What is genomic testing?

Australian Genomics Health Alliance

Your body is made up of cells, and most contain a complete copy of your genetic information (genome). Your genome is made up of 6 billion chemical letters (A, C, T and G). These are the instructions for the growth and development of your body. It is this sequence of chemical letters that makes you unique.

Genomics is the **study of the genome**.

Genomic testing (or genomic 'sequencing') allows us to read the chemical letters that make up your genome. Until recently, doctors and scientists were only able to test one gene at a time. Genomic technology now allows us to test all of your genes at once.

Genomic testing is used to:

- Help **diagnose rare genetic conditions**. Rare genetic conditions affect approximately 1 in 17 people, many of them children. Genomic testing can lead to a diagnosis in 30–50% of people with rare genetic conditions.
- Help families to **access support and services** that they need, and to plan for the future.
- Help health professionals **manage a condition**.
- Provide families with information about **the chance of having another child with the same condition.**
- Inform **care for relatives**. Sometimes, the genomic test result in one person may also be important for the care of their relatives.
- Help direct **cancer management, prediction and prevention**. Understanding the genetic changes that have occurred in a cancer can provide information about prognosis, and can help doctors choose the best treatment.
- Help doctors and scientists **understand complex genetic conditions** involving many genes, and environmental effects.

A genomic test produces **a lot of data**. Each person's genome contains many genetic differences (variants). Most are harmless and do not change how a gene works. Genomic testing is done to find genetic variants that **do change how a gene works and cause genetic conditions**.

From genomic testing data, teams of scientists and doctors try to identify one or two variants that may be causing a genetic condition. This is a complex and time-consuming task, and many checks are in place to ensure results are interpreted correctly.

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There are several possible outcomes of a genomic test:

- Finding a variant that is the cause of a person's condition.
- No gene variants found that could explain a person's genetic condition.
- Finding a variant of unknown significance (VUS). The effect of this variant is unknown.
- Finding a variant for an unrelated condition (incidental finding).

There are some potential risks to consider relating to genomic testing:

- **Incidental findings** – In genomic testing, we are looking at many genes at once, so there is a small chance doctors will find a variant in a gene that is not related to your health condition. This could give you health information that you may not want to know. You can discuss this with your doctor before you have the test and choose not to find out.
- **Insurance** – In Australia, genomic testing will not affect your health insurance policy. However genomic testing in you or your child may affect how easy it is for you or other family members to get income protection, travel or life insurance; or the price of your premium.

Doctors and researchers are still learning about what genes do and how they work. If a diagnosis doesn't happen today, your genomic information could be looked at in the future as our understanding improves.

The data from the genomic test can be stored, and can be re-examined again over time, either to answer the original question in light of new information or to answer new questions.

It is important to remember that genomic testing is not a general health test. It will not identify all variants that could contribute to health problems that may develop in the future.