



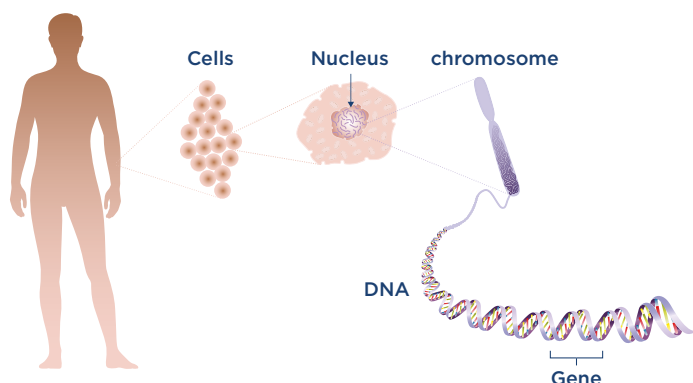
## What is genomic testing?

A genomic (or DNA) test is a medical test that examines the genome. Our genome is like an instruction manual for the body. It contains instructions that tell our body how to grow and develop.

The human genome is made up of 23 pairs of **chromosomes**, like the chapters in a book. These chromosomes are found in every cell of our body (Figure 1). Each of these chromosomes – or chapters – contains thousands of sentences. These sentences are called genes, and each gives a different instruction to the body. The letters that make up each word within the gene are the letters of our genetic code.

The spelling in our genes contributes to everything about us – how we look, the colour of our eyes or hair, and how our bodies function. Just like written instructions, a **spelling change or variation** in one of our genes might mean that the message is not read or received properly. These spelling changes are also called ‘variants’. Every person has many variants (changes) in their genes, and most of them do not affect the way our bodies work or how healthy we are.

**Figure 1. Chromosomes, DNA and genes**



Adapted from Health Education England Genomic Education Program: <https://www.genomiceducation.hee.nhs.uk/>

Sometimes having a variant in a gene can affect our health. Genomic testing looks for variants that are known to cause problems with the way our bodies develop and work and, sometimes, the way our brain develops and works. These types of variants are called pathogenic variants. Health conditions that are caused by pathogenic variants are called **genetic conditions**.



## Why is genomic testing done and how might it help us?

Genomic testing is done for many reasons. It can:

- Help to diagnose rare genetic conditions
- Help families to understand their child and their child’s condition, find specialist support and services, connect with and be supported by families with the same condition, and plan for the future
- Help families to advocate for their child
- Help resolve uncertainty and manage complex feelings
- Help health professionals optimise the management of health conditions and know what problems to look out for
- Help families **plan future pregnancies**
- Help families to find out whether other members of the family could develop the same condition.



### What is involved in genomic testing?

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Before a sample is taken for genomic testing, you will be asked to give consent for the testing to be done. DNA for genomic testing is usually obtained either from a blood sample (taken with a needle from a vein) or a saliva sample (taken by running a swab along the inside of the cheek).

The samples will then be sent to a laboratory. It may also be possible to use DNA that came from a blood or saliva sample that was taken for a previous genetic test. The laboratory will then analyse the DNA to look for variants in genes that are known to cause relevant health conditions.

When a child is being tested, it is often helpful to compare the spelling of their genes with both of their biological parents. This is called a trio test. In a trio test, the spelling of the parents' genes is used to help interpret any unique variants identified in the child, but is not the focus of the analysis.



### Can genomic testing provide an answer for every family?

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A diagnosis is usually found for around 40% of children (4 out of 10) who are thought to have a genetic condition.



### How long will it take before we receive a result?

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Genomic testing usually takes 3–6 months. It depends on where the sample is sent and how busy the laboratory is. You will be contacted once the result is available. You should check with your doctor if you have not received results within 6 months.

If you have not yet received a result and are planning a family or are pregnant, it is a good idea to tell your doctor that this testing has been done. You should also let your doctor know if there are any changes to your family health history or your contact details while you are waiting for the results.



### What results might we receive?

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When the laboratory has finished looking for changes in the genetic code that might have caused the condition you were tested for, they will send a report to your doctor. Possible results you might receive are:

- **Positive result:** A variant has been identified that is thought to be the cause of the condition. You will be told the name of the gene and the specific change that has been found.
- **Uninformative (negative) result:** No variants have been found that explain the condition. Not finding a variant that is causing the condition does not mean that the condition is not genetic. As we learn more in the future, it may be possible to go back and look at the data again, or repeat the test, to find an answer.
- **A result of uncertain significance:** A variant has been found in one or more genes, but it is not known whether this is the cause of the condition. This result is also known as a variant of unknown significance (VUS). The significance of this variant can sometimes be determined by looking at whether other family members with or without the same condition also have the same variant in their genetic code. Looking at the data again, or repeating the test, in the future may allow scientists/doctors to find out if the variants are significant or not.
- **An unexpected or incidental finding:** A variant has been identified in a gene that is not related to the condition for which the testing was done. These types of variants are not found very often (less than 1% of tests conducted in NSW). An example is a variant that is related to a different condition, such as cancer or heart disease. These are typically reported back to you if the information could alter your family's future health management.



### What effect will the test results have on other members of my family and on future children?

Sometimes, the genomic test results can show that the same genetic condition may occur again in future pregnancies. The chance of this happening depends on how the genetic variant is passed on or 'inherited'. You can discuss this with your doctor or another health professional if your child is found to have a genetic condition.

Genomic test results may also show that other family members could have a child with the same condition. In this case, it is important to inform other family members, so they can decide whether to arrange an appointment with their doctor to discuss testing. A **genetic counsellor** can also help you to pass on information to other family members.



### Who can access the test results?

Results are stored in your medical record and sent to your doctor. The results are confidential and cannot be released without your permission, unless required by law.



### Is there anything else I should know?

- The findings from genomic tests can reveal unexpected information about **family relationships**. For example, that the parents are not the biological parents or that the parents are blood relatives.
- **A positive result may not provide all the answers.** A genetic diagnosis cannot always tell us how much a genetic condition will impact on a person, whether the condition will progress, or whether other health conditions will develop. The diagnosis may not change the way the person is cared for if no specific treatment is currently available.
- **A positive result may give you information you do not want to hear.** It is possible the test may suggest that a person will experience future health problems, their condition may worsen, or rarely, that the condition will shorten their life.

- A genomic test is **not a general health test** and will not identify all gene changes that could cause health problems in the future.
- Rare incidental findings could affect your future applications for certain types of insurance such as **life, income protection or trauma insurance policies**. Health insurance is not affected by the results.
- You may be asked in the future if your genetic data can be used for **scientific research** studying the genetic condition in your family. It is your choice whether or not you wish this to happen.



### Can I change my mind about testing once the sample has been sent to the lab?

You can withdraw consent at any time by telling your doctor. If the testing is already complete, the report will be stored in your medical file, but you can ask to not be informed of the result.



It is important to understand the benefits and risks of genomic testing before a test is arranged.

Talking it over with family members, a close friend, your doctor or a genetic counsellor may help if you are unsure about whether genomic testing is right for you or your family

To print more copies of this fact sheet and access links to the underlined topics, go to [www.genetics.edu.au](http://www.genetics.edu.au) and search for 'About genomic testing'

*With thanks to Melbourne Genomics for their contribution to this resource*