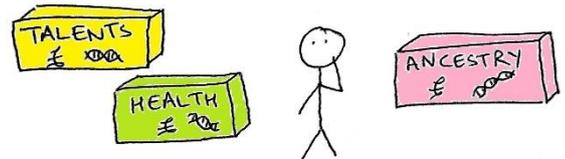


Considering buying a genetic test? – Information for the public, patients and their families

There are lots of companies that sell genetic tests directly to the public. Sometimes people ask the BSGM for its opinion on these tests, and we note that some companies signpost their customers to the BSGM for clinical advice. ***It is important that people think carefully before buying genetic tests, especially tests that claim to provide information about health. If you have questions about the results you might find it difficult to access NHS support.***



It is especially important to be very cautious about seeking further interpretation of ‘raw data’ from direct-to-consumer genetic tests. The BSGM is aware of multiple cases where people have found results in direct-to-consumer genetic test raw data that look medically concerning but that have turned out to be completely wrong.

Here are some key questions that the BSGM recommends you consider before buying a genetic test:

Why do you want the test?

- I think I might have a genetic condition.

Genetic tests have different levels of thoroughness and it's important that you choose the right one to answer your question.

If based on your personal or family history, you think you or your family might be affected by a particular genetic condition, talk to your GP about whether referral to an NHS clinical genetics service might be useful.

Home genetic tests often only take a superficial look at particular genes – they aren't designed for diagnosing genetic conditions.

For example, a popularly available direct-to-consumer *BRCA* test checks for three key variations (differences in the genetic code) in the *BRCA1* and *BRCA2* genes that predispose to breast and ovarian cancer, but there are thousands of other cancer-predisposing variations in these genes that the test wouldn't look for. The test would miss around 80% of people with a cancer-predisposing variation in *BRCA1* or *BRCA2* – they'd need a different, more detailed test instead.

- I want to know about my risk of developing disease in the future.

Genetic tests are getting better at finding answers for people affected by disease. They're not so good at predicting future disease in people who are currently healthy. This is partly because there's so much we still have to learn about how genetic factors work in different people.

To date, most research into the effects of particular genetic variations has been done with people and families affected by health problems, looking into why those health problems happened for them. The effects might be very different if the genetic variation is found in a person with no personal or family history

of the relevant health problem – there might be protective genetic or environmental factors that mean the variation is less likely to cause disease in that person. In many cases, we don't know what the risk would be yet, just that it might be lots lower, and might therefore mean that drastic interventions such as surgery are not indicated.

It's also important to remember that for common diseases, genetics is very rarely the whole story. Lifestyle factors matter a lot, and unlike genetics, you can often do something about them.

Home genetic tests can't give you certainty about your future disease risks – a negative result doesn't mean you won't develop a disease, and a positive result doesn't mean you definitely will.

Lack of evidence around genetic tests screening for future disease, and frequent inaccuracies in results from home genetic testing, mean that your NHS clinical care is unlikely to change based on a home genetic test result. If you want extra tests or screening based on a home genetic test result, you might have to pay for them privately.

What would happen if you got a result you were worried about?

If you were worried or concerned by the result of a direct-to-consumer genetic test, does the testing company have people you could talk to about this? Are they sufficiently qualified to talk about your concerns (for example, registered genetic counsellors), and would you have to pay to talk to them?

It might naturally be worrying or concerning to be told that you have a higher genetic risk of developing a particular health condition. Generally, for common health conditions, having a high genetic risk doesn't mean you'll get the condition – and for some direct-to-consumer genetic 'high risk' results it will sometimes be much more likely that you'll never get the condition than that you will.

You might find it hard to access support on the NHS – GPs are advised to be cautious about commenting on the results of direct-to-consumer genetic tests because these are often low quality, and often there's no clear evidence to support making any changes to standard clinical care.

If you wanted to discuss your home genetic result in depth with a health professional, you might have to do this privately. This might involve paying for extra genetic tests (for example, confirming your result in an accredited laboratory to see if it's actually right) so do think about this beforehand as it could be expensive.

If you're thinking about having a test, look carefully at all the health risks that the company claims to test for. Ask yourself, 'if my result came back as high risk, how would I feel about it? How would I get the support I needed? How much might it cost me?'

As an example, some companies sell tests that look at a genetic risk factor for developing Alzheimer's disease (APOE4). People might be attracted to testing because they would find it reassuring to hear that they don't have the genetic risk factor. However, some people will be told (perhaps unexpectedly) that they do have the genetic risk factor. Sometimes people regret having had the test, and worry every time they forget something that it's because they're developing dementia. There are many people with the genetic risk factor who never develop dementia (and others who do not have the genetic risk factor who do develop it). NHS care and lifestyle recommendations wouldn't change based on the results of the test, so it's important to think through whether having the test would help you or not.

It's also important to remember that some direct-to-consumer genetic tests might give you non-health-related information that might be difficult to come to terms with, for example showing that family genetic relationships aren't what you expected.

What implications might the test have?

- **False reassurance about your health** – direct-to-consumer genetic tests are often very limited, and it's important that you don't take a 'nothing found' result as meaning that you can't get a condition, or can't

have a high genetic chance of developing a condition. ***If you have symptoms or a family history that you are worried about, you need to get this checked out in the same way that you would if you had never had the test.***

- **False alarm about your health** – sometimes, test results are completely wrong (especially if found by looking at ‘raw data’ from direct-to-consumer tests). Other test results might be easy to over-interpret – for example being told you have a ‘high risk’ of a disease can understandably be worrying, but sometimes when you look at the statistics behind it a ‘high risk’ isn’t actually much higher than the average risk.
- **Finding out a genetic risk** – your test might show that you have a higher genetic risk of a particular condition (though sometimes these results will be inaccurate – see above). The way this affects you will depend on various factors, including what the condition is, and the way you tend to deal with this sort of information. Some people might worry a lot and find it hard to stop thinking about whether they might be starting to experience symptoms of the condition. Other people might feel it is useful to find out, especially if there are steps they can take to detect the condition early, or to reduce the chance of it developing.
- **Family links** – we share our genetic code with our biological relatives. This means that if you find a genetic health risk by buying a genetic test, this might have implications for your relatives. If you’re thinking about buying a genetic test, we encourage you to talk to your family about it. It’s also worth noting that asking your family about their medical history will often give you a clearer (and cheaper) idea of your own genetic health risks than buying a genetic test. Some people having genetic tests find that the genetic relationships in their family aren’t what they expected.
- **Insurance** – as a general rule, you don’t need to disclose the results of predictive genetic tests when applying for insurance (i.e. you only need to tell insurance companies about health problems you or your family actually have or have had, not genetic tests looking at your risk of developing health problems in the future (except in very specific circumstances)). However, though insurers shouldn’t ask about predictive genetic tests, they might ask about any medical appointments or investigations you’ve had or that you’re waiting for. So for example, if you buy a genetic test that says you have a higher risk of developing a heart condition, and you have a heart scan to check for the condition, you might have to tell the insurance company that you’re waiting for or have had this check, and they might decide to make your premiums more expensive, or not to offer you insurance.
- **Putting your genetic data online** – in buying a genetic test, you provide the company with a lot of information about you (and potentially about your close family too). For example, the company might have your genetic data, any information you have told them about your health, and your name, address and contact details from your billing information. Companies should explain to you how they use, store and protect your data – for example, whether it might be used in research, and whether they might sell it to other companies or researchers. It’s not always clear who might be able to access your data. For example, law enforcement officials have used databases from home genetic tests to identify new suspects in unsolved crimes.

Further information

<https://www.genioz.net.au/page/community-resources/>

This webpage includes resources designed to help you think through decisions about buying genetic tests. They were developed by the Genioz study in Australia, and most of the points they raise are also very relevant and helpful for people in the UK.

<https://www.bmj.com/content/367/bmj.l5688>

This link is for a medical article and podcast that discuss direct-to-consumer genetic tests. These are mainly aimed at health professionals but cover important pitfalls with these tests, and key questions to consider in advance of buying a test.

Leaflet written December 2019