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When a genomic (DNA) test does not provide an answer



Key points

- Genomic testing cannot always find the cause of a health condition
- An uninformative result does not mean that the condition is not genetic (related to a change in the DNA)
- It may be possible in the future to repeat the test or re-analyse the data
- This result may also be described as *'negative', 'inconclusive' or 'no clinically significant variant identified'*



Why was genomic testing done?

Genomic (DNA) tests are performed to work out whether a particular health condition is caused by a change in a person's **genome**. Our genome is found in every cell in our body and is like an instruction manual for our bodies. The 'sentences' in this instruction manual are called **genes**, and each gives a different instruction to the body. Genes work together to tell our bodies how to grow, develop and function.

Changes in our genes are known as **variants**. Sometimes having a variant in a gene can affect our health, learning or physical development. Health conditions that are caused by variants in genes are called **genetic conditions**.

Genomic testing may find the variant(s) responsible for a health condition. However, in many cases a genetic cause cannot be found to explain a person's condition or symptoms.



Why wasn't a result found?

There are many possible reasons why the genomic test did not find a cause for the condition. It may be that:

- The test did not look at the gene that is linked with the condition.
- The gene causing the condition has not yet been identified or is not understood.
- The variant causing the condition cannot be found by the test. Sometimes the gene linked with the condition is examined but the change in the gene cannot be found using current testing methods. If the condition is caused by the combined effects of minor variants in two or more genes, this may not be detected easily.
- The health condition does not have an underlying genetic cause. The symptoms may have been caused by environmental factors, a **combination of environmental and genetic causes**, or other unknown causes.

An uninformative result does not mean that the condition is not genetic, just that a genetic cause could not be found using current ways of testing

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What happens now?

As technology improves and new genes are discovered, it may be possible to go back and look at the data again, or repeat the test, to find an answer. Although no answer has been found right now, one may be possible in the future.

If you have any questions about the result or additional testing, you can ask the health professionals who gave you the genomic test result. Your healthcare team will continue to care for you or your family member.

The genetic health professionals at a clinical genetics service can provide further information, advice and support, including **genetic counselling**. Details of your local clinical genetics service can be found at the **NSW Centre for Genetic Education**.

Sometimes it is possible to participate in research studies or patient registries. More information about these may be available from the support services below or the doctor who ordered the test for you.



Support

These organisations can provide support when a diagnosis cannot be found:

- **Genetic Alliance Australia**
- **Syndromes Without a Name (SWAN)**
- **Rare Voices Australia**

This fact sheet should not replace a consultation with a specialist healthcare professional.