The GRACE project

Medical uses of genetic testing

Kaustubh Adhikari:

In our last video, we answered the question ‘What is genetics?’ and talked about how changes in our genetic instructions can influence our health.

But you might be wondering, how can our knowledge of genetics actually help us?
Well, you may have heard of genetic testing.

Genetic testing looks at a person’s genetic information for changes which can tell us about their characteristics or chance of developing certain diseases.

This is usually done by taking a small sample of blood or saliva, which is sent to a special laboratory where the DNA is analysed.

There are many different types of genetic tests, so the right type of test for each person will depend on what information is needed.

In this video, we’ll talk about some of the different types of medical genetic tests, including tests which tell us about our own health, those used in pregnancy, and others used for medical research.

Sophie Roberts:

If someone’s experiencing unexplained symptoms diagnostic testing can confirm or rule out whether their symptoms are caused by a particular genetic disorder.

Getting a diagnosis can be a big step in the right direction to give people answers about their health and work out the best treatment options to manage their condition.

On the other hand, predictive testing can predict a person’s future risk of developing a disease before they show any symptoms, which may be suspected if a genetic condition runs in their family.

Some diseases are caused by a single genetic change, so having that change means a person will probably end up getting the disorder later, even if they’re unaffected when tested.

However, most common diseases like heart disease and cancer are much more complex.

Scientists have found many genetic changes which may increase or decrease a person's risk of developing certain diseases.

Predictive tests can estimate a person's genetic risk of getting a disease by looking for these changes in their genetic information.

But having one or more of these changes doesn't definitely mean someone will go on to develop a health condition.

This is because overall risk depends on a combination of many genetic changes and the influence of our environment and lifestyle choices, such as air pollution or activity levels.

Still, understanding genetic risk can help people make informed decisions about their health, such as having frequent screenings to detect a disease early on when it's more treatable.

Preventative treatments and lifestyle changes may also reduce the chance of someone developing the disease in the first place, delay its onset, or prevent serious complications.

You might've come across predictive genetic testing in the news after hearing about Angelina Jolie's experience.

Jolie knew she had a family history of breast and ovarian cancer, having lost her mother, grandmother, and aunt.

Studies have found breast and ovarian cancers can have a major genetic cause in some people, with changes in the BRCA genes massively increasing their risk.

Jolie took a preventative screening test and found she'd inherited the faulty genetic change, which meant she had an 87% chance of breast cancer and 50% chance of ovarian cancer in her lifetime.

Knowing this allowed her to make the decision to have preventative surgeries which greatly reduced her future risk, and sparked a worldwide conversation about genetic testing.

Echo Dyer:
Another type of predictive testing, called pharmacogenomic testing, aims to predict how someone may respond to certain drugs based on their genetic makeup.

This information can help healthcare providers choose which types of drug treatments will be most effective in treating a person's health condition and cause the fewest side effects.

Sophie Roberts:
Carrier screening is used to test whether a couple could pass a genetic condition on to their children, even if they don't have symptoms themselves.

This can happen when someone unknowingly carries a change in one copy of a gene that, on its own, doesn't usually cause symptoms.

But if two carriers for the same genetic change have a child together, there's a 1 in 4 chance of their child getting an altered gene copy from both parents, and having the disorder themselves.

Knowing if someone and their partner are carriers for the same genetic condition can be helpful when making important decisions about having children.

This is especially relevant for those who have a family history of a genetic condition, are closely related to their partner, or belong to an ethnic group with a higher risk for certain disorders.

Echo Dyer:
For those at higher risk of having a child with a serious genetic condition, preimplantation testing alongside in vitro fertilisation, otherwise known as IVF, can be a useful option.

These tests check early embryos for certain genetic abnormalities, so only those with a low risk of having a genetic disorder are chosen, which increases the chance of a successful pregnancy.

Sophie Roberts:
There are many genetic tests which give information on a baby's health during pregnancy and soon after birth.

Prenatal testing is offered during pregnancy to tell whether a baby is likely to have a certain genetic condition that could affect their health and development.

Prenatal screening tests check for a few conditions caused by big genetic changes, such as having an extra chromosome.

These tests are completely optional and non-invasive, meaning they won't hurt a baby, but can't give a definitive 'yes' or 'no' answer.

If screening tests find a baby is at higher risk of having a genetic condition, parents can choose whether to have a further diagnostic test, which gives clearer answers but is more invasive.

For some people, knowing the chance that their unborn baby has a health condition can help them make informed decisions about their pregnancy, further testing, and treatment options.

Mahfuzur Khokan:
Although most babies are born healthy, some may have health conditions which are not obvious right away.

When a baby is a few days old, quick and harmless newborn screening tests can check for certain serious but treatable genetic conditions.

This means babies with these conditions are identified early on so treatment can start as soon as possible to improve their health and prevent them from developing problems in the future.

Sophie Roberts:
The genetic tests we've talked about so far only give information on the health of the user. But what happens when researchers look at genetic data from lots of different people together?

Millions of volunteers around the world, including healthy individuals and those with health conditions, have participated in genetics research by choosing to share their genetic data.

This has helped scientists learn more about how genetic changes can cause certain diseases, and led to the development of more effective tests and treatments for many genetic disorders.

It's clear just how useful genetic testing can be in providing important information about our health.

The decision to have genetic testing is a personal one, so it's important to weigh up the potential benefits and risks to make the best decision for yourself and your family.

The more people who get involved in genetic testing, the more we can learn about the role of genetics in health and disease, to help us make advances in healthcare which benefit everyone.