

Arranging a test

- 1 Your medical specialist will have completed a Cardiac Genetics Diagnostic Test Request Form.
- 2 Payment and collection of your blood sample is arranged by contacting 1800 010 447. Your blood sample is then collected at a Douglass Harly Moir Pathology collection centre.
- 3 We will arrange for an HGSA-qualified genetic counsellor to contact you to discuss testing.
- 4 The results of your test are reported to both your requesting clinician and genetic counsellor in 6-8 weeks by Bioscientia, a Sonic Healthcare laboratory.
- 5 Post-test counselling by an HGSA-qualified genetic counsellor is available to you, and can be arranged by your counsellor.

For further information, including scientific and peer-reviewed publications, please refer to our website, www.sonicgenetics.com.au/cardiac or call us on 1800 010 447.

Why do I need to sign a consent form?

As the outcome of genetic testing may have medical and psychological implications for you and your family, we require pre-test genetic counselling and informed consent prior to testing. This is arranged by Sonic Genetics and is provided to you as part of your cardiac genetic test.

There are some aspects of genetic tests that make them different to other types of medical tests. Many doctors, laboratories, professional societies and regulatory agencies worldwide require the patient's consent for genetic testing be formally obtained before the test is performed. The main reason for this requirement is that the result of a genetic test can:

- ▶ Provide information about your current disorder and carry implications for your future risk of other disorders
- ▶ Carry implications for your relatives regarding their risk of a familial disorder
- ▶ Influence decisions made by an insurance company regarding an application from you or a relative for some types of insurance, e.g. life insurance. Please note this does not apply to health insurance.
- ▶ Potentially allow a doctor to provide information about the genetic risk of disease in your family to your relatives, irrespective of your consent. There are strict restrictions on when and how this might be done; further details are available on our website.

For this reason, we require your informed consent for familial cardiac genetic testing. This consent will be discussed and obtained during your pre-test counselling session. Post-test counselling is also available, and can be arranged by your counsellor.

What does the test cost?

The current prices for our tests are provided on our website. At this stage, these costs are not rebated by Medicare or private health insurers. These tests involve exhaustive assessment of many different genes. Once a mutation has been found, relatives can be tested for the presence or absence of the specific mutation already identified in the family, and this is much faster and less expensive.

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Cardiogenetics

Information for Patients



This leaflet has been written for a person with a cardiovascular disorder who is considering genetic testing. The decision to have such testing should not be taken lightly. It is essential that the issue be discussed with an experienced medical specialist or genetic counsellor.

Further information can be found on our website and at the Australian Genetic Heart Disease Registry.

www.sonicgenetics.com.au
www.heartregistry.org.au

What is a cardiovascular disorder?

The heart pumps blood to the lungs and then to the body. Abnormalities in the heart or in the large veins and arteries taking blood to and from the heart are described as 'cardiovascular disorders'.

Cardiovascular disorders can be due to:

- › Abnormal structure of the heart from birth (congenital heart disorder)
- › Abnormal weakness of cardiac muscle (cardiomyopathy)
- › Abnormal cardiac rhythm (arrhythmia)
- › Progressive abnormalities in the large arteries leading to the lungs (pulmonary hypertension) or to the rest of the body (aortic disorder)
- › Other abnormalities in medium-sized arteries (vasculopathies)
- › Inherited high cholesterol (hyperlipidaemia) or abnormalities in cholesterol metabolism

In each case, the disorder can be due to a combination of inherited genetic errors, environmental factors and chance. The significance of genes, environment and chance will vary with different disorders and in different patients.

Do cardiovascular disorders run in families?

A 'familial cardiovascular disorder' is a disorder that runs in the family due to an inherited genetic error. If an inherited genetic error is the major underlying cause of your cardiac disorder, the abnormal gene may have been inherited by your relatives. Relatives who have inherited the abnormal gene may also be at risk of developing the disorder.

A diagnosis of a familial cardiovascular disorder does not mean that every person in the family will be affected, or that they will be affected in the same way. However, it does mean that there is an increased risk of relatives being affected and knowing that risk is an opportunity to act and reduce the impact of the disorder.

What is the purpose of genetic testing?

The purpose of a genetic test is to identify the underlying genetic error responsible for your cardiovascular disorder. If a genetic error is identified, this would indicate that your disorder is indeed familial, and that your relatives are at risk of carrying the same genetic error. Your relatives could then seek genetic testing if they wish. The identification of the underlying genetic error can also provide your medical specialist with more information to guide recommendations for your treatment.

We do not know all of the genes which can cause a specific disorder, so if the genetic test fails to identify an abnormal gene in you, this does not exclude the possibility that your disorder is familial.

What does the genetic test involve?

Cardiac genetic testing with Sonic Genetics involves having a blood sample collected from you, the affected person. The genetic material in your blood sample is then examined to identify an error in any one of the genes associated with your diagnosis.

The laboratory may also report genetic errors that are known to cause familial disorders, and may include genetic errors that can cause non-cardiac disorders.

The number of genes tested, and their identities, will vary with the disorder. They will also change over time, as our understanding of cardiac genetics improves. You can find the list of genes we currently test for each disorder on our website.

What will be done with the result of a genetic test?

Your doctor will tell you about the result of your test. If one or more genetic errors are found, your doctor will outline how this information explains your diagnosis and the medical implications for you.

The identification of a genetic error paves the way for your relatives to have genetic testing to clarify their risk. Genetic testing of an unaffected relative can raise a number of significant medical, psychological, legal and financial considerations, and so professional pre-test genetic counselling is essential.

