What is genomic testing?

Australian Genomics Health Alliance

Your genome is your entire genetic information:

6 billion chemical letters

that make up the instructions for your growth and development, and make you unique.

Genomics is the study of the genome.

Genomic sequencing

allows us to read the chemical letters that make up your genome. It can be used as a medical test.

It is particularly useful to...

... help diagnose **rare genetic conditions.**

- Rare genetic conditions affect around 1 in 17 people, many of them children.
- A diagnosis can help make treatments better and help families plan for the future.
- Between 30% and 50% of rare disease patients receive a diagnosis through genomic testing.

... help **cancer** management, prediction and prevention.

- Understanding the genetic changes that have caused cancer can provide information about prognosis.
- It can help doctors choose the best treatments.

A genomic test produces vast amounts of data.

Each person's genome contains millions of genetic differences called **variants.**

Most of them are harmless.

From the data, teams of scientists and doctors try to identify 1 or 2 variants that may be causing a medical condition.

This is a complex and time-consuming task, and many checks and balances are in place to ensure the test is done correctly.

What are the possible results of a genomic test?

One or more gene variants are found to explain the condition for which the test was done.

No gene variants are found to explain the condition for which the test was done.

A variant is found in genes associated with the condition but the significance is not known (variants of unknown significance).

A gene variant is found for an unrelated condition (incidental finding).

The data from a genomic test can be stored.

It can be re-examined again over time, either to answer the original question in light of new information, or to answer new questions.

We are still learning about what genes do and how they work.

We expect that more patients will receive a diagnosis in the future, as the technology and our ability to interpret genomic information improve.

For more information, visit *genomicsinfo.org.au*

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