to mean "die" (line 321), she clarified that she made this translation in her head and didn't say it out loud, as it was obvious. In relation to sharing the bad news, Mary reported "that was the truth of the matter. But, but the means in which it was delivered was overly dramatic" (lines 323-324). Reference to "that's just drama" (line 265) and direction to "leave out the dramatics" (lines 267-268) can be seen and has been highlighted with the commentary on the previous extract, Extract 10.2. Mary appeared to express preference for being told facts. This interpretation can be further supported when considering this idea within the context of Mary's whole accounts. At another point, Mary praised another doctor, remarking he was "really factual" (line 360), and "spoke to use just about the facts" (line 361).

Taking together, Extracts 10.2 and 10.3, Mary reflected on both 'how' and 'where' herself and her husband were told information relating to their child's health. We may interpret the place, as well as the looks, words and perceived thoughts of the medical professionals that can leave indelible marks on parents' memories. Mary made this guidance very clear "I was thinking those people have just heard *every* medical thing that's happening to our child and that's unfair…you should be given at least a small bit of privacy" (lines 327-329). Mary proposed other options that were available for the professional ("should be given at least a small bit of privacy or bring us over to a corner, you know, away from where there's other parents at least…if there's no room available, to deliver information like that", lines 329-331). The choice of words Mary used here may serve to highlight she is looking for minimal efforts even to maintain privacy with phrases such as "at least" (line 329, 330), "small" (line 329), and "a corner" (line 330).

In Extract 10.4 below, Mary appears to be considering what is useful to know. We have insights into Mary's views on this from elsewhere in her narrative (e.g., "I strongly believe should always be told the extend of what's happening for their child", lines 266-267, Extract 10.1; *7.3 Researcher Reflection,* within the GET*, 'Entering the world of diagnosis: "There's nothing wrong but everything is wrong"').*

Extract 10.4

Mary / Alexander:

902	P	things like we've been told in the last year, you know, his heart condition's
903		gonna come back, em, and this was a bit of shock to us, like we knew
904		it was always a possibility but we didn't know it was a certainty and we
905		got told this is a certainty, even though his heart is perfectly healthy
906		right now and that nearly killed him before so then we were like, we were
907		totally shocked in the appointment, he just kinda came out with it,
908		and it's not Dr.[X] anymore^
909	R	Ok
910	P	it's eh, Dr.[X] and **he just kinda said it to us, off the cuff** and we were like
911		you know, he just said you know, he, he is gonna have that back,
912		there will come a day where we're going back to his medications and
913		**you don't think then to ask any questions because you're just so**
914		**surprised by this statement^**
915	R	Shocked
916	P	**^em, yeah, but at the same time then you know, there's no real answers**
917		**so I said like when, like when, when will this happen, I don't know**, you
918		know
919	R	Mmm

In Extract 10.4 above, Mary appears to be evaluating the usefulness of being told Alexander's heart condition will retain when they don't know when ("You're just so surprised by this statement, but at the same time then you know, there's no real answers…when, when will this happen, I don't know", lines 914-917).

In the Extract 10.5 below, we return to an earlier point near the entry to Mary's account, where she recounted Alexander's birth. Below, we see Mary's first use of "I'll never forget" (line 57):

Extract 10.5

Mary / Alexander: "where's my baby, what's wrong you know, what's happening"

45	P	no (.) no (.) no (.) yeah exactly yeah so em we were you know that was

very traumatic because we were **rushed straight down** to theatre and there were a lot of people coming at me with **questions, questions, questions** and of course you have to go in on your own and then they forgot to go get Jason and you know I was just lying there in this complete state of shock em being operated on and the-the doctor said "where is this woman's husband? This baby's about to be born" and someone **ra:n** to get Jason and **I was just lying there** going what is happening (laughing) and it was just em it was a bit of mayhem (laugh) you know

R Yeah (nod)

P And then when he was born eh she took him out and she said "**it's a boy (.) a very small boy"** she said you know **and I'll never forget** kind of em you know her saying that and I was like <what does she mean> and I couldn't see-she did hold him up apparently but I didn't see it so she must have just done it for a quick second because she could obviously see that he was very sick em

R Ok

P **They just took him away from** me and I was kinda lying there going **"where's my baby, what's wrong you know, what's happening"**

In Extract 10.5 above, the repetition of "questions, questions, questions" (line 47-48) appear to echo the intensity of the moment. The choice of verbs "rushed straight" (line 46) and the extended "ran" (line 52) convey the urgency of the situation. The repetition with the addition of the noun modifier "small", magnified with the adverb "very" in the reported speech "it's a boy, a very small boy" (lines 55-56) appear to emphasize Alexander's size. Mary surmised the event as a "bit of mayhem" (line 53). Mary's reference to her position "I was just lying there going what is happening" (line 52) appear to depict her vulnerability and helplessness in this moment. Insights into Mary's thoughts during this time again serve to convey to the reader a deeper closeness to Mary in this moment. The words "just took him away from me" (line 61) in reference to a mother's new-born child being taken away are undoubtedly emphatic. As far back as the animal kingdom, Greek mythology, and biblical stories, a baby taken from one's mother is, by evolution, traumatising. The imagery conjured up in this quote is reminiscent to the sense of impending doom as permeating Mary's account (as previously discussed in *9.5 Researcher Reflection,* in relation to the referenced study of the 'Mother and Child' within Picasso's 'Guernica', 1937).

In the following extract, Mary details the events after Alexander's birth. Mary's inclusion of gesture and demonstration (lines 84-85; line 95), to accompany her description of how she briefly got to "kiss his head for one second" (line 85) strengthen the vividness of the description for the reader and may serve to signify the detail with which this moment can be recalled in Mary's mind's eye:

Extract 10.6

Mary / Alexander: "I'll never forget his face"

83	P	So, yeah, that really affected me. So basically then Alexander was em
84		brought straight to ICU so I got to hol-or I got to just kinda (demonstrating
85		palm to cheek) kiss his head for one second and Jason got to hold him for
86		one second and then em like the doctor that was at my initial delivery em
87		downstairs, he walked in and he looked at Alexander and I could just see
88		**his face was just *shocked*** and he was just like "(in breath) oh my god" you
89		know, **"this child is very sick"** and **I'll never forget his face** and he was a
90		beautiful man (laughing) not that it's relevant but **he was the most**
91		**beautiful man I'd ever seen** and I was just like
92	R	Oh, god (smiling)
93	P	**Why is his face looking like** that, you know. This can't be good, you know
94	R	Yeah
95	P	**I really remember his face** (gesturing hands) just being so shocked em, so
96		then obviously I was left there for another while or you know they were
97		finishing off my operation and em Jason was brought out and I was just
98		left lying there you know not really understanding what had just
99		happened and em then I was in theatre and I just remember waking up
100		like and Jason was there and I was like "where is he?" like "where is
101		Alexander?" you know, because I had no experience of what would
102		happen in that scenario and I was wanting skin-to-skin and I was wanting
103		to breastfeed and you know do all those wonderful natural things

In Extract 10.6 above, Mary also recollected the look on the doctor's face when he saw Alexander and his words "'this child is very sick'" (line 89). Mary emphasised the memory of his look, with "I'll never forget his face" (line 89) and "I really remember his face" (line 95), again accompanied with a gesture. The reference to the quality of his face as "beautiful" (line 90) shows the clarity of detail with which Mary can mentally resurrect the doctor's expression.

10.2 Researcher Reflection

What I find striking to notice in Mary's reflections, is the vividness with which she can recount the events surrounding Alexander's birth, at the time of the interview, 7 years on from the time of his birth. I wonder if this phenomenon (i.e., sustaining detailed memory) is more generalisable beyond a mother's experience of rare disease for their child in Ireland to other witness experience of family acute illness?

In the penultimate episode of *This is Us* (Brownell & Koch, 2021), an American television series, the protagonist Rebecca Pearson, is facing her last moments of life, of which Alzheimer's has been the disease which ultimately leads to her death. We, the audience, are invited into the character's mind in her final hours. This outbound journey from life is depicted as on board a moving train on its way towards the final destination, representative of end of life. On this train ride, Rebecca encounters people who represent those who have made significant lasting impressions and added meaning to her life. Amongst them is Dr. Katowski, Rebecca's obstetrician who delivered her three children, one of whom died at birth. During this intense moment of grief and trauma surrounding the loss of a child, Dr. Katowski gave the following advice to Rebecca's husband:

"You take the sourest lemon that life has to offer and turn it into something resembling lemonade" (Brownell & Koch, 2016, 35:21).

Dr. Katowski and the above quote is referenced throughout the series and 'nodded' to on Rebecca's final train ride. This fictional depiction may echo my participant's experience of the power and lasting impact of healthcare professionals and their words, encountered during moments of life-defining significance.

I will now move to look to examples, in relation to the subtheme '*Looks that speak volumes and words I'll never forget'* from Claire's account. Early in her narrative, Claire recounted what she was told at Rose's birth (e.g., "after she was born the paediatric doctor took her, gave her a check over and handed her back to me in my arms and used the sentence 'you have a perfect baby girl'", lines 39-41, as examined previously in relation to the subtheme of '*Searching for an answer or "firefighting" moment by moment?*' in GET, *'Entering the world of diagnosis: "There's nothing wrong but everything is wrong".* See*'*). Similar to Mary for Alexander, Claire presented her memory of the words spoken to her at the time of Rose's birth.

In Extract 10.7, Claire recalled the words of the junior doctor which appeared to be the stimulus for her switching from "[healthcare professional] mode" (line 847) into "[healthcare professional] tutor" (line 847-848) mode. This scenario was discussed previously within GET 3, *'The world of mothers in healthcare systems: "You adjust to it because you have to, because your child needs you to",* subtheme '*Maternal role and responsibility: balancing trust and* onus', subsection 9.2.2 *Switching between mother and therapist mode.* Part of this extract has been reproduced below for its relevance to the current subtheme, as Extract 10.7:

Extract 10.7

Claire / Rose: "Don't you dare refer to my child or any patient as interesting"

845	P	stint after six months and he's sitting back and he's going "oh,
846		interesting, interesting, interesting" and I *sna:pped* well out out of
847		speech therapy mode and back into, actually I think I went into speech
848		therapy tutor? Kind of mode (laughter)
849	R	Practice educator hat (laughter)
850	P	**Don't you *DARE* refer to my child or any patient as *interesting.***
851		**Interesting. Don't you dare**, I said this is my child. **I don't want to hear**
852		**it's an interesting case to you** and I *dissolved* back into tears again
853		(laughter) and all of a sudden I was out of that mode and I was back
854		into "oh, I don't want my child to be *interesting.* I want my child to be
855		really boring" (laughter) you know, am, so that (laughter) yeah, kind of
856		fell out of that then again but no in that moment of knowing that there

In Extract 10.7 above, Claire recalled the dialogue between herself and the doctor, as she remembers it (lines 845-846; lines 850-851). This interaction appears to have been highly emotive for Claire, as exemplified through the use of increased volume, emphasis, prolongation repetition (e.g., "*interesting"*, line 850; "I don't want to hear it's an interesting case to you", lines 851-852; "don't you *DARE…Don't you dare*", lines 850-851) and the choice of words (such as "*sna:pped*" line 846; "*dissolved*", line 852; "dare"). Perhaps, the grave level of emotion attached to the interaction, and to the situation, contributed to persevering this memory for Claire. Claire recounted an emotive an encounter (i.e., "I kind of got very, *irate",* line 282) with a nurse with a similar level of detail (see earlier Extract no. 9.16, introduced within the subtheme of GET three, '*Maternal role and responsibility: balancing trust and onus')*. Noteworthy, of relevance to the current subtheme, within Extract 9.16is Claire's use of the phrase "I just remember" (line 271-272) and reference to the exact time of the interaction ("about 2-3 in the morning" line 272).

10.3 Researcher Reflection

I wonder is there a pattern evident in Claire's narrative, in that she appears to close her eyes when speaking about strong memories or circumstanced recalled with weighted emotion. Several examples of 'closed eyes' (marked in bold and parenthesis) from throughout Claire's narrative, for the purpose of considering the potential significance of the occurrence with regards to the meaning attached to her utterances, are presented below:

- *Description of her daughter – thick description*

  "(..) Rose is **(eyes closed)** the most wonderful girl in the world (laugh)" (line 3)

- *Worst week admittance in hospital, start NG tube-feeding, identification of seizure condition - vivid painful memory*
  "I-the-we-I **(eyes closed)** like that week is (.) like burned into my brain, so" (line 100)

- *Description of seizure – emphasis, vivid / painful memory*
  "cluster, massive **(eyes closed)** cluster of seizures that just went on and on" (line 185)

- *Starting NG tube feeding – painful / vivid memory*
  "she was completely, nearly, atonic, em and we had to put in an NG tube, **(eyes closed),** em, we had to put an NG tube in, she-we were admitted." (lines 224-225)

- *Significant time – realisation not safe to orally feed child*
  "right down into the 70s, her coughing and spluttering, and I remember saying to the nurses **(eyes closed),** this child is not safe to feed, I can see" (lines 233-234)

- *Recalling interaction with nurse who Claire descripted "really got under [her] skin" (line 266)*
  "remember, about 2 or 3 in the morning, and I was like that kind of **(eyes closed)** (laugh) switching off (eyes open) the mammy part (gesturing pushing to the side with both hands) and just turning on and I go, really? that's what the speech therapist is going to do, and how" (lines 272-275)

- *Moment of decision-making around NG tube*
  "looked at me and she just said, [Mum] do you think we need an NG tube and I just **(eyes closed)** burst into tears and I said to her (eyes open) please don't make me make that decision" (lines 287-289)

- *Diagnosis and first birthday*
  "beginning. That was heart-breaking, and getting this diagnosis yes of [seizure condition] but why, we didn't know, em and the the, her first birthday because it was just **(eyes closed)** it, you know, I just didn't know what we were celebrating, I mean we were celebrating her but" (line 312-315)

- *Hopes for diagnosis through participation in study, recalls less favourable odds for finding out*
  "Getting a, getting a reason (.) mattered. Em, an::d, I::, yeah we got into that study and then I think **(eyes closed)** at the, they kind of meeting with the researcher for that we were told,

there is a, a large p-, I think it's like two thirds of people that go through that, will find out (.) something. One third won't. The won't find anything in a third" (lines 575-579).

- *Significant moment – returning to work*
  So I started back to work, was the big thing, em, I went back to work, I had taken **(closed eyes)** all of my maternity leave, as much as I could" (likes 617-618)

- *Perhaps emphasis on distinguishing feature – this same feature is one of the commonalities that identifies Rose with the phenotype expression of her diagnosis.*
  "and we can- I can find commonalities, like from really frivolous things like Rose has the (.) world's **(closed eyes)** most incredible eyelashes" (lines 1053-1054)

Like Claire and Mary, Robyn reports on words remembered as spoken by the paediatrician (as previously referred to in Extract 8.14, within *'The world of rare disease: What's in a name?'*, GET 2, subtheme "*Not fitting in with the "ASD Moms… or Down syndrome Moms)* as seen in lines 40-41, in the extract below "look, look around you, your baby's not so bad, and be, be, be happy with what you have 'cause there's other kids in here who are":

Extract 10.8

Robyn / Nathaniel

39	P	couple of, of em, of initial, eh, meetings with him and eh, he was fine, except
40		he was kind of like**, "look, look around you, your baby's not so bad (.) and be,**
41		**be, be happy with what you have 'cause there's other kids in here who are",**
42		so I was, but look, we didn't know, so I was like, maybe he's not, like I don't
43		know, anyway, because [county]'s not in the [regional health area] which we

Later in her narrative, Robyn returned to the words of the doctor, saying "I do think often of what the paediatrician said, the VERY first time, saying that be, be grateful for what you have, you don't have a child with you know, rea:lly bad special needs, and I don't, because" (lines 289-291). It may be interpretated that the paediatrician's words had a lasting impact on shaping how Robyn perceives her living experiences, or what she feels is permissible to see it is. There is an underlying tone of 'fortune' or 'gratitude' that transcends throughout Robyn's narrative. The language used by Robyn to refer to her situation appears to contribute to this sense of "luck" and wanting to avoid sounding "super-dramatic".

10.4 Researcher Reflection

Continuing on from the idea discussed above in relation to the overall sense of 'luck' or 'fortune' which can be felt in Robyn's account, I have presented below a more linguistic analysis of the phrases used which I feel contribute to this underlying tone felt in her narration.

- "I know that's people, not most people's experience but we were very, very lucky" (lines 77-78)
- "I was very lucky that eh, my husband's aunt, she has a playschool" (lines 245-246)
- "I was lucky to have people living around" (line 314)
- "we are lucky and unlucky" (line 359) in relation to Nathaniel attending a small school
- "I am lucky, Nathaniel is progressing huge and he's doing really well" (line 667)
- "we got off lightly" (line 759)

Robyn referenced being "lucky" eight times throughout the interview.

Similarly, Olivia discussed the response from healthcare professions when they sought advice desperately to manage their child's behaviours. The advice referenced in lines 747-748 "you just need to be more positive because there are children out there who can't walk and your child can walk so you just need to look on the, you know, at the positives" was received by Olivia as "obviously it was wildly fucking unhelpful" (line 751), (as previously introduced in Extract 7.24 within the subtheme of '*Dismissal and maternal questioning: Being "pooh-poohed" - "I must be going crazy*" of 'Entering the world of diagnosis: "There's nothing wrong but everything is wrong", GET 1).

Examples of words remembered are also evident within Judy's account. Some of which have already been presented in relation to other subthemes, such as recalling the interaction with the CDNT manager (see Extract 8.7) and in relation to the 'diagnostic moment' ("kind of more or less blasé…I'm diagnosing him today with [RDD]", lines 124-128, see Extract 7.6).

10.3 Chronology: Moments remembered in timelines

This second subtheme of this GET 'Etching of the mothers' inner worlds: "I'll never forget" is '*Chronology: moments remembered in timelines'*, which refers to how the participants appear to remember and frame their memories in terms of specific timepoints. This phenomenon is common across the majority of the participants' accounts and a more prominent feature within Claire, Mary and Judy's accounts, in relation to significant events for the mothers. For Mary, this appears to be Alexander's birth. For Claire this is the first week in hospital, and for Judy, the 'diagnostic moment'.

10.5 Researcher Reflection

I wonder what factors are similar in Claire, Mary and Judy's experiences that may be associated with the level of detail recalled within their recollections of significant moments. Mary and Claire shared the experience of living in hospital. Mary cited the trauma specifically in the experience of Alexander's birth, which she in part attributes to the shock or change in plan (*"*it was very traumatic for me, em, because I really wasn't-it was like the complete opposite of of eh what I *wanted* y;know *to* happen basically" lines 33-35; "the complete other extreme" line 38) as discussed in *9.3.1 Maternal Trauma.* Similarly for Judy, the 'diagnostic moment' can be considered her unexpected entrance into the world of diagnosis.

For Mary, her descriptions of the events surrounding Alexander's birth appear to be notable in her account. Elements of Alexander's birth have been discussed within various subthemes at this point and will not be revisited within this subtheme.

Of note for Claire, she recalled the events of a week in hospital, July 2019, beginning when she first identified Rose was having a seizure at five months of age, from Monday through to Sunday morning. Claire takes us through a detailed account of the occurrences that week ("that week is like burned into my brain", line 100).

10.6 Researcher Reflection

Claire's narrative in particular appears as structured around a chronological time framing. I have re-organised some elements from Claire's account to present events described with the time referent given, as shown in table below.

Time Referent	Event
Overdue	Overdue (lines 33-38)
Born	"Apgar scores were nine and ten" (lines 32-33)
First couple of days	"fine" (line 42) "just challenges establishing breastfeeding" (line 43)
First 24 hours after birth	"very sleepy" (line 44)
Home from hospital	"she was doing all the things that a baby should do" (lines 49-50) "probably something niggling about Rose" (line 53)
About 11 weeks old	"a gunky eye" (line 68)
"11 weeks that day" (line 74)	"she's not really looking at stuff" (line 75)

Rose at five months, the beginning of July 2019 (lines 98-99) **"that week is like burned into my brain" (line 100)**	**Monday** – "my husband was very worried, more about me" (line 107); "we both agreed that there was stuff we kind of were expecting her" (line 112); "keep an eye on Rose, we'll go the GP, we'll get a paediatric review and I need to go to the GP urgently and see if I need to start [xxx]" (lines 119-121) **Tuesday** – "she had done this weird thing" (lines 122-123) **Wednesday** – "she started doing that movement again" (line 127) "it was a weird day getting onto the GP and phone the GP going, I think my daughter is having seizures" (lines 134-135) "they said, just go to A&E" (line 149) "called it infantile spasms" (lines 150-151) "clusters of seizures a few times a day" (line 160) "got to get cannulas in" (line 161) **Thursday** – discussion about timings to get to another hospital in Ireland, for an EEG scheduled Friday morning. **Friday** – "call an arrest" (lines 193-194); "that was the lowest point where we thought she was, I actually thought she was gone" (lines 197-198); "[seizure condition]". Admitted to hospital. **Saturday night** – "she was conked and we had to wake her again and I just remember talking to the nurse going, this is not making any sense. We're waking her to cause the seizures and separate to that, there's you know, the clear risk of aspiration" (lines 259-263) **Sunday morning** – NG tube was inserted. "please don't make me make the decision to put an NG tube in my child" (line 291); "Claire your daughter needs an NG tube" (lines 294-295)
July, August, September	Hospital
Week or two later	Repeat EEG
November	Hospital
Christmas and New Year's, up to first birthday	Six or seven weeks in hospitals Seizure control PEG fitted 26th December, Stephen's day – Claire informed of diagnosis. "the doctor told us she thought Rose had [RDD X] and I had gotten to [RDD X] myself from Googling" (lines 432-434) 28th December – Claire having to disclose diagnosis to husband "I have to break this devastating news to him" (line 453)

First birthday	"we had the most beautiful doll, beautiful doll, who like a baby Annabell, you know, peed and pooed and that was it we couldn't get anything else out of her" (lines 221-223)
June 2020	Revision of diagnosis to [RDD Y]

I will now present in detail, commentary on Claire's recollections of Rose's first week in hospital (as introduced in the *10.5 Researcher Reflection* above). Claire began with Monday's events which could be summarised by "my husband was very worried, more about me" (line 107) and they "both agreed that there was stuff we kind of were expecting [Rose to be doing]" (line 112). At this point there were still possible explanations given for why Rose was not meeting anticipated milestones, "maybe she's not a smiley baby, first time parents, didn't really know" (lines 112-113). Claire discussed how she considered whether the problem was with herself (as previously discussed in *7.3 Dismissal and maternal questioning: Being "pooh-poohed" - "I must be going crazy")*.

Claire described how there was ambivalence about whether there was something ("and then she'd have moments where she would smile and you'd go, okay" lines 116-117). On Tuesday, "she had done this weird thing" (lines 122-123). Claire described and demonstrated the thing and states at this point "I didn't know what she was doing" (line 126). On Wednesday, Rose started the movement again and this time Claire recalled she "knew" Rose was having a seizure (as shown in Extract 10.9 below):

Extract no. 10.9

Claire / Rose

129	P	her lying on the floor and I looked at her face and as soon as I looked at
130		her, **I knew she was having a seizure, I just knew** by the (facial
131		expression) you know, this, this (demonstrating) tensing of the body
132		was a seizure, and **that kind of, was the, what started us (.) down the**
133		**road.** Em, so, yeah like I remember, and again you kind of go, it was a

This Wednesday is demarcated as the day that started the journey "that kind of, was the, what started us down the road" (lines 132-133, in the extract above).

Claire described the sequelae of events that followed, including "There was a lot of toing and froing trying to get in contact with the GP. This wording "toing and froing", "trying", "getting hold of" and "eventually" all appear to capture the struggle in the process in accessing the doctor. Further events of Wednesday are detailed. The seizures being named "infantile spasms" (line 151) in A&E, having

an MRI. Thursday was detailed as being centred around considering how best to facilitate the 11 o'clock EEG booked for Friday in the children's hospital in another region of the country.

In the following extract, Claire narrated the events of Friday morning (e.g., "she had a *massive* cluster, *massive* (eyes closed) cluster of seizures", lines 184-185):

Extract 10.10

Claire / Rose

176	P	terrible piece of advice. So we hoped in the car, **four in the morning**,
177		**lashing rain**, with our child, having seizures the whole way to [county],
178		driving at, I don't know what speed, am, landed in [hospital], it wasn't that
179		strict at all. Like if **she hadn't slept until 12 or 1,** they would have been
180		fine about it but we kind of had been told, this is your slot, these slots
181		are like gold dust, if you don't take it at 11, that's it. Anyway, landed in
182		and between the going at four in the morning and keeping her awake, we
183		know now that that a seizure trigger was the lack of sleep, the crying, the
184		everything and **we had the EEG at 11 o'clock** and **she had a *massive***
185		**cluster, *massive* (eyes closed) cluster of seizures** that just went on and on
186		and on and on for well over half an hour, just one after another, after
187		another and when it kind of was all over and you know, **by 12 o'clock**
188		when we had disconnected her from the machine, she was, well

Of relevance to the current subtheme of '*Chronology: moments remembered in timelines',* Claire's reference to specific times of the day is what is highlighted for attention in including Extract 10.10, such as "four in the morning, lashing rain" (lines 176-177), "we had the EED at 11 o'clock" (line 184), "by 12 o'clock…" (line 187). Extract 10.11 below continued to describe the events which occurred on the Friday (e.g., "I couldn't hear her breathing and I just remember screaming call an arrest, call an arrest…" lines 192-193):

Extract 10.11

Claire / Rose: "I just remember screaming call an arrest…"

191	P	I was in a hospital, I just clicked into work mode and I just looked and she
192		was, I had never seen her so white and I couldn't hear her breathing and
193		**I just remember screaming call an arrest, call an arrest, call an arrest**
194		**now, call an arrest, where's your phone, call an arrest,** we need a team
195		here now (.) anyway, the neur-the neuro team came in, she was being
196		checked, she was fine, she was fine but she had just, you know, really
197		gotten, that was the lowest, that was the lowest point where we

198 thought she was, **I actually thought she was gone** em, and eh they told

The events in the extract below still occurred on the Friday. Use of "screaming" and the repetition of "call an arrest" five times appears to convey the hysteria in the moment. Of relevance to the current GET 'Etching of the mothers' inner worlds: "I'll never forget", Claire's recalled in line 193 "I just remember". "Claire finished with an evaluative comment "I actually thought she was gone" (line 198).

In addition to that first week in hospital, Claire highlights two marked days. She identified the day they got the diagnosis of a named seizure condition, and Rose's first birthday as the "two saddest days" (line 310). Claire also denoted her experiences around "we had to put an NG tube in" (line 225) as "one of the hardest things" (line 227).

Disclosing the diagnosis to her husband is named as another significant event remembered by Claire. In the Extract 10.12 below, Claire relayed how her brother-in-law came into visit with her husband and when her husband, Tom, went to the bathroom Claire disclosed the diagnosis in a brief couple of minutes to prepare her brother-in-law that her husband is going to need to talk about (i.e., "Dominic they think she has [RDD X]..", line 451):

Extract 10.12

Claire / Rose

447 P ^em (.) and my *brother*-in-law (laughing) was home for Christmas, they
448 came back up, Tom and Dominic came up on the 28th because he was
449 flying home to Dubai and I remember Tom went to the toilet and
450 Dominic was there and I just, had two minutes, and I just vuh-blu:
451 (demonstrating word vomit**) "Dominic they think she has [RDD X],** Tom's
452 going to drop you to the airport now, when Tom comes back to the
453 airport, I have to break this devastating news to him, it's going to break
454 his heart, you *RING* (pointing) him when you land because he's going to
455 need to talk about it and" (laughing) I basically just everything that I
456 needed, which was, **I needed to talk about it,** I needed someone to, to

In the final lines of the extract above, Claire also recalls how she needed somebody to process it with her. In terms of the subtheme, '*Chronology: Moments remembered in timeline',* Claire recalled the date of the interaction recounted above as the 28th (line 448) and she again, includes specific dialogue, as she remembers it. The reference to Christmas as well is suggestive of occasions which may help to frame the mothers' memories.

For Judy, the events surrounding the 'diagnostic moment' (including those proceeding and successive to) for Declan seem to constitute a significant moment, recalled with attention to minute detail (as have been in discussed in relation to Extracts 7.6 and 7.5 of her transcript, in *7.2. Searching or "firefighting" moment by moment*?). Examples of the specifiers in her narration surrounding this event have been re-presented in isolation here, as is relevant to illustrating the subtheme at hand, including "there was ferocious flooding" (line 106), "absolutely awful weather" (line 110), "I was given that diagnosis back that day in March" (line 368), "I think I'd rang, this was a Friday…" (line 320).

10.4 Summary

Within this GET*, 'Etching of the mothers' inner worlds: "I'll never forget"',* the subtheme '*Looks that speak volumes and words I'll never forget'* attended to instances where the mothers referenced remembering or never forgetting, and where they included recalled dialogues and interactions with healthcare professionals within their narrations. This phenomenon was seen across all mothers' accounts. I chose extracts which I felt best embodied the subtheme to present and discuss. I also examined how the words or looks remembered by the mothers may have contributed to how they made sense of their situations, at the time, and how they may *carry* these memories into how they *continue* to understand their experiences in the present, in relation to the phenomenon of diagnosis for their child. The subtheme of '*Chronology: moments remembered in timelines'* referred to the detail and specificity which some of the mothers recounted apparent significant moments, within in their own life timelines framed by times of year, days, or milestones.

Chapter 11: Living within a Sociocultural world: Constructions and Perceptions of 'disability' - "Why do I have to use the word disorder to describe my child?" (GET 5)

11.1 Introduction

The GET of '*Living within a sociocultural world: Constructions and perceptions of 'disability' - "Why do I have to use the word disorder to describe my child?"*'[34] provides a window into the social context within which the mothers are living, about the their own, and others, views on disability and diagnosis. Some divergence is evident between the mothers' viewpoints surrounding the harm or benefit of diagnostic 'labels', as will be explored in the subtheme *'Labelling'*. The next subtheme *'How do you tell people?'* considers how the participants navigated telling family, educators and colleagues about their child's diagnosis. The final subtheme '*Onlookers: "To me he's a beautiful child inside and out, no matter, disability or not, you know but other people don't understand it"* involves how the mothers cope with the lack of understanding of disability in their communities. This GET includes remembered dialogues with grandparents, neighbours, other parents and strangers, adding a multiplicity of voices. Reference to participants' individual factors may help to illuminate influences on how the mothers interact within their sociocultural world, such as geographical location and prior family experience of 'disability'.

Table 11.1

Overview of Subthemes in GET 4

General Experiential Themes (GETs)	Subthemes
Living within a sociocultural world: Constructions and perceptions of 'disability': "Why do I have to use the word disorder to describe my child?"	*Labelling* *How do you tell people?* *Onlookers: "To me he's a beautiful child inside and out, no matter, disability or not, you know but other people don't understand it"*

11.2 Labelling

The subtheme of *labelling* refers specifically to the practice of naming through diagnosis and the belief that doing so will accompany negative connotations for the person to whom the label

[34] This theme resonates with what Mishler (1984) described in relation to the Voice of the Lifeworld (VoL) that "a patient's contextually-grounded experiences of events and problems in life, timing of events and significance are dependent on biographic situation and position in social world" (p. 103).

is applied such as stigma, social exclusion, exacerbation of mental illness (Cooksey & Brown, 1998; Timimi, 2014; Watson, 2019)[35].

In the Extract below, Olivia expressed a strong abhorrence for healthcare professionals' reluctance to diagnose (i.e., "I don't what it is, they're just like 'hey, let's just wait five years and see" lines 1076-1077):

Extract 11.1

Olivia / Anna: "the labelling thing drives me fucking spare…it's not like it follows them around like a scarlet letter"

1070	P	feeling like, em, and there's, aw then there's this whole "oh we don't want
1071		label the child with the, oh we don't want" uch, <**the labelling thing drives me**
1072		**fucking spare>** do you know what I mean? Am, and I get it yeah, there's a
1073		huge increase in diagnosis, **blah blah blah blah blah**, but like if you diagnose
1074		a child with whatever, em you know, **it's not like it follows them around like**
1075		**a scarlet letter the rest of their life,** do you know what I mean? This whole
1076		negativity around diagnosis (.) **I don't what it is, they're just like "hey, let's**
1077		**just wait five years and see** (.) is it, is it actually what we think it is or is it this
1078		other thing" and you're like (puzzled facial expression), **I understand you**
1079		**don't want to misdiagnose but in the process of doing that you just (.) make**
1080		**people suffer,** do you know what I mean

In lines 1071-1072 above, the use of strong language and low volume in the phrase "<the labelling thing drives me fucking spare>" and the repetition of "blah blah blah blah blah" (line 1073) appear to convey Olivia's intense disregard for the preservation of diagnosis for fear of labelling. Olivia's suggestion that diagnosis is not a "scarlet letter"[36] (line 1075), appears to execute satirical hyperbole in order to depict the absurdity of concerns regarding labels.

Elsewhere in Olivia's narrative, in relation to interpreting Anna's behaviours early on, Oliva shared "I just thought she was just a bit of an oddball to be honest and sure we're a family of oddballs, wouldn't be that big of a deal, you know what I mean (smile)" (lines 111-113). This comment was conveyed with a smile and seemingly 'light' intentions. The determiner "just" used twice in the

[35] As discussed in *2.4 Diagnosis: Shaped by the Individual and Society.*

[36] The 'scarlet letter' is a reference to a large red 'A' a that served as an allegory in Hawthorne's (1850) fictional novel whereby a woman is sentenced to wear a large red A to present herself as an 'adulterer' for public shaming and humiliation. A modern film 'Easy A' (Gluck, 2010) was inspired by Hawthorne's (1850) original work.

quote above may be interpreted as showing how 'trivial' being an "oddball" would be for Olivia. Olivia also referenced in her narrative that there was a family history autism, and intellectual disability, and that some genetic testing had been done within the family already. Olivia's position on disability or diagnosis may be understood as having been influenced by her own family experiences (i.e., Gadamar's, 2004, 'horizons of understanding' and Heidegger's, 1927/2008, notion of 'Dasein'). Olivia's acceptance or comfort with difference may be considered similarly to her assertions provided around special education schools. She reported "I don't see what the negativity around it is, if it's the best place, it's the best place" (line 670-671).

11.1 Researcher Reflection

Olivia's comment "I understand you don't want to misdiagnose but in the process of doing that you just (.) make people suffer" (lines 1078-1080) erects to mind for me the question of medical ethics and the discussion by Reiser (1980), as introduced in *Researcher Reflection 10.1,* within the subtheme *'Looks that speak volumes and words I'll never forget'.* How much should doctors tell patients? In his paper, Reiser (1980) concludes "the physician must choose, and act, before all the facts are in" (p. 841). This thinking appears to be in harmony with Olivia's strong desire for being told diagnostic hypotheses from medical practitioners even if such diagnosis may later prove to be incorrect. Similarly, Mary, Claire and Judy all seem to express preference for transparency, as shown within the subtheme *'A fragmented "jigsaw" puzzle: lack of transparency and co-ordinated care' (7.4).*

Robyn's belief surrounding labelling appears to directly oppose Olivia's statement that "it's not like it [diagnosis] follows them around like a scarlet letter the rest of their life" (lines 1074-1075). In contrast for Robyn, she appears to see harm in labelling.

In the Extract 11.2 below, Robyn appears to hint at concerns regarding the permanence and longevity of diagnoses (i.e., "would it stay with him for the rest of his life?", line 558; "how damaging is that to him when he's 14 or 15 and he's grown into himself" line 559). The phrase 'rest of his/their life' is used by both Olivia and Robyn in demonstration of the opposite effect[37].

[37] Berkovits et al. (2020) specifically explored perspectives of adolescents surrounding their autism diagnosis. Their findings provide some support for Robyn's concerns, as cited in Extract 11.2. Return to Section *2.5 Labeling Theory* for discussion on some of the literature surrounding stigma associated with diagnostic labels, including further details on Berkovits et al.'s (2020) study.

Extract 11.2

Robyn / Nathaniel: "would it stay with him…"

556	P	had it, like I said my husband has the gene, is I'm wondering if people know
557		that he has this genetic diagnosis and people don't really understand what
558		that is, **would it stay with him for the rest of his life**, even if he, you know
559		**how damaging is that to him when he's 14 or 15 and he's grown into himself**
560		**and he's doing great socially and they're still like he's that kid with that**
561		**weird diagnosis**
562	R	Okay
563	P	So that's the reason why we're not
564	R	Yeah
565	P	We don't know what to expect for the future you know

Robyn's reflections in Extract 11.2 above are linked with her views on questioning the purpose of diagnosis (as introduced in Extract 8.2 within the subtheme '*Rare disease: "a series of letters and numbers that doesn't mean anything to anyone"'* within the GET, 'The world of rare disease: What's in a name?').

11.2 Researcher Reflection

Are diagnoses retractable? Robyn's question about the permanency of diagnosis reminded me of a question a parent asked me in the clinical setting in relation to her son who had recently gotten new diagnoses of autism and dyspraxia. Her question was, when did the previously assigned diagnosis 'global developmental delay' no longer apply to her son? Do the new diagnoses of autism and dyspraxia pronounce the former diagnosis of global developmental delay obsolete? In considering these questions, I wonder do we have clearly defined borders in healthcare about when one diagnosis replaces another or when diagnoses coincide and exist in plurality? If these borders are defined, do we explain them to those whom we support? Because it matters. In Walsh et al. (2018), Patricia, a woman who received a diagnosis of Asperger's syndrome in mid-adulthood, explores her thoughts on when this diagnosis was expulsion from the medical vernacular with its removal from the Diagnostic Statistic Manual-Fourth Edition (DSM-IV, 1994).

Robyn extended her commentary on consideration to the lasting effect of diagnosis to include "like I said, we live in a super small town so (laughing)" (line 568). This is reminiscent of 'Dasein' (Heidegger, 1927/2008; Revisit footnote 33 for definition) and may imply Robyn's societal context contributes her views on the harm of diagnosis.

Robyn also referenced timing and its relation to the value of diagnosis. She discussed "if he stopped improving then maybe that diagnosis [ASD] is for him and maybe it gives him clarity in regards to maybe why he's a little bit weird, like right now we don't need it" (lines 418-420). It appears Robyn placed importance in questioning the purpose of diagnosis. She reflected "For any diagnosis, you have to look at, well is there a benefit to it, what's the actual purpose for it, you know so, and for that I feel at the moment all the diagnoses people are doing is to try to tick a box" (lines 440-442). It seems she viewed diagnosis as important for accessing services which she regards as an unfortunate reality (Robyn's experience of educators urging her to solicit an autism diagnosis for Nathaniel was previously discussed in relation to the subtheme '*Rare disease: "a series of letters and numbers that doesn't mean anything to anyone"')*.

In the extract below, Robyn provides insights into her perceptions of society's interests and motivations in diagnosis (e.g., "people are *OBSESSED* with getting an ASD diagnosis…because it opens *so* many doors" (lines 198-201):

Extract 11.3

Robyn / Nathaniel: "people are OBSESSED with getting an ASD diagnosis"

195	P	Not really, and a lot of the things, say with his diagnosis, in terms of the
196		genetics side of it, it is all about, that there may be some ASD tendencies or
197		some traits, and the child, your child will grow around to them, so (.) but I
198		have felt, in just in my own experience, that **people are *OBSESSED* with**
199		**getting an ASD diagnosis.**
200	R	Mmm (nodding)
201	P	And because it opens *so* many doors and so, for parents, because you're not
202		constantly fighting for things, you're like at least I have this form and this is
203		what it says^

The phrase *'obsessed with diagnosis'* is repeated later in Robyn's narrative ("they're freaking obsessed with diagnosis, obsessed with diagnosis that they understand about and it's *purely,* and it is driven from the right place in that they just want to get services, it's just the only way you can get services", lines 770-772). This repetition appears to emphasise how strongly Robyn senses the desire for diagnosis amongst others. Similarly, Robyn's interpretation of diagnosis practice and culture in her surrounding social world, can be seen in her reflections, "people have said and we're not diagnosis centric but *everybody* is still diagnosis centric because it just make it easier" (458-459) and "the diagnosis is still the most important thing, that you need to get" (lines 713).

For Mary, whose son does not have a unifying diagnosis, she regarded diagnosis would be helpful in aiding other's understanding of Alexander (Mary's hopes for what diagnosis could bring were explored in depth within the subtheme of *'Searching for answers or "firefighting" moment by moment?",* of the GET *'Entering the world of diagnosis: "There's nothing wrong but everything is wrong"').*

In Extract 11.4 below, Mary offered how having an overall diagnosis would be helpful in explaining to others about their family experience (of relevance also to the next subtheme of '*How do you tell people?*'), "at least then people can go off an look that up and kind of say ok, …this is what the lads are going through", (lines 1064-1065). In the second half of Extract 11.4 below, Mary described how learning Alexander was autistic has been helpful (e.g., "huge help for family to understand Alexander", lines 1068-1069); "for people to stop giving us this unwarranted unneeded advice on our parenting", lines 1069-1070):

Extract 11.4

Mary / Alexander: "a huge help for family to understand Alexander"

1060	P	think sometimes as well you know, **if you did have an overall diagnosis**
1061		**your family and your friends would understand it better too,** you know,
1062		if you could say, you know, Alexander has [X] disease, which
1063		is likely, what he has, you know, possibly what he has, although we can't
1064		prove it, you know, at least then **people can go off and look that up** and
1065		kind of say ok, you know this is what the lads are going through with
1066		Alexander and this is, you know, his experience, and you know it gives
1067		people something. Even I remember when we discovered Alexander was
1068		autistic, like that **actually was a huge help for family to understand**
1069		**Alexander**, em and **for people to stop giving us this unwarranted**
1070		**unneeded advice on our parenting**, you know

In her continued narrative, Mary discussed further the benefits of "being able to say Alexander is autistic" (line 1074) in terms of waiting times and accessing services. Mary offered the conclusion, given her experience of the usefulness of an autism diagnosis that "I can only imagine havin' a proper overall diagnosis would have to help" (lines 1077-1078). Mary's use of the *'proper'* as a precursor for 'overall diagnosis' returns to mind the question of the legitimacy of 'undiagnosed' or 'syndrome without a name' (SWAN) (as contemplated previously in *8.2. Researcher Reflection).* Consistent with the ideas expressed within Mary's whole narrative, it may be concluded 'undiagnosed' or 'SWAN' cannot be afforded sufficient status as diagnostic labels in and of themselves.

Similarly, Olivia, in her account expressed sorrow and deep upset in learning her daughter Anna did not reach the criteria for a diagnosis of autism. Olivia held hopes that such a diagnosis would aid their understanding of how to support Anna and to access appropriate school placement. In Olivia's subsequent written update, we learned Anna had since been diagnosed with autism (as was previously introduced in *8.4 Learning to live: "we just need to live our life now",* within the GET *'The world of rare disease: What's in a name?'*). Oliva advised how such diagnoses provided "road map of where to go next." (line 14w) and "will enable us to better understand her needs and find support" (line 15w).

For Judy, a recurrent assertion within her narrative was the importance of language class for Declan. She referred to language class as the *'silver lining'* (lines 538 and 924) in receiving a late diagnosis as a diagnosis would have precluded Declan from being able to access language class. Judy professed significant indignation at this fact, reporting it as "criminal" (line 934), "it's not fair, it's not right" (line 935), and "it's wrong" (line 949) accompanied with shaking her head. Judy described this issue in relation to autism class, "they're not bad enough for autism but they're not good enough for language class, and you know, they're falling between the cracks" (lines 963-965).

The first line of the Extract 11.5 below, "he's always special but now he's special, special" (line 1070) is Judy's recollection of Declan's siblings remark when she shared about Declan's new genetic diagnosis (further insights from the sibling voice will be explored in the following and final GET *'The world of the sibling: "Second fiddle"').* Extract 11.5 has been included within the current subtheme of '*Labelling'* given Judy's outlooks on personhood and disability which can be impressed:

Extract 11.5

Judy / Declan: "he's always special but now he's special, special"

1070	P	**He's always special but now he's special, special,** so I was like yeah, yeah,
1071		he's been upgraded (laugh) am so that's they way, you know, that they
1072		would have put it, am, but no, from a day-to-day, **he's still Declan**, and you
1073		know somebody said to me, you have to remember, he's still Declan and he's
1074		still the same, like it's not, you know that am one day he was this, and
1075		tomorrow he's this way, it's just explaining what, what he always was
1076		you know, am, he really is, and was the same little boy, am, and that you
1077		know I think I had to be reminded of that too myself

Judy's declaration "he's still Declan"[38] (line 1072, repeated in line 1073), I think carries much weight in illuminating Judy's view of diagnosis. They are an embodiment of person-first thinking. Declan is not his diagnosis, first and foremost, he is Declan. Claire addressed the issue of labels in relation to conversations with others about her daughter, as will be detailed and discussed under the following subtheme *'How do you tell people?'.*

11.3 How do you tell people?

The subtheme *'How do you tell people?'* is named using Claire's words, who referenced "how do you tell people" (line 1164) as one of "the two *big* things" (line 1162), in relation to talking about Rose. Of the participants, navigating how to tell people features most prominently within Claire's account. Robyn explored this topic in terms of when to tell Nathaniel's siblings about their brother's diagnosis, which she considered in relation to genetic heritance and family planning. Judy also explained how she supports family and friends understanding of Declan's rare diagnosis (i.e., through a comparison to Down syndrome, see Extract 8.19, as discussed in *Chapter 8, GET 2: The world of rare disease: What's in a name? Not fitting in with the "ASD Moms… or Down syndrome Moms".)*

Throughout Claire's narrative she discussed on multiple instances how she and her husband navigated returning to work, starting a new job and how to talk to colleagues about Rose. Claire referenced going to back to work as "the big thing" (line 617). She described how she was working part-time in a clinic and mostly working alone, except for interactions with clients.

In the extract below, Claire described a difference in her experience when returning to the post working in hospital, which she had occupied prior to Rose being born. Claire described there was need for an element of almost 'rehearsal' in describing how she would respond to comments about Rose ("not practice, but I'd kind of consciously make the decision to say" lines 640-641):

Extract 11.6

Claire / Rose: "oh no, no, she has complex needs, anyway, am, cup of tea?"

634	P	know, I'm a hospital therapist, and em, when I was up there, you know
635		everyone, "god you're back (.) how long have you been gone". It was like
636		two years, I've be- she, she's two or she's nearly two at that stage or
637		just shy of two and eh, "oh my god, she must be an absolute handful
638		at this stage, running around the place", and, you know, this that and the

[38] This reminds me of words in Grue's (2021) memoir recalling his parents ascertains that "For us, you were always just Jan" (p.23).

other and I'd find myself going "no::, eh" and I just used to say things like oh, eh and I'd like, not practice, but **I'd kind of consciously make the decision to say "oh no, no, no, Rose has additional needs", "Rose has** special needs", "oh no, em, Rose has complex needs", em, you know, and like **"oh no, no, she has complex needs, anyway, am, cup of tea?"** and it's not that I didn't want anyone to ask, it's just that I**, I felt like I was doing her a disservice by not talking about her the way other parents get to talk about their kids, about her likes, her needs, what she's into**, what she, you know, **and then people would be uncomfortable with, "oh, sorry"** "*sorry?*" (perplexed facial expression), don't be sorry, oh my god, no, **if you met Rose, you would not be sorry**, you know (laugh) there's nothing to be sorry about but it, it was just, just *hard* to, yeah, to just somehow say this to people

Within the above extract, Claire's own evaluations serve to do a lot of the interpretative sense-making work. Claire recounts how she would say "oh no, no, no, Rose has additional needs", "oh no, no she has complex needs, anyway, am, cup of tea?". Claire clarifies it's not that she didn't want others to ask but that she felt a conflict between feeling like she was doing Rose a disservice by not getting to talk about Rose the way other parents get to talk about their kids (e.g., "about her likes, her needs, what she's into", line 646) and also, a wish to not make others feel uncomfortable (e.g., "and then people would be uncomfortable with, "oh sorry").

In the Extract 11.7 below, in comparison, Claire described the experience for her husband, Tom, in starting a new job with colleagues with whom he did not have pre-established relationships with. Claire recounted discussions between herself and her partner about how to talk about Rose (e.g., "if you don't say anything Tom, there's two things, one is that it seems like you are ashamed or this is a secret, or this is lesser…and second of all, people will ask you questions…they'll talk about their kids school plays…and *then,* somewhere along the way…they'll find out that your experience of parenting is different and they'll do the whole 'oh my god, I feel terrible…", lines 664-673):

Extract 11.7

Claire / Rose: "how do you tell people"

P people and they know me because I worked with them, so when they'd say, and if I'd go "Rose, no, Rose has additional needs", I don't know, I kind of had already established a relationship with them so I think I found it easier, whereas Tom was trying to make new relationships, new colleagues at work and he, he really struggled with ***how* do you tell people**. It wasn't for anything, and I remember saying to him, **if you don't**

665 **say anything Tom, there's two things, one is that it seems like you are**
666 **ashamed or you know, this is a secret, or this is lesser,** or something and
667 you don't want to say it which I *KNOW* is *NOT* how he thinks at all,
668 and he kinda "that's not-", I said and, and **second of all, people will ask**
669 **you questions and they'll make comments and they'll talk about their**
670 **kids school plays and sports day and all this kind of stuff and *then*,**
671 **somewhere along the way, down the line, they'll find out that your**
672 **experience of parenting is different and they'll do the whole, "oh my**
673 **god, I feel terrible** and here was me talking about my child, sure, poor
674 Tom hav-" and I **said it creates, the total wrong picture, they will then**
675 **think, he doesn't want to talk about it and he's ashamed,** and I said, I
676 know that's not true and the only reason Tom didn't want to tell them
677 because he didn't kind of want to do what I did, explain everything in
678 negative terms
679 R Yeah
680 P And also, **he didn't want them to feel like they *couldn't* talk about their**
681 **kids (laughing)**

In the recalled dialogue within Extract 11.7 above, it appears Claire was highlighting to Tom how others would feel in response to what he says regarding Rose. Claire elucidated if Tom did not mention Rose has additional needs, "it creates, the total wrong picture, they will then think, he doesn't want to talk about it and he's ashamed" (lines 674-675), whereas Tom did not explain everything about Rose because "he didn't want them to feel like they *couldn't* talk about their kids" (lines 680-681).

11.3 Researcher Reflection

Claire's considerations regarding how herself and her husband should talk to her colleagues about Rose reminds me of 'politeness theory' (Brown & Levinson, 1978). Politeness theory maintains that we have two different types of 'face', positive and negative, and that in interactions we are appealing to one or another of these types. In considering how the conversation will make her colleagues feel, Claire may be said to be trying to avoid a 'negative face-threatening act' (Brown & Levinson, 1978) for her colleague. In considering how herself or her husband, Tom, will come across if they don't reference Rose's differences, may be said to be attending to protect the couple's 'positive' face.

In the extract below, Claire discussed her varied position having taken a new job and being Tom's situation of how to tell new colleagues about Rose ("I'm now in Tom's position, I'm now the new person", line 689).

Extract 11.8

Claire / Rose: "disorder, delay, difference, I don't know…not that I don't want to talk about it, it's not part of the conversation"

700	P	doesn't even have a *na:me* and like it, you know, [seizure condition] still is
701		nearly the easiest thing, I don't say it because it's not necessarily what
702		she has, but [RDD Y] neurodevelopmental (.) disorder? (searching facial
703		expression), again, **why do I have to use the word (.) *disorder***
704		**(searching facial expression) to describe my child, you know, [RDD Y]**
705		**neurodevelopmental (.) I don't know, you have to put in a 'd' there,**
706		**disorder, delay, difference, I don't know (laugh)**
707	R	Yeah, how does the term end.
708	P	How does the term *end* and what does that mean. So, you're still back to
709		n::: buduhbuduh you know, so we're still back to,
710		Rose has complex needs, everybody has complex needs, Rose has add-
711		U:H, so it's still a hard one to, to, I've actually started saying and it
712		only happened *this week* that I got talking to one of my new
713		colleagues and she was asking about, you know, your kids, and oh it's
714		busy, and a three year old and a one year old, and you know, oh god,
715		.hh, something about the trampoline and going trying to keep that
716		three year old in off the trampoline and I was like, well, actually, that's
717		not an issue because **my daughter has a neurodevelopmental**
718		**disability**
719	R	Okay
720	P	And I kind of said it, and I went neurodevelopmental dis-, **it's a rare**
721		**genetic thing** and I just left it at that and the person I was talking to
722		was like "oh ok" and we moved on, and that's all I expected, like I
723		wasn't expecting the whole *"do you want to talk about it?",* like, no
724		I don't (laughing), **not that I don't want to talk about it, it's not part of the**
725		**conversation**

I used some of Claire's words from lines 703-706 in the extract above ("why do I have to use the word, disorder to describe my child, you know, [RDD Y] neurodevelopmental, I don't know, you have to put in a 'd' there, disorder, delay, difference, I don't know") to form part of the title of this GET *'Living within a sociocultural world: Constructions and perceptions of 'disability' - "Why do I have to use the word disorder to describe my child?".* In 11.8 above, Claire concludes how she recently described Rose ("my daughter has a neurodevelopmental disability", lines 717-718); "a rare genetic thing", lines 720-721) and the person accepted this and moved on. Claire's recollected "It's not part of the conversation" (lines 724-725) (i.e., talking about Rose's 'genetic thing').

11.4 Researcher Reflection

In the extract above, Claire's reflections on how the sentence 'neurodevelopmental' ought to finish ("how does the term *end* and what does it mean", line 708) may be considered as adding to the larger conversation of terms of the language we use to talk about people with disabilities. In their paper, Mousavi et al. (2020) discuss how 'disability', as term is contributing to stigma and disempowering those to whom it is applied. The authors suggest using the prefix 'para' instead of 'dis' to denote 'para-ability' instead. From Claire's account, we learn she used 'neurodevelopmental disability' and 'rare genetic thing' most recently to talk about Rose. 'Thing' might be considered a preferred, and less disabling term than those beginning with 'd' (see lines 705-706) by Claire. The ideas contemplated by Claire in the section above also relate to considerations to labels. However, given her consideration to labels is in direct relation to telling people about Rose, I have chosen to represent the data here as opposed to within the earlier subtheme of *'Labelling' (11.2).* Both subthemes however are within the overall GET, *'Living within a sociocultural world: Constructions and perceptions of 'disability' - "Why do I have to use the word disorder to describe my child?"'.*

In Extract 11.9 below, Claire described the varied reactions of others in response to hearing about Rose's differences. She recalled two separate conversations, one is with her former manager, a fellow speech and language therapist who works in disability (see lines 740-755) described by Claire as "a real black and white person" (lines 740-741), and one with a neighbour (see lines 756-759):

Extract 11.9

Claire / Rose: "How you break the news or what people say…depends on the conversation, depends on the context but really the label doesn't…have a huge impact on that"

738	P	*do* ask, am:: (..) I don't mind, like I don't mind talking about it at all, em
739		and how people ask and what people say can de-depend. Like I have a,
740		**one of my old managers** who I (.) *adore*, and is just you know, she's a
741		**real black and white person** and has been great to me and you know and
742		all the trials and tribulations of being in your twenties and heartbreaks
743		and all the rest, saw me through them all, I remember
744	R	I've been there (laughing)
745	P	(laughing) when my, when Rose, when [xxx] "Claire, I heard (..) I heard"
746		and I go "yeah, yeah" and she goes "Claire, what (..) w-how bad? Just
747		tell me, how bad." And I went "mm, bad?" and I was like "immobile,
748		probably non-verbal, uncontrollable epilepsy, tube-fed, mm", then she
749		goes "oh. Pretty bad then" and I go "okay, bad" (laughing) "okay bad"
750		and I didn't get upset about it, I was like I know you, I know where you're

coming from, I know what you're trying to ask me, you're like just tell me, I'm a [healthcare professional] as well, I just need to know (.) the picture (gesturing out square) and I knew that behind all that, there was no judgement, there was no, she wouldn't think **"ah, tragic"** because she's (.) like **disability is her *thing* you know**, so **I didn't *mind* whereas *someone else* will ask and I remember a neighbour here started talking about the burden, such a burden, such an awful burden**, I was like (gesturing fist) *no::* (laughing) you don't get to say that, like she's not, she's a blessing, my god, **it was so *judgy* you know, so (.) it depends. How you break the news or what people say,** I've kind of learned to- **depends on the conversation, depends on the context but really the label doesn't, doesn't have a hu:ge impact on that**, ultimately, even though it's really what I was chasing down, it still doesn't mitigate that conversation *which* I have no trouble *having,* if anyone wants to know, I'll tell you everything, *anything in the world* about [RDD Y], you wanna know it, no problem (laughing) I'll tell ya (laugh) em, but it just, sometimes I think people don't know and *I* don't know how to progress that on

In Extract 11.9 above, Claire reflected in relation to the conversation with her manager, "I knew that behind all that, there was no judgement, there was no, she wouldn't think 'ah, tragic' because she's, like disability is her thing you know, so I didn't mind" (lines 753-755). Claire introduced the comparison in the next word with "whereas" (line 755) which appears to signal Claire's discontent with the neighbour's response. Claire expressed strong rejection for the neighbour's use of the word "burden". Repetition of the word three times in line 757 may function to depict how strongly Claire was appalled or insulted by the use of the words to refer to Rose or the family's situation ("you don't get to say that, like she's not, she's a blessing, my god, it was so judgy", lines 759-780). In Claire's concluding evaluations of this extract, Claire highlights *'how do you tell people?',* "depends on the conversation, depends on the context but really the label doesn't, doesn't have a huge impact on that, ultimately" (lines 761-762). This reflection may be interpreted as for Claire, the context of diagnosis is of more relevance in the sociocultural world than the category (i.e., 'label').

For Robyn, she introduced the issues of heritability in the case of rare disease in terms of in utero testing, and when to tell the child and siblings regarding likelihood of reoccurrence (which will be discussed as part of the next GET, *'The world of the sibling: "Second Fiddle".*

11.4 Onlookers: "To me he's a beautiful child inside and out, no matter, disability or not, you know but other people don't understand it"

The subtheme *'Onlookers: "To me he's a beautiful child inside and out, no matter, disability or not, you know but other people don't understand it"'* is named using Mary's words within her descriptions of how other people in the community react to her son Alexander. This subtheme addresses perceptions from others, as understood and told through the mothers' experiences of interacting with family and other people in their social worlds.

In the Extract 11.11 below, Mary commented about the challenges specifically in terms of Alexander having an 'invisible' condition in that people may not anticipate him to respond differently ("looks perfectly normal" line 1052):

Extract 11.11

Mary / Alexander: "because he looks perfectly normal"

1036	P	**people would come up to him** when he was younger and there were like
1037		"oh hello" and then he wouldn't talk back because he couldn't talk back
1038		and you know, you would be just kinda like, you know, you would end
1039		up having to explain to them or you know, say I just eventually just
1040		started saying hello back to them, like, oh hello, yeah this is Alexander,
		…
1043		that **people are a little bit (.) afraid maybe, you know, because Alexander**
1044		**is autistic** and you know he does have a moderate-well he's scoring in
1045		the moderate range for an intellectual disability and you know, he can
1046		get really squealy and he, if he's excited he'll be leaping in his buggy and
1047		you know doing all kinds of funny noises and you know, people, some
1048		people just kinda like at you, like you know, like oh my god what's wrong
1049		with that child and they have no concept of, em^
1050	R	Mmm
1051	P	^of his disability at all **because he looks perfectly normal**, you know if
1052		you look at Alexander, he's a beautiful child and you know he just looks^

The use of "people" (line 1036) might imply the experience with onlookers approaching Alexander to say hello was a series occurrence and not an isolated event. The pause in line 1043, "people are a little bit (.) afraid maybe, you know, because Alexander is autistic", may be suggestive of Mary trying to think of the word to best articulate her interpretation of people's reaction to Alexander. Alternatively, the pause may hint at Mary's sadness at this public reaction.

Elsewhere in Mary's account, she offers further insights into her experiences of onlookers, in offering a recounted experience of bringing Alexander to a play group (i.e., "they're looking at your child kinda going, what's wrong with him you know, and there's nearly a fear to even ask you, you know, like why is he just sittin' there spinin' a plate in a corner you know, and watching it from different angles", lines 566-570). Mary's use of "nearly" in "nearly a fear" (line 568) and "a little", as seen in "a little bit (.) afraid" (line 1043) above, may signal some trepidation in using the word 'fear'.

In the extract below Mary appears to move from this tentative position in asserting "they're afraid of it" in line 1059 extract below.

Extract 11.12

Mary / Alexander:

1056	P	Yeah, yeah, you know I think they're, I think people are really shocked
1057		then when they actually see (.) *who* he is, em, and and **to me he's a**
1058		**beautiful child inside and out, no matter, disability or not, you know but**
1059		**other people don't understand it**, you know, **they're afraid of it** and I

11.5 Researcher Reflection

Does Mary's experience of others' responses to Alexander in the community reflect something about our culture in understanding difference? How do we respond to something we don't understand? What do we consider acceptable to remark on or ask? Does this experience reflect wider universal, general attitudes and beliefs. The insights provided in Mary's accounts here may be considered to add to understandings gleaned from Claire's accounts, in relation to how other respond with pity ("poor Tom", line 673-674, Extract 11.7) and an assumption of burden (Extract 11.9).

Where Mary described 'fear' as the predominant emotive response from onlookers, Claire referenced 'pity'. A strong rejection of 'pity' may be felt throughout Claire's account, as referred to in the previous subtheme of *'How do you tell people?',* extract 11.9 with a rejection of Rose's neighbour assumption of Rose as a "burden" (line 757). Throughout Claire's account, as exemplified through the examples discussed in this GET, it is clear that Claire does not want Rose to be defined in terms of a "disability". Claire began her account by introducing Rose with a presentation of who she is, as a person, "Rose is (closed eyes) the most wonderful girl in the world (laugh)…Rose is three, she loves music, she loves Reggae music in particular, em, which we discovered, eh she loves very, she's a real foodie" (lines 3-13, as introduced in *6.4.2 Entry Point for Claire, Mother to Rose*).

When reflecting on talking about Rose with colleagues, Claire spoke about how she feels it would be a "disservice" (line 643, as seen in Extract 11.6 above) to not talk about her like other parents get to talk about their child. Previously in her account, Claire recalled "I found I was describing Rose based on what she couldn't do and that was not what I wanted to do. I wanted to give the kind of description I gave you at the start" (lines 521-523; as also referred to in *6.4.2 Entry Point for Claire, Mother to Rose).*

The Extract 11.13 below presents an example of Claire's strong prohibition on Rose being looked upon with pity by her grandparents (e.g., "CUT out poor Rose", line 1088):

Extract 11.13

Claire / Rose: "CUT out poor Rose…there's no poor Rose here, cut out that shut"

1083	P	So my mother, eh, so there's still a little element of tragedy across the
1084		board which we don't have a lot of time for, em, **we have *explicitly***
1085		**banned the phrase 'poor Rose'** because it was a constant referral to
1086		and how's poor Rose today, there's poor Rose on the floor, poor Rose
1087		sitting down there, I see poor Rose and we just went WHERE'S the
1088		poor Rose coming from? **CUT out poor Rose**, Rose is having the crack
1089		over there, em, playing with her toys, she's not playing with them
1090		(shaking head) in the same way- you know she's enjoying the music
1091		like everybody- **there's no poor Rose here, cut out that shit, there's no**
1092		**poor.** So my Mum has just been doing with us for the last little while em,

In Extract 11.13 above, the strength and force of Claire's rejection can be obviously felt in the use of emphasis ("we have *explicitly* banned" line 1084), increased volume ("WHERE'S the poor Rose coming from?", lines 1087-1088, "CUT out", line 1088), and repetition of phrases ("cut out", and "there's no poor"). Claire expressed, in contrast, Rose does get to be referred to as 'poor Rose' when she is unwell with the example given of a current bad E.coli bug. Later in her narrative, Claire elaborated "there's a lot of aww, aww (demonstrating downturned lower lip and head tilt) sitting and looking. She just wants to play, you know, so there can be elements of tragic but she's *better* about it" (lines 1101-103).

Claire's views on disability can be further discerned through examining her recollected dialogue with her father, as represented in the Extract 11.14 below (e.g., "stop trying to think of Rose being on the same path as another child but further back, Rose's got a *whole* other path all of her own", lines 1122-1124):

Extract 11.14

Claire / Rose: "Rose's got a whole other path all of her own"

1118	P	^and and less maybe towards Rose, per say, and then my Dad is just,
1119		you know, he'll keep saying things like well they did say she'd be
1120		delayed by a year and a half so if you think she's three now and a year
1121		and a half, there's some one and a half year olds that aren't walking
1122		and I just keep trying to go no, Dad, no, **stop trying to think of Rose**
1123		**being on the same path as another child but further back, Rose's got a**
1124		***whole* other path all of her own** that she's going to follow along and
1125		**she's going to go miles and miles down her *own* path** and it's just not
1126		going to look like another kids and he can't *really* (..) you know, he's still
1127		kind of trying to process that a little bit, em, and does the diagnosis

It appears from Extract 11.14 above, Claire's father was trying to frame Rose's development as delayed along a typical developmental pathway. Claire's response clarifies the uniqueness of Rose's journey, "she's going to go miles and miles down her *own* path" (line 1125).

We can abstract further insight into Claire and Tom's views on disability, life, and living from examining the sentiments presented towards the end of her account, with "what will be, will be" (line 1247 and line 1248 within the subtheme; as previously introduced in Extract 8.21 in the subtheme *"Learning to live: "we just need to live our life now",* GET 1).

In terms of this GET *'Living within a sociocultural world',* Robyn discussed the social withdrawal that occurred for her as a result of having a child who is different. In her account, she reflects, "you kind of stop visiting other parents who don't have, who don't have it because you've got the child whose got a book of pictures that he brings with you" (lines 320-322). The following Extract 11.15, gives us further insight into the change to Robyn's social activities and reasoning for this (e.g., "you kind of stop going to birthday parties…because a lot of the time they're like 'oh and is he walking?..."):

Extract 11.15

Robyn / Nathaniel

328	P	over it a small bit now, is that **you kind of stop going to birthday parties**
329		**and you stop doing those things because a lot of the time they're like "oh**
330		**and is he walking?",** "no.", "oh, does he not like to talk, how
331		are you? What age are you?" and you're like (rolling eyes upwards) uch stop
332		asking him questions, he can't answer you, you know (laughing)

Throughout Robyn's account, she made several references to "weird". In relation to how Nathaniel was currently viewed ("that **weird** kid in the class", line 382-383; line 499) and concerns for how he may *continue* to be viewed if others knew of his rare genetic diagnosis ("how damaging is that to him when he's 14 or 15 and he's grown into himself and he's doing great socially and they're like he's that kid with that **weird** diagnosis", lines 559-561; see Extract 11.2). Robyn offered that diagnosis may be validating for Nathaniel, depending on his progress ("if he stopped improving then maybe diagnosis is for *him* and maybe it gives him clarity in regard to maybe why he's a little bit **weird**", lines 417-419; see discussion further to Extract 11.2)[39]. Robyn also used weird in her description of how AIM (The Access and Inclusion Model) viewed Nathaniel's diagnosis in providing supports ("they look at it and go oh it's this **weird** genetic brain thing, we don't know what that is so obviously (laughing)" (lines 148-149). Whilst the above reflections offer understanding into how Nathaniel is perceived by "onlookers", pertaining to the current theme, there is some overlap with insights on Robyn's views on diagnostic labels (as discussed within the earlier subtheme *'Labelling',* within this GET).

Similar to Claire's description of Rose provided at the opening of her narrative (see *6.4.2. Entry Point for Claire, Mother to Rose)*, Judy provided insight into Declan's personality at the end of the interview, as seen in the Extract 11.16 below:

Extract 11.16

Judy / Declan

1182	P	he's a great personality and everybody who meets him would say that, you
1183		know, he's a real determined, what'll I say, he's a real level, great level of
1184		determination, you know, has a real 'well, you know, this is me, deal with
1185		it kind of attitude am so it's great do you know, and he's not a bit shy and
1186		he's not a bit kind of you know, reclusive, or you know like, when he's
1187		transferring onto mainstream, at least I kind of know well sure, he will
1188		just you know, he will be able to hold his own in class, he's not going to be
1189		kind of too shy, I hope, and you know, personality does, I mean we have

The above reflection from Judy, appears a fitting way to conclude this subtheme, '*Onlookers: "To me he's a beautiful child inside and out, no matter, disability or not, you know but other people don't understand it", by* offering a window into Judy's view of her son, as Declan.

[39] Some of the literature surrounding the potential labelling and benefits of an autism diagnosis for the individual was introduced in *3.4.4 Validating Experiences for the Individual ('Inner') and Society ('Outer')*.

11.5 Summary

The first subtheme of this GET, *'Labelling',* explored the mothers' varied perspectives on the so called 'diagnosis debate'[40] in terms of the mothers' varied views on the harm and benefits of diagnostic labels, relative to their child. The issue of how to tell other people about their child's diagnosis or needs was illuminated in the subtheme *'How do you tell people?'*. Most understanding on this topic was gleaned from analysis of Claire's account, who concluded 'context' and 'conversation' were ultimately more relevant on this matter than diagnostic 'label'. The final subtheme of '*Onlookers: "To me he's a beautiful child inside and out, no matter, disability or not, you know but other people don't understand it"',* presented the mothers' perceptions of others' reactions towards themselves and their children. This subtheme also offered suggestions into the mothers' own views of disability.

[40] Re-visit footnote 8 for expansion on the term.

Chapter 12: The World of the Sibling: "Second fiddle" (GET 6)

12.1 Introduction

The mothers' accounts provide a window into the perceived experience for siblings. Some of the mothers include recalled conversations had and overheard, which contribute insights pertaining to the 'sibling voice'. '*The world of the sibling: "Second fiddle"'* also includes the mothers' standpoint on the sibling's role, contemplations, and recollections on telling the sibling about their brother or sister's diagnosis or difference. The participants also spoke about the lack of available supports for siblings of children with rare or undiagnosed conditions (which has been introduced in the subtheme, *'Not fitting in with the "ASD Moms… or Down syndrome Moms"',* in ''*The world of rare disease: What's in a name?', GET 2).*

Table 12.1

Overview of Subthemes in GET 6

General Experiential Themes (GETs)	Subthemes
The world of the sibling: "Second fiddle"	*Experience of the sibling: 'Sibling voice'*
	Role of the sibling
	Telling the sibling about their brother or sister's diagnosis or difference

12.1 Researcher Reflection

I directly elicited insights into the sibling experience during the semi-structured interviews. I have included my invite to the mothers to hear of the sibling experience within the extracts in this chapter. The rationale for this is to provide context and transparency for the reader, as similarly described in *6.4. Entry to the Mothers' Accounts*. Olivia was the only participant who referred to the impact of Anna's behaviours on her brother, Jude ("her brother couldn't even go to school because she was up screaming all night", lines 262-263) before I explicitly probed for her thoughts in relation to the sibling experience, as pertaining to the phenomenon in question (i.e., "how do feel in terms of it affecting your family, what about Jude? How do you think he feels or is aware of the situation?", lines 839-840, Extract 12.1 below). In her subsequent written update, Olivia also volunteered information on Jude, stating "her relationship with her brother has improved enormously" (lines 4-5w). I considered what it might mean for the majority of mothers in my study, that they did not spontaneously volunteer information on the sibling experience. Should it be interpreted that the sibling experience is outside of the experience of the mothers in relation to the research question? Or is it that the mothers did not consider the

information as relevant, based on aims of the research, as outlined in the participant information leaflet? Regardless of the potential reasons, I decided to include the *World of the sibling: "second fiddle"* as a GET given the common experiences reported by the participants and the potential implications of the findings. In keeping with quality markers of IPA, I have included these details in order to remain transparent on my decisions making process throughout the analysis.

12.2 Experience of siblings: 'Sibling Voice'

As referenced in *12.1 Researcher Reflection* above, Olivia referenced the impact of Anna's behaviour on her brother Jude. Olivia's reference was in the context of detailing the impact of Anna's behaviour on whole family functioning. Olivia elaborated on the experience for Jude further to my direct probe (lines 839-840), as shown in the extract below:

Extract 12.1

Olivia / Anna / Jude

839	R	And how do you feel in terms of it affecting your family, em, you know,
840		em, what about Jude? How do you think he feels or is aware of the situation
841	P	It's *very* difficult for him, *very* difficult for him, he's just the most
842		sweetest and understanding child known to man but like for example, during
843		Covid, every time she went into the hospital, I'd have to go in with her,
844		because you have to stay with the child 24/7 in the particular ward that she
845		used to go into, the lady bird ward, there's no, they don't have enough
846		nurses, so you have to stay with the child at all times, you can't leave them at
847		*any* time. So once you get admitted, you're locked into the room essentially,
848		because you can't even go out into the hallway, em, ooo (turning in response
849		to child presenting figurine), em

Olivia elaborated that "we'd swap him at the door of [hospital] because only one parent was allowed" (lines 863-864), as seen in the extract above. Here, Olivia discussed how Covid-19 regulations within the hospital at the time, impacted on Jude's experiences. In the following extract, Olivia reflected on her idea of wanting to turn a trip to the hospital into a positive family trip for the family:

Extract 12.2

Olivia / Anna / Jude

872	P	Night night. We tried to turn a trip to [county] into a holiday?, because she
873		had an appointment in [hospital]. So we tried to turn it into like a little
874		holiday, and it turns out that she doesn't like traveling anywhere (laugh) but
875		we didn't really know that, because we hadn't really been anywhere

876 since [hospital], em so we were like oh we'll have one small
877 appointment at [hospital], and then you know, we'll go to the zoo and
878 we'll have loads of fun and we'll stay in a hotel, we'll get the train and it'll be
879 great fun (..) <it, was, a, nightmare> em, you know, in fairness to her, I don't
880 know why I didn't think that it would be a nightmare, I just, I don't know why
881 I thought she would enjoy it, it was stupid of me really. Em, she screamed^

In lines 880-881, it is evident Olivia is critical of her previous hopes for a family holiday. In the lines that follow Extract 12.2, Olivia added further details into Jude's experience of that trip in relation to his sister's distress "he was in the bath, with a bath bomb, sticking his head, sticking his ears under the water because he said he didn't want to listen to her scream anymore before it was making him sad (.) you know, em" (lines 892-985). In addition to discussing impact on his daily life and family activities, Olivia describes Jude's expectations for his sister, as exhibited in the extract below (e.g., "he was going to chase them and they were going to be giggling and it was going to be the most fun he ever had", lines 905-906):

Extract 12.3

Olivia / Anna / Jude: "…it was going to be the most fun he ever had and he's never been able to play chasing with her"

904 P ^he just, he just, **he was going to play chasing with them, there was going to**
905 **be a fat baby running around in a nappy and he was going to chase them**
906 **and they were going to be giggling and it was going to be the most fun he**
907 **ever had and he's never been able to play chasing with her** and he can play
908 chasing with my niece and she loves playing chasing with him even though
909 she's younger and she plays all these games with him that his sister doesn't
910 and for a very long time she, this has only stopped recently, she took a
911 sudden weird dislike to him, I don't know why, like she wouldn't hug him,
912 you know she wouldn't, she barely hugs him now to be honest with you but,
913 em (.) the (.) she took a dislike to anybody hugging him. If he sat down on the
914 couch, then she'd flip out, if I tried to give him a hug, she'd flip out, and like
915 not in like a screaming way, in a like going over and head butting or biting
916 him kind of way and he'll just like, "why doesn't she like me" (long pause)

Olivia introduced her niece into the dialogue in line 908 above, whom she referenced used to love playing chasing and games with Jude. Olivia divulged that this niece no longer plays with Jude, as described in the later lines 910-916 of Extract 12.3 above. Following the long pause referenced in line 916 I offered some words of support for Olivia, at which point she was observed to become upset and wipe tears from her eyes. This is the only time in the interview where Olivia became

visibly upset. I wonder if this may signal to the significant emotional weight of Jude's upset for Olivia. This viewpoint may be further supported when comparing the content of Olivia's subsequent written update (see *12.1 Researcher Reflection).*

Mary discussed "the emotional hurt" (line 992) for Alexander's brother, Evan, in Extract 12.4 below (e.g., "it really impacts on him, when Alexander was in pain and he's crying all day, and he attacks Evan…", line 988):

Extract no. 12.4

Mary / Alexander / Evan: "the emotional hurt"

980	R	And how do you think for eh, did, em, Alexander's brother then, how do
981		you think he thinks about or understands his older brother
982	P	Yeah (.) Evan is four, and em he's a very very smart boy, very clued in,
983		very emotionally aware and you know we're very honest with him, like I
984		obviously don't, em, share the bigger stresses with him you know but
985		**he just takes Alexander as he is, you know, that's my brother** and he
986		doesn't know any different, you know he doesn't know what it's like to
987		have a typically developing sibling, you know em so he just sees Alexander
988		as his brother. You can **see it really impacts on him, when Alexander**
989		**was in pain and he's crying all day, you know, em and he attacks Evan**,
990		now the two of them aren't dissimilar in size, Alexander's very small
991		still, em you know, so he's not actually really going to hurt Evan very
992		much, **it's more the emotional hurt^**

Above, Mary also provides insight into her interpretation of how Evan views his brother (i.e., "he just takes Alexander as he is, that's my brother and he doesn't know any different…he just sees Alexander as his brother", lines 985-988).

In Extract 12.5 below, Mary described various instances which have a "massive impact on Evan" (line 999) and where they can see visible impact of his experiences with his brother surrounding toileting (e.g., "he's holding his poo, he's soiling himself a lot", line 1011):

Extract 12.5

Mary / Alexander / Evan: "Evan will be second in line for anything…it's not easy (laughing) like because you're trying to balance everything"

994	P	^like Evan gets very upset when Alexander, does something like that
995		and I of course try and explain it to him that em, you know, Alexander's
996		in pain and Mammy's tell-has given out to Alexander and told him, he's

not allowed to hurt Evan, you know and (.) but it is very hard for a four year old to (.) deal with, you know, em Alexander had recently big mental health issues [X]. Now, that em, that has a **massive impact on Evan** because then Evan is left alone, you know, em, he's left on his own in the sitting room or you know, he's sat in front of the television and and we're, you can't leave Alexander when he's like that, you know, so you need two people all the time and a lot of the time, you know, **Evan will be second, like in line for, any, anything you know,** em, he's very good at making himself first in the line which is good (laughing) you know, he doesn't shy away (laughing)

R Makes his way up there (laughing)

P (laughing) he really does, yeah. And like **we're very conscious of you know his needs and try to look after him as best we can** but you know, we do see it coming out em, in things like his toileting, like you know, he's withhold-, you know **he's holding in his poo**, he's soiling himself a lot, and like this is a very smart child that knows well^

R Mmm

P ^when he needs to go and it's an emotional impact on him and I'm **trying to get him help**, em, you know I've got him referred to Psychology and I'm doing parenting courses and stuff to try and help him with that as well but you know, because I don't want to turn around in another five years and see this massive impact that's you know, like that say living with Alexander has had on him, em, and then it's a little bit harder maybe to (.) to kind of reverse, you know, the damage that's been done^

R Yeah

P ^or whatever you know, so we're trying to now, em, kind of **nip it in the bud and try and, you know, have ways to help Evan to cope as well, em so it's not easy (laughing) like because you're trying to balance everything you know**

R Yeah

P Yeah, yeah, but it is hard on him for sure

In the opening lines of this extract, Mary shared how she explains Alexander's actions to Evan (lines 994-998). Mary continued to talk about how the recent mental health difficulties for Alexander have resulted in Alexander requiring supervision, and Evan being left alone (lines 998-1003). In lines 1008-1009, and 1015-1026, Mary described how she tries to support Evan and prevent future consequences for him as a result of living with Alexander (e.g., "nip it in the bud…have ways to Help Evan to cope", lines 1023-1024). Mary referenced "Evan will be second, like in line for, any,

anything" (line 1004-1005). To the same effect, Olivia, and Judy both use the term "second fiddle" to refer to Anna and Declan's siblings. This phrase denotes part of this GET.

In the extract below, Robyn similarly described their efforts to not have Nathaniel be the "centre of everything" (line 469-477):

Extract 12.5

Robyn / Nathaniel / Siblings: "we've tried really hard that Nathaniel isn't the centre of everything"

466	R	And what about for Nathaniel's, I think you said brothers is it, how^
467	P	Yes
468	R	^how do you think they (.) consider
469	P	So, em, yeah, we've, we've tried really hard, maybe not successfully all the
470		time, **we've tried really hard that Nathaniel isn't the centre of everything** in
471		the whole house because he *is*, you know, I can say that as a parent is that in
472		the beginning we had, we had so many appointments for speech and language
473		and OT, and physio, we had an appointment every week and we're trying to
474		do all the speech and language, the kids are learning Lámh , the kids are
475		doing um, understanding PECS and you know so as much as I say he's not the
476		centre of everything, he really *is*, in terms of our day to day, um, and so we
477		really, really try for it not to be that

In the above reflection, Robyn listed the many therapy appointments and activities as something that makes Nathaniel "the centre of everything" (line 470) in their day-to-day life. Similarly, Judy spoke to the "extra time and attention" (line 988) Declan gets and having to explain the equipment Declan gets are part of therapy, not toys. The phrase "time and attention" is repeated by Judy twice more in her narrative.

In the extract below, Judy discussed how she interprets her teenage daughter feels in the situation (e.g., "she *resents* sometimes", lines 997-998), pertaining to the '*sibling voice',* as within the title of this subtheme (i.e., '*Experience of the sibling: perceived and 'sibling voice''*. She used the analogy of "second fiddle" to describe how she imagines her daughter feels (line 999):

Extract 12.6

Judy / Declan / Siblings: "I think she does feel second fiddle"

994	P	You know and and, I think fourteen-year-old to be honest struggles more
995	R	Mmmm
996	P	Am am, I just think you know being in the teenager year and she's a girl
997		you know she's kinda, not that she resents her brother but she *resents*

998 sometimes the time and the attention and the the energy that the
999 appointments require and **I think she does feel second, second fiddle,** you
1000 know to a certain point of view, am and I actually had her booked into one
1001 of these sibling workshops things and Covid of course, Covid yeah stopped it

Judy recalled advise she heard and executes to support Declan's siblings, in Extract 12.7 below (e.g., "If they have something, well I'll have your back the same way that I'll have Declan's back", lines 1031-1032):

Extract 12.7

Judy / Declan / Siblings: "If they have something, well I'll have your back the same way that I'll have Declan's back"

1027 P I do remember listening to a very good podcast, a parenting podcast a
1028 while back and they mentioned you know, they were talking about this
1029 issue and something struck with me and eh they said, once they they, we'll
1030 say the typical child has am, knows, and I said this to them you know,
1031 **if they have something, well I'll have your back the same way that I'll have**
1032 **Declan's back**, you know, if you need something, you'll get all that time and
1033 attention too. So**, I *hope* that that's the way that they understand it,** you
1034 know, am. It's it's, but it is, it's tough on them, am, and I did say to

Judy's emphasis on 'hope' (line 1033, Extract 12.7 above) appears to echo the sentiment captured by "try" in Robyn's evaluation, "we try to do it all together, I don't know how successful we are in that but we do, we do try" (lines 513-514), and similarly in Mary's (i.e., lines 1015, 1024, Extract 12.1).

Elsewhere in her account, in response to whether the name made a difference to Declan's siblings, Judy detailed that there are some more hospital appointments since the diagnosis and as an outcome of those, the diagnosis may have more of an impact on Declan's siblings. Ultimately, she concluded "on a day-day-level, no, it's just a name for what Declan has" (1068). The value of diagnosis on Judy's siblings will be explored further in the final subtheme of this GET, '*Telling the sibling about their brother or sister's diagnosis or difference'.* Judy recounted the response from one of Declan's siblings further to telling him about Declan's rare disease diagnosis, "He's always special but now he's special, special, so I was like yeah, yeah, he's been upgraded (laugh)" (lines 1070-1071). This is similar to how Mary described Evan sees Alexander (see Extract 12.1, lines 985-956). Robyn recounted a conversation she overheard between one of Nathaniel's brothers and his friend, which offers insight into how he sees his brother, as below (i.e., "you have no idea how amazing Nathaniel is", lines 486-487):

Extract 12.8

Robyn / Nathaniel / Siblings: "you have no idea how amazing Nathaniel is"

485	P	and I overheard my eldest son talking to one of his friends and his friends
486		were like why doesn't he talk and he was like **you have no idea how amazing**
487		**Nathaniel is**, Nathaniel was told he couldn't walk and then he actually
488		walked because he wouldn't give up, you know so

12.3 Role of the sibling

This subtheme focuses on the mother's interpretation of the siblings in relation to their brother or sister with a rare or undiagnosed condition.

For Claire, Rose's brother was very young at the time of the interview (approximately one year old). Claire's insights may suggest a mutually beneficial relationship for both Rose and her brother, "the greatest gift that we could give to Rose would be a sibling and the greatest gift that this sibling could have is Rose" (lines 1149-1150).

Robyn's position on roles and responsibilities of Nathaniel's brothers can be understood through examination of an "incident" she described, specifically in attending to lines 508-511 (e.g., they're not there to babysit him"), in the extract below:

Extract 12.9

Robyn / Nathaniel / Siblings: "it's not their role"

499	P	uch there's that weird kid in the class and he's related to me, em we did have
500		a <very big incident> in the class, em, this is the stance I took on it, not
501		sure if it was the right or wrong, stance, is em, Nathaniel, he'd very very
502		rarely have an accident, he wet himself in class and the teacher took [X],
503		my eldest son out of his class to change him during the school day and I and
504		he was real embarrassed about that
505	R	Mmm
506	P	Mostly embarrassed in case one of his friends figured out what he was doing
507	R	Yeah
508	P	And I then did go into the school and said, I don't want the kids being
509		disrupted with anything because it's not, **it's not their role**, they're not there
510		**to babysit him,** they're not there to change their brother in school, you have
511		an SNA and you have a teacher, figure it out, you can't bring him out to do it

In the Extract 12.10 below, Judy explained the role she attributes to Declan's siblings in relation to what she feels they need to know surrounding Declan's diagnosis, "they're not his parents, they're his siblings and it's a different level of information they require but you know the plan is certainly as they age and get older and understand, you know, am that they will be informed" (lines 1021-1023):

Extract 12.10

Judy / Declan / Siblings: "they're not his parents, they're his siblings"

1017	P	^they might, I suppose, have the broader view of well like, you know,
1018		obviously he can't speak the same as other people, he was sicker as a baby
1019		compared to other children and things like that , am but the ins and outs of
1020		it, we're not sitting down and giving them the whole shebang, you know,
1021		**they're not his parents, they're his siblings and it's a different level of**
1022		**information they require but you know the plan is certainly as they age and get older**
1023		**and understand, you know, am that they will be informed** and I think even
1024		just themselves to be able to name it and say well this is why he gets that extra
1025		attention you know it's not a, you know, a favour- you know the favourite
1026		child of whatever, it's he does need the time and attention, you know, and

Judy's reflection above shows she considers what information they need to know and that the level of understanding they require will evolve over time.

12.4 Telling the sibling about their brother or sister's diagnosis or difference

Evidence for this subtheme will be extracted from Robyn and Judy's accounts, only. Given the older age of Declan and Nathaniel's siblings, in comparison to the other siblings, it is likely they are the only two mothers in the study who can offer insights into the subtheme at hand.

Robyn discussed how she addressed questions Nathaniel's siblings had regarding his development, as presented in Extract 12.11 ("why doesn't he talk or why doesn't he talk like the other kids", lines 479-480; "like does he have special needs" line 493) below:

Extract no. 12.11

Robyn / Nathaniel / Siblings: "does he have special needs"

479	P	And we then, em, a lot of the time they'd have questions around well **why**
480		**doesn't he talk or why doesn't he talk like the other kids** and em, we would
481		say, well you know what, em, Nathaniel was said and I'll tell you in a second
482		what the, we always say Nathaniel was said he'd never walk and he tried so

hard and now he's walking and the same with his talk, speech is that he will, you know so, we have to try super hard and really help him get there

…

P Yeah, so it was, just real interesting you know so em, but now [X] is twelve and he would ask now, he's like **does he have special needs** and I'm like do you know [X] most people have some kind of special need (laughing) you know so, but it's because he's like one of my friends said that he has special needs and like that it's a dirty word and I was like, uch [X], that child probably has special needs too (laughing) so it's not something that's but I do feel that since he's started school the kids are probably more aware that uch there's that weird kid in the class and he's related to me, em we did have

The inclusion of the sibling's questions, and comments of their peers, within the mothers' accounts adds additional voices and further breadth to the data.

In Extract 12.12, Judy shares that she is also a sibling of a brother with additional needs (line 975) but how she was unaware of 'his *official* diagnosis' (line 976):

Extract no. 12.12

Judy / Declan / Siblings

R And you said as well about em Declan's siblings, so how do- are they aware of the name? [RDD] or do you think it means anything to them? (..) about Declan

P Yeah, we, I suppose, we just, I take a very open approach, am, you know, I, I grew up with a brother with special needs as well and to this day, **I actually couldn't tell you what his *official* diagnosis is**, you know, we just always knew that he had special needs, am, so I've been very- not that I sit them down at the table and have a very formal conversation look you're brother got this diagnosis today, you know, they're only eleven and fourteen but certainly yeah like I mean they're aware that he has, that he has speech delays and he was going to language class. They, they were aware of like we'll say, like he has DLD, he has verbal dyspraxia, am, you know and I would, you know I think, I think in my own way I would disseminate information as I felt they needed it, so like they are aware now that he has this diagnosis of [RDD], am they know that he needs to go to [hospital] just to be kind of, you know have an overview up there, am, you

In the extract above Judy shares that she is also a sibling of a brother with additional needs. Judy advised she does not know her brother's "*official* diagnosis". The addition of, and emphasis, on the

precursory '*official'* may hint that for Judy, the diagnostic label or name is less relevant from the sibling's perspective. Judy initially informed Declan's siblings on his needs and later she informed them of Declan's exact diagnosis. Judy advised she provides information as she feels it is needed (line 984, and as seen earlier, in lines 1021-1023, Extract 12.10, above).

12.2 Researcher Reflection:

As signposted within the main text above, Judy revealed her brother has special needs. This new information gives the reader further insight into her horizon of understanding (Gadamar, 2004) and lifeworld. Judy was recruited to take part in the current study given her role as a parent of a child with a rare neurodevelopmental condition. Judy also can identify as the sibling of a person with special needs. This revelation, I think, grounds the readers, in reminding us that Judy does not exist only within the parent-child dyad being explored, but in other relationships, such is the notion of 'Dasein' (Heidegger, 1927/2008) and 'fusion of horizons' (Gadamar, 2004). Like the researcher, Judy's experiences are lived through the context of, and can't be detangled from, her prior experiences.

In the Extract 12.13 below, Judy detailed for the researcher exactly how she described Declan's rare diagnosis to his siblings (e.g., "it's a rare enough thing", line 1009), as follows:

Extract 12.13

Judy / Declan / Siblings

1004	P	And then she kind of says to me, "well I'm not going to therapy to talk about
1005		my pro-" (using voice as if speaking as her daughter) you know she's (laugh)
1006		kinda like this am but look we do the best we can, **we muddle along as**
1007		**best we can**, am you know I tell them about well you know, he needs to go
1008		to [hospital] just so that they can check him out and that that's where the
1009		expert is and **it's a rare enough thing** and that's just where he needs to go
1010		am, it doesn't mean that he's going to be staying in hospital, it doesn't mean
1011		he's going to be getting operations, you know just that we need kind of need
1012		to link in with this person, they're the best in the country (.) for this thing
1013		am, you know and am, yeah, so that's kind of yeah the way we present it
1014		to the siblings so they certainly are- they would be aware that he has a
1015		diagnosis^

The line "we muddle along as best we can" (line 1006) above may illuminate the philosophy by which Judy lives (a concept as explored earlier in the subtheme *'Learning to live: "we just need to live our life now"* within the first GET *'Entering the world of diagnosis: "There's nothing wrong but everything is wrong").*

Robyn was the only participant who spoke about the implications surrounding genetic hereditability, specifically in relation to talking about diagnosis with siblings, as seen in Extract 12.14 (e.g., "I don't know at 18 would we sit down and say do you want to get yourself tested for this or not", lines 538-547)":

Extract 12.14

Robyn / Nathaniel / Siblings: "a future thing" (line 579)

533	P	em, yeah so I think they're, I I do wonder because it is a genetic diagnosis and
534		I did ask about whether to get the two oldest boys tested and they said we
535		couldn't do that until they were, it would have to be optional for them to do
536		it, em, but really it would be, and again, I suppose I don't know is it something
537		that we would, so **we don't freely throw around that diagnosis with the kids,**
538		they wouldn't know that it's there, I don't know at 18 would we sit them down
539		and say do you want to get yourself tested for this or not
540	R	Mmm
541	P	If you're thinking of having kids, do you want to get yourself tested, I'm not
542		really sure whether we would go down that route or not

For Robyn, she identified "we don't freely throw around that diagnosis with the kids" (line 537). This is in conflict with Judy's orientation, in that Declan's siblings know about his diagnosis, which is similarly rare and genetic in origin. The word "throw" used by Robyn above may imply a level of carelessness, which may be suggestive of the fragility which Robyn feels such a rare genetic diagnosis ought to be handled with. In the lines that follow this extract, Robyn concluded "I think we'll cross that bridge when we come to it so I'm not really sure (laughing)" (lines 546-547). Robyn cast these considerations as "a future thing" (line 579).

12.5 Summary

In summary, the majority of the mothers, Robyn, Mary, Judy, and Olivia all interpret that the siblings feel second place in comparison to the time and attention their siblings' demand. Their accounts inform the sixth GET identified in the data of *'The world of the sibling: "Second fiddle"'.* In the first subtheme '*Experience of the sibling: Perceived and 'sibling voice''* we see how Judy, Mary, Robyn and Olivia, interpret the impact on the sibling and how the sibling views Declan, Alexander, Nathaniel and Anna receptively. Included within this subtheme, is the mother's endeavours to try to support the siblings. Within the subtheme of *'Role of the sibling'* we examine the stance the mothers adopt on what the sibling *should* and *should not* be responsible for. Within the final subtheme, I presented Judy and Robyn's differing perspectives on what and how to talk to their

siblings about their brother's rare genetic diagnosis. Extracts and insights on the sibling experience are limited within Claire's account, given Rose's brother was an infant at the time of the interviews. The implications of the identification of this GET in the data contributes to the overall discussion and implications of findings as dealt with in the final chapters of this thesis.

Chapter 13: Discussion - Summarising the Research and Situating the Findings within the Literature

13.1 Review of Study's Aims

This study set out to explore how the phenomenon of 'diagnosis' is experienced by mothers of children who have a rare or undiagnosed neurodevelopmental condition in Ireland today. Literature on the experiences of parents whose children have rare or undiagnosed conditions is particularly lacking in Ireland, so this study aimed to contribute novel insights to the field. I anticipated such insights might have implications for education, training and clinical practice which could ultimately improve healthcare services. Specifically, I aimed to enhance understanding on how the construct of 'diagnosis' is perceived by mothers within the context of their child with a rare or undiagnosed neurodevelopmental condition, how 'diagnosis' may interact with daily life for these families, what importance these parents attribute to 'diagnosis', and to understand any unique challenges for this group. I also considered my research might provide insights into parental perspectives of broader constructs including disability and parenthood. (Please refer to *1.2 Research Questions* for a detailed listing of the main and sub-research questions).

13.2 Introduction to Discussion Chapters

I will present three discussion chapters. In this chapter, Chapter 13, I will first summarise the original contribution of the research, specifically how my research adds novel interpretations from the particular perspective of mothers of children with rare or undiagnosed conditions in Ireland (*13.3 Summary of Findings: Novel Insights and Trajectory and Body of Experience*). Next, I will consider the findings within each GET, relative to the current literature, and explore how they may help to inform and contribute to existing premises when considered in the context of the same or related fields (*13.4 Situating the 'World of* Diagnosis' to *13.9 Situating the 'World of Siblings')*.

In Chapter 14, I will offer theoretical reflections regarding implications of the study for the concept of 'diagnosis'. I will discuss the study as an exemplar of IPA and what it may add to surrounding the theory of hermeneutic phenomenology. I will also include a final researcher reflection, from the vantage point of having reached the end stages of the research.

In the final chapter, Chapter 15, I will offer possible practical implications of the research for healthcare communication, services and procedures, education and policy innovations and developments. I will also discuss limitations of the study, recommendations for further research and culminate with an overall conclusion.

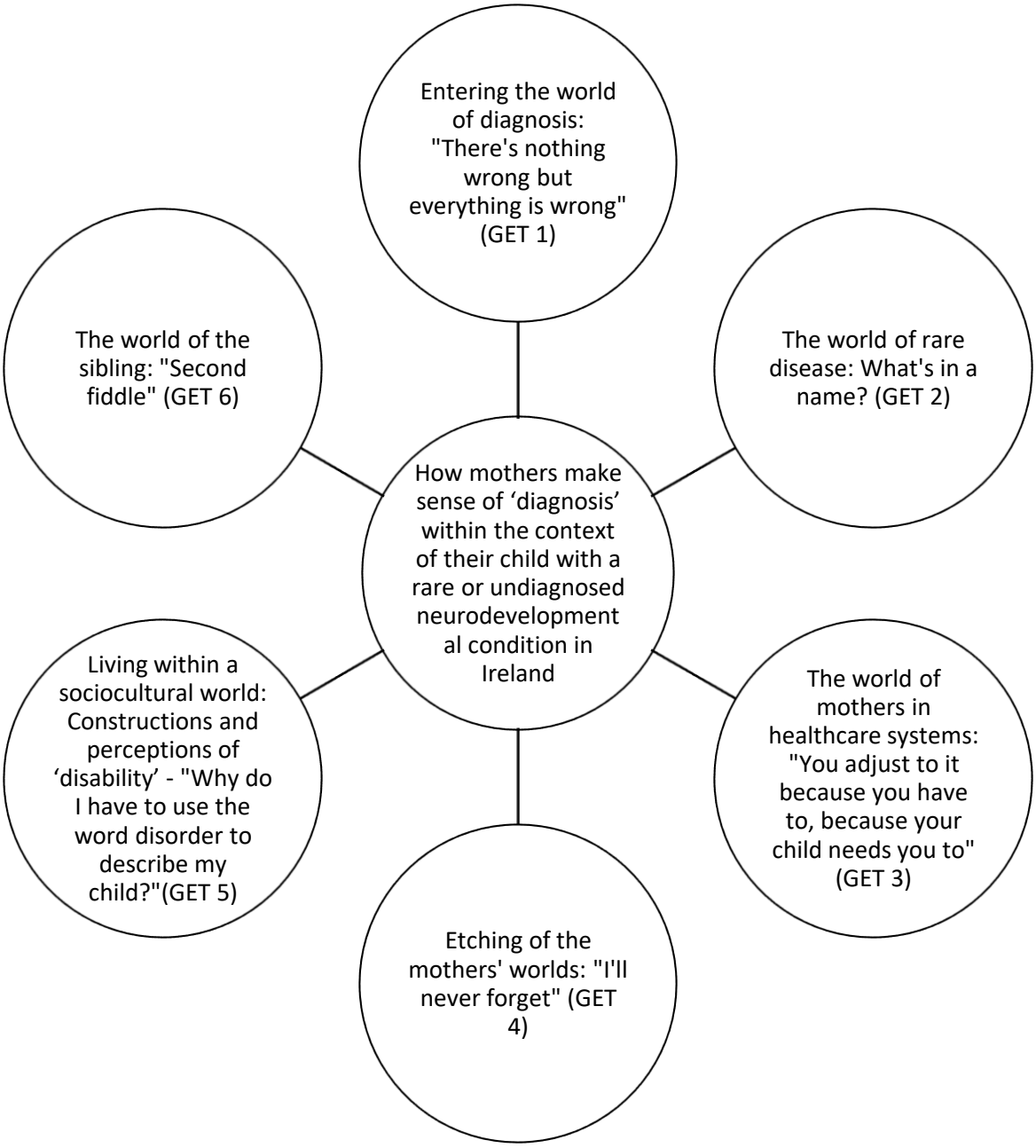
13.3 Summary of Findings: Novel Insights and Trajectory and Body of the Mothers' Experience

This study offers novel insights into the phenomenon of study by presenting in-depth accounts of five mothers' experiences, living in the Irish healthcare system and their own sociocultural context. IPA afforded me the opportunity to provide a discrete lens on the witness experience of mothers of 'diagnosis' for their children with rare or undiagnosed conditions. This perspective has historically been marginalised due to the 'rare' nature of the conditions. I believe one of the most distinctive findings to be in relation to GET 4 'Etching of the mothers' worlds: "I'll never forget". I interpret reference to how the mothers *remember* in relation to the words spoken and 'looks' given by others, specific timelines, and memorable moments, is a more nuanced finding compared to similar studies in the area of childhood illness, rare disease, and the wider illness experience. This study is also unique with regards to its methodology, primarily an IPA study with distinct inclusion of aspects of Narrative Analysis (i.e., presentation of individual story summaries and detailed narrative senses for each participant). The study's findings allowed the stories of the participants be heard in an individual matter and also in a collective way. How the study's methodological considerations have added to the findings will be discussed in the next chapter, under Section 14.3 *IPA as Permitting Entrance into the Participants' Lifeworlds.*

In terms of summarising the mothers' collective experiences, the following figure, Figure 13.1, provides a visual depiction of the six general experiential themes identified from my analysis of the data (this is a re-production of Figure 6.1, as previously presented, to support the reader in considering the findings, as a whole).

Figure 13.1

Graphical Depiction of General Experiential Themes in response to the Research Question 'How is the phenomenon of 'diagnosis' experienced in Ireland today by mothers within the context of their child with a rare or undiagnosed neurodevelopmental condition in Ireland?



The main findings from this research show how the mothers make sense of 'diagnosis' for their child with a rare or undiagnosed neurodevelopmental condition. The experience seems to involve entrance into a series of progressive worlds. The mothers' journeys move from pregnancy and birth through a period of *'entering the world of diagnosis' (GET 1).* For Claire, Mary and Olivia, this time

is marked by searching for an answer. In contrast, Robyn received notice of a medical complication during pregnancy and a genetic diagnosis for Nathaniel when he was around one year. For Judy, she was shocked by an unexpected diagnosis when Declan was 6 years old, having been unaware that they were looking for a unifying diagnosis. Claire and Olivia in particular described the experience of when there was something wrong but nothing wrong, leading to maternal self-doubt and questioning. Olivia spoke about having her concerns dismissed by healthcare professionals. The mothers at large attested to the lack of transparency and co-ordinated care within healthcare systems. At different points, the mothers moved into '*the world of rare disease*' (GET 2) which encompasses challenges associated with the rarity, anonymity or absence of diagnosis. All mothers describe a palpable difference between the supports and services available to themselves as parents, and their family, in comparison to groups of children with other more commonly understood diagnoses, such as autism or Down syndrome. The mothers unanimously declare a strong desire to be connected to other mothers of children in the same situation, attributing this as one of the main reasons for wanting a diagnosis. The mothers also contemplate how to *live* within this world, in the knowledge of a rare diagnosis or in the absence of unifying diagnosis.

'The world of mothers in healthcare systems' (GET 3), already having entered the worlds of diagnosis and rare disease (whether identified or undiagnosed) focuses on how the mothers learn to advocate for their children's needs through knowledge gained from navigating healthcare systems up to this point. There is tension in the adopted mother role and the perceived boundaries between parent and professional accountability. On the one hand, the mothers express a desire to be listened to, but this is juxtaposed with a pressure felt by the mothers in being responsible for managing their child's care. The experience of becoming institutionalised is reflected on by Mary and Claire. The subject of maternal trauma, and switching between parent and healthcare professional, are each themes informed uniquely by a single participant. The next GET '*etching of the mothers' worlds*' (GET 4) involves the enduring impact of looks and words of healthcare professionals and the vivid details with which the mothers remembered significant moments.

'*Living within a sociocultural world: Constructions and perceptions of disability'* (GET 5) is informed by how the mothers live and interact within society and their social worlds in relation to their child with a rare or undiagnosed condition. This GET explores the mothers' positions on generally recognised debates in the field of diagnosis and disability, namely issues on the value or harm of diagnostic labels and how to talk to other people about your child's needs or diagnosis. This theme provides an entrance into the mothers' views on disability and the language of disability at large as well as delivering insights from multiple other perspectives, through reported dialogues as recalled by the mothers. The final world is the '*the world of the sibling'* (GET 6). The majority of the mothers

acknowledge that the siblings fall second place in line, with regards to their sibling with a rare or undiagnosed condition by the nature of the time and attention they demand. The world of the sibling includes the mothers' interpretations of how the siblings make sense of or view their sibling. The mothers provide guidance on what they believe constitutes the sibling role, and whether and how to talk to the sibling about their brother or sister's diagnosis.

In summary, the findings provide novel insights into the trajectory and body of experience for mothers of children with rare or undiagnosed neurodevelopmental conditions in Ireland. The study offers detailed reflections on how the mothers *enter* into the world of diagnosis, further *funnel* into the discrete world of rare disease, *become* mothers of children with rare or undiagnosed conditions, how they *remember* and *recall* their experiences within their multiplicity of words, *live* within the sociocultural world and consider their other children. The world of the sibling highlights the need to recognise an experience as having an impact on the whole family unit.

13.4 Situating the *'World of Diagnosis'*

This section relates to 'entering the world of diagnosis' (GET 1) which deals with how the mothers came to enter the realm of diagnosis (i.e., peri-natal diagnosis, normal pregnancy and birth, not meeting developmental milestones, lengthy hospital stays or frequent GP visits). This is a time where mothers either have an answer at the outset, are explicitly seeking an answer, or given a diagnosis without expectation. This theme includes reflections on the processes at the moment of diagnosis or receiving news about their child's health.

It is relevant to consider some of the existing literature on 'breaking bad news' and on the 'diagnostic moment' (Jutel, 2019) here. I would like to highlight the distinction between these two terms. 'Breaking bad news' has been described as informing a patient of any bad information regarding their health which may pose significant changes to or limitations upon their quality of life or which may negatively alter the person's view of the present or future (Fallowfield & Jenkins, 2004). The 'diagnostic moment' (Heritage & McArthur, 2019; Jutel, 2019a), however, refers specifically to the first time the diagnosis is named by a professional. The diagnostic moment is afforded the power of demarcating life into two distinct periods, life before, and after diagnosis (Fleishman, 1999; Jutel, 2019a).

Tobin (2006) produced a framework for conceptualising 'breaking bad news' in the context of giving and receiving cancer diagnosis, entitled '*Tripartite Transition: A Process of Inclusive Knowing'*. Tobin (2006) concluded that 'breaking bad news' was experienced across a trajectory, with three themes, moving from 'disturbance of the everyday world', 'surfacing within the lived world', and

'embodiment within the lived word'. My participants' experiences as described in '*entering the world of diagnosis'* can be understood as similar to Tobin's (2006) theme of *'Disturbance of being in the everyday world'.* Tobin's subtheme of '*suspected-knowing – 'knowing and yet not'* (2006, p. 149) echoes the sense of "there's nothing wrong but everything is wrong" found in my study. Tobin (2006) also speaks to the *'dismissive mantra – 'don't worry''* (2006, p. 247), which assimilates with what the mothers in my study described within the subtheme *'Dismissal and maternal questioning: Being "pooh-poohed" - "I must be going crazy".*

Similar to the idea of 'suspected knowing' (Tobin, 2006), literature on adolescents' experience of juvenile idiopathic arthritis[41] (JIA) reports on the experience and impact of 'diagnostic uncertainty' (Hoffart & Wallace, 2023; Wakefield et al., 2023). Wakefield et al. (2023) report findings from one mother speaking to this experience: "[The] challenge was that the believability, the invisibility of it, going through multiple providers, trying to convince them, and you knowing your child best, and what was going on, and still not being heard" (p. 347). This quote echoes the 'relief' of sorts as described by Claire in my study on the confirmation of something being wrong when she first identified her daughter Rose was experiencing seizures. In Wakefield et al. (2023), one mother described her perception of the source of her daughter's frustration as "not being believed" (p. 347) and the sense of validation that came with diagnosis for her daughter in affirming "she's not crazy" (p. 347). Mary in my study speaks a lot about the importance of being believed and listened to in interactions with healthcare professionals so that she does not feel she has to prove herself or made to feel 'mad' or 'crazy'.

It is evident from reviewing the literature, parents' experience surrounding illness for their child, irrespective of the origin or type of illness, is commonly represented as comprising both an 'inner' and 'outer' experience. Jacoby et al. (2018) identified eight themes in relation to parental experiences of end-stage renal failure for their children. The authors separated the themes under two over-arching headings, denoting the 'int***ra***subjective experience' (for example including loneliness, responsibility and guilt) and 'int***er***subjective experience' (for example including lack of understanding, the health-care system: between satisfaction and disappointment). Lewis et al. (2010) explored parents' experience of raising a child with no known diagnosis in the UK and similarly divided the parents' experience into the 'inner emotional' and the 'outer sociological' experiences. In relation to parents' experience of receiving lethal fetal diagnosis, Cote-Arsenault and Denney-Koelsch (2011) identified themes across two dimensions, the 'personal pregnancy

[41] Defined by Wakefield et al. (2023) as a "childhood autoimmune, inflammatory condition with associated chronic pain, but with well-defined diagnostic criteria" (p. 341).

experience' (for example including 'grieving multiple loses' and 'my baby is a person') and 'interactions of others' (for example including 'fragmented health care' and 'disconnected family and friends'. Within my findings, 'the world of mothers in healthcare systems' and 'living within the sociocultural words' may similarly be considered as representing the 'inner' journey of mothers and 'outer' interactions within the sociocultural world. I consider these two worlds as inseparable and cannot be disentangled. Both the inner and outer experiences undoubtedly interact with one another and influence how the mothers perceive and respond within their lifeworlds.

Nicholl (2008) explored the experiences of mothers in caring for children with complex needs in Ireland. Similar to my research, Nicholl's (2008) study set out to fill a known gap in the literature surrounding needs of children with complex conditions, including rare, progressive, and life-limiting disorders to advance the development of Irish healthcare services. Nicholl (2008) conceptualised the mothers' experience as being discernible across a series of worlds "an inside world" at home, an "outside world" and a "going-between" world (2008, p. 115). Nicholl identified eight dimensions (will be elaborated on in *13.5 Situating 'the world of mothers in healthcare systems')*, common to all worlds but which may be more evident in some worlds than others.

In the context of parental experience of rare disease for their child in Canada, Baumbusch et al. (2018) defined "the diagnostic journey" (p. 82), as comprising seeking, receiving diagnosis and post-diagnosis support. This trajectory is more commonly referred to by other authors as the 'diagnostic odyssey' (Bauskis et al., 2022, p. 233). Moore (2021) examined the experience of people with young onset dementia (YOD) in Ireland. Moore (2021), similar to Baumbusch et al. (2018), identified various stages in the process involving *'pathway to diagnosis and disclosure of the YOD', 'experience of living with YOD'* and *'the personal sphere and YOD'.* In my study, I have considered the diagnostic journey as involving a multitude of worlds, moving from entering the world of diagnosis, the world of rare disease, becoming a mother in healthcare systems, the sociocultural world, and the world of the sibling.

Marsh et al. (2018) explored fathers' experience of the diagnosis of intellectual disability for their child. This study aimed to fill gaps in the current literature base by providing insights into fathers' experiences within the Irish context which the authors state were limited. Of the 10 fathers included in the study, four of the participants had an unknown diagnosis with one query diagnosis of autism and the remaining six children had a diagnosis of Down syndrome. This study similarly found fathers wanted more information and were disappointed with healthcare systems in receiving diagnosis on their own and ambivalence from the GP. Lack of co-ordination in healthcare for those with rare or undiagnosed conditions is supported in the literature (Currie & Szabo, 2019;

Lewis et al., 2010). Skotko and Bedia (2005) explored parents' experiences of how diagnosis is delivered in Spain. The authors identified a want for candid disclosure of clinical hypothesis (even in absence of confirmed diagnosis), privacy, and information at the time of diagnosis, and timely direction to community support services. A desire to know what healthcare professionals are thinking even if they do not know, well presented information and a private space and access to a parent support group are all findings reported within my study. This point links to the ideas as discussed by Reiser (1980) on the history of truth-telling practices in medicine and guidance from Boyd (1996), who advised practitioners to give as complete and honest accounts as possible, in respect of medical ethics.

Smits et al. (2023) identified common needs for parents of children with rare disease in the Netherlands, including for example empathic communication, a desire for information, and for parents to be prepared to ask questions of healthcare professionals surrounding their child's care. Chiaraluce (2018) explored the influence of the subconscious assumed family and gender reals in relation to how parents navigate "doing family" in America, where their child has a diagnosis of autism. In Chiaraluce's (2018) study, a participant, Sarah, mother to six-year-old boy with diagnosis says "If I knew we were going to get an autism diagnosis, I would have at least asked my husband to come. So, I remember walking out of the doctor's alone, in shock, and then driving home silently, still in shock" (p. 2890). These findings resonate with the findings from my study regarding Judy's reflections on the moment of diagnosis. Examining the process and challenges of diagnosis in other conditions shows how my research findings may have applicability beyond the topic being studies (i.e., rare or undiagnosed conditions) to contribute to a body of literature and understanding of 'diagnosis' at large. In terms of considering what meaning diagnosis has for parents, Avdi et al. (2000) looked at parents' experience of an autism diagnosis. Avdi et al. (2000) reported benefits such as supporting the diagnostic announcement to others, access to services, validating own concerns, and understanding their child and harm such as worry of stigma by diagnosis. The authors concluded similar conflict amongst meaning for parents in terms of "complex and multiple meanings that parents employ around the diagnosis of their child" (Avdi et al., 2000, p. 252). This conflict is similar to the finding in my research that the participants expressed varied views with regards to the "labelling" effects of diagnosis or the purpose it holds for them. The possible influence of my research on adding understanding of the phenomenon of 'diagnosis' will be discussed further in *14.2 Reimagining Diagnosis as a Triadic Concept: Introducing Diagnosis as a 'relative phenomenon'.*

13.5 Situating the *'World of Rare Disease'*

This section relates to GET 2 'the world of rare disease' which explores mothers unique challenges specifically in relation to their child having a rare or undiagnosed condition. Commonalities amongst the mothers' experiences involve questioning what meaning does a rare diagnosis offer, lack of understanding of rare disease in education and healthcare settings, consensus on desire for a designated support group and coming to a way to live life.

Bauskis et al. (2022) explored the parental experience regarding the diagnostic journey in relation to their child with an undiagnosed condition in Australia. In Australia, there is an established charity SWAN Australia for parents of children with an undiagnosed condition, founded in 2012. Bauskis et al. (2022) findings, includes parents' reflections on the benefits of having such a group, "We (in the support organisation) were all on the same page'" (p. 7); "'We can connect through stories and what we've gone through and if they've tried something different, and just to have a little bit more explanation like why he was …so short in height because no one has been able to explain that. So…it's just little things.'" (p. 9-10). Similarly, Carmichael et al. (2015) advise on the need for service providers to connect parents to support services for those with undiagnosed conditions, in the United States of America. Smits et al. (2022) echo the same the need for social supports for parents of children with rare or complex conditions in the Netherlands. All the mothers in my study reported on a lack of support for parents and siblings of children with rare disease in comparison to more recognised conditions (as discussed within the subtheme '*not fitting in with the "ASD Moms…or Down Syndrome Moms"').* My findings add rich insights informing the need for a specially dedicated parent group for parents of children with rare or undiagnosed conditions, and specified sibling supports, in the Irish context, akin to those available for more commonly understood diagnoses.

The desire for and benefits of a peer support group following diagnosis is a commonly reported finding in the existing literature, in relation to a wide variety of other contexts. The literature includes examples relating to the first-hand experience of illness and the witness experience (i.e., where a family member is affected by diagnosis), such as those with autism (Postavaru et al., 2022), neurodevelopmental disorders (Simon et al., 2022), Klinefelter syndrome, (Bourke at al., 2014), Parkinson's disease (MacPhail, Hanson, & Kuhnke, 2022), people with a diagnosis of monogenic diabetes (Guan, Maloney & Pollin, 2020), and people with young onset dementia (Moore, 2021).

What can be taken, when viewing my findings within the global research, is that there are commonalities between parents' experiences across international healthcare systems. Research, conducted internationally, exploring parents' experience of rare disease and the 'diagnostic odyssey' have indicated a need for family-focused and better integrated and co-ordinated care

(Bauskis et al., 2022; Carmichael et al., 2015; Currie & Szabo, 2019; Baumbusch et al., 2018; Smits et al., 2022). Dharssi et al. (2017) assert that rare disease is a "global public health issue" (p. 1). The authors evaluated rare disease policies, referred to as "National Rare Disease Plans" (p. 3) from 11 countries, excluding Ireland, against five paradigms of patient needs including improving coordination of care, diagnostic resources, access to treatments, patient awareness and support, and promoting innovative research. The authors concluded that implementation of rare disease plans is unequivocal across countries. Somanadhan et al. (2022) conducted research to identify research priorities for those living with rare disease within the Irish context. My research may contribute insights from parents' real-life experience in Ireland which may help to inform some of the identified priority areas including *"support at the time of diagnosis", "education and training"* and *"patient voice"*. Similar to challenges described by the mothers in my study, lack of understanding and knowledge of rare disease amongst healthcare professionals and those in education settings, is echoed in the literature (Currie and Szabo, 2018; Wakefield et al., 2023). This challenge is supported by my findings and discussed in particular, most visibly and vividly by Judy, in relation to her interactions with school and disability teams in relation to lack of knowledge or understanding of her son Declan's rare diagnosis (as discussed mostly within the subtheme of '*Rare disease: "a series of letters and numbers that doesn't mean anything to anyone").*

Smits et al. (2022) also identified a need for 'empathetic communication' and 'psychological support' (p. 1) amongst common needs found for parents of children with a rare disease or complex condition in the Netherlands. These findings resonate with my findings, as discussed most explicitly by Mary in relation to directions for healthcare professionals. In relation to 'empathetic' communication, Mary speaks about needing doctors *that care,* are kind, caring and factual in how they present information (as mostly explored within the subtheme '*Looks that speak volumes and words I'll never forget'*). In relation to direction for psychological support, Mary commented on the trauma she experienced in relation to Alexander's birth (as discussed within the subtheme 'Finding your voice, lessons learned').

Pollard et al. (2021) conducted focus groups to explore what parents value regarding outcomes of genetic testing for their children in Canada and the UK. Their study identified projected expectations and preferred outcomes for genomics to diagnose rare disease in children including "improved management strategies, reduced stress and anxiety, increased knowledge, access to community" (p. 3). Uncertain or inaccurate results was identified as a potential negative. In contrast, Mary, Claire, Olivia express preference for professionals to share what they know or do not know, even if they have to retract that finding later (as mostly discussed within the subtheme *'Labelling',* see section 11.1).

My study offers some insights into participants' experiences of genetic testing in relation to including lack of informativeness on what tests are being done and outstanding, delays in carrying out genetic testing, preparing parents for the diagnostic meeting, and follow-up information and care. My study also adds to considerations in the case of heritable conditions regarding genetic sharing information of diagnosis with the child's sibling in relation to genetics with their considerations for having children in the future.

In relation to the world of rare disease, Spillman et al. (2017) set out to explore stories of illness from first-hand and witness (i.e., parents' experience of illness in relation to their children) perspectives of illness experience in the context of undiagnosed conditions. The authors used Frank's (1995, 2013) narrative illness types to describe the most predominant underlying narrative in the participants' stories. Spillman et al. (2017) found the *voice of chaos* coexists with being undiagnosed. The chaos narrative is defined by lack of control and not foreseeing life getting better (Frank, 1995). Although my study's findings also point to identified challenges with the experience of being undiagnosed, I would not consider the over-arching climate of chaos. There are notable moments where the participants reference a need to "live" and pause the search for diagnosis. This finding perhaps shows the mothers' ability to adopt a new narrative to facilitate living, over the lack of control associated with no diagnosis. Refer to previous discussion in *6.2 Participant Story Summaries* and within the subtheme *'Learning to Live' (8.4).*

13.6 Situating the '*World of Mothers in Healthcare Systems*'

This section relates to GET 3 'the world of mothers in healthcare systems'. Interpretations of the mothers' experiences within this theme consider the tension between assumed mother roles and responsibilities, gender and parent roles, and lessons learned from navigating healthcare interactions.

The multiplicity of roles occupied by mothers when they have a child with a rare or undiagnosed condition is well documented in the research. Currie and Szabo (2019) identified that as a result of fragmented care mothers are required to adopt roles as *advocates, case managers, and medical navigators.* Similarly, Bauskis et al. (2022) found parents of children with undiagnosed condition occupy roles of *'navigator, expert and advocate'* (2022, p. 12). Nicholl (2008) completed a detailed study on mothers' experience of caring for their child with complex needs in Ireland, as previously referenced. Nicholl (2008) identified eight distinct dimensions of care-giving including for example *'normal mothering', 'technical care-giving', 'pre-emptive care-giving', 'constant communication',* showcasing the complexity of parenting in this context. Kaniamattam and Oxley (2022) explored the varied roles of mothers in caring for their children with developmental disabilities and

associated complex communication needs in South India. The authors identified roles including *'tutors', 'case managers', 'disciplinarians', 'nurses'* and *'orchestrators of home'.* Mothers are again considered to ascribe these roles to accommodate for a lack of professional understanding and fractured care. The mothers in my study add complementary data to this finding. The multiple roles of mothers are most evident with the theme of *"Searching for* an answer or "firefighting" moment by moment?" and the GET under discussion. Examples from my findings which complement findings in the existing research include reports of services as a "jigsaw" puzzle, fighting and firefighting, monitoring symptoms, taking responsibility to ask questions of professionals, educating others on your child's condition.

Swallow et al. (2011) compared fathers' and mothers' experience in developing skills in managing their child's long-term medical condition. The study found that both parents participated in care-giving. The authors identified fathers tended to occupy the 'protector' role and worried more about long-term health and well-being of the children, in comparison to mothers who occupied concern within current clinical issues and managing relationships with professionals. Dabrowka and Pisula (2010) found an increased level of parental stress in mothers of autistic preschool children in comparison to fathers with no difference found amongst parents of children with Down syndrome. Exploring stress and coping styles of the mothers was beyond the scope of the current research. However, my study adds thick descriptions into differentiated gender and parent roles within the subtheme of '*Maternal role and responsibility: balancing trust and onus'.*

Fisher and Goodley (2007) identified three narratives in the stories of mothers of children with disabilities and discuss how each of these narratives influenced care-giving. Fisher and Goodley (2007) conclude "*interwoven and multi-layered narratives reflect complex lifeworlds and suggest that mothers' understandings of their child's disability are constantly open to renegotiation and flux"* (p.76). This study highlights narrative as a relevant gateway into understanding how the experience of disability for their child is being lived by parents, so that consideration to alternative narratives, may be supported and nurtured. The idea that the mothers in my study are actively living an unfolding narrative is evident in my analysis.

A consideration of Western cultural portrayals of ideal motherhood has a place across all themes, in that the preconceived notion of what a mother ought to be and ought to do may influence the participants' internal consciousness, external societal expectations, and the perpetual interplay between those inner and outer voices. The construction of ideal motherhood will be discussed more wholly within *13.7 Situating 'the sociocultural world'.*

13.7 Situating *'Etching of the Mothers' Inner Worlds'*

This section relates to GET 4 'Etching of the mothers' inner worlds' which discusses how the mothers remember significant moments in terms of the words and looks of healthcare professionals and others. This theme also relates to how participants remember significant moments with immense specificity and in relating to time.

The lasting impact of healthcare professionals during interactions with patients has been considered in terms of sharing information and communicating bad news practices in medical care. Over four decades ago, Reiser (1980) reviewed the medical debacle of sharing information with patients, and to what extent truth should be shared in order to do least harm in medical practices over time from ancient Greek, medieval times to the modern-day physician (as referred to in *Researcher Reflections 10.1* and *11.1).* Reiser (1980) discussed the challenges for practitioners who opt to divulge potentially upsetting health news with patients. Reiser (1980) advised physicians must consider effective communication, one that supports and acknowledges the psychological burden of the news, and which affords time in the process of disclosure. More recently, Waxler et al. (2013) discussed recommendations for practice on the diagnostic process in relation to the experience of families receiving a diagnosis of Williams syndrome. In recommendations for practice, the authors call healthcare providers to consider "remember your words will have a lasting impact on the family" (Waxler, p. 540).

Jutel (2019a) also explores historical thinking on sharing diagnosis with patients and how much to share. Jutel refers to the diagnostic moment as "a moral, not a clinical, stance" (2019, p. 66) which involves giving of a '*truth'* (p. 66). Jutel (2019a) concludes that *whole* truth telling is a clinical expectation in modern day medicine, as opposed to an issue for the physician to consider. The participants in my study speak of their desire for being told the truth (as discussed in subtheme *'Searching for an answer or "firefighting" moment by moment?").* My study contributes insights into the diagnostic process for rare disease and provides considerations for how practice could be improved to support the child and family experience within this context.

Within my research, the subtheme '*Etching of the mothers' inner worlds'* resonates with other ideas presented in the current disability literature. In examining parents' experiences of children with Intellectual Disability, Bostru et al. (2009) identified a theme of 'time orientation – past, present or future?' (p. 97). Additionally, Roberston (2015) provided an anecdotal account of her experiences mothering an autistic teenager, Ben. She forefronts the temporality of her experience. She wrote:

> Notions of time are central to the discourse around disability, both in terms of medical discourse and definitions but also the cultural discourses that surround disability. In fact, living with disability often opens up a temporal focus that wasn't there previously. There is diagnosis time, prognosis time…curative time…relapse or remission, and there is developmental time, that imaginary ideal trajectory of human development based on finding the mean of able-bodied/minded people's experiences. (Roberston, 2015, p. 3)

13.8 Situating the *'Sociocultural World'*

This section relates to the GET of 'Living within a sociocultural world: Constructions and perceptions of 'disability' - "Why do I have to use the word disorder to describe my child?". This theme includes consideration of the mothers' view of labelling, experience of other's perceptions of their child and the mothers' own apparent views on disability.

Chiaraluce (2018) introduced the term "Doing Family" in reference to the impact of cultural ideations of the normal or ideal family on parents of children with a diagnosis of autism. For participants in Chiaraluce's (2018) study, support groups are considered a place of relief from experiences of "otherness" which families outside of the normative family model reside. Chiaraluce (2018) identifies a need for families to reconstruct narratives that move away from pre-defined normative constraints, as living within heteronormative constraints of what it means to 'do family', leads to "social exclusive and marginalization" (p. 2903). Similarly, Robertson (2015) spoke about the constraints of the 'ideal motherhood'. In my study, Claire in particular expressed strong rejections of pity which may inform research on parental interpretation of disability (as discussed mostly within the theme '*Onlookers: "to me he's a beautiful child inside and out, no matter, disability or not, you know but other people don't understand it"').* Goodley et al. (2019) note "d*isability is not a flaw, an individual tragedy nor a whispered recognition of another's embodied failing or a shameful family truth*" (p. 973). This sentiment appears to be reflected in Claire's account.

Of relevance to the consideration of the influence of culture on experience, Choi et al. (2005) explored how mothers experienced mothering relative to constructs of ideal motherhood embedded in society. Choi et al. (2005) found that expectations of ideal motherhood contributed to feelings of inadequacy amongst mothers which led to greater endeavours to conceal this to offer a portrayal of "supermum, superwife, supereverything" (p. 167). How having a child with disabilities interrupts this framework is considered within feminist disability studies. In my study, the mothers' experience of entering the world of diagnosis may be better understood when considered within the context of ideal motherhood personifications. Malacrida (2009)

acknowledges that expectations of ideal motherhood are unrealistic and unattainable for any mother, irrespective of additional needs or differences for the child. Although it was beyond the scope of my research to explicitly explore how the mothers in my study were influenced by these predominant existing narratives, the influence of these narratives may aid understanding into the mothers' thoughts and actions as discussed across GETs.

13.9 Situating the *'World of Siblings'*

This section relates to GET 6 'the world of the sibling', as told through the mothers' perception of the sibling experience, considered role of the sibling and whether or how to talk to siblings about their brother or sister's diagnosis.

The sibling relationship has been identified as one of most enduring throughout a person's life, outlasting the length of the parent-child and spouse relations (Tomeny et al., 2017). This astute observation brings a stark awakening to examine this relationship, how it may be impacted when one sibling has an additional need. Existing representations of the sibling experience are referred to by others as "silent voices of siblings" (Gera, Martin & Zahra, 2021, p. 77) and "forgotten child" (Mandleco & Webb, 2015). This is similar to my finding of the siblings as "second fiddle".

Some studies have explored the experience of a homogonous groups of siblings, for example, siblings of autistic children (Leedham et al., 2020; Moss, et al., 2019; Pavlopoulou et al., 2022) or siblings of children with 22q11 deletion syndrome (Goodwin, Alam & Campbell, 2017). Other studies have compared the experience of a heterogenous cohort of siblings belonging to two sample groups determined by their sibling's diagnosis. For example, Manleco and Webb (2015) examined the sibling experience of living with a sibling with Down syndrome in comparison an autistic sibling. Studies also vary according to the age of the siblings at the time of participating in the study. The age of the sibling at the time of research is an important factor in attending to how experiences are lived and made sense across the lifespan. Considering research with older adults (such as Goodwin et al., 2017; Moss et al., 2019; Tomeny et al., 2017) may also help to inform recommendations for practice to support siblings of children with disabilities growing up. Reflecting on views from adult siblings to inform current practice for child siblings, is similar to how we may refer to adult experiences of getting a diagnosis of autism to understand why we might want to talk to our children about their diagnosis.

Gera et al. (2021) spoke to children whose siblings presented with varied disabilities living in Malta. The authors concluded siblings of children with disabilities have unique, and often unmet needs. The implications of the study recommended that siblings also require attention from their parents,

and services for information to understand their siblings' difficulties, and to provide peer support. The finding that siblings would benefit from peer support services is commonly cited in the research (Goodwin et al., 2017; Leedham et al., 2020; Mandleco & Web, 2015). Don Meyer is considered the father of the 'Sibshop' model (Meyer & Vadasy, 2008). Sibshops are facilitated in disability services internationally, including within practice in Ireland. The aims and benefits of such groups have been documented. Limited published research on the models facilitated in Ireland. An example is reflections of a Sibshop in Southern Ireland (i.e., County Cork), involving three disability services, namely Enable Ireland, Brothers of Charity, COPE Foundation (D'Arcy et al., 2005).

Jacoby et al. (2018) coined a term called the 'ill unit' which serves to demarcate the emotional space of a child's illness which is occupied by the child and his or her parents. Taking account of the existing research and my findings, I would like to extend this term to include the siblings as occupying space with this 'ill unit'. Carpenter et al. (2004) describe a week of training days that took place in USA July 2002. This article showcases how a need for professionals to work with the *whole* family have been around and visible for over 20 years. The take-home provocation from the week was that "families of children with special needs must be recognised, involved and celebrated" (Carpenter et al.,, 2004, p. 75). During the week, restricted views of parents as encompassing family were torn and reconfigured to include siblings, grandparents and other family members. The conference was open to family and professionals. Some days were aimed towards particular members, for example a day for mothers ('Mum's the Word'), and adult siblings ('A Lifelong Journey'). Smits et al. (2022) also identify a need for "family-focused" care (p. 4) in their study on parent experiences of children with rare disease. My study provides nuanced insights into the sibling voice (as recalled by the mothers), navigating issues such as supporting the sibling, talking to the sibling of diagnosis and roles, in the particular case of rare or undiagnosed conditions in the Irish context. My study's findings provide further argument and evidence for services to attend to the needs of the whole family in care, including the sibling and specifically the allocation of services to facilitate sibshops.

13.10 Summary

In this chapter, I have provided a summary of existing research as is relevant to situating my findings. In the following chapter, I will turn to examine more closely what my study may add to understanding the construct of diagnosis, and any methodological implications as an exemplar IPA study.

Chapter 14: Discussion - Theoretical Considerations

14.1 Introduction

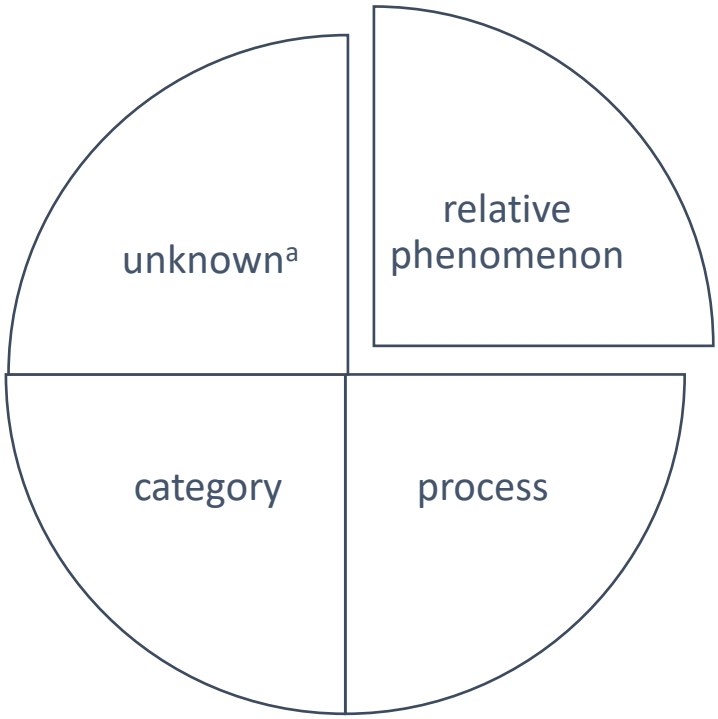
As previously outlined, in this chapter I will discuss the possible theoretical contributions of my study in adding to existing understandings surrounding the construct of diagnosis. I will suggest an expanded definition for 'diagnosis' based on the study's findings. I will also consider how the application of IPA allowed me to explore the phenomenon under question (i.e., how is the phenomenon of 'diagnosis' experienced in Ireland today by mothers of children who have a rare or undiagnosed neurodevelopmental condition?). I will specifically address the inclusion of elements of narrative research, as complementing IPA, in allowing the mothers' stories to breath (to adopt Frank's (2010) use of the phrase). I will conclude with a final piece of researcher reflection at this point, at the close of writing process.

14.2 Reimagining Diagnosis as a Triadic Concept: Introducing Diagnosis as a 'relative phenomenon'

Diagnosis has been defined by Blaxter (1987) and is regarded in the literature as a dual process, owing to both 'category' and 'process' (as first introduced in *2.2 Concept and Construction of 'diagnosis': What is 'diagnosis').* My interpretation of the data suggests reimagining this definition of diagnosis to include a third property, diagnosis as a 'relative phenomenon' Thus, diagnosis may be defined as at least a 'triadic' concept. I use 'at least' to appreciate the possibility that my study's findings are not finite or definitive and to remain open to further qualities or dimensions of diagnosis (see Figure 14.1 below for graphical representation). Blaxter (1978) introduced the idea of diagnosis as 'category' and 'process' in the case of alcoholism which she terms a "'social' diagnosis" (Blaxter, 1978, p. 9). She described diagnosis as category as pertaining to "a list of diseases" (p. 9) and process pertaining to "the thing the physician does: the conclusion reached, or the act of coming to that conclusion" (p. 9). Similar to how Blaxter (1978) used alcoholism as a vehicle to evidence new ideas about diagnosis, I consider my topic of rare or undiagnosed neurodevelopmental conditions provided a suitable medium to allow new understandings of diagnosis to breathe. My research findings may also offer new insights for diagnosis as 'category', and as, 'process', in addition extending to include diagnosis as a 'relative phenomenon'. Each of which, I will now explore in detail.

Figure 14.1

Graphical Depiction highlighting the Re-framing of 'Diagnosis' as a Triadic Concept



Note. 'Relative phenomenon' appears as an incoming sector to illustrate the 're-framing' of Blaxter's (1978) definition of diagnosis as a dual concept consisting of both 'category' and 'process'.

[a]The 'unknown' sector is represented to account for the possibility that my study's findings do not suggest a finite or definitive representation of diagnosis as a concept and accounts for possibility of further dimensions to be uncovered (see 14.2 above).

This study may contribute to understanding of diagnosis as *category* in terms of the category of rare disease and undiagnosed conditions. Blaxter (1978) did highlight that diseases vary according to their certainty, referencing "syndromes with unknown etiology" (p. 10) as the least defined. My data addresses the quandary of where there is less known about the disease origin or trajectory as is the case for rare diseases or undiagnosed conditions, do these titles afford the title of 'category'? This consideration is complementary to what Blaxter (1978) referred to as problems in the application of diagnosis as category or process in the case of a social diagnosis, where the diagnostic label does not accompany suggestions for management. The mothers' considerations to their experiences, as explored in the subtheme '*Rare disease: "a series of letters and numbers that doesn't mean anything to anyone*" (in *'The world of rare disease: What's in a name?',* GET 2)*,* mostly informs illuminations on this topic (see *14.1 Researcher Reflection* for further discussion).

Interpretation of the data may also provide suggestions for diagnosis as *process*, in terms of how information is shared, preparing families for the diagnostic meeting, follow-up care and healthcare communication within breaking bad news or the diagnostic moment. Implications for education and practice will be discussed in Chapter 15.

The advent of diagnosis as a 'relative phenomenon' is a concept that was highly evident throughout iterative examination of the data. As has been referenced many times throughout this thesis, the influence of 'Dasein' (Heidegger, 1927/2008) and 'horizons of understanding' (Gadamer, 2004) on how the mothers appear to be making sense of their experiences, and in turn, how I as a researcher interpret their sense-making, is undeniably apparent. The advent of diagnosis as a relative phenomenon is complementary to Brown's (1995) work owing to the 'social construction' of diagnosis. My research may heavily exemplify Brown's (1995) propositions that disease is differentiated from illness, and people classified as having the same disease will indeed *experience* illness differently. Brown (1995) discusses factors which influence the experience of illness including personality, race, class, sex, ethnicity, social support, viewed interference with activities of daily living (including occupational and social activities), and worldviews. My findings confirm various properties of diagnosis that were considered in the literature review (i.e., diagnosis as malleable, in that it is not fixed, may be revised or abolished; facilitative or prohibitive; less or more legitimate; accepted or denied)**.** Examination of the findings suggest the notion of 'relative' includes four interrelated dimensions of *experience, space, time and telling*, each of which I will expand upon below.

14.2.1 Relative to Experience

Experience of diagnosis may refer to the specifics of the situation, medical context, navigations in healthcare, social, education and occupational sites, both past and present. Diagnosis is relative to the historic landscapes with which the mothers came to enter experiences with their child, including their sociocultural upbringing, prior personal and family experiences, personal viewpoints. My study also supports a common finding in relation to the experience of diagnosis at large, that diagnosis 'requires company', so to speak. The study found all mothers expressed a strong desire for diagnosis to enable access to a group of parents who could share their common experiences. The mothers described wanting the same peer supports for the child's siblings.

14.2.2 Relative to Space

My study suggests diagnosis as a family event, with possible social and emotional consequences for family members. This notion is evidenced by the influence of diagnosis for their child on the mothers (as most clearly seen in 'The world of mothers in healthcare systems: "You

adjust to it because you have to, because your child needs you to"', GET 3) and in reference to impact on the sibling (as most notably seen in 'The world of the siblings: "second fiddle"). As discussed in the preceding chapter, taking account of the existing research, I am suggesting extension of what Jacoby et al. (2018) coined the 'ill unit' term to include the siblings as occupying space with the parent and child. Space also refers to the influence of beliefs omnipresent in certain cultures and geographical areas.

14.2.3 Relative to Time

The length of the 'diagnostic journey' and age at which diagnosis is given all appear to contribute to a person's experience of diagnosis. Timing also appears to influence the value and purpose attributed to diagnosis and is relative in the sharing of diagnosis. My findings suggest timing may influence the gradient of information shared at a particular time (e.g., in relation to what a sibling needs to know regarding their sibling's diagnosis at a particular point). Diagnosis might also be influenced by time in the way it is remembered (as mostly informed by the discussion pertaining to GET 4 'Etching of the mother's inner worlds: "I'll never forget"'). The idea of diagnosis as being remembered links with the notion of 'retrospection'. Diagnosis often affords understanding in hindsight to provide explanation for previous symptoms that came before the diagnostic moment. Diagnosis is often considered from this perspective of 'looking back' with its attendant ever-evolving thoughts and emotions.

14.2.4 Relative to Telling

I have highlighted on numerous occasions within the analysis chapters where the participants appear to be making further sense of their experiences, in real-time, through the process of telling in dialogue with the researcher. The mothers also referred to how they navigated telling others. Reflections on how the mothers spoke about their children appeared to afford insight into their own stance on disability and parenthood, which may have been brought to conscious awareness for the mothers themselves through telling of diagnosis and the response of the listener. My findings also suggest context as being important to how the mothers tell, which is dependent on who they are speaking with (i.e., level of knowledge on the topic of the other person, new or previously established relationship).

14.1 Researcher Reflection

Is 'syndrome without a name' (SWAN) a sufficient diagnostic label? The fact that the majority of mothers in my study were actively seeking a diagnosis or "answer" for their child (i.e., Claire, Mary, Olivia) in the absence of a named diagnosis, would imply that a "syndrome without a name" is not sufficient. However, when we consider what the participants *want* from diagnosis,

does this alter the value of 'syndrome without a name' or 'SWAN' as a diagnostic category. For example, Claire spoke of wanting to find access to support group as being her primary motive for seeking diagnosis. She also wanted a diagnosis to be able to explain about her daughter in interactions with others in a way that did not involve listing things her daughter could not do. If we do two things: (1) provide a dedicated support network for parents of children and their families with SWAN and (2) enhance public and professional knowledge of the term 'SWAN', can we be said to have afforded legitimacy to the 'SWAN' diagnosis? Thus, by extension, in doing so, are we supporting families, for whom a more 'specific' diagnosis is not possible, to end the 'search' and reach a 'category' of diagnosis and enable progression to learn to live with diagnosis. However, diagnosis is usually considered as something which directs further actions in terms of treatment as opposed to an exercise within itself (Blaxter, 1978). If SWAN does not carry information surrounding treatment or disease progressions, does the value of its category again fall into question? Additionally, the possibly transient or temporary nature of a SWAN diagnosis must too be considered. With continuing life sustaining medical advancements, research, and growing European and global rare diseases databases, what was previously identified as genetic difference of unknown origin may later be identified as relevant chromosomal deletions or duplications. I wonder how knowledge of the possibility of a future 'more etiologically based' diagnosis, may cast shade on the legitimacy of 'SWAN' as a diagnostic category?

14.2 Researcher Reflection:

I will provide a recent anecdote which I think exemplifies the embodiment of such influence on my current views towards health. I myself am of Ashkenazi Jewish descent and would be familiar with Yiddish phraseology. Accounting for Heidegger's notion '*Dasein*' (1927/2008), I cannot deny the potential influence of family roots and upbringing within a Jewish household in terms of contributing to my 'fusion of horizons' (Gadamar, 2004) relating to contributing to my views on health, illness, and family.

When encountering a woman who had fallen on the street, I stopped to attend to her and offer support and be a force of calm while waiting for the ambulance. In attempts to comfort the woman, I exclaimed "my Granny would call this a 'kepara'", a Yiddish phrase to which I offered an English translation "to prevent something worse from happening". The use of 'kepara' in this context illuminated how use of language can aid in making sense of the occurrence of illness. I think this example might also hint at the application of my study's finding that 'diagnosis' is a 'relative phenomenon'. This example highlights how religion has a place in contributing to the way in which individuals and whole families make sense of illness. Considering pre-historic influences

on my position, as above, is aligned with hermeneutic phenomenology in bringing to conscious attention one's own views to acknowledge how these [my] views may influence interpretation.

14.3 IPA as Permitting Entrance into the Participants' Lifeworlds

My research contributes to the literature on witness illness narratives offering unique experience-near rich qualitative data which, as Smith (2011) noted can serve to "shine light on the phenomenon" (p. 7). The phenomenon in this case, being what diagnosis means for mothers' experience the phenomenon of diagnosis in relation to their child with a rare or undiagnosed condition. In hermeneutic phenomenology, phenomena are said to be understood through 'being' (Husserl, 1970) and achieved through the consciousness. The 'hermeneutic circle' (Heidegger, 1927/2008), as previously defined, is an iterative process, which involves circular examination of the data moving from the parts to the parts within the whole, to understand the data. In hermeneutic phenomenology, it is this process which is said to provide rigor for the validity of the findings. Using IPA, allowed me in to interact with the participants-in-context and their relatedness to the experience being studied (Larkin et al., 2006). In fulfilment of IPA's commitment to convey a story (Nizza et al., 2021), through attending to the idiographic and convergence and divergence, I was able to present the experience of five mothers and a collective account of the experiences of mothers, at large.

Boden et al. (2019) describe the "interview arc" (p. 223), as used in the relational mapping interview approach, in the context of how drawings can be used to complement more traditional semi-structured interviewing within IPA. This interview arc is organised around a series of "touch points" (p. 224), namely "mapping the self", "mapping important others", "standing back" and "considering change" (p. 224). Inadvertently, my interview data could be seen to be re-organised along these touch points. 'Mapping the self' may include participants' references to their own thoughts and emotions on their own experiences. 'Important others' includes reference to significant others in terms of important people in the participants life and what Boden et al., (2019) referred to as "people who are part of the participants' quotidian social landscape but who may not be well known to them" (p. 225). I consider these people to comprise the 'characters' in the participants' living stories, in line the comparisons woven throughout my analysis of the participants' interpretations to film and theatre. 'Standing back' relates to evaluative comments, such as reflections recounted by the participants and those made live through the unfolding dialogue with the researcher during the interviews. 'Considering change' looks to the "ideal future" as described by Boden at al. (2019, p. 226). I interpreted what mothers would like others to know and presented these as clinical implications from the findings, which I will detail in Chapter 15.

IPA's attention to the 'linguistic' form of data, allowed me to transcend the language of the mothers' accounts into deeper meaning to better understand the phenomenon of study. Aristotle lauded metaphors, in saying "ordinary words convey only what we know already; it is from metaphor that we can best get hold of something fresh". The power of metaphors to transcend meaning is well recognised (Boden et al., 2019; Svendler 2009; Neefjes, 2022). Neefjes (2022) found parents used war metaphors such as 'fighter', 'hero' or 'trooper' (p. 427) to express feelings of pride about their children, where the child was living with a life-limiting condition. Cited examples include "courageously fighting their disease". Several metaphors used by the mothers in my study have been previously explored for their possible intended meanings, where they have been identified, within the findings' chapters[42]. Farr and Nizza (2019) and Smith (2011) discuss methods used to capture experiences over time using IPA. Attention to paralinguistic features and reported dialogue (as in *5.9.2 More Than Five Voices,* within the findings chapters at large and most explicitly within *10.2 Looks that speak volumes and words I'll never forget)* also afforded rich insights into the mothers' experience.

The employment of IPA as my chosen qualitative methodology has enabled me to illuminate how mothers' experience of the phenomenon of 'diagnosis' for them at one point in time, along their journey of parenting a child with a rare or undiagnosed neurodevelopmental condition in the Irish healthcare context. These findings contribute to a growing, yet limited, body of research into parents' experience of rare or undiagnosed conditions in Ireland. This study is unique in terms of its potential to add true, unfolding narratives and present experience-close findings, as afforded through the chosen methodology of IPA. IPA's recognition of the horizon of understanding of the researcher as influential in the double-hermeneutic process, adds to the unique quality of the data. In essence, this is a novel study in that it presents findings from a small number of mothers' interviews, in relation to their own *particular* experience of the phenomenon of diagnosis for their *particular* child with a rare or undiagnosed condition, as made sense of at a *particular* moment in time by the mothers and by me, as a researcher. This attention to the 'particular' is the gift of IPA's idiographic commitment. The phenomenological aspect of IPA enables understanding of the human experience, as it is being lived and reflected upon, through unstable relational and temporal paradigms. This is manifested in the findings with suggestion of diagnoses as a 'relative

[42] For example, Olivia's metaphor of "jigsaw" used to describe the lack of co-ordination within healthcare (as can be seen in Chapter 7, Extract 7.26, line 1028); Judy's description of Declan's father's understanding of diagnosis as similar to a father seeing the line on a pregnancy stick (Extract 9.12, lines 1142-1145); Claire's description of the hospital room as a "glass box" (line 379; see discussion in Section *9.3.2 '"Living in hospital": Maternal experience of becoming institutionalised'*).

phenomenon', which is influenced by experience, space, time and telling for any individual. The data from the individual interviews also afford the opportunity to capture the instability of perception of experience, given the participants readily describe their shifting priorities and hopes at different points along their journey.

IPA has philosophical underpinnings in phenomenology, hermeneutics, and ideography (Smith et al., 2021), which combine to deliver a methodology that attends to interpreting the meaning of a living experience for a particular individual, living with a particular time, and accounts for the shades of prior experiences and culture on the participants' perceptions of their current lifeworld. I considered and was cognisant of making the best effort to attend to quality indicators for qualitative research (Nizza et al., 2021; Yardley, 2000; Younas et al., 2023). In attending to these dimensions, I hope to have provided thick descriptions and a detailed, reflective analysis which augmented the transferability of the findings to other situations (Hays & McKibben 2021; Younas et al., 2023).

Somanadhan et al. (2021) reference the special ethical considerations needed to avoid a breach in confidentiality for the participants given the rare nature of their child's conditions places them in a more identifiable position. My study could be seen as an exemplar on how to manage data which could be potentially identifying for the participant, given the rare nature of the conditions. The challenge is to preserve the unique, individual quality to the data while honouring the ethical obligation to protect the identity of not only the participant, but significant others including the child themselves, siblings, healthcare professionals. The way this was achieved in my study could provide an example for others engaging in qualitative research with those with rare conditions.

14.3.1 Exemplar in IPA: Borrowed insights from Narrative Inquiry

In terms of more nuanced applications of IPA, I drew on both narrative analysis and Frank's (1995, 2013) work on illness narratives and typologies. I provided five individual stories (see *6.2 Participant Story Summaries)* which intended to summarise the key moments of each mother's own story, their main thoughts and sense-making with respect to diagnosis for their child. This method felt like a transparent and faithful way to honour the uniqueness of each mother's experience in relation to the phenomenon being studied. Riggs and Coyle (2002) provided precedence for what they termed 'analytic strategy' (p. 5). In their IPA study, the authors primarily presented a detailed case-by-case analysis, followed by an overview which attended to conclusions that could be made from looking across cases. Having considered this method, in line with guidelines for best practice in achieving quality IPA, I decided to instead present a series of participant story summaries as an

introduction to a detailed presentation of GETs as identified through analysis across cases identifying convergence and divergence.

As introduced in *3.3 Illness Narrative: Definitions and Considerations*, Frank (1995, 2005) proposed three main narrative types and attested these were not intended to be precluding. Other researchers have introduced variant types in relation first-hand and second-hand illness (e.g., Benbenisye et al. 2008; Harrington, 2008). In the table below, I have summarised the main narrative 'sense' felt through my iterative analysis of the participant interviews. The narrative 'sense' is intended to be different to the story summaries (as discussed above) and more closely aligned with Frank's definition of narrative 'type'. Frank (1995) defined 'narrative type' as it was defined by Arthur Frank as "the most general storyline that can be recognised underlying the plot and tensions of particular stories" (p. 75). I think inclusion of these narrative 'sense' is important to provide the reader with entry to the soul of the mothers' narratives. I have chosen to reframe 'type' as 'sense' in an endeavour to hint at the emotive force and aura that can be felt within the underlying events regarding diagnosis.

Table 14.1

Overall Narrative 'sense' felt within each of the Mothers' Accounts

Participant	Narrative 'Sense'
Robyn, Mother to Nathaniel	Position of luck or fortune with careful consideration towards diagnostic labels.
Claire, Mother to Rose	Preservation of personhood and rejection of assumed tragedy associated with disability.
Mary, Mother to Alexander	Living in fear of dying and searching for answers to help.
Judy, Mother to Declan	Grievances for the diagnostic processes balanced with understanding and a spiritual appreciation of blessings and fate.
Olivia, Mother to Anna	Abhorrence towards public services and looking for direction on how to cope with challenging behaviours in the absence of diagnosis[a]. Gratitude and acceptance in the knowledge of continued challenges and not having all the answers[b].

[a]As pertaining to Olivia's spoken account, provided during the semi-structured interview.

[b]As pertaining to the Olivia's subsequent written account.

In summary, IPA afforded me the opportunity to answer the research question at hand. IPA's underlying framework of hermeneutic phenomenology allowed the stories to breathe, in a way that shone light on the commonalities and differences within the participants' experiences whilst maintaining attention to the idiosyncrasies within the participants' experiences. Considering ideas from narrative research, which were congruent with IPA's methodology, aggregated my ability to

present the mothers' accounts as living 'stories'. This accompanied narrative lens contributes a considered application of IPA to the literature basis. I feel the coalescence of IPA and NA within this study permitted me to present findings which both preserve and illuminate the *individuals* and their *'stories',* as well as attending to the collective experience of these mothers.

14.4 Closing Researcher Reflection: What Does it all Mean Looking Back?

As has already been discussed at length within this thesis, IPA as a methodology recognises the integral role of the researcher in interpreting the participant's sense-making of the phenomenon being studied (Smith & Nizza, 2021; Starr & Smith, 2011). Inclusion of the researcher as an active agent in the sense-making process, aligns with hermeneutic phenomenology methodology (Laverty, 2003). Thus, in adopting a *hermeneutic phenomenological approach* to the research question, my position as a twenty-nine year old practising Speech and Language Therapist, clinician working within an interdisciplinary CDNT, identification as cisgender female, my religious upbringing and views, and currently having not experienced being a mother, amongst other cultural-historic and living experiences, confluence to fuse my own personal horizons of understanding (Gadamar, 2004). This fusion of horizons constitutes the lens through which I conducted all elements of this research project from design to analysis, interpretation and reflection. Further details on my 'kaleidoscope of understanding' was discussed previously in *4.4.1 'Researcher Stance: My 'Kaleidoscope' Through which I View the World; The Shape-Thrower which Crafts my Research Framework'.*

On reaching the end of this research journey, I find the words of the participants in my study, and the literature I engaged with throughout the research project ever prominent in my daily practice and clinical interactions with families and colleagues. Diagnosis as a process, through clinical hypothesis testing and assessment, and diagnosis as a construct, in terms of what it means for individual children and families, are conversations I navigate daily in interactions with families. When I meet with a new family, I enter with them on their own, unique family journey. I am invited to understand how they came to be 'mothers in healthcare systems'. For some families they are on the journey, before entering the world of diagnosis. For others they may be in the world or in the process of entering (i.e., for example through the process of Autism diagnosis), may be entering or within the rare disease world. I attend to the parents' journey and aim to hear where they are and meet them at that space. I am conscious of my potential lasting influence on the parents' etchings of their own lifeworlds. I am hyperaware that my words, looks and actions may leave enduring memories, positive or negative on the families I work with. While I acknowledge this knowledge carries a great weight as a professional, it is something that I think paramount for clinicians and healthcare workers to be able to monitor and reflect on our own practice.

Chapter 15: 'So What?' Practical Implications and Conclusions

15.1 Introduction

In this chapter, I will suggest practical implications from the findings, address certain limitations of the study with direction for how these may be addressed by future research, and end by offering an overall conclusion.

15.2 Implications of the Findings

IPA research can be considered a method and methodology which enables understanding of how people make sense of a particular phenomenon, and thus afford entry into their lifeworlds. Taken together, the findings of this study may provide implications for education and training for undergraduate medical and healthcare professional students and continuing professional development for practitioners and educators. In highlighting how mothers make sense of 'diagnosis' in relation to their child with a rare or undiagnosed condition, this study provides suggestions for healthcare procedures, communications, and services to support the attainment of what Mishler defined as "human***e*** healthcare" (1979, p. 3).

In the sections to follow and based on the implications of the findings of my study, some recommendations are suggested within the contexts of (i) healthcare communication, (ii) healthcare services and procedures, including considerations of the diagnostic process *per se,* (iii) healthcare education and finally, and (iv) policy innovations and developments.

15.3 Healthcare Communication

The findings suggest how healthcare professionals could significantly enhance their communication with children and families. These suggestions include conscious attention to the words and looks healthcare professionals use in interactions with children and families, with consideration to how these looks and words may be perceived and remembered (i.e., impact and *enduring* impact). This study found parents prefer when professionals listen, believe and act on early parental concerns in relation to the child's health and development. At the same time, while professionals should listen to parents, there needs to be balance between placing too much responsibility or burden on parents to monitor or report upon their child's health. In terms of transparency, professionals should favour candid and factual communication. Professionals should be open about clinical hypotheses, even if not confirmed, so that families can be informed and understand what tests are being carried out or outstanding, and *why.* The findings might suggest mothers' desire to be given advance notice of phone reviews and appointments, and of what appointments will entail so that they can make necessary accommodations and prepare both

logistically and emotionally. Professionals should consider the privacy of the physical setting for discussion with parents surrounding their child's health, and care management. Professionals should also consider the health literacy[43] of parents (or by extension to other fields, people / other family members) they work with and how to support the accessibility, understanding and application of oral and written information provided. Healthcare communication and services and procedures are inextricably linked. Therefore, further suggestions for communication will be included within Section *15.4 Healthcare Services and Procedures.*

15.3.1 'Reflective Toolkit': A Suggestion

In order to support the implementation of the study's findings, I suggest the creation of a reflective toolkit for healthcare professionals working with this cohort to improve self-awareness and attendance to parents' preferences for care. A suggested title for this toolkit is *'Medical, Health and Social Care Professional Reflective Practice Toolkit: Entering the World of 'Diagnosis': For use when working with Children with Rare and Undiagnosed Conditions and their Families, in Ireland.*" The toolkit would be based on the findings of this doctoral study and congruent with other research findings in the field and considered evidence based. I propose maintaining some direct quotations from the current study to preserve the voice of the parents and *'closeness'* in directly reaching and addressing the professionals. Affording time and space to examine clinical care and healthcare communications in clinical supervision meetings may also help to bring clinician's conscious attention to this important aspect of humane care and to effect and maintain positive change on service.

I have included a sample of what may be included in this toolkit. Figure 15.1 below presents a quadrant indicating mothers' desirable and non-desirable qualities of healthcare professionals and communication of healthcare professionals, with consequent outcomes for the child and family. The participants' direct words accompany such suggestions.

[43]Health literacy has been defined as the "cognitive and social skills which determine the motivation and ability of individuals to gain access to, understand and use information in ways which promote and maintain good health" (Nutbeam, 1998, p. 357).

Figure 15.1

Doctors that care: What it means for me, my child and my child's health[a]

Desirable Qualities and Practices	**Consequent outcomes for child and family**
• Listen. "Doctors that do listen are the actually really good doctors" • "Kind nature" • "Calming" • "Really gentle" • "Really factual" • Avoids drama. "Spoke to us just about the facts". Remove sensational language. • Be aware of your looks. "Looks speak volume to parents" • Remember the person above all. Be conscious to avoid referring to a child and family as "interesting" • Are caring and care. "You just need doctors that care about your child, regardless of their abilities" • "Get things done" • Honest. "Say I don't know if you don't know because then at least, you know, you trust these doctors more". • Give your best efforts. • "Are they going to believe me?". Trust parents' instincts, take parents at their word and trust in what they are saying. • Knowledge and experienced. • Comfortable in supporting children with complex needs. "Had a brilliant way with him". Knows how to be playful. • Demonstrate inclusion, equity, and equality. Treat the child with the value they deserve to be treated with. • Playful and pleasant with the child. • Time bountiful. Professionals who give you time.	• When you care, "I know I don't have to persuade [him] to do [his] best" • Parents can "relax a bit" • When you listen, "there's some reassurance in that, that I've relayed all my concerns to you and you as a medical professional are ok with those concerns" • Better medical outcomes from testing and procedures • More pleasant experience and associations for their child in attending medical appointments and interactions. Avoids or lessens child trauma. • Parents feel more "comfortable and safe" with a doctor who follows the formula.
Non-desirable qualities and practices	**Consequent outcomes for child and family**
• Harsh delivery. • Brushes you off. • Doesn't listen to you. • Avoidance. "Talks about an issue like a politician" • Feel like you have to prove what you are saying is true.	• "Make you feel like you're a bit mad" • "I start to doubt myself and wonder am I imagining these things?" • Won't get proper measurements. • Enduring trauma and negative associations for the child. This has repercussions when the child must attend future procedures.

[a]*Health is defined as "a state of complete physical, mental and social well-being and not merely the absence of disease or infirmity" (WHO, 1948).*

15.4 Healthcare Services and Procedures

In terms of ensuring optimal healthcare services and procedures for families accessing care, the findings of this study suggest several requirements. Access to a specifically dedicated support group in Ireland for children, and their families, affected by rare or undiagnosed neurodevelopmental conditions is a unanimous need identified by all mothers in the study. Disability services should provide supports for siblings of children with rare or undiagnosed conditions. This may look like further resources allocated into running *Sibshops* (Meher & Vedasy, 2008) so that these can be facilitated at more frequent intervals within yearly calendars, creation of peer networks for school age and young adult siblings to support establishing connections, and a forum for open discussion and provision of appropriate information (monitored and facilitated by healthcare professionals). Access to psychoeducation supports such as parent education and training could also be made available with regards to how to consider and support siblings. Where appropriate and indicated, access to one-to-one direct psychology services could be available for siblings.

The promotion of co-ordinated, transparent, timely and time-bountiful care has also been suggested from the findings. Transparent care might include better communication with families on processes, such as sharing of reports and correspondence pertaining to their child amongst professionals within and across involved agencies, informing families of onward referrals, and explanation of clinical rationale and working hypotheses. Co-ordinated care might include the linking of various elements of a child's care and sharing of professional opinion to present a clear and agreeable evaluation to parents. Timely and time-bountiful care may include timely responses to families (which may be acknowledgement that a raised concern is being followed up on, even if answers are not yet available at the time of communication) and affording sufficient time for duration of appointments to hear family views. Provision of timely care will ultimately rely on staffing and resources.

Follow-up emotional support for parents who have had traumatic (i.e., child in ICU) or lengthy periods of living in hospital is indicated. Further support for managing implications on family planning associated with genetic conditions would also be desirable. This support would include great access to genetic counselling for families. Ireland's campaign for rare disease, is asking for greater resources within genetics services in Ireland. A suggestion was to employ genomic resource associates to provide triage supports in reviewing referrals to free up genetic counsellor time and to facilitate shorter waiting lists for families to access these services.

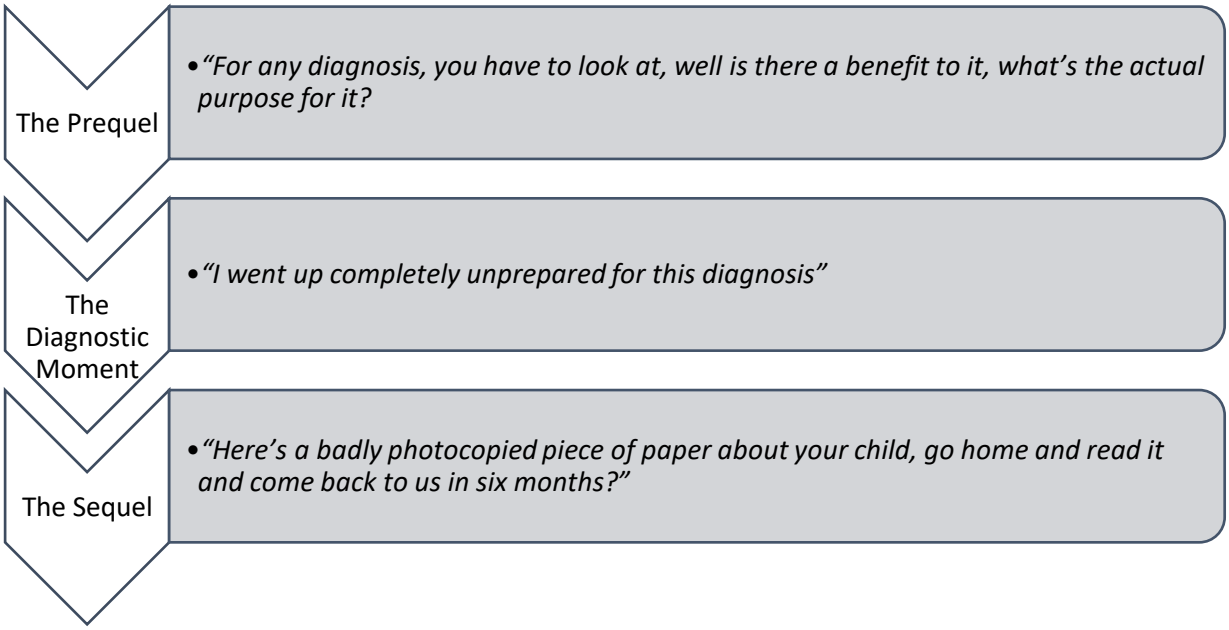
More specifically, in terms of *the diagnostic process,* I propose the following points to be considered. Application of these suggestions may extend beyond the context of rare or undiagnosed childhood conditions to include other diagnostic practices in the case of autism, developmental language disorder, and may even extend to adult diagnoses and first-person experience of illness for adulthood late diagnosis or adult-onset disease / conditions.

Firstly, practitioners should be transparent with families about their working clinical hypothesis, even if these hypotheses are still being tested and explored, as honesty has been expressed as a preference for families, even in the knowledge of diagnostic uncertainty. Secondly, in relation to preparing families for the diagnostic meeting, parents should be advised to consider bringing a support person with them and planning for transport to the appointment (as also indicated in *15.3 Healthcare Communication)*. Further to the diagnostic meeting, parents should be afforded time for processing and reflection; they should be given a determined pathway for how they can return to the professional who gave the diagnosis, following this period, to discuss or clarify any concerns or questions. Thirdly, when being informed about their child's diagnosis, parents should be given timely well-presented and balanced information regarding the named rare disease; such information may support parents in telling family or others about the child's diagnosis. Finally, parents should be directed to established supports in their local and wider communities as pertaining to the *whole* family, namely supports for the child (such as accessible leisure activities and peer networks), the parents and any siblings.

In relation to the *Reflective Toolkit,* introduced in Section 15.3.1 above, I have proposed a reflective checklist for the diagnostic procedure. The procedure is divided into three phases, 'the prequel' (time before diagnosis), 'the diagnostic moment' (pertaining to the moment of informing on diagnosis) and 'the sequel' (the follow-up actions not intended to be a 'tick-the-box'). Completion of this checklist is not intended to be an administrative exercise. Instead, it is intended to be an exercise in reflective practice as defined in the HSCP Reflective Practice Statement (2019, p.3) as "a process by which you stop and think about your practice, consciously analyse your decision-making, draw on theory and relate it to what you do in practice." The aim of this checklist is to encourage professionals to 'stop and think' about the child and family who they are working with and to reflect on their own practice, with the ultimate aim of enhancing the quality of our services in interactions and practice when supporting children and families with rare disease. The findings of this study suggest a need to be candid, clear, and transparent with families in communication about procedures pertaining to the diagnostic process (and indeed interactions for the child within the medical or clinical sphere). I have included a draft of this checklist in Figure 15.2 below.

Figure 15.2

Reflective Practice Checklist: 'Entering the World of Diagnosis'



The Prequel

☐ Have you understood your child or family's journey before the point you are meeting? ☐ Have you considered your child or family's thoughts and beliefs on diagnosis? What does it mean to the child and family? What are the perceived values attached to diagnosis? ☐ Have you informed parents about what testing you are carrying out? ☐ Have you communicated with parents what testing you are carrying out in a language that makes sense to them?	*"We would love to have a diagnosis, just purely for because, for that reason that we could go and find that group"* *"Why give another diagnosis just for the sake of it?"* *"Having a diagnosis to me would, would, well in some ways it might [help], it depends on what it is?"* *"At least with a diagnosis you'd know, roughly, maybe, what to expect, what's coming"* *"I wasn't aware that they were even testing for this RDD"*

The Diagnostic Moment

☐ Have you informed parents in preparation for the diagnostic meeting? ☐ Have you informed parents in preparation for the diagnostic meeting? ☐ Have you suggested a parent consider bringing a supportive person to the meeting? ☐ Have you considered and discussed with the parent whether it is appropriate for the child to attend this meeting? Have you suggested the parent may consider alternative arrangements for childcare? ☐ Have you considered the space for you are going to have the meeting? ☐ Have you considered the wording you are going to use? ☐ Have you prepared to be factual about the information you have? ☐ Have you prepared to be honest about what you do or do not know?	*"A heads up that something big is coming down the line"* *"I was on my own"* *"Declan was pulling at me for something"* *"I was distracted with him"* *"In front of everybody, in the war, told us how sick our child was. There was parents, I will never forget, there was parents standing right behind us"*

The Sequel

☐ Have you provided opportunity for the parent to return with any questions or concerns after they have digested the information? On that day, on the successive days? ☐ Have you provided information that is presentable? ☐ Have you provided parents with information on relevant support groups? ☐ Have you considered direction for sibling supports? ☐ Have you discussed options for emotional support or counselling or where to access these services?	*"You're there, you're responding, but you're not actually processing"* *"You should be given 15 minutes to kind of go away and have a chat amongst yourselves and come back with questions"* *"I had to chase and almost fight to get the answers to those questions"* *"Got handed a leaflet, that was very badly photocopied"* *"Here's a contact number, here's a support group"* *"I feel strongly about actually that they should come to you and say you know, this has been really difficult, do you need some help?"*

15.5 Education in Healthcare

The findings of this study also have implications that may inform educational practices in both undergraduate and graduate teaching and continuing professional development. For example, the curriculum of medical and healthcare professional courses should include information on, and attention to, rare or undiagnosed conditions. Directing students and healthcare professionals to available parent supports and information, such as the previously referenced national rare disease plans, clinical pathways for the most common rare diseases (HSE, 2023) and professional knowledge sharing groups (i.e., European Reference Networks) is pivotal to prepare students and professionals for appropriate work with this client group. Moreover, greater education and training, specifically on elements of humane care, as detailed at the opening of this section (15.2 *Implications of the Findings*), should be included within teachings on rare and undiagnosed disease, and in relation to general clinical practice. Instruction on the origins and theoretical constructions of 'diagnosis' (such as a 'triadic concept' as specifically exemplified in this thesis) should be included within curricula. Additionally, healthcare professionals should be supported to engage in continuing professional development, including being informed on the advancements in the field of rare disease. Finally, clinical supervisors and management should advise professionals of local campaigns and new research in the area.

Involvement of parents in teaching within college curricula (such as parents speaking on their witness illness experience and experience of healthcare systems and 'diagnosis') and on programs for continuing professional development, would also support providing future and practising medical and healthcare professionals with understanding of parents' and families' experiences. Such understanding may lead to more harmonious and compassionate healthcare. Consultation with parents on directing further research in the field is also indicated. Public patient involvement in IPA research may include for example, consultation on how to recruit and include certain groups (i.e., such fathers, and siblings of children with rare or undiagnosed conditions), and perhaps to help guide what interview probes to consider.

15.6 Policy Innovations and Developments

The findings from my study come at an opportune time when the needs of people living with rare disease and their families are being campaigned for, globally, and notably within the Irish context (as illustrated within 3.2 *Current Landscape*). My findings may contribute mothers' perspectives on the meaning of 'diagnosis' in relation to their child with a rare or undiagnosed condition, in Ireland, to inform the new *National Rare Disease Plan* (Department of Health, 2023, "Minister for Health announces development of new National Rare Disease Plan", para 3). The study's findings might also make a case for the creation of, and adherence to, local standard

operating procedures and pathways within childhood acute, social care, community, and other services. Such procedures and pathways may refer to considering the sibling experience, signposting counselling and psychology supports at various points with a parent and family's journey, and reserving space for reflection on *'humane'* healthcare communications and practices within clinical supervision (all as detailed more specifically in 15.4 *Healthcare Services and Procedures* above).

15.7 Study Limitations and Direction for Further Research

Although this study afforded the researcher the opportunity to explore in-depth the experiences of *mothers* and obtain evidence on the 'mother' role specifically, further research examining the meaning of diagnosis for other family members and healthcare professionals who occupy a shared "lived world" (Larkin et al., p. 183), would be useful in triangulating understanding on the phenomenon. Further research is needed to understand how the phenomenon of study is made sense of by fathers, other family members as carers, and siblings. The experience of fathers of children with disabilities is underrepresented in research (Marsh et al., 2018; Mitchell & Lashewicz, 2019). It would be pertinent to explore the topic of enquiry further from the father perspective (i.e., what is *their* perceived role?). How do *fathers* make sense of the experience of rare or undiagnosed experience for their child? How do fathers perceive the *mothers' role*? Such research would add to understanding of illness as a whole family experience (as previously detailed, specifically in relation to extension of Jacoby and colleagues, 2018, term 'ill-unit' on p. 237). Furthermore, my study offered some insights into the sibling experience from the parent perspective (i.e., 'witness' accounts). Interviews with siblings would provide first-hand accounts of the sibling experience in relation to the phenomenon under study. Examining the experience, knowledge, and confidence of healthcare providers supporting families with rare or undiagnosed neurodevelopmental conditions, would also help facilitate patient-professional understanding, preferences, and priorities for healthcare interactions.

The study presents how the experience of the phenomenon under investigation is lived by five particular mothers. Although this study attends to practices surrounding humane care (as defined by Mishler, 1979; 2005), it does not consider in much detail, ethics of social justice which Mishler (2005) describes looks at inequality within healthcare in terms of race, ethnic and class variables. It would be useful to address the topic with a more diverse range of mothers, including a consideration as to how the concepts of diagnosis and illness are lived and made sense of by other cultures and languages.

Future researchers could consider developing a *'Medical, Health and Social Care Professional Reflective Practice Toolkit: Entering the World of 'Diagnosis': For use when working with Children with Rare and Undiagnosed Conditions and their Families, in* Ireland' (as introduced in Section 15.3 above) using a co-design process in its creation to incorporate multiple perspectives such as the six-step co-design process (Boyd et al. 2010, 2012) or Experience-Based Co-Design (EBCD; Donetto et al., 2015). The latter EBCD, is said to be underpinned by: (1) participatory action research; (2) user centred design; (3) learning theory; and (4) narrative-based approaches to change (Donetto et al., 2015), and might provide suitable method. Fylan et al. (2021) describe the use of EBCD as a healthcare improvement tool. Co-design methods facilitate the involvement of multiple stakeholder groups (i.e., family and healthcare professionals) to work together to identify priorities, implement change and reflect on and celebrate the achievements.

Future researchers might also consider use of a constructivist grounded theory (GT) approach (e.g., Charmaz, 2012; Timonen et al., 2018) to explore process and generate theory as to why the phenomenon at hand is experienced as such (i.e., 'What factors influence how the diagnostic odyssey is experienced by parents of children with rare or undiagnosed conditions?'). The usefulness of GT to present experiences in healthcare has been demonstrated in the literature (e.g., Foley & Timonen, 2015). Potential challenges in addressing the current phenomenon of study may include access to a large enough sample to reach 'theoretical saturation' (Timonen et al., 2018).

15.8 Conclusion

In summary, my findings contribute original insights to a very limited body of qualitative research within the realm of rare disease and offer unique insights through in-depth exploration of how five mothers' make sense of the phenomenon of 'diagnosis' in relation to a rare or undiagnosed disease for their child, in the Irish context. My interpretation of the findings suggest that the mothers' experience can be understood across the six related themes, as already discussed:

(1) *Entering the world of diagnosis: "There's nothing wrong but everything is wrong"*
(2) *The world of rare disease: What's in a name?*
(3) *The world of mothers in healthcare systems: "You adjust to it because you have to, because your child needs you to"*
(4) *Etching of the mothers' inner worlds: "I'll never forget"*
(5) *Living within a sociocultural world: Constructions and perceptions of 'disability' - "Why do I have to use the word disorder to describe my child?"*
(6) *The world of the sibling: "Second fiddle"*

The mothers' experiences appear to vary in terms of how they come to *enter* the world of diagnosis, whether they were searching for an answer, given a pre-natal or unexpected diagnosis. The findings suggest mothers experienced varying levels of being dismissed by healthcare professionals, and maternal self-questioning in relation to unidentified needs for their child. In terms of *rare disease*, the mothers provided consensus on their wish to access to specifically dedicated support group for parents for children with rare or undiagnosed conditions, and sibling supports. The common experience of lack of knowledge surrounding rare disease and the challenges of this in terms of accessing services, education and in telling others about the child is evidenced. Additionally, the findings suggest the mothers commonly experience and report on a difference to how diagnosis is experienced for their family in comparison to more understood, *'mainstream'* diagnoses. The pursuit for diagnosis and how useful it may be, appears to be *relative* to the individual family and variable along the individual mothers' diagnostic journey. The study contributes to the 'labelling' debate with some mothers finding labels useful for access to services, while others noting their potential harm if they are maintained when they no longer apply.

This study presented a nuanced methodology, using Interpretive Phenomenological Analysis as the primary methodology, with borrowed insights from Narrative Analysis, to enable a more tangible entrance into the mothers' lifeworlds. The study also provided a re-defining of 'diagnosis' as a phenomenon characterised by at least three dimensions: 'category', 'process' and, additionally, from the findings of this study, *'relative'.* Diagnosis as a *relative* phenomenon speaks to the influence of experience, space, time and telling on how 'diagnosis' is experienced. Whilst the findings suggest there is a common trajectory for mothers of children who have rare or undiagnosed conditions, through a series of progressive worlds, there maintains unique elements for every individual family pertaining to their own fusion of horizons and worldview, which may be understandable as their narrative 'sense'. This 'sense' does not appear to be static and continuously evolves through ongoing interactions within their lifeworlds.

The implications of the study include suggestions for healthcare communication, services and procedures, and education and policy innovations and developments, where in Ireland supports for those with rare or undiagnosed conditions are underdeveloped or lacking. Implications for healthcare may extend beyond the context of study to other childhood diagnoses and indeed adult diagnoses and first-person experience of illness. In terms of healthcare communication and procedures, suggestions are made for greater attention to the words and non-verbal behaviours (e.g., looks, facial expressions) used when speaking with parents and families; preference for transparent and honest communication; 'boundary-ing' of parent and professional roles; better co-ordination of care, and specifications regarding the diagnostic process (i.e., preparing families

for an appointment, providing a private setting and opportunity for follow-up care). As well as knowledge on the common experience of families affected by rare or undiagnosed conditions, healthcare professionals should seek to understand each family's own needs, preferences, and perspectives on 'diagnosis', viewing these areas as evolving on a continual basis.

Limitations of the study might include the fact that the phenomenon was explored from the position of mothers only. In terms of future research, it would be useful to extend the current findings by examining the phenomenon of study from a multi-perspectival dimension (including fathers, other family carers, siblings, and healthcare professionals) to support further advancements.

In sum, ultimately this research offers unique entry into mothers' lifeworlds, relative to how they make sense of '*diagnosis'* for their child with a rare or undiagnosed condition, whilst preserving the individual experience of *each* mother yet at the same time giving voice to their collective experience. This study also affords new layers of meaning to the term diagnosis as *relative* to experience, space, time and telling.

References

Aftab, A. (2022, May 23). *Neurodiversity and the social ecology of disability.* Psychiatric Times, *39*(5). https://www.psychiatrictimes.com/view/neurodiversity-and-the-social-ecology-of-disability

American Psychiatric Association. (1987). *Diagnostic and statistical manual of mental disorders* (3rd ed., revised [DSM-III-R]). American Psychiatric Publishing, Inc.

American Psychiatric Association. (1968). *Diagnostic and statistical manual of mental disorders* (2nd ed. [DSM-II]). American Psychiatric Publishing, Inc.

American Psychiatric Association. (1994). *Diagnostic and statistical manual of mental disorders* (4th ed. [DSM-IV]). American Psychiatric Publishing, Inc.

American Psychiatric Association. (2013). *Diagnostic and statistical manual of mental disorders* (5th ed. [DSM-5]). American Psychiatric Publishing, Inc. https://doi.org/10.1176/appi.books.9780890425596

American Psychiatric Association. (2022). *Diagnostic and statistical manual of mental disorders* (5th ed., text revised [DSM-5-TR]). American Psychiatric Publishing, Inc. https://doi.org/10.1176/appi.books.9780890425787

Arantzamendi, M., Garcia-Rueda, N., Carvajal, A., & Robinson, C. A. (2020). People with advanced cancer: The process of living well with awareness of dying. *Qualitative Health Research, 30*(8), 1143-1155. https://doi.org/10.1177/1049732318816298

Archibald, M. M., Ambagtsheer, R. C., Casey, M. G., & Lawless, M. (2019). Using Zoom videoconferencing for Qualitative Data Collection: Perceptions and Experiences of Researchers and Participants. *International Journal of Qualitative Methods, 18,* 1-8. https://doi.org/10.1177/1609406919874596

Austin, J. L. (1975). *How to do things with words* (2nd ed.). Harvard University Press.

Avdi, E., Griffin, C., & Brough, S. (2000). Parents' constructions of the 'problem' during assessment and diagnosis of their child for an autistic spectrum disorder. *Journal of Health Psychology, 5(*2), 241-254. https://doi.org/10.1177/135910530000500214

Basel, D., & McCarrier, J. (2017). Ending a diagnostic odyssey: Family education, counselling, and response to eventual diagnosis. *Paediatric Clinics of North America, 64*(1), 265-272. https://doi.org/10.1016/j.pcl.2016.08.017

Baukis, A., Strange, C., Molster, C., & Fisher, C. (2022). The diagnostic odyssey: insights from parents of children living with an undiagnosed condition. *Orphanet Journal of Rare Diseases, 17*(233). https://doi.org/10.1186/s13023-022-02358-x

Baumbusch, J., Mayer, S., & Sloan-Yip, I. (2018). Alone in a crowd? Parents of children with rare diseases' experiences of navigating the healthcare system. *Journal of Genetic Counselling, 28*(1), 80-90. https://doi.org/10.1007/s10897-018-0294-9

Beach, W.A. (2001). Introduction: Diagnosing "lay diagnosis". *Text & Talk, 21*(1-2), 13-18. https://doi.org/10.1515/text.1.21.1-2.13

Benveniste, S., Goldzweig, G., & Jacoby, R. (2020). Illness narratives through the eyes of parents with children with end-stage renal disease. *Qualitative Health Research, 30*(12), 1798-1806. https://doi.org/10.1177/1049732320938040

Berkovits, L. D., Moody, C. T., & Blacher, J. (2020). "I don't feel different. But then again, I wouldn't know what it feels like to be normal": Perspectives of adolescents with autism spectrum disorder. *Journal of Autism and Developmental Disorders, 50,* 831-843. https://doi.org/10.1007/s10803-019-04309-1

Blashfield, R. K., Keeley, J. W., Flanagan, E. H., & Miles, S. R. (2014). The cycle of classification: DSM-I through DSM-5. *The Annual Review of Clinical Psychology, 10*, 25-51. https://doi.org/10.1146/annurev-clinpsy-032813-153639

Blaxter, M. (1978). Diagnosis as category and process: The case of alcoholism. *Social Science and Medicine, 12,* 9-17. https://doi.org/10.1016/0271-7123(78)90017-2

Blaxter, M. (2009). The case of the vanishing patient? Image and experience. *Sociology of health and illness, 31*(5), 762-778. https://doi.org/10.1111/j.1467-9566.2009.01178.x

Blume, S. (1994). Making the Deaf hear: The cochlear implant as promise and threat'. *Medische Antrhopologie*, *6*, 108-121.

Bourke, E., Snow, P., Herlihy, A., Amor, D., & Metcalfe, S. (2014). A qualitative exploration of mothers' and fathers' experiences of having a child with Klinefelter syndrome and the process of reaching this diagnosis. *European Journal of Human Genetics, 22*, 18-24. https://doi.org/10.1038/ejhg.2013.102

Bouwman, M. G., Teunissen, Q. G. A., Wijburg, F. A., & Linthorst G. E. (2010). 'Doctor Google' ending the diagnostic odyssey in lysosomal storage disorders: Parents using internet search engines as an efficient diagnostic strategy in rare diseases. *Archives of Disease in Childhood, 95*(8), 642-644. http://dx.doi.org/10.1136/adc.2009.171827

Boyd, J. W. (1996). Narrative aspects of a doctor-patient encounter. *The Journal of Medical Humanities, 17*(1), 5-15. https://doi.org/10.1007/bf02276310

Boyd, H., McKernon, S. & Old, A. (2010). *Health Service Co-design: Working with patients to improve healthcare services*. Auckland: Waitemata District Health Board.

Boyd, H., McKernon, S., & Old, A. (2012). Improving healthcare through the use of co-design. *The New Zealand Medical Journal, 125*(1357), 76-87.

Braiden, H., Bothwell, J., & Duffy, J. (2010). Parents' experience of the diagnostic process for autistic spectrum disorders. *Child Care in Practice, 16*(4), 377-289. https://doi.org/10.1080/13575279.2010.498415

Braun, V., & Clarke, V. (2021). Can I use TA? Should I use TA? Should I *note* use TA? Comparing reflexive thematic analysis and other pattern-based qualitative analytic approaches. *Counselling and Psychotherapy Research, 21*, 27-47. https://doi. org/10.1002/capr.12360

Braun, V., & Clarke, V. (2022). Conceptual and design thinking for thematic analysis. *Qualitative Psychology, 9*(1), 3-26. https://doi.org/10.1037/qup0000196

Brown, P., & Levinson, S. C. (1987). Politeness: Some universals in language usage. In E.N. Goody (Ed.), *Questions and politeness: Strategies in social interaction* (pp. 56-289). Cambridge University Press.

Brown, P. (1995). Naming and framing: The social construction of diagnosis and illness. Journal of Health and Social Behaviour, Spec No, 34-52.

Burke, P. (2004). *Brothers and Sisters of Disabled Children.* Jessica Kingsley Publishers.

Campbell, F. K. (2009). Contours of ableism: The production of disability and abledness. Palgrave Macmillan. https://doi.org/10.1057/9780230245181

Cardiff and Vale University Health Board. (2022, October 26). *Britain's first SWAN clinic offers hope for patients in Wales with syndromes so rare they don't have a name*. https://cavuhb.nhs.wales/news/latest-news/britains-first-swan-clinic-offers-hope-for-patients-in-wales-with-syndromes-so-rare-they-dont-have-a-name/ https://cavuhb.nhs.wales/news/latest-news/britains-first-swan-clinic-offers-hope-for-patients-in-wales-with-syndromes-so-rare-they-dont-have-a-name/

Carmichael, N., Tsipis, J., Windmueller, G., Mandel, L., & Estrella, E. (2015). "Is it going to hurt?": The impact of the diagnostic odyssey on children and their families. *Journal of Genetic Counselling, 24*(2)*,* 325-335. https://doi.org/10.1007/s10897-014-9773-9

Carpenter, V., Addenbrooke, M., Attfield., & Conway, S. (2004). 'Celebrating families': An inclusive model of family-centred training'. *British Journal of Special Education, 31*(2), 75-80. https://doi.org/10.1111/j.0952-3383.2004.00332.x

Chatman, S. (1978). Story and discourse: Narrative structure in fiction and film. Cornell University Press.

Charlick, S., Pincombe, J., McKellar, L., & Fielder, A. (2016). Making sense of participant experiences: Interpretative phenomenological analysis in midwifery research. *International Journal of Doctoral Studies, 11,* 205-2016. https://doi.org/10.28945/3486

Charmaz, K. (2009). Shifting the ground: Constructivist grounded theory methods. In J. M. Morse, P. N. Stern, J. Corbin, B. Bowers, A. K. Charmaz & E. A. Clarke (Eds.), *Developing Grounded Theory: The Second Generation* (pp. 127-193). Routledge.

Charmaz, K. (2012). The power and potential of grounded theory. *Medical Sociology Online, 6,* 2-15.

Charmaz, K. (2020). “With constructivist grounded theory you can’t hide”: Social justice research and critical inquiry in the public sphere. *Qualitative Inquiry*, *26*(2), 165-176. https://doi.org/10.1177/1077800419879081

Charmaz, K., & Thornberg, R. (2021). The pursuit of quality in grounded theory. *Qualitative Research in Psychology, 18(3),* 305-327. https://doi.org/10.1080/14780887.2020.1780357

Choi, P., Henshaw, C., Baker, S., &Tree, J. (2005). Supermum, superwife, supereverything: Performing femininity in the transition to motherhood. *Journal of Reproductive and Infant Psychology,* 23(2), 167-180. https://doi.org/10.1080/02646830500129487

Cooper, R. (2005). Classifying madness: A philosophical examination of the diagnostic and statistical manual of mental disorders. Springer.

Criminal Law (Insanity) Act 2006 (IRE)

Criminal Law (Insanity) Act 2010 (IRE)

Crotty, M. (1998). *The Foundations of Social Research: Meaning and Perspective in the Research Process.* Allen and Unwin.

Cudjoe, E. (2022). Using diaries with interpretative phenomenological analysis: Guidelines from a study of children whose parents have mental illness. *International Journal of Qualitative Methods, 21,* 1-9. https://doi.org/10.1177/16094069221084435

Currie, G., & Szabo, J. (2019). “It’s like a jungle gym, and everything is under construction”: The parent’s perspective of caring for a child with rare disease. *Child Care Health Development, 45*(1)*,* 96-103. https://doi.org/10.1111/cch.12628

D’Arcy, F., Flynn, J., McCarthy, Y., O’Connor, C., & Tierney, E., (2005). Sibshops: An evaluation of an interagency model. *Journal of intellectual disabilities, 9*(1), 43-57. https://doi.org/10.1177/1744629505049729

Denman, K., Smart, C., Dallos, R., & Levett, P. (2016). How families make sense of their child’s behaviour when on an autism assessment and diagnosis waiting List. *Journal of Autism and Developmental Disorders, 46*(11)*,* 3408-3423. https://doi.org/10.1007/s10803-016-2873-7

Department of Health. (2014). *National rare disease plan for Ireland 2014-2018.* https://assets.gov.ie/37342/da70fc6fadd24425b98311e679f4406b.pdf.

Department of Health. (2023, February 23). *Minister for Health announces development of new National Rare Disease Plan* [Press release]. https://assets.gov.ie/37342/da70fc6fadd24425b98311e679f4406b.pdf

Dharssi, S., Wong-Reiger, D., Harold, M., & Terry, S. (2017). Review of 11 national policies for rare diseases in the context of key patient needs. *Orphanet Journal of Rare Diseases, 12*(63), 1-13. https://doi.org/10.1186/s13023-017-0618-0

Dilthey, W. (1976). *Selected Writings* (H. Rickman, Ed., Trans. And Introduction). Cambridge University Press.

Donetto, S., Pierri, P., Tsianakas, V., & Robert, G. (2015). Experience-based Co-design and Healthcare Improvement: Realising Participatory Design in the Public Sector. *The Design Journal, 18*(2), 227-248. https://doi-org.elib.tcd.ie/10.2752/175630615X14212498964312

Duchan, J. F., & Kovarsky. (2005). *Diagnosis as cultural practice.* Walter de Gruyter GmbH & Co. KG.

Duchan, J. F., & Kovarsky, D. (2005). Introduction. In J. F. Duchan, & D. Kovarsky (Eds.), *Diagnosis as cultural practice* (pp. 1-13). Walter de Gruyter GmbH & Co. KG.

Dyck, E., & Russell, G. (2020). Challenging psychiatric classification: Healthy autistic diversity and the neurodiversity movement. In S.J. Taylor, & A. Brumby (Eds.), *Healthy minds in the twentieth century: In and beyond the asylum* (pp. 167-187). Palgrave Macmillan. https://doi.org/10.1007/978-3-030-27275-3_8

Easton, A., & Atkin, K. (2014). Understanding narratives: A beacon of hope or pandora's box? In S. Weatherhead & D. Todd (Eds.), *Narrative approaches to brain injury* (pp. 1-26). Karnac Books Ltd. http://dx.doi.org/10.4324/9780429477508-1

Eilers, N. (2020). A critical disability studies reading of Beauty and the Beast: Detournement in pedagogical practice. *Journal of Media Education, 12*(2), 54-63. https://doi.org/10.23860/JMLE-2020-12-2-5

Eldershaw, P., Mayan, M., & Winkler, A. (2007). Through a Painted Window: On Narrative, Medicine, and Method Interview with Arthur W. Frank Conducted by the International Institute for Qualitative Methodology EQUIPP Students, November 16, 2005. *The International Journal of Qualitative Methods, 6*(3), 121-139. http://dx.doi.org/10.1177/160940690700600302

Engel, G.L. (1977). The need for a new medical model: A challenge for biomedicine. *Science, 196*(4286), 129-136. https://doi.org/10.1126/science.847460

Epilepsy Ireland. (n.d.). *Working with epilepsy.* https://www.epilepsy.ie/content/working-epilepsy

European Reference Networks. (n.d.). *European reference networks.* https://health.ec.europa.eu/european-reference-networks/overview_en

EUROPLAN. (2010). *Recommendations for the development of national plans on rare diseases guidance document*. http://www.europlanproject.eu/Resources/docs/2008-2011_2.EUROPLANRecommendations.pdf

EURORDIS. (n.d.). *What is a rare disease?* https://www.eurordis.org/information-support/what-is-a-rare-disease/

EURORDIS Rare Diseases Europe. (2011, October). *Position paper: Patients' priorities and needs for rare disease research 2014-2020.* http://download2.eurordis.org/documents/pdf/what_how%20_are_disease_research.pdf

Fallowfield, L., & Jenkins, V. (2004). Communicating sad, bad and difficult news in medicine. *Lancet*, *363*, 312–319. https://doi.org/10.1016/s0140-6736(03)15392-5

Fisher, P., & Goodley, D. (2007). The linear medical model of disability: Mothers of disabled babies resist with counter-narratives. *Sociology of Health & Illness, 29*(1), 66-81. https://doi.org/10.1111/j.1467-9566.2007.00518.x

Fleischman, S. (1999). "I am…, I have…, I suffer from… A linguist reflects on the language of illness and disease." *Journal of Medical Humanities, 20*(1), 1-31. http://hdl.handle.net/10822/922707

Fogelman, D. (Writer), & Ficarra, G., & Requa, J. (Directors). (2016, December 6). Pilot (Season 1, Episode 1) [Television series episode]. In D. Fogelman (Executive Producer), *This is Us.* NBC Studios.

Fogelman, D., Dorsey, J., & Bauman, D. (Writers), & Olin, K. (Director). (2022, May 17). The train (Season 6, Episode 17) [Television series episode]. In D. Fogelman (Executive Producer), *This is Us.* NBC Studios.

Foley, G., Timonen, V., Conlon, C., & O'Dare, C. E. (2021). Interviewing as a vehicle for theoretical sampling in grounded theory. *International Journal of Qualitative Methods, 20,* 1-10. https://doi.org/10.1177/1609406920980957

Foster-Galasso, M. L. (2005). Diagnosis as an aid and a curse in dealing with others. In J. F. Duchan, & D. Kovarsky (Eds.), *Diagnosis as cultural practice* (pp. 17-31). Walter de Gruyter GmbH & Co. KG.

Frank, A.W. (1994). Reclaiming an orphan genre: The first-person narrative of illness. *Literature and Medicine, 13*(1), 1-21. https://doi.org/10.1353/lm.2011.0180

Frank, A.W. (1995). *The Wounded Storyteller.* The University of Chicago Press.

Frank, A.W. (2007). Just Listening: Narrative and deep Illness. In S. Krippner, M. Bova & L. Gray (Eds.), *Healing stories: The use of narrative in counseling and psychotherapy* (pp. 21-40). Puente Publications.

Frank, A.W. (2010). *Letting stories breath: A socio-narratology*. The University of Chicago Press.

Frank, A.W. (2013). *The wounded story teller: Body, illness and ethics.* (2nd ed.). The University of Chicago Press.

Fylan, B., Tomlinson, J., Rayno, D. K., & Silock, J. (2021). Using experience-based co-design with patients, carers, and healthcare professionals to develop theory-based interventions for safer medicines use. *Research in Social and Administrative Pharmacy, 17,* 2127-2135. https://doi.org/10.1016/j.sapharm.2021.06.004

Gale, J. (2010). Discursive analysis: A research approach for studying the moment-to-moment construction of meaning in systemic practice. Human Systems: The Journal of Therapy, Consultation & Training, 21(2), 7-37.

Genetic Alliance UK. (2018). 'Like trying to read a map in the dark': Syndromes without a name, service use and further research. London: Genetic Alliance UK and Birmingham Children's Hospital, Birmingham Women's and Children's NHS Foundation Trust.

Gera, J.V., Martin, G. M., & Zahra, A. J. C. (2020). An insight into the lives of young siblings of disabled children in Malta. *Disability & Society, 36*(1), 58-80. https://doi.org/10.1080/09687599.2020.1712188

Gilbert, P. (2009). *The compassionate mind: A new approach to life's challenges.*

Constable and Robinson.

Gill, V. T., Pomerantz, A., & Denvir, P. (2010). Pre-emptive resistance: patient's participation in diagnostic sense-making activities. *Sociology of Health and Illness, 32*(1), 1-20. https://doi.org/10.1111/j.1467-9566.2009.01208.x

Gilligan, V., Johnson, M., & Maclaren M. (2008-2013). Breaking Bad [Television Series]. Culver City, California: Sony Pictures Television.

Gillman, M., Heyman, B., & Swain, J. (2010). What's in a name? The implications of diagnosis for people with learning difficulties and their family carers. *Disability and Society, 15*(3), 389-409. https://doi.org/10.1080/713661959

Gimenez-Lozano, C., Paramo-Rodriguez, L., Cavero-Carbonell, C., Corpas-Burgos, F., Lopez-Maside, A., Guardiola-Vilarroig, S., & Zurriaga, O. (2022). Rare diseases: Needs and impact for patients and families: A cross-sectional study in the Valencian Region, Spain. *International Journal of Environmental Research and Public Health, 19*(16). https://doi.org/10.3390/ijerph191610366

Glatzer, R. & Westmoreland, W. (Directors), & Lutzuz, L.. Brown, J., & Koffler, P. (Producers). (2014). *Still Alice* [Motion picture]. United States: Sony Pictures Classics.

Gluck, W. (2010). *Easy A* [Film]. Screen Gems.

Goldspink, S., & Engward, H. (2019). Booming clangs and whispering ghosts: Attending to the reflexive echoes in IPA research. *Qualitative Research in Psychology, 16*(2), 291-304. https://doi.org/10.1080/14780887.2018.1543111

Goodley, D. (1998). Supporting people with learning difficulties in self- advocacy groups & models of disability. *Health and Social Care in the Community, 6(*5), 438-446. https://doi.org/10.1046/j.1365-2524.1998.00136.x

Goodley, D. (2013). Dis/entangling critical disability studies. *Disability & Society, 28*(5), 631-644. https://doi.org/10.1080/09687599.2012.717884

Goodley, D. (2016). Disability Studies: An Interdisciplinary Introduction. 2nd ed. Sage.

Goodley, D., & Tregaskis. (2006). Storying disability and impairment: Retrospective accounts of disabled family life. *Qualitative Health Research, 16*(5), 630-646. https://psycnet.apa.org/doi/10.1177/1049732305285840

Goodley, D., Lawthorn, R., Liddiard, K., & Runswick-Cole, K. (2019). Provocations for Critical Disability Studies. Disability and Society, 34(6), 972-997. https://doi.org/10.1080/09687599.2019.1566889

Goodwin, J., Alam, S., & Campbell, L. E. (2017). 'At the end of the day, it is more important that he stays happy': An interpretative phenomenological analysis of people who have a sibling with 22q11.2 deletion syndrome. *Journal of Intellectual Disability Research, 61*(9), 888-898. https://doi.org/10.1111/jir.12397

Gray, D. E. (2001). Accommodation, resistance and transcendence: Three narratives of autism. *Social Science & Medicine, 53*(9), 1247-1257. https://doi.org/10.1016/s0277-9536(00)00424-x

Grob, G. N. (1991). Origins of DSM-1: A study in appearance and reality. *American Journal of Psychiatry, 148*(4), 421-431. https://doi.org/10.1176/ajp.148.4.421

Hamilton, J. G., & Robson, M. E. (2019). Psychosocial effects of multigene panel testing in the context of cancer genomics. *Hastings Center Report, 49 Suppl 1(*Suppl 1), S44-S52. https://doi.org/10.1002/hast.1016

Hawthorne, N. (1850). *The Scarlet Letter, A Romance.* Ticknow, Reed & Fields.

Health (Regulation of the Termination of Pregnancy) Act 2018 (IRE).

Health Service Executive. (2022). *National strategy for accelerating genetic and genomic medicine in Ireland.* https://www.hse.ie/eng/about/who/strategic-programmes-office-overview/national-strategy-for-accelerating-genetic-and-genomic-medicine-in-ireland/national-strategy-for-accelerating-genetic-and-genomic-medicine-in-ireland.pdf

Health Service Executive. (2022, May). *Policy framework for service delivery of children's disability network teams.* https://www.hse.ie/eng/services/list/4/disability/progressing-disability/pds-programme/documents/policy-framework-for-children-s-disability-network-teams.pdf

Health Service Executive. (2023). *Care Pathways for Rare Diseases.* *https://www.hse.ie/eng/services/list/5/rarediseases/care-pathways/*

Heidegger, M. (2008). *Being and Time* (D. F, Krell, Trans.). Harper Collins. (Original work published 1927).

Hellawell, D. (2006). Inside-out: analysis of the insider-outsider concept as a heuristic device to develop reflexivity in students doing qualitative research. *Teaching in Higher Education, 11*(4), 483-494. https://doi.org/10.1080/13562510600874292

Highmore, F., Williamson, S., & French, R. (Executive Producers). (2017-present). The Good Doctor [Television Series]. ABC Studios.

HSCP CPD Sub-Group (2019, October). *Reflective practice statement.* HSE. https://iicms.ie/wp-content/uploads/2020/06/13-HSE-HSCP-Reflective-Practice-statement-Oct-2019.pdf

Huynh, S., McCrimmon, A., & Strong, T. (2020). The change in classification of Asperger Syndrome: An exploration of its effects on self-identity. *The Qualitative Report, 25*(2), 379-398. https://doi.org/10.46743/2160-3715/2020.4122

Husserl, E. (1927). *Phenomenology*. For Enclyopaedia Brtianica (R. Palmer, Trans.) (Revised). Encylopaedia Britannica, Inc.

Husserl, E. (1980). *Phenomenology and the foundations of the sciences* (T.E Klein & W.E Pohl, Trans.). Martinus Hijhoff Publishers. (Original work published 1952).

Information for the undiagnosed. (n.d.). Health Service Executive. https://www.hse.ie/eng/services/list/5/rarediseases/undiagnosed.html

Irish Association of Speech and Language Therapists (2021). *Standards of practice for speech and language therapists on the management of feeding, eating, drinking and swallowing disorders: Working with neonates and babies.* https://www.iaslt.ie/media/vn4bt2m1/iaslt-feds-neonates-and-babies-final-april-2021.pdf

Jaarsma, P., & Welin, S. (2012). Autism as natural human variation: Reflections on the claims of the neurodiversity movement. *Health Care Analysis, 20*(1), 20-30. https://doi.org/10.1007/s10728-011-0169-9

Jacoby, R., Benveniste, S., & Goldzweig, G. (2018). Illness as experienced by parents of children with end-stage renal disease: The "Ill Unit". *Illness, Crisis & Loss, 29*(3), 205-219. https://doi.org/10.1177/1054137318800874

James-Scooter, M., Walker, C., & Jacobs, S. (2019). An interprofessional perspective on job satisfaction in the operating room: A review of the literature. *Journal of Interprofessional care, 33*(6), 782-794.

Jernigan, D., Fernandez, S., Pensyl, R., & Shangping, L. (2009). Digitally augmented reality characters in live theatre performances. *International Journal of Performance Arts and Digital Media, 5*(1), 35-49. https://doi.org/10.1386/padm.5.1.35_1

Johnson, S.K. (1928). Some aspects of dramatic irony in sophoclean tragedy. *The Classical Review, 42*(6), 209-214. http://www.jstor.org/stable/701419

Jones, C. M., & Beach, W.A. (2005). "I just wanna know why": Patient's attempts and physicians' responses to premature solicitation of diagnostic information. In J.F. Duchan and D. Kovarsky (Eds.). *Diagnosis as Cultural Practice* (pp. 103-136). Walter de Gruyter GmbH & Co.

Josselson, R. (2004). The hermeneutics of faith and the hermeneutics of suspicion. *Narrative Inquiry, 14*(1), 1-28. https://psycnet.apa.org/doi/10.1075/ni.14.1.01jos

Jutel, A. (2009). Sociology of diagnosis: A preliminary review. *Sociology of Health and Illness, 31*(2), 278-299. https://doi.org/10.1111/j.1467-9566.2008.01152.x

Jutel, A. (2019a). *Diagnosis: Truths and tales.* University of Toronto Press.

Jutel, A. (2019b). Diagnosis: A critical social reflection in the genomic era. *Ciencia & Saude Coletiva, 24*(10), 3619-3626. https://doi.org/10.1590/1413-812320182410.34502018

Kaniamattam, M., & Oxley, J. (2022). Unpacking the varied roles of mothers of children with developmental disabilities in South India. *Disability & Society, 37*(1), 38-62. https://doi.org/10.1080/09687599.2021.1918540

Kant, I. (1970). *Critique of Judgement* (W. S. Pluhar, Trans. and Introduction & M. J. Gregor, Forward). Hackett Publishing Company, Inc.

Kawa, S., & Gordano, J. (2012). A brief historicity of the diagnostic and statistical manual of mental disorders: Issues and Implications for the future of psychiatric canon and practice. *Philosophy, Ethics, and Humanities in Medicine, 7*(2), 1-9. https://doi.org/10.1186%2F1747-5341-7-2

Kilty, S. (2000). Telling the illness story: The healing power of words. *The Patient's Network, 5(*3), 17-18. http://dx.doi.org/10.1080/1756073X.2016.1149303

Knepper, K. N. K., & Arrington, M. I. (2018). Parent's narratives in an online PHPV forum: Toward a typology of caregiver illness narratives. *Illness, Crisis & Loss, 26*(4), 316-329. https://doi.org/10.1177/1054137316667594

Kocak Tufan, Z., & Kayaaslan, B. (2020). Crushing the curve, the role of national and international institutions and policy makers in Covid-19 pandemic. *Turkish Journal of Medical Sciences, 50*, 495-508. https://doi.org/10.3906%2Fsag-2004-167

Kole, A., & Hedley, V. (2021). Recommendations from the Rare 2030 Foresight Study: The future of rare diseases starts today. EURORDIS-Rare Diseases Europe. http://download2.eurordis.org/rare2030/Rare2030_recommendations.pdf

Kovarsky, D., Snelling, L. K., & Meyer, E. (2005). Emotion and objectivity in medical diagnosis. In J.F. Duchan and D. Kovarsky (Eds.). *Diagnosis as Cultural Practice* (pp. 263-276). Walter de Gruyter GmbH & Co.

Ladd, P. (2002). *Understanding Deaf Culture: In Search of Deafhood*. Multilingual Matters, Ltd. https://doi.org/10.21832/9781853595479

Laverty, S.M. (2003). Hermeneutic phenomenology and phenomenology: A comparison of historical and methodological considerations. *International Journal of Qualitative Methods, 2*(3), 1-29. http://dx.doi.org/10.1177/160940690300200303

Leedham, A., Thompson, A. R., Smith, R., & Freeth, M. (2020). 'I was exhausted trying to figure it out': The experiences of females receiving an autism diagnosis in middle to late adulthood. *Autism, 24*(1), 135-146. https://doi.org/10.1177/1362361319853442

Lewis, C., Skirton, H., & Jones, R. (2010). Living without a diagnosis: The parental experience. *Genetic Testing and Molecular Biomarkers, 14*(6), 807-815. https://doi.org/10.1089/gtmb.2010.0061

Linton, K. F. (2014). Clinical diagnoses exacerbate stigma and improve self-discover according to people with autism. *Social Work in Mental Health, 12*, 330-342. http://dx.doi.org/10.1080/15332985.2013.861383

Loring, M., & Powell, B. (1988). Gender, race and DSM-III: A study of the objectivity of psychiatric diagnostic behaviour. *Journal of Health and Social Behaviour, 29*(1), 1-22. https://doi.org/10.2307/2137177

Madeo, A. C., O'Brien, K. E., Bernhardt, B. A., & Biesecker, B. B. (2012). Factors associated with perceived uncertainty among parents of children with undiagnosed medical conditions. *American Journal of Medical Genetics Part A, 158A*(8), 1877-1884. https://doi.org/10.1002/ajmg.a.35425

Mandleco, B., & Webb, A. E. M. (2017). Sibling perceptions of living with a young person with Down syndrome or autism spectrum disorder: An integrated review. *Journal of Specialists in Pediatric Nursing,* 20(3), 138-156. https://doi.org/10.1111/jspn.12117

McMullan, J., Crow, A. L., Bailie, C., Moore, K., McMullan, L.S., Shamandi, N., McAneney, H., & McKnight, A.J. (2020). Improvements needed to support people living and working with a rare disease in Northern Ireland: current rare disease support perceived as inadequate. *Orphanet Journal of Rare Diseases, 15*(315), 1-14. https://doi.org/10.1186/s13023-020-01559-6

Mead, N., & Bower, P. (2000). Person-centredness: A Conceptual Framework and Review of the Empirical Literature. *Social Science and Medicine, 51*(7)*,* 1087-1110. https://doi.org/10.1016/s0277-9536(00)00098-8

Mitchell, J. L., & Lashewicz, B. (2019). Generative fathering: a framework for enriching understandings of fathers raising children who have disability diagnoses. Journal of family studies, 25(2), 184-198.

Mishler, E. G. (1979). Meaning in context: Is there any other kind? *Harvard Educational Review, 49*(1), 1–19. https://doi.org/10.17763/haer.49.1.b748n4133677245p

Mishler, E. G. (1984). *The discourse of medicine: The dialectics of medical interviews.* Cambridge University Press.

Mishler, E. G. (2005). Patient stories, narratives of resistance and the ethics of humane care: a la recherche du temps perdu. *Health: An Interdisciplinary Journal for the Social Study of Health, Illness and Medicine, 9*(4), 431-451. http://www.jstor.org/stable/26649690.

Moore, V. (2021). *"I'm struggling but I'm not suffering" The lived experience of persons with young onset dementia in Ireland: An Interpretative Phenomenological Analysis* [Doctoral thesis, Trinity College Dublin]. Trinity Access to Research Archive. http://hdl.handle.net/2262/96362

Moss, P., Eirinaki, V., Savage, S., & Howlin, P. (2019). Growing older with autism – The experiences of adult siblings of individuals with autism. *Research in Autism Spectrum Disorders, 63,* 42-51. https://doi.org/10.1016/j.rasd.2018.10.005

Muecke, F. (1983). Foreshadowing and dramatic irony in the story of dido. *The American Journal of Pilology, 104*(2), 134-155. https://doi.org/10.2307/294288

Muir, E. (2016). *The rare reality - an insight into the patient and family experience of rare disease.* Rare Disease UK. https://www.raredisease.org.uk/media/1588/the-rare-reality-an-insight-into-the-patient-and-family-experience-of-rare-disease.pdf

Murray, M. (2008). Analysing narrative accounts. In M. Murray (Ed.), *Narratives, health and illness: A collection* (pp. 99-120). https://www.academia.edu/7886708/Narratives_Health_and_Illness_a_collection_2008_

Neefjes, V. (2022). Metaphors and decision making in parental blogs about their children with life-limiting diseases: who's afraid of the war metaphor? *Medical Humanities, 49*(3), 427-435. https://doi.org/10.1136/medhum-2022-012507

NGO Committee for Rare Diseases. (2018, May 16). *Information on the challenges of living with a rare disease*. https://www.ngocommitteerarediseases.org/what-is-a-rare-disease/

Nicholl, H. (2008). *An exploration of mothers' experiences in caring for children with complex needs* [Doctoral thesis, Trinity College Dublin]. Trinity Access to Research Archive. http://hdl.handle.net/2262/77056

Nutbeam, D. (1998). Health promotion glossary. *Health Promotion International, 13*(4), 349-364. https://doi.org/10.1093/heapro/13.4.349

Parry-Jones, B., & Soulsby, J. (2001). Needs-led assessment: the challenges and the reality. Health and Social Care in the Community, 9(6), 414-428.

Pavlopoulou, G., Bruns, C., Cleghorn, R., Skyrla, T., & Avnon, J. (2022). "I often have to explain to school staff what she needs". School experiences of non-autistic siblings growing up with an autistic brother or sister. *Research in Developmental Disabilities, 129,* 1-14. https://doi.org/10.1016/j.ridd.2022.104323

People (DPP) v Alchimionek [2019] IECA 49.

Picasso, P. (1937). *Guernica.* [oil on canvas]. Museo Reina Sofia, Madrid.

Portway, S. M., & Johnson, B. (2005). Do you know I have Asperger's syndrome? Risks of a non-obvious disability. *Health, Risk & Society, 7*(1), 73-83. https://psycnet.apa.org/doi/10.1080/09500830500042086

Postavaru, G., McDermott, H., Biswas, S., & Munir, F. (2022). Receiving and breaking bad news: A qualitative study of family carers managing a cancer diagnosis and interactions with healthcare services. *Journal of Advanced Nursing, 79*(6)*,* 2211-2223. https://doi.org/10.1111/jan.15554

Rabbitte, K., Prendeville, P., & Kinsella, W. (2017). Parent's experiences of the diagnostic process for girls with autism spectrum disorder in Ireland: An Interpretative Phenomenological Analysis. *Educational & Child Psychology, 34*(2), 54-66. http://dx.doi.org/10.53841/bpsecp.2017.34.2.54

Rare Diseases Ireland. (2021). *Call for Ireland to support adoption of UN Resolution Addressing Challenges of Persons Living with Rare Diseases & their Families*. https://rdi.ie/wp-content/uploads/2021/09/Michael-Martin-UN-Resoultion-4-Rare-Diseases.pdf

Rare Disease Day. (2023). *What is rare disease day?* Retrieved 6 June 2023 from https://www.rarediseaseday.org/what-is-rare-disease-day/.

Reframing Autism. (2023). *Neurodiversity-affirming language: A letter to your child's support network.* Retrieved from https://reframingautism.org.au/neurodiversity-affirming-language-a-letter-to-your-childs-support-network/

Reiser, S. J. (1980). Words as Scalpels: Transmitting Evidence in the Clinical Dialogue. *Annals of Internal Medicine, 92(6),* 837-842. https://doi.org/10.7326/0003-4819-92-6-837

Rimes, S. (Executive Producer). (2005-present). Grey's Anatomy [Television Series]. Shondaland.

Roberts, J. S., Christensen, K. D., & Green. (2011). Using Alzheimer's Disease as a model for genetic risk disclosure: Implications for personal genomics. *Clinical Genetics, 80*(5), 407-414. https://doi.org/10.1111/j.1399-0004.2011.01739.x

Robertson, R. (2015). Out of time: Maternal time and disability. *Studies in the Maternal, 7*(1), 1-13. http://dx.doi.org/10.16995/sim.194

Rosell, A. M., Pena, L. D. M., Schoch, K., Spillmann, R., Sullivan, J., Hooper, S. R., Jiang, Y., Mathey-Andres, N., Goldstein, D. B., & Shasi, V. (2016). Not the end of the odyssey: Parental perceptions of whole exome sequencing (WES) in pediatric undiagnosed disorders. *Journal of Genetic Counselling, 25*(5), 1019-1031. https://doi.org/10.1007/s10897-016-9933-1

Royal College of Speech and Language Therapists. (2022, May 26). *Dysphagia – Clinical information for SLTs | RCSLT*. RCSLT. https://www.rcslt.org/speech-and-language-therapy/clinical-information/dysphagia/

Rowe, L., & Kidd, M. First do no harm: Being a resilient doctor in the 21st century. Maidenhead: McGraw-Hill.

Sanders, C., & DeBlois, D. (Directors), & Spencer, C. (Producer). (2003). Lilo and Stitch [Motion Picture]. Walt Disney Pictures and Walt Disney Feature Animation.

Sanders, L. (2009). *Every patient tells a story.* Broadway Books.

Savin-Baden, M. & Major, C. H. (2013). *Qualitative research: The essential guide to theory and practice.* Routledge.

Scott, W. (1990). PTSD in DSM-III. *A case of the politics of diagnosis and disease. Social problems, 37*(3), 94-310. http://dx.doi.org/10.1525/sp.1990.37.3.03a00020

Simon, J., Hyde, C., Saravanapandian, V., Wilson, R., Distefano, C., Besterman, A., & Jeste, S. (2022). The diagnostic journey of genetically defined neurodevelopmental disorders. *Journal of Neurodevelopmental Disorders, 14*(27). https://doi.org/10.1186/s11689-022-0943

Skotko, B., & Canal Bedia, R. (2005). Postnatal support for mothers of children with Down syndrome. *American Association on Mental Retardation, 43*(3), 196-212. https://doi.org/10.1352/0047-6765(2005)43[196:psfmoc]2.0.co;2

Smith, J.A. (2004). Reflecting on the development of interpretative phenomenological analysis and its contribution to qualitative research in psychology. *Qualitative Research in Psychology, 1*(1)*,* 39-54. https://doi.org/10.1191/1478088704qp004oa

Smith, B., & Sparkes, A. C. (2008). Changing bodies, changing narratives and the consequences of tellability: A case study of becoming disabled through sport*. Sociology of Health and Illness, 30*(2), 217-236. https://doi.org/10.1111/j.1467-9566.2007.01033.x

Smith, J. A., & Osborn, M. (2008). Interpretative phenomenological analysis. In J.A. Smith (Ed.), *Qualitative Psychology: A Practical Guide to Research Methods* (pp. 53-80). SAGE Publications. http://dx.doi.org/10.1002/9780470776278.ch10

Smith, J. A., Flowers, P., & Larkin, M. (2009). *Interpretative phenomenological analysis: Theory, method and research.* SAGE Publications Inc.

Smith, J. A., & Osborn, M. (2015). Interpretative phenomenological analysis as a useful methodology for research on the lived experience of pain. *British Journal of Pain, 9*(1), 41-42. https://doi.org/10.1177%2F2049463714541642

Smith, J. A., Flowers, P., & Larkin, M. (2021). *Interpretative phenomenological analysis: Theory, method and research* (2nd ed.). SAGE Publications Inc.

Smits, R. M., Vissers, E., te Pas, R., Roebeers, N., Feitz, W.F.J., L.M., van Rooij, I.A.L.M., de Blaauw, I., & Verhaak, C.M. (2022). Common needs in uncommon conditions: A qualitative study to explore the needs for care in pediatric patients with rare diseases. *Orphanet Journal of Rare Diseases, 17*(1), 153. https://doi.org/10.1186/s13023-022-02305-w

Somanadhan, S., Nicholson, E., Dorris, E., Brinkley, A., Kennan, A., Treacy, E., Atif, A., Ennis, S., McGrath, V., Mitchell, D., O'Sullivan, G., Power, J., Lawlor, A., Harkin, P., Lynch, S. A., Watt, P., Daly, A., Donnelly, S., & Kroll, T. (2020). Rare Disease Research Partnership (RAinDRoP): A collaborative approach to identify research priorities for rare diseases in Ireland. *HRB Open Research, 3*(13). https://doi.org/10.12688/hrbopenres.13017.2

Somanadhan, S., Bristow, H., Crushell, E., Pastores, G., Nicholson, E., Kroll, T., Larkin, P. J., & Brinkley, A. (2021). IMPACT study: Measuring the impact of caregiving on families and healthcare professionals of children and adults living with mucopolysaccharidoses in Ireland. *Therapeutic Advances in Rare Disease, 2,* 1-16. https://doi.org/10.1177%2F26330040211020764

Sparks, R. (1964). "Diminished responsibility" in theory and practice. *The Modern Law Review, 27*(1)*,* 9-34. http://dx.doi.org/10.1111/j.1468-2230.1964.tb02785.x

Spekman, N. J., & Roth, F. P. (1988). An intervention framework for learning disabled students with communication disorders. *Learning Disability Quarterly, 11*(3)*,* https://doi.org/10.2307/1510769

Spillman, R.C., McConkie-Rosell, A., Pena, L., Jiang, Y., Undiagnosed diseases network, Schoch, K., Walley, N., Sanders, C., Sullivan, J., Hooper, S. R., & Shasi, V. (2017). A window into living with an

undiagnosed disease: Illness narratives from the undiagnosed diseases network. *Orphanet Journal of Rare Diseases, 12*(1), 1-11. https://doi.org/10.1186/s13023-017-0623-3

Soanes, C., & Stevenson, A. (Eds.). (2004). Mise en scene. In *Concise Oxford English Dictionary* (11th ed.). Oxford University Press.

Svendler, N. (2009). Children's embodied voices: Approaching children's experiences through multi-modal interviewing. *Phenomenology & Practice, 3*(1), 80-93.

Suopis, C., & Carbaugh, D. (2005). Speaking about menopause: Possibilities for a cultural discourse analysis. In J. F. Duchan and D. Kovarsky (Eds.). *Diagnosis as Cultural Practice* (pp. 263-276). Walter de Gruyter GmbH & Co.

Surís, A., Holliday, R., & North, C. S. (2016). The evolution of the classification of psychiatric disorders. *Behavioural Sciences, 6*(5), 1-10. https://doi.org/10.3390/bs6010005

Swallow, V., Lambert, H., Santacroce, S., & Macfadyen, A. (2011). Fathers and mothers developing skills in managing children's long-term medical conditions: how do their qualitative accounts compare? *Child: Care, Health and Development, 37*(4), 512-523. doi:10.1111/j.1365-2214.2011.01219.x

Swoboda, D. A. (2008). Negotiating the diagnostic uncertainty of contested illness: Physical Practices and Paradigns. *Health, 12*(4), 453-478. http://www.jstor.org/stable/26649871

Fletcher-Dallas, R. (2021). What does SWAN or being undiagnosed mean? SWAN UK. https://www.undiagnosed.org.uk/support-information/what-does-swan-or-being-undiagnosed-mean/

Tan, N.C. (2020). Comment from the field: "New directions in Critical Disability Studies": Postgraduate Symposium, University of Sheffield. Journal of Literacy & Cultural Disability Studies, 14(3), 1757-6466.

The Lancet Diabetes Endocrinology (2023). Rare diseases: Individually rare, collectively common. *The Lancet Diabetes Endocrinology, 11*(3), 139. https://doi.org/10.1016/S2213-8587(23)00042-6

Thomas, C. (2007). *Sociologies of disability, 'impairment', and chronic illness: Ideas in disability studies and medical sociology.* Palgrave.

Timman, R., Roos, R., Maat-Kievit, A., & Tibben, A. (2004). Adverse effects of predictive testing for Huntington disease underestimated: Long-term effects 7-10 years after the Test. *Health Psychology, 23*(2), 189-197. https://doi.org/10.1037/0278-6133.23.2.189

Timimi, S. (2014). No more psychiatric labels: Why formal psychiatric diagnostic systems should be abolished. *International Journal of Clinical Health and Psychology, 14*, 208-215. https://doi.org/10.1016/j.ijchp.2014.03.004

Timonen, V., Foley, G., & Conlon, C. (2018). Challenges when using grounded theory: A pragmatic introduction to doing GT. *International Journal of Qualitative Methods, 17*, 1-10. https://doi.org/10.1177/1609406918758086

Tomeny, T. S., Ellis, B. M., Rankin, J. A., & Barry, T. D. (2016). Sibling relationship quality and psychosocial outcomes among adult siblings of individuals with autism spectrum disorder and individuals with intellectual disability without autism. *Research in Developmental Disabilities, 62,* 104-114. https://doi.org/10.1016/j.ridd.2017.01.008

Trix, F. (2015). Documenting awareness of the cultural process of diagnosis: Letters of recommendation for medical school faculty. In J. F. Duchan and D. Kovarsky (Eds.). *Diagnosis as Cultural Practice* (pp. 241-262). Walter de Gruyter GmbH & Co.

Vance, N. (2021). Labeling Theory. *Salem Press Encyclopedia*. *Salem Press Encyclopedia*. Grey House Publishing. https://search-ebscohost-com.elib.tcd.ie/login.aspx?direct=true&db=ers&AN=89185563

Van Manen, M. (2017). But is it phenomenology? *Qualitative Health Research, 27*(6), 775-779. https://doi.org/10.1177/1049732317699570

Wakap, S. N., Lambert, D. M., Olry, A., Rodwell, C., Gueydan, C., Lanneau, V., Murphy, D., Le Cam, Y., & Rath, A. (2020). Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. *European Journal of Human Genetics, 28,* 165-173. https://doi.org/10.1038/s41431-019-0508-0

Wakefield, E.O., Belamker, V., Sandoval, A., Puhl, R. M., Edelheit, B., Zempsky, W. T., Rodrigues, H. A., & Litt, M.D. (2023). Does diagnostic certainty matter? Pain-related stigma in adolescents with juvenile idiopathic arthritis. *Journal of Paediatric Psychology, 48*(4), 341-351. https://doi.org/10.1093/jpepsy/jsac092

Walsh, I. P., Delmar, P., & Jagoe, C. (2018). "It's not the Asperger's that causes the anxiety, it's the communication: Person-centred outcomes of hope and recovery in a cultural-clinical borderland". *Topics in Language Disorders, 38*(2), 108-125. http://dx.doi.org/10.1097/TLD.0000000000000149

Walton, T. (2008). *Clinical Psychologists' experience of facilitation trainees' learning through supervision – an IPA study* [Doctoral dissertation, University of Limerick]. Research Repository University of Limerick. https://researchrepository.ul.ie/articles/thesis/Clinical_psychologists_experiences_of_facilitating_trainees_learning_through_supervision_an_IPA_study/19828645

Ward, A. J., Murphy, D., Marron, R., McGrath, V., Bolz-Johnson, M., Cullen, W., Daly, A., Hardiman, O., Lawlor, A., Lynch, S. A., MacLachlan, M., McBrien, J., Ni Bhriain, S., O'Byrne, J. J., O'Connell, S. M., turner, J., & Treacy, E. P. (2022). Designing rare disease care pathways in the Republic of Ireland: A co-operative model. *Orphanet Journal of Rare Diseases, 17(*162), 1-14. https://doi.org/10.1186/s13023-022-02309-6

Waxler, J. L., Cherniske, E. M., Dieter, K., Herd, P., & Pober, B. R. (2013). Hearing from parents: The impact of receiving the diagnosis of Williams Syndrome in their child. *American Journal of Medical Genetics Part A, 161(*3), 534-541. https://doi.org/10.1002/ajmg.a.35789

Weiss, R.S. (1994). *Learning from strangers: The art and method of qualitative interview studies.* The Free Press.

Werkhoven, S., Anderson, J. H., & Robeyns, I. A. M. (2022). Who benefits from diagnostic labels for developmental disorders? *Developmental Medicine & Child Neurology, 64*(8), 944-949. https://doi.org/10.1111/dmcn.15177

WHO Interim Commission. (1948, July 19-22). *Official Records of the World Health Organization No. 2: Summary Report on Proceedings, Minutes and Final Acts of the International Health Conference Held in New York.* World Health Organization.

WHO Director-General's opening remarks at the media briefing on COVID-19 - 11 March 2020. (2020, March 11). World Health Organisation. https://www.who.int/director-general/speeches/detail/who-director-general-s-opening-remarks-at-the-media-briefing-on-covid-19---11-march-2020

World Health Organization. (2022). *ICD-11: International classification of diseases* (11th revision). https://icd.who.int/

Wrigley, O. (1996). *The politics of Deafness.* Gallaudet University Press.

Zoom Video Communications, Inc. (2016). *Security guide.* https://d24cgw3uvb9a9h.cloudfront.net/static/81625/doc/Zoom-Security-White-Paper.pdf

Appendices

Appendix A: Research Ethics Committee Full Approval Letter

Trinity College Dublin
Coláiste na Tríonóide, Baile Átha Cliath
The University of Dublin

Application: Academic Year 2019/20
Application Code: HT32

Applicant/Supervisor Name: Milofsky, Beth RESEARCH PG/Dr Irene Walsh

Title of Research: Exploring the experience of 'diagnosis' for parents of children with diagnosed, and undiagnosed*, neurodevelopmental conditions, within an Irish health system and society.

Date of this letter: 30-04-2020

Dear Beth,

Your submission for ethics approval for the research project above was considered by the Research Ethics Committee (REC), School of Linguistic, Speech and Communication Sciences, Trinity College Dublin on 30.04.20 and has been approved in full.

Please note:

(i) that on completion of research projects, applicants should complete the *End of Project Report Form* (which can be found at: https://www.tcd.ie/slscs/research/ethics/) and submit one signed hard copy to the School Office (Room 4091, Arts Building) as well as an electronic copy (to slscs@tcd.ie);

(ii) the REC requests that you attend, in particular, to your commitments as regards the storage and destruction of data arising from this research, in keeping with REC policy and General Data Protection Regulation (GDPR) guidelines.

We wish you every luck with your research,

Best wishes,

Professor Kathleen McTiernan
Chair, Research Ethics Committee
School of Linguistic, Speech and Communication Sciences

Scoil na nEolaíochtaí Teangeolaíochta, Urlabhra agus Cumarsáide, Coláiste na Tríonóide, Baile Átha Cliath 2, Éire
School of Linguistic, Speech & Communication Sciences, Trinity College, Dublin 2, Ireland
T 353 (0)1 896 1560
slscs@tcd.ie
www.tcd.ie/slscs

Appendix B: Participant Information Leaflet (PIL)

Trinity College Dublin
Coláiste na Tríonóide, Baile Átha Cliath
The University of Dublin

TRINITY COLLEGE DUBLIN
SCHOOL OF LINGUISTIC, SPEECH AND COMMUNICATION SCIENCES

Participant Information Leaflet

Exploring the experience of 'diagnosis' for parents of children with diagnosed, *and* undiagnosed*, neurodevelopmental conditions, within an Irish health system and society.

**Where undiagnosed refers to a collection of symptoms believed to have a genetic origin, where genetic testing was unable to identify a genetic cause.*

Principal Investigator / Researcher	Beth Milofsky, Speech & Language Therapist, B.Sc. in Clinical Speech and Language Studies. Email: milofskb@tcd.ie CORU Registration no. SL016242
Research Supervisor	Dr. Irene P. Walsh, Director of Research, SLSCS, Associate Professor in Speech & Language Pathology, Department of Clinical Speech & Language Studies (CSLS), School of Linguistic, Speech & Communication Sciences, Room 107, 7-9 South Leinster Street, Trinity College Dublin, Ireland Email: ipwalsh@tcd.ie Tel: + 353 1 896 2420/1588 CORU Registered www.coru.ie CORU Registration no. 018391
Funder	Partial funding received from Dublin Maccabi Association (sum of €1000), Tel: +353 87 2240783. Email: info@dublinmaccabi.com. Remaining cost of research project self-funded.
Data Controllers	**Trinity College Dublin (for research data)**
Data Protection Officer	**Data Protection Officer Secretary's Office Trinity College Dublin Dublin 2**

You are being invited to take part in a research study that is being done by Beth Milofsky at Trinity College Dublin.

Before you decide whether or not you wish to take part, please read this information sheet carefully. You should understand the risks and benefits of taking part in this study so that you can make a decision that is right for you. You may wish to discuss it with others. Ask us if there is anything that is not clear or if you would like more information. Take time to decide whether or not you wish to take part. Thank you for reading this.

This leaflet has five main parts:

Part 1 – Information about the Study
Part 2 – Information on how your data will be used and stored
Part 3 – Information about Costs, Funding and Approval
Part 4 – Future Research
Part 5 – Further Information

Part 1 - The Study

Why is this study being done?

I am doing this study to explore how parents, of children who present with diagnosed and undiagnosed complex conditions, living in Ireland, experience 'diagnosis'. This study specifically aims to:

- Explore the experience of parenting a child who has a diagnosed or undiagnosed neurodevelopmental condition.
- Identify what is unique about the experience of parenting children with diagnosed and undiagnosed conditions.
- Investigate what importance 'diagnosis' holds for parents of children with diagnosed and undiagnosed conditions in Ireland.
- Inform health professionals on the parental experience to enhance clinical management of affected families and children.
- Inform the establishment of streamlined services within the Irish healthcare system.
- Contribute patient experiences to guide the development of government policy and legislation.
- Reflect on the culture and perceptions surrounding 'diagnosis' in Ireland.

Why have I been invited to take part?

You have been invited to take part because you are a parent of a child who presents with (i) diagnosis of Down Syndrome OR (ii) a complex condition that is undiagnosed. You have not previously or are not currently working with the researcher in carrying out direct interventions with your child.

Do I have to take part? Can I withdraw?

You don't have to take part in this study. *It is up to you to decide whether or not to take part.* If you decide not to take part it won't affect your current or future contacts with the researcher, access to clinical services or support networks. This research project is entirely unrelated to any other activities or involvements you may share with the researcher.

You can change your mind about taking part in the study and opt out at any time even if the study has started. If you decide to opt out, it won't affect your current or future contacts with the researcher, access to clinical services or support networks. You don't have to give a reason for not taking part or for opting out. If you wish to opt out, please contact Beth Milofsky, Principal Investigator, milofskb@tcd.ie, who will be able to organise this for you.

What happens if I change my mind?

You can change your mind at any time by contacting Beth Milofsky, Principal Investigator, milofskb@tcd.ie. If you choose not to continue to take part, this will not affect your current or future contacts with the researcher, access to clinical services or support networks in any way. If you wish, you can ask for your data to be destroyed. If you request this, we will destroy all data that are still in our possession. We will no longer use or share your data for research from this point onwards. However, it will not be possible to destroy data already used in research studies prior to this time.

What will happen to me if I decide to take part? What will I need to do?

If you agree to participate, you will be invited to attend a once-off individual interview with the researcher. This interview will be approximately 1-1.5 hours in duration. The location will be chosen based on your convenience (i.e. your home, the Department of CSLS or other place of convenience for you). The interviews will be scheduled between May-September 2020.

You will be offered the opportunity to attend a subsequent optional focus group with up to 9 other parents, of children who present with a diagnosed or undiagnosed neurodevelopmental condition. Participation in this group will involve providing an introduction to yourself and your child, and taking part in a group discussion around 'diagnosis'. Discussion will be facilitated by the researcher. The duration of the meeting will be approximately one hour. Location of this meeting to be determined. The focus group will be scheduled after the completion of the individual interviews, estimated to take place between August-October 2020.

Individual interviews and focus groups will be recorded using audio-recording devices.

Are there any benefits to taking part in this research?

Taking part in this study may have the following potential benefits for you:

- Platform to share your experiences
- Opportunity to hear others' experiences
- Potential to contribute to others' (e.g. healthcare professionals, researchers) understanding of the parental experience in order to reform current thinking, professional practice, healthcare services, policy and legislation in Ireland.

Research using your data and information may result in the beneficial outcomes listed below. This research project is being carried out over a minimum three year period so the benefits of the research may not be seen for several years.

- Enhance understanding of the parental experience amongst other parents, professionals, researchers and the community.
- Inform the reform / establishment of streamlined services within the Irish health care systems
- Provide support and information to other families experiencing similar situations
- Add new accounts to the Irish research literature base

Are there any risks to me or others if I take part? What will happen if something goes wrong?

There is a risk that a connection to your identity could be made. Great care will be taken to ensure the confidentiality of all data and the risk to participants of a breach of confidentiality is considered very low. If something did go wrong (i.e. incidents, breaches or suspected breaches1) we would report it to the Data Protection Officer at Trinity College Dublin.

There is potential risk of inconvenience, emotional distress, discomfort, stress, anxiety and fatigue for you. The researcher is a qualified speech and language therapist who has counselling skills to support you during the interview/ focus group should you become uncomfortable or upset. This will help to reduce the risk.

If you feel any negative affects following or associated with participating in this research, please contact your GP, allocated social worker or other identified support network.

1 A personal data breath means 'A breach of security leading to the accidental or unlawful destruction, loss, alteration, unauthorised disclosure of, or access to, personal data transmitted, stored or otherwise processed.' - Article 4 EU General Data Protection Regulation 2016/679.

Part 2 - Data Protection

How will my data be used?

Data from this research project may be published in future in scientific/medical/linguistic/educational journals. You will not be able to be identified in any reports or publications unless you have given your explicit consent for this. The original stored data will only be available to the researcher and research supervisor. The pseudononymised recordings may be available to postgraduate and doctoral students, researchers and professors in national and international hospitals, academic and research institutions engaged in similar work for the purpose of health research. Portions of the pseudononymised recordings may be played in linguistics classes or during conference presentations, or written transcriptions may be made for teaching purposes or for linguistic analysis. If you agree to your data being used in future research and in teaching, your consent form will be held until the data is no longer in use.

What information about me (personal data) will be used as part of this study?
Will my medical records be accessed?

Your name and contact (number or email address) will be required in order for you to contacted by the researcher as part of this research study. You may provide your home address if you express your home as your preferred location for the interview to take place. By agreeing to participate, you are providing sensitive personal data regarding your child's diagnosis. It is your choice what sensitive personal data you share during the individual interview and focus group about your child's medical diagnosis and needs, opinions surrounding diagnosis. Your medical records will not be accessed.

Who will have access my personal data? What will happen to my personal data?

All the personal data that we collect about you during the course of the research will be kept strictlyconfidential and will only be accessible to Beth Milofsky, Principal Investigator, and Dr. Irene P. Walsh, Research Supervisor. All of your personal data will be stored in Ireland. There will be no disclosure of the personal data unless that disclosure is required by law or you have given explicit consent to the disclosure. If you agree to us sharing the information you provide with other researchers (e.g. by making it available in a data archive) then your personal details will not be included unless you explicitly request this. Data that can identify you will be kept for a minimum of seven years from the date of completion of the researcher's relevant degree and publication of the research. Anonymised or coded data will be kept for a minimum of five years to allow us revisit the data at a later time. After this time period your personal data will be archived. If you agree to your data being used in future research and in teaching, your consent form will be held until the data is no longer in use.

The pseudonymised recordings may be available to postgraduate and doctoral students, researchers and professors in national and international hospitals, academic and research institutions engaged in similar work for the purpose of health research. Portions of the pseudonymised recordings may be played in linguistics classes or during conference presentations, or written transcriptions may be made for teaching purposes or for linguistic analysis. If you do not consent to your personal information being stored for possible future research related to the current study, you will still be able to participate.

Will my personal data be kept confidential? How will my data be kept safe?

Your privacy is important to us. We take many steps to make sure that we protect your confidentiality and keep your data safe. Here are some examples of how we do this:

Any information or data which is obtained during this research which identifies you will be treated confidentially. Once audio data has been transferred from the audio recording devices to the researcher's individual password protected computer and to an encrypted external hard drive, the audio files will be deleted from the transportable audio recording device. The data will then be made anonymous so as to hide your identity. This will be done by allocating a pseudonym to all participants. All original files will be transferred to an encrypted hard drive, which will be stored in a locked press in the research supervisor's office, and in an encrypted file in a folder on the research supervisor's computer on the Trinity College Dublin computer network. Any files containing identifiable information will then be deleted off the researcher's the laptop, so that only pseudonymised data remains. Pseudonymised data will be stored on the researcher's individual password protected computer. Hardcopies of pseudonymised data will be stored securely in the researcher's home.

Only the researcher and research supervisor will have access to the original raw data. Only the researcher and research supervisor will have access to the code key that links personal data to participant pseudonyms. This key will be stored in the research supervisor's office, in a locked press, in a separate location to the stored raw data, in the Department of CSLS. The researcher and research supervisor will keep a signed log to record any processing of any research data.

You will be offered the opportunity to review the transcript from your own individual interviews and edit any information, which you believe to be identifying or misinterpreted.

Any publications of this research will include pseudonyms only to protect the confidentiality and anonymity of the participants.

All individual researchers involved in this project have been trained in data protection law and are bound by professional code to maintain confidentiality. A risk assessment and data protection impact assessment has been carried out, indicating a low level risk. If something did go wrong we would report it to the Data Protection Officer at Trinity College Dublin.

What is the lawful basis to use my personal data?

According to data protection legislation[2], we are required to inform you of the legal basis for using your personal data. The tasks we are performing are considered to be in the public interest[3]. Some data that is defined as more sensitive (e.g. information about your child's diagnosis, your experience and opinions of diagnosis) is being used for scientific purposes[4].

What are my rights?

You are entitled to:

- The right to access to your data and receive a copy of it
- The right to have your data transferred to another organisation or 'data controller'
- The right to restrict or object to processing of your data
- The right to object to any further processing of the information we hold about you (except where it is de-identified)
- The right to have inaccurate information about you corrected or deleted
- The right to request deletion of your data

[2] The European General Data Protection Regulation (GDPR)
[3] Article 6(1)(e)
[4] Article 9(2)(j)

By law you can exercise these rights in relation to your personal data, unless the request would make it impossible or very difficult to conduct the research. You can exercise these rights by contacting the researcher, Beth Milofsky at milofskb@tcd.ie or the Trinity College Data Protection Officer, Secretary's Office, Trinity College Dublin, Dublin 2, Ireland. Email: dataprotection@tcd.ie Website: www.tcd.ie/privacy

Part 3 - Costs, Funding and Approval

Has this study been approved by a research ethics committee?

This study is awaiting the outcome of its' application to the Stewarts Care Research Ethics Committee.

Who is organising and funding this study?

Partial funding received from Dublin Maccabi Association (sum of €1000), Tel: +353 87 2240783. Email: info@dublinmaccabi.com. Remaining cost of research project self-funded.

Is there any payment for taking part? Will it cost me anything if I agree to take part?

No, we are not paying participants to take part in the study. It may cost you to travel to take part in the individual interview and / or focus group.

Part 4 - Future Research

Due to the nature of this research it is very likely that other researchers may find the data collected to be useful in answering future research questions about parent's experience and views regarding 'diagnosis'. We will ask for your explicit consent for your data to be used in this way. You do not have to agree to have your data available for future research. Future research will only take place if it has research ethics approval.

Part 5 - Future Information

Who should I contact for information or complaints?

If you have any concerns or questions, you can contact:

- Principal Investigator: Beth Milofsky. Email: milofskb@tcd.ie.
- Data Protection Officer, Trinity College Dublin: Data Protection Officer, Secretary's Office, Trinity College Dublin, Dublin 2, Ireland. Email: dataprotection@tcd.ie Website: www.tcd.ie/privacy
  Under GDPR, if you are not satisfied with how your data is being processed, you have the right to lodge a complaint with the Office of the Data Protection Commission, 21 Fitzwilliam Square South, Dublin 2, Ireland. Website: www.dataprotection.ie

Will I be contacted again?

If you would like to take part in this study, you will be asked to sign a Consent Form at the start of the data collection episode (i.e. individual interview and focus group). At this time, you will be given a copy of this information leaflet and the signed Consent Form to keep. If you wish to participate, please contact the researcher (Email: milofskb@tcd.ie) or research supervisor (Email: ipwalsh@tcd.ie Tel: + 353 1 896 2420/1588).

Appendix C: Individual Interview Template

Trinity College Dublin
Coláiste na Tríonóide, Baile Átha Cliath
The University of Dublin

TRINITY COLLEGE DUBLIN
SCHOOL OF LINGUISTIC, SPEECH AND COMMUNICATION SCIENCES

Individual Interview Template

Exploring the experience of 'diagnosis' for *parents of* children with diagnosed, *and* undiagnosed*, neurodevelopmental conditions, within an Irish health system *and society*.

**Where undiagnosed refers to a collection of symptoms believed to have a genetic origin, where genetic testing was unable to identify a genetic cause.*

Individual Interview: Guiding Probes

Tell me about your child
Tell me about your family
How did your child get his/her diagnosis?
Can you tell me about your experience of receiving/searching for a diagnosis?
How did you feel on receiving/not receiving a diagnosis?
What do you think about the importance of diagnosis? · Is it helpful or restricting?
Any specific challenges with having/not having a diagnosis? Across different contexts, such as · Health professionals/health systems · Education systems · Community and social settings · Legal systems
How important was/is it for you to have a diagnosis?
Can you tell me a bit about what the word 'diagnosis' means to you?
What importance does your child's diagnosis hold for you?
Can you tell about what you think of 'diagnosis' in general?
How does 'diagnosis' fit into your family?
Is there anything you would like people in the community to know?
Is there anything you would like health professionals to know?

Appendix D: Consent Form – Individual Interview

Link to online form:

https://forms.office.com/Pages/ResponsePage.aspx?id=DQSIkWdsW0yxEjajBLZtrQAAAAAAAAAAAAO__dEnKjNUMEhTTUc3STFDVDlYVjkwTElIWTEwUDJNSS4u

Appendix E: PIL Summary Page

Trinity College Dublin
Coláiste na Tríonóide, Baile Átha Cliath
The University of Dublin

Participant Information Leaflet – Summary Page

Trinity College Dublin, School of Linguistic, Speech and Communication Sciences

"Exploring the experience of 'diagnosis' for parents of children with diagnosed, and undiagnosed*, neurodevelopmental conditions, in Ireland."

**Where undiagnosed refers to a collection of symptoms believed to have a genetic origin, where genetic testing was unable to identify a genetic cause.*

You are being invited to take part in a research project being conducted by Beth Milofsky, Speech and Language Therapist, at Trinity College Dublin. Before you decide whether or not you wish to participate, you should read the accompanying detailed participant information leaflet. **Please don't hesitate to contact the researcher or research supervisors if you have any questions, or to express your interest in participating:**

Researcher:
Beth Milofsky
Email: milofskb@tcd.ie

Research supervisors:
Dr. Irene Walsh
Email: ipwalsh@tcd.ie
Tel: + 353 1 896 2420/1588

Dr. Caroline Jagoe
Email: cjagoe@tcd.ie
Tel: +353 1 896 4029

Why is this study being done? This study aims to (i) explore the experience of parenting a child who has a diagnosed or undiagnosed neurodevelopmental condition (ii) identify what is unique about these experiences, and (iii) what importance the concept of 'diagnosis' holds for parents in the Irish context. The study aims to inform health care professionals and others on these parental experiences to inform Irish healthcare systems and influence the development of policy and legislations.

Why have I been invited to take part? You are invited to take part if your child has a (i) diagnosis of Down Syndrome, or (ii) a complex neurodevelopmental condition that has been undiagnosed.

What happens if I decide to take part? You will be invited to attend an individual interview with the researcher. This will last around 60-90 minutes. This interview may take place using an online video conferencing platform (i.e., Zoom). If in-person interviews are permitted within National Public Health guidelines to prevent the spread of Covid-19, the location will be a place that is convenient for you. You will be invited to attend an optional follow-up focus group with other parents. You will be required to read and sign a consent form before taking part in the interview and focus group. There is no payment for your participation.

Do I have to participate? No. It is your choice whether or not you want to participate. You can change your mind and opt out at any time.

Are there benefits to taking part? This study provides an opportunity for you to share your experiences with others and to hear others' experiences. This may enhance understanding of the parental experience and inform services.

Are there risks if I take part? There is a low risk that a connection to your identity could be made. Great care will be taken to ensure the confidentiality of all data. There is a risk of emotional distress, discomfort, stress, anxiety and fatigue. There are factors in place to reduce this risk.

How will my data be used? You will not be able to be identified in any reports or publications. The original data will only be available to the researcher and research supervisors. Pseudonymised recordings (i.e., with no identifying information) may be available to others engaged in similar work.

How will my data be stored? Once pseudonyms (i.e., alternative names to protect identity) have been assigned to participants, the original data will be deleted from the researcher's computer and stored securely in Trinity College Dublin. Pseudonymised data will be stored securely on the researcher's individual password protected computer and in the researcher's home. Data that can identify you will be kept for a minimum of seven years from the date of the completion of the researcher's degree and publication of the research. After this your data will be archived.

Full ethical approval has been granted for this study by the Research Ethics Committee, School of Linguistic, Speech and Communication Sciences (SLSCS), Trinity College Dublin.

Appendix F: Transcription Conventions

Table B

Transcription Conventions – Symbol and corresponding Meaning

Transcription Conventions	
Symbol	*Meaning*
()	Analyst commentary on non-verbal observations
(.)	Brief pause
(..)	Significant pause
[xxx]	Transcription uncertain
[X]	Redacted information to protect anonymity of participants or referenced others
Italics	Said with emphasis
UPPER CASE	Said loudly
.h	Out breath. Number of h's corresponds to length of outbreath.
?	Rising intonation
Wor-	Speech cut off or stopped
:	Lengthened
^	Continuing speech

Appendix G: Sample of Clustering Experiential Statements to Form Personal Experiential Themes for one Participant, namely Olivia

Olivia's Story

Personal Experiential Themes

Cluster of experiential statements

- *"it was wildly fucking unhelpful"* (line 753). Disgust for public health services
    - Who to trust?
    - (Medical procedures – effect on parent / child)
    - Contradicting findings and recommendations from healthcare professionals
    - Disappointment
    - Assessment of need process
    - Dismissal – displacement of parental concerns from medical professionals
    - Complaint
    - Parent role to protect child – parent forced to take control – advocacy / legal actions – fight for services – boundary between parent knowing best to support child's development vs. limitations, parent is not professional
    - Public vs. private health sectors
- *"the lack of sleep and the challenging behaviours are the biggest difficulty"* (lines 453-454). Challenging behaviours
    - Looking for advice – "how do I keep her safe while also encouraging her independence?" (lines 487-488); "I mean I was asking them for help" (line 473)
    - Examples
- *"we would love to have a diagnosis"* (line 632). Benefits of a known diagnosis
    - Peer assimilation and sharing - Support for family, sibling
    - Education
    - Future
- *"sure we're a family of oddballs, wouldn't be that big of a deal"* (lines 112-113). Concept of normal and representation of disability
    - Influence of family circumstance and viewpoints
    - Difference vs. disorder – realm of normalcy. 'something was going on' – in retrospect

Experiential statements

- When something was first noticed – something was not right – there's something
- Genetic testing
- Family history
- Referrals
- Service access
- Health and social care professionals
- Healthcare dismissal – lack of action of medical professionals – unsatisfaction with healthcare services – displacement of parental concerns from medical professionals, failed attempt 'reassurance' – disregard from medical professionals as 'typical' – lack of co-ordinated care – contradictions amongst professionals – conflicting opinions - who to trust – lack of consensus across services - disappointment

- Diagnosis
- GP visits
- Retrospection – hindsight
- Covid-19
- Effect on sibling (probed by researcher)
- Conscious / awareness of listener perspective
- Influence of public – supermarket outing
- Difference vs. disorder – realm of normalcy
- Specificity in memories – timelines – words of healthcare professionals
- Medical procedures – effect on infant / child – conflicting findings
- Systems in healthcare – procedures – referrals – waiting times – access to services
- Parent role to protect child – parent forced to take control – advocacy / legal action – jigsaw – fight for services – boundary between parent being empowered role in supporting child's development vs. limitations, parent is not professional
- Public vs. private sector
- Symptoms
- Behaviours of concern
- Lack of follow-up care
- Community supports
- Influence on whole family
- Sleep disturbance
- Medication
- Early intervention
- Rare disease – SWAN diagnosis
- Lay diagnosis – hypothesis
- Education
- Financial impact
- Intervention
- Acute services
- PDS
- Parental concerns
- Looking for help
- Advice from HSCPs – specificity – seeking advice
- Complaint
- Misdiagnosis – change in diagnosis
- Future planning
- Assessment of need process
- Deceit – unhelpful advice from HSCPs
- Time
- SLT interventions
- Equipment
- Legal action
- Child likes and dislikes – food
- Access to information – information on your child
- Advice to parents
- Advice to healthcare professionals
- Family support systems