PATIENT FACT SHEET

GENOME ONE®

GENETIC TESTING FOR HEART CONDITIONS

What are genetic heart conditions?

Genetic heart conditions are heart conditions that have an underlying genetic cause. A person's genes can influence the way the heart works, including its structure, strength, and how it beats. A change (variant) in one of the genes responsible for these functions can alter the way that the heart works, and may increase a person's risk of developing a heart condition. These genetic variants are usually inherited from a parent, but in some rare cases they may occur for the first time in that person.

There are three main types of genetic heart conditions:

- Aortopathies affect the large blood vessel that carries blood from the heart to the rest of the body (the aorta). Most of the time, aortopathies cause the aorta to become enlarged, making it weak and more likely to tear.
- Arrhythmias are disruptions to the heart's usual rhythm. This may result in the heart beating too fast, too slow, or with an irregular beat.
- Cardiomyopathies affect the heart muscle, causing it to become too weak or too thick. This can make it harder than usual for the heart to pump blood.

Genetic heart conditions are inherited in families, usually in a pattern known as autosomal dominant inheritance. This means that each parent with a variant causing a genetic heart condition has a 1 in 2 (50%) chance of passing the genetic variant on to each child that they have.

The symptoms of genetic heart conditions can vary greatly between family members, and can range from mild to severe.

Why has my specialist recommended this test?

Your symptoms, medical test results, and possibly your family history indicate that you might have a genetic heart condition.

The genetic test by Genome.One will analyse a group of genes associated with a specific type of heart condition. Your specialist selects the test that is appropriate for you, and can give you more information about the condition or conditions that you have been offered testing for.

What are the possible outcomes?

The genetic test offered by Genome.One involves looking at many variants in many genes to help identify the ones that might cause heart conditions. It's possible that you are the first person in your family to be offered genetic testing, or you might have been offered genetic testing because there is a relative in your family who is already known to have a genetic variant that is causing a heart condition. There are three potential outcomes to consider:

- VARIANT FOUND
  A variant is found in a gene that causes a genetic heart condition. We recommend you talk to your specialist about how this may impact you, your treatment and your family. Other family members may now be referred to a genetic specialist and offered genetic testing to determine if they have or have not inherited the same variant.
- NO VARIANT FOUND
  No variants were found in any of the genes tested. This does not change your clinical diagnosis or ongoing management. There may still be a variant in a gene not analysed in this test or as yet unknown to science. Your specialist can request to have your genome reanalysed in the future to see if additional variants have been discovered that might be relevant to you.*
- VARIANT OF UNCERTAIN SIGNIFICANCE (VUS)
  A genetic variant is identified in a gene, but there is uncertainty about whether it actually causes a genetic heart condition, either in you or in others. This does not change any clinical diagnosis you may have. Your specialist can request that the VUS be reviewed in the future to see if more information about its meaning has been identified.*

*additional fees may apply

What are the benefits of finding a genetic variant?

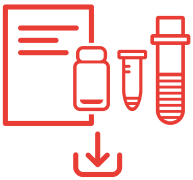
Genetic heart conditions can be passed on through your genes, meaning that there may be implications for both yourself and close family members to consider.

- Possible benefits for me
    - o Confirmation of a genetic heart condition your specialist may suspect you have or are at risk for
    - o Information that you and your specialist can use to make decisions about monitoring and treatment options specific to the diagnosis
- Possible benefits for my family
    - o Predictive testing in family members to determine if they have or have not inherited the same variant
    - o The option for earlier clinical screening in relatives who may have inherited the same variant, as well as earlier diagnosis and treatment if relevant
    - o The avoidance of unnecessary treatment in those who have not inherited the same variant
    - o Guidance for family planning (Genome.One does not offer preimplantation genetic testing)

How do I arrange a test?

1. Your specialist will refer you for testing by completing the Genome.One request form.

2. Your specialist will arrange for a genetic health professional to talk to you about the practicalities and implications of genetic testing. If you decide to proceed with the test you will need to sign a patient consent form.



3. Next you need to provide a blood sample at any pathology collection center that will be sent to Genome.One for testing.

4. Your results will be summarised in a detailed medical report that will be sent to your specialist within 8-12 weeks. Your specialist will then discuss the report with you.

How much does the test cost?

Please contact Genome.One for current pricing. Contact details are available on the back of this sheet.

Tests offered by Genome.One are not currently eligible for Medicare benefits and are not covered by private health insurance.

What happens next if a variant is identified?

Your specialist is the best person to talk to about your genetic testing results and how they influence your ongoing management.

There may also be implications for your family members, and genetic counselling is recommended for you and your family. Genetic counselling is helpful when talking to your family about your genetic test results and the implications for them.

Genetic counselling for heart conditions

The Human Genetics Society of Australasia recommends genetic counselling when you undergo genetic testing for heart conditions. Genetic counselling aims to provide you and your family with information and support as you work through the decision to have genetic testing, and can help you understand and adjust to the genetic results. Your specialist will be able to refer you to a genetic counselling service.

Genome.One can offer genetic counselling to individuals both before and after undergoing genetic testing with us at no additional cost. Please contact us if you would like to access this service.

Additional information about genetic counselling and genetic testing can be found in Fact Sheet 6 on the Centre for Genetics Education website
www.genetics.edu.au

How is my genetic information stored?

Your blood or saliva sample is held securely in the Genome.One laboratory in accordance with applicable law and standard laboratory practice.

Your personal information is securely stored in paper-based and electronic records in the Genome.One laboratory in accordance with the Genome.One privacy policy and applicable law. Your genomic data is held on servers in the United States of America for the purposes of storage and highly specialised data processing and analysis. However, before any data is transmitted to the USA, information identifying you is removed and replaced with a secure code. Only staff at Genome.One have the ability to link the code with your personal information.

Information from your test will not be shared with other people without your permission, except where this is required or permitted by law. Information from genetic testing is important for medical research about the genetic causes of disease. When providing consent, you have a choice whether your deidentified data can also be shared with researchers or included in research data banks.

The full Genome.One privacy policy is available at
www.genome.one/privacy-policy

How will genetic testing affect my insurance?

Under current Australian law, your private health insurance will not be affected by your decision to have genetic testing, or by any result of genetic testing.

However, genetic testing can have implications for your ability to obtain risk-rated insurance policies, including life insurance, disability insurance and income protection insurance. This could include denial of cover, exclusion from cover or increased premiums.

More information on how genetic testing affects your insurance can be found in Fact Sheet 20 on the Centre for Genetics Education website
www.genetics.edu.au

Patients outside of Australia should obtain advice about the implications of genetic testing for insurance in their home country.

Contact details

Genome.One

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P **1300 G-NOME-1 (1300 466631)**

E **enquiries@genome.one**

Further information and support

Heart Foundation Australia
www.heartfoundation.org.au

Australian Genetic Heart Disease Registry
www.heartregistry.org.au

Centre for Genetics Education Fact Sheets
www.genetics.edu.au/publications-and-resources/facts-sheets

FACT SHEET 6: Genetic counselling
FACT SHEET 20: Life insurance and genetic testing
FACT SHEET 57: Cardiomyopathies
FACT SHEET 58: Primary arrhythmogenic disorders

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