



Genetic testing

Science background for teachers

What's the issue?

Do your genes hold secrets that could change the way you live your life? Scientists are developing a range of tests which could tell you your risk of developing genetic conditions from Huntington's disease to cancer. But under what circumstances would you want to know?

Key terms

DNA stands for deoxyribonucleic acid. It is the molecule that carries the genetic instructions that make you you, and me me. It is our DNA which makes us very similar to and yet different from each other.

Genes are sections of DNA. They carry information which determines, among other things, your health, the way you look and how your body works. Genes carry instructions from one generation to the next.

Genetic conditions are caused by gene mutation. **Recessive conditions** only develop when both parents pass on a gene mutation to their child, whereas **dominant conditions** develop if only one parent passes on the mutated gene.

Scientists launched the **Human Genome Project** in 1990 with the aim of reading the chemical sequence of human DNA (the **genome**). They published the complete genome in 2003, and analysis is gradually identifying genes associated with different conditions and diseases.

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What's all the fuss about?

Tests for rare genetic conditions such as cystic fibrosis or Huntington's disease have been available for many years. You might want to know the chance of passing on these genes to your children.

Now scientists are getting a better understanding of how small variations in people's genes affect their health. One day, genetic tests may allow us to predict the chance of someone developing almost any disease.

But would you want to find out about all the potential diseases you could get? Could you cope with whatever the result is? Would knowing the statistical *likelihood* of developing a condition really be useful? And should we be concerned about who could get access to our results?

Key statistics

- One in three people will develop some form of cancer during their lifetime.
- One in 2500 people of north European origin are affected by cystic fibrosis.
- Scientists have found 10,000 diseases that are caused by a problem with a single gene.

How do genetic tests work?

A small blood sample or a few cheek cells are collected with a brush or cotton swab. The sample is then sent to a lab which tests for specific genetic conditions. The results are finally reported back to the person's doctor, who is able to explain the results and provide medical and psychological support if necessary.

What's the difference between single-gene conditions and multifactorial conditions?

Single-gene conditions are caused by mutations in just one gene. They usually have a clear pattern of genetic inheritance, which makes them easy to diagnose with a genetic test.

In contrast, a genetic test can only reveal a statistical likelihood of developing a multifactorial condition. Examples of multifactorial conditions include diabetes, asthma and high blood pressure, which are all caused by a combination of different genetic variations and environmental and lifestyle factors. They often run in families and are much more common than single-gene conditions, but don't have a clear pattern of genetic inheritance. This makes precise diagnosis and predictions of likelihood difficult.

What do genes have to do with...

...cystic fibrosis?

Cystic fibrosis is a recessive single-gene condition. It mainly affects a person's lungs and digestive system, by clogging them with mucus. This makes it difficult to breathe and digest food. Cystic fibrosis can usually be treated with physiotherapy, exercise and medication, so life expectancy depends largely on access to healthcare.

...sickle-cell anaemia?

Sickle-cell anaemia is a recessive single-gene condition. It affects the red blood cells, causing pain in the chest and joints. Although the symptoms can be treated there is currently no cure. However, carrying the sickle-cell gene (i.e. inheriting the recessive gene from just one parent) provides some protection from the disease malaria.

...Huntington's disease?

Huntington's disease is a dominant single-gene condition that affects a person's ability to think, talk and move. There is currently no cure. A test can tell people whether or not they have inherited the altered gene, but cannot predict when they will start to develop symptoms, which usually happens between the ages of 30 and 50.

...diabetes?

There are two main types of diabetes. Type 1 diabetes develops when the insulin-producing cells in the body have been destroyed and the body is unable to produce any insulin. Scientists are currently uncertain about exactly what causes this to happen. Type 2 diabetes develops when the body can still make some insulin, but not enough, or when the insulin that is produced doesn't work properly (known as insulin resistance). The risk of developing type 2 diabetes may be increased by certain genes as well as lifestyle factors such as body fat and smoking.

...breast cancer?

Scientists have identified that age and lifestyle are not the only factors that can increase the risk of developing breast cancer. People with certain breast cancer genes have a 55 to 85% chance of getting this disease in their lifetime. No genetic test is 100% accurate, but people with a strong family history of breast cancer are often advised to take a test to determine whether they have one of these 'high risk' genes. If the test is positive a number of options are available, including regular screening or surgery.

Who might choose to have genetic tests?

Anyone can choose to take a genetic test, but some groups are more likely to make this choice, for example:

People with a family history of a multifactorial condition

If the test reveals they have inherited a condition such as diabetes that can be 'risk reduced', their doctor can recommend appropriate lifestyle changes.

People with a family history of a single-gene condition

If the test reveals that a person will develop an as yet undiagnosed genetic condition in the future, counselling, support and planning can begin before symptoms start to appear.

Couples with a family history of a single-gene condition who are considering having children

The test can identify the risk of a couple passing the condition onto their children if both of them carry a recessive genetic condition or if one partner carries a dominant condition. If the result is positive they could choose not to have a baby, screen embryos or go ahead with the pregnancy anyway.

You could discuss...

- What are the potential impacts of having a test?
- Who else might be affected by your decision to have a genetic test? Your siblings? Your own children or parents?
- Who should have access to your test results? Only you? Your doctor? Your school? Your employer?
- Should everyone be tested for genetic conditions? Do you think you could cope with the results?
- Should parents and carers be allowed to have their children tested?

Personal viewpoints

Dr Jess Buxton, genetics researcher at Imperial College London (September 2010):

'Some of this research may lead to new genetic tests, but mostly it will lead to a deeper understanding of how our bodies work, and what happens when we get ill. This will help scientists develop new ways of preventing and treating serious illnesses such as diabetes, schizophrenia and cancer.'

Matt (age 22): 'I chose to get tested to see if I have Huntington's disease so I can make informed decisions about my life. For instance, having children I can make sure that they're not at risk using procedures such as screening the embryos for Huntington's disease.'

Chrissy (age 15): 'At my age, I don't think you would be able to cope, knowing that you have a disease that will be there all your life. Knowing could stop me from doing the same things as my friends.'

Baz (age 12): 'I chose not to get tested despite knowing my mum and sister have the gene for ataxia, partly out of fear of what it might mean for me and the immediate consequences, that is, how I would react to knowing. I'm not sure the way I live my life or my outlook would change knowing that I was going to be affected by this in the future.'

Links to the Science Museum

The *Who am I?* gallery investigates the science of who you are, focusing on genetics and brain science:

sciencemuseum.org.uk/educators/whats_on_for_your_group/who_am_i

The *Making the Modern World* gallery presents a reconstruction of the double-helix model of DNA built by Francis Crick and James Watson in 1953 using some of the original metal plates:

sciencemuseum.org.uk/educators/whats_on_for_your_group/galleries/making_the_modern_world

The *Who am I?* website provides images, animations and more information about genes and current genetic research:

sciencemuseum.org.uk/who_am_i

The Science Museum's resources for educators include activities to help you explore genetics in the classroom:

sciencemuseum.org.uk/educators/genetics

Further information

The Wellcome Trust's Sanger Institute is Europe's largest genome centre providing public access to the views and concerns of leading researchers in the field and information about the ongoing analysis of the Human Genome Project:

yourgenome.org

The Wellcome Trust's *Big Picture* (January 2010) explores the topic of your genetic identity:

wellcome.ac.uk/stellent/groups/corporatesite/@msh_publishing_group/documents/web_document/WTDV027300.pdf

The UK Genetic Testing Network is a collaborative group of genetic testing laboratories, clinicians and commissioners of NHS genetic services. The network's website provides information about genetic tests:

ukgtn.nhs.uk/gtn/Home

Cancer Research UK is the world's leading charity dedicated to beating cancer through research. It provides various kinds of information about cancer addressed at patients:

cancerhelp.org.uk/type/breast-cancer/about/risks

The article 'Spit parties: Genetic testing becomes a social activity', published in the *Guardian* (18 September 2008), presents the limitations of using a genetic test from a commercial company:

guardian.co.uk/science/blog/2008/sep/18/genetic.testing

The article "'DIY genetics tests' guidelines published' from BBC News (4 August 2010) presents some discussion about initial guidelines for companies that offer genetic tests directly to customers:

bbc.co.uk/news/health-10844495

For this activity and many more, visit sciencemuseum.org.uk/educators

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