ETHICAL AND LEGAL ISSUES IN HEALTHCARE

The right not to know: Issues in Genetic Testing

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Nuala O Faolain's comment in her recent final radio interview with Marian Finucane, "As soon as I heard I was going to die, the goodness went from life", highlights the negative effect that personal health information may potentially have on an individual.

Notwithstanding arguments that individuals who have such forewarning about their prognosis have an opportunity to 'do things they might not otherwise have done' or even, in some instances, to avail of life-prolonging treatment, the comment reminds us that information about one's health status can be dramatically negative in a manner that is additional to but separate from impending physical deterioration. This raises the question of whether there might be a legitimate 'right to not know'.

Respect for autonomy is generally defined as the right to consent to or refuse a healthcare intervention and is considered to be a core principle in healthcare today. Fundamental to the exercise of consent is that the patient be appropriately informed about the healthcare decision to be made. Hence, at face value, a right 'not to know' would be incompatible with respect for autonomy as it would entertain a paternalistic approach by the healthcare practitioners involved. This creates a dilemma when attempting to balance society's reservations regarding paternalism in healthcare with the interest a patient might have in not knowing healthcare-related information about them. Knowledge is an important element in expressing one's autonomy, and easily accessible screening services are central to patient satisfaction in this regard. The general objectives of screening include that screening services generally be provided only for conditions for which there are treatment or preventative measures available and hence do fit with many aspects of the evolving field of genetic screening in primary care.

Genetic screening

Mapping of the human genome, completed in 2001, provides the template to glean intimate knowledge regarding individuals from tiny samples of DNA. Anyone can obtain their entire DNA profile by taking a swab from the inside of the mouth and availing of a 'confidential' internet-based service. The question is whether access to that information would really be in that person's 'best interests'. Once we have the information, there is no return to blissful ignorance. Genetic data is a past, present and future diary and has the potential to influence lifestyle, reproductive, insurance and employment decisions. Genetic information may be personally predictive and also have implications for relatives. Science currently has the capacity to use genetic testing in the prediction or diagnosis of a single gene disorder. The tests are used to confirm or initiate a medical clinical diagnosis, to ascertain whether the person has a genetic predisposition or susceptibility to a disease or to identify the carrier of a disease. The likelihood is also that such advances may lead to the targeting of therapy to the patient in question.

Genetic screening is not uniform in terms of complexity or implications of the results. Some diseases may be widely screened for in primary care whereas others will require specialist or secondary care settings. Approximately 4,500 diseases are genetically based. In some cases, as with Huntington's disease, if a patient carries the gene and lives long enough (usually to the fifth decade) he will develop the disease. Tuberculosis is different in that there is no gene for tuberculosis, but there are genes that make people more or less resistant. More commonly, genetic testing will indicate a predisposition to developing a disease, e.g. in hereditary breast cancer or hereditary colon cancer. A specific genetic alteration can be detected in an affected relative. This specific genetic alteration may then be screened for in healthy relatives to identify whether they also have a predisposition to the disease. It must be emphasised that a positive test result identifies a predisposition, not that the cancer will develop. People react differently to such information, e.g. some women who have the 'predictive' gene for BRCA1/2 decide to have annual breast examination whereas others choose to have healthy breasts removed. Genetic screening results may therefore have a much greater impact on an individual than, for example, blood pressure or cholesterol screening. Such information can cause significant stress, especially where the condition is untreatable or preventative measures involve radical surgery such as double mastectomy. It is becoming increasingly evident that some people would simply prefer to exercise their right 'not to know'.

It is generally agreed that genetic screening should not be undertaken without pre-screening counselling which is considered critical to a patient being 'appropriately informed' in the consent process. The irreversible nature of acquiring such information means that people need to be aware of the consequences of both having a test done, and, in some cases, the separate decision as to whether to access the results of the test. Decisions as to what is an acceptable disorder for which to screen must be made in collaboration with the patient. In the USA pre-screening genetic counselling is recommended best practice. However, in Europe, there are wide variations in practice.

Family Matters

Personal autonomy may be in conflict with the need for family members to know details of an individual's genetic profile. The potential value of family genetic information to other family members, in order to exercise their own right to take preventive action, complicates matters. In terms of precedence, it can be argued that the taking of a family history for patient use and for third parties, e.g. insurance companies, is common practice for which consent from other family members is rarely obtained. Family conflicts, which have the potential to influence the consent process, may arise, e.g. where crusaders within families pressurise relatives into facilitating group predictive tests, where new information may contradict previous advice within the family, where an individual family member does not wish to reveal diagnosis or genetic nature of disease to family or where the are confidentiality issues about non-paternity. The principle of 'therapeutic privilege' facilitates the aversion of harm by disclosure where the potential harm to another substantially outweighs the patient's claim to confidentiality, and all reasonable efforts have been made to avoid such a breach of confidentiality without explicit consent. In a nutshell this is a reminder that there is no absolute right to confidentiality.

A patient may also wish to exercise the right not to know to avoid risk of discrimination, but such objectives are not without challenge either. While the Disability Act 2005 makes the processing of genetic data for insurance or employment an offence under the Data Protection Acts 1998 and 2003, there is no appropriate relevant legislation to prevent 'voluntary' pre-employment genetic testing in Ireland. Once a person knows the result of a genetic test, he/she will thereafter have to include that information when applying for personal insurance cover such as critical illness or permanent health insurance applications. Additional concerns surround the potential for genetic testing to fuel eugenic practices or the stigmatisation of ethnic groups.

Patients are entitled to be autonomous in their healthcare-related decision making but the process by which they would be assured opportunity to achieve 'appropriately informed' status before undergoing genetic screening is unclear. 'The right to remain in ignorance about

one's genetic make-up should not be mistaken for a waiver of informed consent'. (Andorno, 2004). The requirement to provide pre-counselling before genetic screening is in the patient's best interests but internet and mail-order facilities provide access to the test results without the provision of the 'essential' screening service. Family claims to genetic data may lead to a patient's right to refuse sharing results of genetic screening to be over-ruled, but such breaches of confidentiality naturally require to be monitored. When children or mentally incompetent adults are being considered for genetic screening, it raises numerous other issues for review. The key questions that must be asked, however, is how a healthcare professional can know that a patient would prefer not to know without specifically asking him/her and if the practical issue of whether it is possible to assure that an individual remains 'ignorant' – especially when the diagnosis is a family affair. Indeed Laurie suggests that 'rights' is too strong a terminology to use and that a better approach might be 'to talk of the interest that individuals might have in not knowing' (Laurie p.265).

Community Pharmacy and Patient Good

Community pharmacy practice is increasingly involving itself in the provision of screening services which, when provided in a professional manner, increase our ability to care for our patients. Professionally focussed genetic testing would generally require the addition of specific skills and services, including the availability of appropriate pre-screening counselling of a type not typically available in primary care. Pharmacy needs to constantly monitor that any screening services it provides are appropriate to the setting, in the best interests of the patient and supported by appropriate competencies. This is the means by which we avoid prematurely or unprofessionally removing the 'goodness from life' for any of our patients.

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