Is informed choice in genetic testing a different breed of informed decision-making?

A discussion paper

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Abstract

Traditionally genetic counselling has promoted a non-directive approach to patients' decision-making but the feasibility of this has been questioned. Unlike most branches of medicine, which are shifting away from a paternalistic model, genetic counselling is approaching shared decision-making from a different perspective. There are certain features of genetic counselling and genetic testing which may complicate the drive towards shared decision-making and informed choice: 1. Genetic test results can have broader implications than non-genetic test results. 2. Genetic test results may be perceived by the patient differently to non-genetic test results. 3. Carrier status for autosomal recessive conditions may be difficult for patients to conceptualize. 4. Decisions in genetic counselling are often multiple and sequential. 5. Most information in genetic counselling is based on probabilities and uncertainties. Each of these features is discussed in relation to achieving shared decision-making in genetic testing and the implications for genetic counsellors are described. The points raised, however, have broader implications for medicine as several of the features, although central to genetic testing, are not entirely unique. Lessons learnt from genetic testing and genetic counselling in achieving shared decision-making could help develop methods of promoting informed choice in other medical arenas such as cancer screening.

Introduction

The doctor-patient relationship is slowly evolving from a paternalistic one towards a partnership where health professionals share their biomedical viewpoint with the patient's personal values and experiences in order to reach a mutual decision.¹ Decision-making in a medical context can extend across a spectrum from the traditional attitude of 'Doctor knows best' through to the patient acting as a consumer who

is supplied with sufficient information and reaches a decision alone (Fig. 1). Historically, genetic counselling has attempted to adopt a stance towards the consumer-choice end of this spectrum. Genetic counselling aims to be non-directive, simply providing patients with information regarding their risk of a genetic disorder and options for managing that risk, and seeking to support patients adjust to their risk of a specific genetic disease.² Such a non-directive approach can be seen as an effort to distance

Figure 1 The spectrum of decision-making in medical settings (adapted from Coulter⁷).

genetic counselling from its historical associations with the eugenics movement and demonstrate its primary role in offering patients informed choices.

However, several commentators have questioned whether non-directive genetic counselling is either appropriate or feasible. Clarke argues that in the field of prenatal testing the very offer of a test implies a recommendation to accept the test and proceed to termination of pregnancy in the event of a positive result.³ In certain clinical circumstances it may be entirely appropriate to recommend a particular course of action if there is strong evidence in its favour and this may extend to advising patients to inform their relatives of their genetic risk. Studies of patients attending genetic counselling show that they expect to be offered advice and help with making decisions.⁴ Furthermore, genetic counselling consultations often include attempts by the counsellor to influence a patient's decision.⁵ In response to the difficulties of providing genuinely non-directive counselling, Elwyn and co-workers have proposed a shift towards a shared decision-making perspective in genetic counselling.^{6,7}

Thus, as most branches of medicine attempt to move away from the professional choice model of decision-making towards shared decision-making, genetic counselling is approaching this model from the other end of the spectrum outlined in Fig. 1. However, there are several features of genetic counselling and genetic testing which make this goal of attaining shared decision-making more difficult than in other medical consultations:

- Genetic test results can have broader implications than non-genetic test results;
- Genetic test results may be perceived by the patient differently to non-genetic test results;

- Carrier status for autosomal recessive conditions may be difficult for patients to conceptualize;
- Decisions in genetic counselling are often multiple and sequential; and
- Most information in genetic counselling is based on probabilities and uncertainties.

In this paper I shall discuss each of these issues in more detail, providing specific examples from a range of different genetic tests, and end by suggesting how genetic counselling might achieve shared decision-making.

The broader implications of genetic tests

Choosing to have a specific genetic test can have implications that extend far beyond the individual's health. It can affect family dynamics by indirectly informing other relatives of their risk, or by creating guilt in individual family members who test negative, and it can alter a person's ability to obtain employment or insurance regardless of the result. This point is clearly illustrated by the decision to be tested for Huntington's disease.

Huntington's disease is a progressive neurological disorder that is characterized by chorea and dementia with onset of symptoms usually occurring in the fourth and fifth decades. It is inherited as an autosomal dominant condition. First-degree relatives of an affected individual therefore have a 50% risk of inheriting the disorder. Predictive DNA testing for the disorder is possible but there is no preventive treatment available for mutation carriers. Thus from a purely biomedical perspective there is no direct health benefit from genetic testing for Huntington's disease. Probably for this reason fewer patients at risk of Huntington's disease have actually chosen to be tested than

predicted from early surveys of patients' intentions.⁸

Nonetheless, despite the lack of any treatment to prevent the onset of Huntington's disease some people have chosen to be tested. The principle reasons stated for this decision are twofold: the reduction of uncertainty (although a positive test cannot predict the likely age of onset of the disease) and the ability to plan one's future. These benefits may underlie the improvement in psychological well-being observed after predictive testing for Huntington's disease even in those found to be at increased risk of the disease.⁹ However, predictive testing for Huntington's disease has been shown to have a number of negative consequences in other aspects of the person's life. It can impair relationships with other members of the family, particularly for those testing negative who may be rejected by their relatives.¹⁰ The results of an individual's test may have direct implications for other family members. For example, if a person chooses to be tested because her paternal grandfather had Huntington's disease, she is indirectly consenting for her father to be tested. Thus a positive result would mean that her father is also positive even if he had chosen not to be tested. Predictive testing for Huntington's disease may also have an impact on a person's ability to obtain employment or insurance. In one survey 46% of people who were not at risk from Huntington's disease reported having difficulties obtaining life insurance following predictive testing.¹¹

Public perceptions of genetic tests

Advances in our understanding of molecular biology and genetic predisposition to disease risk are creating a society influenced heavily by genetic reductionism.¹² Media reports of single genes that 'explain' complex behaviours and diseases only serve to reinforce in the public mind the primacy of genetic explanations over environmental and social aetiologies. Consequently, public perceptions of and responses to genetic tests may be affected by notions of irreversibility and certainty. In reality, many genetic

tests that will arise from the Human Genome Project will infer only an increased risk of developing a specific disease. Genetic determinism could adversely affect patients' responses to advice to change their behaviour or take prophylactic therapy to reduce their risk.

These issues are demonstrated in a study of public perceptions of neonatal screening for familial hypercholesterolaemia.¹³ Familial hypercholesterolaemia is an autosomal dominant disorder of lipoprotein metabolism. The heterozygous state has a prevalence in the UK of 1 in 500 and is associated with substantially increased mortality from ischaemic heart disease. It can be successfully treated with lipid lowering drugs. Parents of children that were identified through a neonatal genetic screening programme for familial hypercholesterolaemia were interviewed to explore their perceptions of the disease. Parents that viewed the disease as a disorder of raised cholesterol perceived the condition as less threatening and more readily treatable than parents who thought the disease had a genetic origin. The findings and implications of this study require confirmation of course and it could be argued that, if counselled appropriately about genetic predisposition, it might be possible to increase a person's motivation to change his/her behaviour.

Understanding carrier status

Genetic screening programmes for autosomal recessive conditions aim to identify either homozygotes to enable early treatment (e.g. hereditary haemochromatosis) or heterozygote 'carriers' to enable prenatal diagnosis for couples who are both carriers (e.g. haemoglobinopathies and cystic fibrosis). In both these circumstances healthy carriers will be identified and the nature of carrying a single mutation for a recessive condition needs to be explained. Conceptualizing carrier status can be problematic and being identified as a heterozygote can have important psychological consequences. In the early 1990s several pilot studies of cystic fibrosis carrier screening were conducted in the UK and the participants followed up for 3 years. In the short

term being identified as a cystic fibrosis carrier resulted in increased anxiety but this dissipated after 3 months. However, at 3 years carriers perceived their health as significantly poorer than non-carriers and 20% of carriers misunderstood the implications of their result.¹⁴ If informed choice in genetic screening for autosomal recessive conditions is to be achieved, methods to communicate adequately the meaning of a 'positive test' must be explored further.

Decision-making in genetic counselling

The decisions that an individual must make when considering a genetic test are often multiple and sequential. The initial decision to enter a genetic screening or testing procedure can result in the individual feeling that they have boarded a train from which they cannot alight. Prenatal screening for Down's syndrome provides a clear example of this process. A pregnant woman and her partner face a series of decisions in relation to Down's syndrome screening. Depending on their geographical location they may be offered different serum screening tests or possibly an ultrasound assessment for nuchal fold thickness. Although arbitrary cut-off levels of risk are used to define a 'positive test' the couple must decide what is an acceptable level of risk for them and balance this with the risk of miscarriage induced by the amniocentesis. Finally, if the fetus is found to have Down's syndrome the couple must make the ultimate decision of whether to terminate the pregnancy based on uncertain evidence about the likely severity of handicap. Thus, throughout this process decisions are being made at multiple levels that relate to health, emotion, social responsibility and finance.¹⁵

Probabilities and uncertainty in genetic testing

A central element of genetic counselling is the calculation and communication of risks. Historically, this has been the evaluation of recurrence risks of rare genetic syndromes. With the advent of predispositional genetic testing for

common diseases, genetic counselling has expanded to include the calculation and communication of more complicated risks. This is best demonstrated in the area of genetic risk assessment for breast cancer. Using epidemiological models it is possible to calculate a woman's risk of carrying a mutation in the breast cancer predisposing genes BRCA1 and 2 based on her family history of breast cancer.¹⁶ From these risk estimates the genetic counsellor can then apply current estimates of the penetrance of a mutation in BRCA 1 and 2 to calculate the risk of developing breast cancer over specific time periods including the woman's lifetime risk. The risk of carrying a BRCA1 or 2 mutation can also be used to discuss with the patient the role of genetic testing. An affected relative must first be tested to identify the specific BRCA mutation in the family. Unaffected relatives can then be offered a predictive test for that specific mutation. If the test is negative then the patient can be informed that her risk of breast cancer is the same as in the general population (1 in 11). If the woman is found to carry the mutation then the counsellor must discuss further probabilities and uncertainties. These include uncertainties about the risk of developing breast and other associated cancers, and uncertainties regarding effective prophylactic treatments and surveillance methods.¹⁷

Conclusions

I have discussed the trend in genetic counselling to move away from a purely non-directive approach to patient decision-making towards a model of shared decision-making. I have demonstrated that there are several special features of genetic counselling and genetic testing that may complicate effective shared decision-making. Genetic tests often have wide-ranging implications for an individual and his/her family. The rise of genetic determinism means that patients could perceive the information generated by genetic tests as more certain and adopt a fatalistic attitude towards their disease predisposition. Carrier status for

recessive conditions is a difficult concept to communicate and new methods are required to reduce the adverse effects of being identified as a heterozygote on an individual's perception of their health. When patients are faced with the offer of a genetic test they may be entering a lengthy, complicated process of balancing different risks and values. Furthermore, they may be presented with a series of numerical risks about the likelihood of carrying a genetic mutation, of developing disease and the uncertainties associated with the range of current treatment and surveillance options available to them.

Of course, some of these features are not unique to genetic testing and the issues discussed can be extended to include non-genetic tests. All medical tests suffer from the problem of false positives and false negatives and there is inherent uncertainty in most medical practice. Communicating risk information is becoming increasingly important throughout the whole of medicine but it is a more central feature of the genetic consultation. Furthermore, decision-making outside of genetics can be multiple and sequential. Discussing results of Down's syndrome screening, for instance, shares several features with discussing results of prostate or cervical cancer screening.

For shared decision-making in genetic testing to be successful, genetic counsellors will require many skills.¹⁸ They will need to be able to explore a patient's understanding of inheritance and disease risk which may differ significantly from the biomedical model of Mendelian genetics.¹⁹ They will need to incorporate this view into a discussion of the nature of the genetic test, the meaning of a positive and negative result and its implications for the family, insurance and employment. The options for managing the patient's genetic risk of disease, including potential disadvantages as well as benefits of treatment, should also be discussed. Within the consultation it will be necessary to employ various strategies to present numerical risk information, although how best to present this numerical information remains unclear.²⁰

It should be apparent that the goal of shared decision-making in genetic testing is a formidable one. Genetic counsellors are approaching it from a tradition of non-directive counselling and may therefore be less resistant to the concept than health professionals who are used to making decisions for their patients. Informed choice in genetic testing has several distinctive features and these will need to be faced by other health professionals as the number of genetic tests available increases. These features are not entirely unique to genetic testing and the development of shared decision-making in genetic testing has implications for most branches of medicine. The ultimate aim of the genetic counsellor should be to help 'clients reach a decision wisely, rather than reach a wise decision'.¹⁵ Lessons learnt from genetic counselling could help develop methods of promoting informed choice in other medical arenas.

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