

Are Genetic Tests Exceptional?

Policy makers have suggested that information about genetic risk factors, which are associated with low risk and for which preventive strategies exist, should not be considered "exceptional" and should not warrant special safeguards, such as data protection or specialist pre-test counselling. There is scant research on how such risk factors are perceived. To explore this we conducted qualitative interviews with 42 participants who had been tested for a low risk genetic susceptibility to deep vein thrombosis.

Our findings suggest that low risk genetic susceptibilities, for which preventive strategies exist, may not necessarily be perceived as exceptional.

Background

The UK Human Genetics Commission has suggested that the argument that all genetic information is "exceptional" and warrants special safeguards, such as additional data protection or pretest counselling, is an outmoded paradigm.

There is little research on whether patients perceive low risk, genetic susceptibilities, for which preventative strategies exist, as special or exceptional or not. Qualitative research on patient experiences of familial hypercholesterolaemia (FH) and genetic susceptibility to haemochromatosis for which preventive strategies exist, has observed that different groups of patients may have significantly different perceptions of the conditions. The perceptions range from feeling indifference and being in control to fatalism and resentment for not being diagnosed earlier.

In light of these discussions, we explored whether patients, who had undergone genetic testing for a low risk susceptibility to deep vein thrombosis (DVT), perceived the test results to be different from other medical information. We also explored how patients used the genetic information to prevent DVTs and whether they thought specialist counselling should be offered prior to testing.

Thrombophilia is a polygenic condition associated with five gene alterations: Factor V Leiden (FVL), Prothrombin, Protein S Deficiency, Protein C Deficiency and Antithrombin Deficiency. It is common (the most prevalent alteration, FVL is present in 1:25 of Caucasians) and is usually associated with low risk. Preventive strategies include avoidance of oral contraceptives and hormone replacement therapy, prophylactic anti-coagulants in high risk situations, caution during long flights and avoidance of overweight and smoking. Thrombophilia receives much less public attention than breast cancer genetics, but it is one of the most common genetic tests in the US and the UK. Testing for thrombophilia can be managed by primary and secondary care clinicians, without necessary involvement of clinical genetics. Thus, thrombophilia offers a good case for exploring how feasible the abstract bioethical and policy propositions about genetic exceptionalism are from a patient point of view.

Exceptional and non exceptional genetic tests

The US Secretary's Advisory Committee on Genetic Testing (SACGT) of the National Institutes of Health and the UK Human Genetics Commission (HGC) have attempted to distinguish between low and high impact genetic tests that require and do not require special legal protection and special consenting procedures. It has been argued that:

- Predictive tests are more exceptional than diagnostic tests.
- Tests that identify conditions with high penetrance (likelihood that the DNA-sequence will lead to an

- illness) are more exceptional than those with low penetrance.
- Tests that identify conditions for which no cure or treatment exists are more exceptional than those that detect conditions that can be treated or prevented.

For example, the genetic test for Huntington's disease, which identifies individuals who will nearly certainly develop an incurable, fatal neurodegenerative disease in mid-life, is considered exceptional.

Genetic exceptionalism is a bioethical concept that argues that genetic test results are different from, and potentially more harmful than, other types of medical information. It has been suggested that genetic information is special, because it is immutable throughout the life course, sometimes determines or predicts a future illness, can lead to social discrimination or psychological anxiety, can be used for other purposes (paternity testing, forensic research) and has implications for others (family members). Because of these features special safeguards, such as anti-discrimination laws and special consenting and counselling protocols, have been established or recommended to protect patients from harm.

Genetic tests for susceptibility to common diseases may identify mostly low and preventable risk. Yet, it is uncertain how patients perceive these risks, how they act upon them in terms of prevention and whether they feel they would benefit from special support. Regardless of high expectations and research activity in this area, the UK National Health Services (NHS) currently offers only two tests that fall within this category: the genetic tests for susceptibility for DVT and for hemochromatosis.

Findings

When we interviewed individuals, who had undergone testing for genetic thrombophilia, it emerged that they did not consider the test special or "unusual", even if it sought to detect for a genetic risk factor. Interviewees stated they thought it was much less serious than, for example, genetic testing for a predisposition to breast cancer or a non-genetic diagnostic test for diabetes.

Participants did not think of thrombophilia testing as exceptional for several reasons:

- Participants perceived DVTs to be **preventable**. They often emphasized that they could "do things" to avoid DVTs.
- Participants viewed DVTs to be **caused by environmental triggers as well as genetic factors**. Many focused preventive action on environmental triggers, such as long flights, the Pill or pregnancy, which they thought had caused their own or a close relative's DVT.
- Deep vein thrombosis was perceived as a **less frightening illness** than cancer.
- Many participants **did not have a strong family history** of DVTs in terms of many relatives with serious clotting episodes.

Wide Range of Understandings

We also discovered that participants' perceptions of the test lay on a wide spectrum of understanding. Some participants understood the test and its implications well; most of these participants were middle-class and well-educated.

Some participants had a fair understanding of the test. For example, they might know they were positive but did not know which marker they had inherited in a situation, where different markers are associated with different risks. These participants were of mixed social class, and they had typically been referred to the test because of family history of DVTs or thrombophilia.

At the least informed end of the spectrum seven participants were unaware of having had a genetic test. These participants were all women, older than other participants and came from the lower social classes; many of them had experienced several DVTs or cerebrovascular events and had poor health.

The research suggests that the test was often introduced into an informal manner, which contributed to poor understanding.

Behavioural Consequences

All female participants, who were aware of having genetic thrombophilia had stopped oral contraceptives and hormone replacement

therapy. Many participants stated that they had decided to take the test in order to be able to advise their daughters to avoid the Pill if they were also found to be positive.

The participants had also taken precautions during long flights. They were aware that lifestyle, such as smoking, being overweight, and not exercising, increased the chance of DVT. However, the majority of participants reported that they had not significantly changed their lifestyles as a consequence of thrombophilia testing.

The less informed participants did not significantly differ from the well-informed ones in terms of the preventive actions they had undertaken. However, less informed participants were uncertain if they were doing the right thing or not.

The participants generally perceived DVTs to be preventable. The preventive strategies the participants chose suggest that they used genetic information to effectuate limited changes in their lives, such as decide about medications or take precautions during flights. The fact that participants did not change their lifestyle is discouraging against the hope that genetic susceptibilities would motivate smoking cessation or healthier diet and exercise.

Genetic Counselling Needed?

Some participants thought genetic counselling was not needed prior to thrombophilia testing, and some participants even thought referring patients to a specialist counselling would unnecessarily frighten them. In light of these answers the suggestion is that genetic testing for susceptibilities to common disease could well be declared not exceptional and managed in mainstream medicine.

However, the responses of the less informed participants, who felt that specialist counselling could have helped them to understand what they were being tested for and what the implications were, cast a doubt on the argument against exceptionalism. The responses showed that the way mainstream medicine currently manages genetic testing produces a subgroup of patients with poor understanding, who can be perplexed, indifferent, worried or entirely unaware of having had a genetic test.

Our findings indicate an emerging way of perceiving genetic tests not as revealing "in depth" truths about health and identity but as "surface" information, which is used to make specific health care decisions. Against this background it could be feasible to offer genetic testing in mainstream medicine. However, the views of the subgroup of patients, who were left with poor understanding and occasionally confused, suggest that mainstream medicine is not currently prepared to manage genetic testing.

It could also be that the particular characteristics of thrombophilia testing account for people's not unduly concerned attitude. It could be that people would perceive a genetic susceptibility test for cancer differently. This underlines the need to evaluate the personal implications of and service provision for any genetic susceptibility tests that may become available on a case by case basis. It is also important to pay attention to the experiences and needs of subgroups, such as those with less education or at high risk.

Notes:

Egenis is the ESRC Centre for Genomics in Society charged with researching the impact of genetic technologies in society. Egenis is part of the University of Exeter, and the ESRC Genomics Network.

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