

Case study: Testing for genetic disorders

Purpose: A genetic disorder is a disorder that is caused by an abnormality (or several abnormalities) in a person's genome. Genetic disorders are often hereditary, which means they are passed down to a child from its parents. The most common genetic disorder is familial hypercholesterolaemia (a genetic predisposition to high cholesterol levels), which is thought to affect 1 in 250 people in the UK. Other disorders, such as cystic fibrosis, are rarer although they can be relatively common in particular ethnic groups; for instance, the incidence of cystic fibrosis in Scotland is almost double the global average. Collectively genetic disorders affect lots of people because there are more than 10,000 different types worldwide. Genetic testing is used to inform people whether they have a disorder, or if there is a chance they might pass a disorder on to their children.



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Approaches to identifying genetic disorders

In the UK, if a person is concerned they might have, or might develop, a genetic disorder based on their family history, they can:

- Go to their GP, who might refer them for genetic testing. Patients usually provide a DNA sample, which is then analysed for a specific abnormality. Usually a single gene will be tested, but whole-genome sequencing approaches are being developed. The 100,000 Genomes Project, for example, aims to study whole-genome sequences from patients with cancer and rare disease
- Use a commercial genetic testing service. In this case, customers collect samples of their own DNA and send them away for analysis and interpretation
- Take no action. Some people decide it is better not to know if they have or might develop a genetic disorder.

UK facts & figures

- More than half a million people in the UK have some kind of genetic disorder
- Home genetic testing kits went on sale in the UK in 2014, with some now costing less than £100
- As of 7 August 2017, the 100,000 Genomes Project had sequenced 33,000 genomes
- The current cost of sequencing a whole genome is around £750. This does not include the cost of interpreting the results and employing a genetic counsellor to help people understand the results.

Arguments in favour of testing for genetic disorders

- Identifying a genetic disorder, or predisposition to a genetic disorder, could lead to appropriate treatment, preventative treatment, or behaviours that reduce the risk of disease
- Effective preventative treatment could lead to reduced costs to health services
- Even where no treatment or risk reduction is available, accessing information about their future health can help people make plans for their lives and influence people's reproductive choices such as whether and when to have children, or whether they should use pre-implantation genetic diagnosis
- Having more data about people's genomes improves research into the links between genes and disease.

Arguments against testing for genetic disorders

- Genetic data about a patient could be shared with or requested by third parties, such as pension or insurance providers, affecting the service people receive from those companies
- Genetic data could be stolen in the event of a cyber security failure
- Customers with a poor understanding of genetics may struggle to make an informed decision about the consequences of using commercial genetic testing services
- The results may be difficult to interpret, which might cause people to worry unnecessarily about potential health concerns
- Vendors are likely to direct concerned consumers to public health services, which increases the strain on public services, especially in the UK where there are insufficient genetic counsellors to meet demand
- People who have undergone genetic testing at birth might not have chosen to have their own genome sequenced.