



yourgenome.org

Sam will be 50 years old next month. He really likes birthdays, and this year (2027) will be a big one in a lot of ways. Sam's father and brother both had major heart attacks in their early fifties and his father died. His brother, Ben, is only four years older than him, so Sam has decided to get some insurance in case of the worst.

The trouble is the insurance company want to see Sam's genotype before they will offer him any cover. Sam's wife, Esther, is against people being genotyped. Sam has never really thought about being genotyped in the past, he's not against it; it just never came up before. Of course, if he takes the test and something bad turns up, the insurance might be expensive or even refused. But that would be the least of his worries. Sam doesn't know which is worse – knowing or not knowing?

Should Sam take the test?

I was so scared when I had my heart attack; I kept thinking of my dad and wondering if I would live as long as him. Sam should have the test – it will put his mind at rest. Or if it turns out to be bad news, we will know what we're dealing with and may be able to prevent him going through the same as me. Forget the insurance; this test could save his life!

Ben
(Sam's brother)

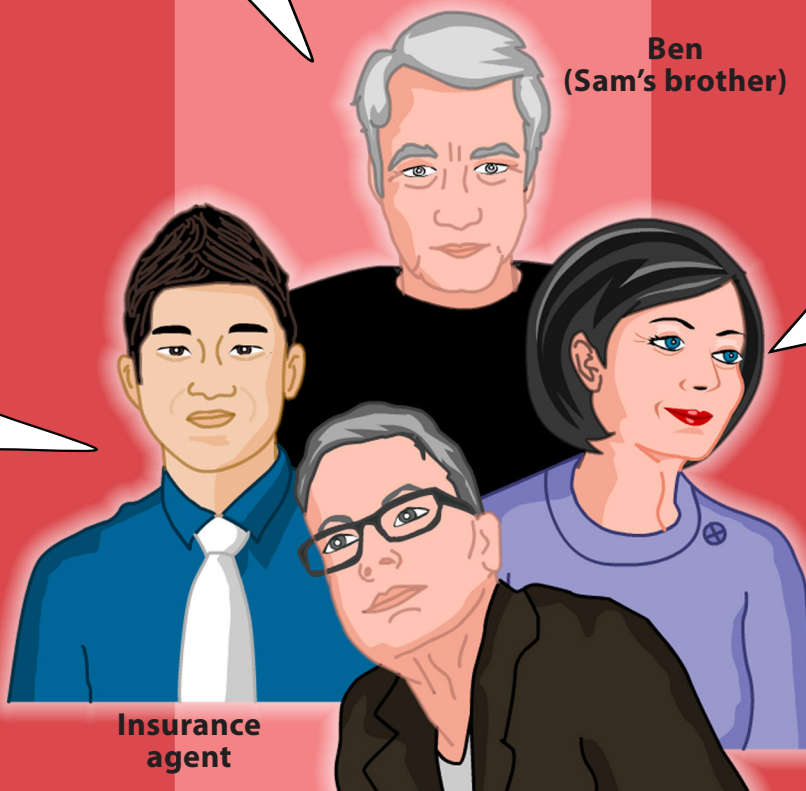
I just don't see the need to have these tests and give them to an insurance company. Sam's genes belong to him and his family. No one has the right to intrude in those relationships.

Esther
(Sam's wife)

"[In 2027] we cannot offer insurance to an individual with Sam's family history without taking extra precautions. This will help us to ensure Sam gets the right insurance policy for his current and future health needs.

Insurance agent

Sam





Is it better to know that you have a genetic predisposition or is it better not to know?

What would the impact of taking the test be on Sam's family? Are his wife's concerns justified?



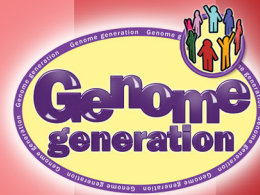
As knowledge about the genetic links to heart disease grows, and doctors have gathered data, should it be made accessible to others?

Should insurers be allowed to ask for this kind of information in the future?



Who should pay for the test? Should it be freely available through national health services, or, if not, is it fair that it is only available to those who can afford it?

Disease risk is often a balance between genes and lifestyle. Should people take responsibility for the impact their lifestyle has on turning a genetic predisposition into a disease?



Our genes

Most of the DNA in human cells is packaged into 46 chromosomes, in 23 pairs. Our genes are arranged along these chromosomes and carry the instructions to make molecules, such as proteins. To function properly, each cell depends on thousands of proteins doing the right job, in the right place, at the right time. Our complete set of DNA, containing all the instructions required to make us, is called a genome.



yourgenome.org



Heart disease and family risk

Although some families suffer more heart disease than others, inherited risk seems to involve many different genes and environmental factors. Scientists have identified over 20 different genes linked to heart disease. Individually, most of these genes do not make a big difference to overall heart disease risk, aside from a few rare exceptions.



yourgenome.org

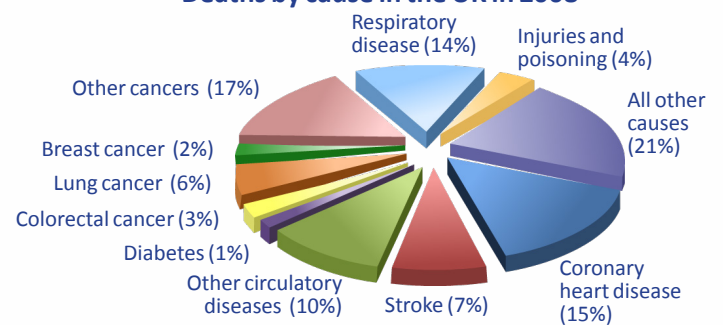


yourgenome.org

Risk factors for heart disease

Rates of heart disease are decreasing, but it remains the UK's biggest killer. It is caused by a complex mixture of environmental and inherited factors. Smoking, physical inactivity, obesity, poor diet and excessive alcohol consumption significantly increase risk. Raised blood cholesterol, raised blood pressure, diabetes and stress are also important risk factors. About 4% of men and 0.5% of women in the UK have had a heart attack.

Deaths by cause in the UK in 2008



Source: British Heart Foundation Statistics Database



yourgenome.org

What happens in a heart attack?

Men over 45 and women over 55 are at increased risk of heart attack. Heart attacks occur when a blood clot blocks an artery and cuts off the blood supply to the heart. This causes a sudden reduction in oxygen supply to the heart muscle. During an attack, areas of the heart muscle die and in severe cases, this can be fatal.



What could genomic analysis mean for heart disease?

In the future, personal genomic screening for heart disease could guide people to the best treatment for their genotype. For example, research suggests that people with particular forms of the *KIF6* gene, which has been linked to heart disease, get more benefit from cholesterol-lowering drugs than people with other genotypes. This could be because they are more vulnerable to the effects of raised cholesterol in the first place.



yourgenome.org

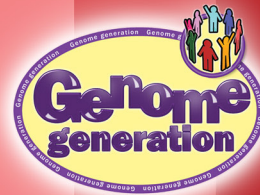


Insurance and predictive genetic tests

Currently there is no law to stop insurers from requesting information from **predictive** tests that have already taken place. However, there is currently a voluntary moratorium between the government and the insurance industry in the UK. It limits the access insurers have to genetic test results, with the exception of tests relating to Huntington's disease. It is unclear what will happen when the moratorium ends in 2017.



yourgenome.org



Insurance and medical history

Predictive genetic tests are used to see if a person is at **risk** of developing a disease, whereas **diagnostic** genetic tests can be used to decide how to treat an **existing** disease.

When calculating risk, insurers take into account medical history (including any **diagnostic** genetic tests), family history and lifestyle.



yourgenome.org

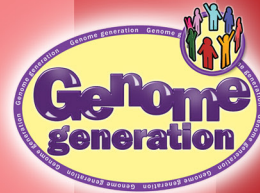


Genetic testing and employment

The 'Equality Act 2010' restricts the use of genetic tests by employers. Employers may only ask for information that is directly relevant to an applicant's ability to carry out the work.



yourgenome.org



yourgenome.org

What is genotyping?

Genotyping tests look for single letter changes in a person's genetic code, at specific locations that are associated with particular diseases or characteristics. Genotyping tests for heart disease have been developed but heart disease risk varies and is a result of complex interactions between several different genes and environmental factors such as lifestyle, diet and exercise. Because of this, some experts question the value of these genotyping tests.



yourgenome.org

Our DNA

Our bodies are made up of around 50 trillion cells. Most of these cells contain a person's genetic information within a long molecule called DNA (deoxyribonucleic acid). This DNA contains the genetic information that we inherit from our parents.

DNA is made up of specific sequences of letters or bases that encode instructions on how to make proteins. These bases [Adenine (A), Guanine (G), Cytosine (C) and, Thymine (T)] are lined up along two strands of DNA. The DNA is coiled into a double helix structure that resembles a twisted ladder.



Gene mutations

A mutation is a permanent change in the DNA sequence. Mutations come in a number of different forms and can change the functioning of our genes. Some mutations may lead to a difference in the amount, or structure, of a protein produced by specific genes. Some mutations may switch genes on at the wrong time, preventing a protein from doing its job, or changing the *way* it does its job.



yourgenome.org