

Information for patients

# Cardiac genetics

Genetic testing for  
cardiovascular disorders



Cardiac genetic disorders can have long-term and significant health implications for both an individual and their biological relatives. Having information about the presence of an abnormality in a gene can assist in ongoing clinical management.

## **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, hypercholesterolaemia) 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 genetic error may have been inherited by your relatives.

## Arranging a test

- 1 Your medical specialist will have completed the appropriate Sonic Genetics test request form.
- 2 Payment (if not eligible for a Medicare rebate) is arranged by contacting 1800 010 447.
- 3 Your blood sample can be taken at any of our pathology collection centres.
- 4 Once the sample is received in the laboratory, if requested by your doctor, we will arrange for a genetic counsellor<sup>^</sup> to contact you to discuss testing.
- 5 The results of your test are reported to your doctor, usually within in 4–8 weeks.
- 6 Post-test counselling by a qualified genetic counsellor is available to you, and can be arranged by your requesting doctor or genetic counsellor.

<sup>^</sup> Terms and conditions apply.

Please refer to [www.sonicgenetics.com.au/patient/genetic-counselling](http://www.sonicgenetics.com.au/patient/genetic-counselling)

## Cost

Please refer to our website for current pricing, [www.sonicgenetics.com.au](http://www.sonicgenetics.com.au). At this stage, only the test for familial hypercholesterolaemia is rebatable by Medicare, subject to specific criteria. All other cardiac genetic tests that we offer are not rebated by Medicare and are not covered by private health insurance. These tests involve exhaustive assessment of many different genes.

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 developing 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.

The decision to have such testing should not be taken lightly. It is essential that the issues be discussed with an experienced medical specialist or genetic counsellor.

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 your genetic test fails to identify an abnormal gene, 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 cardiovascular diagnosis.

The laboratory may also report genetic errors that are known to cause familial disorders. This may include genetic errors that can cause non-cardiovascular disorders.

The genes tested will vary with the disorder. They will also change over time, as our understanding of cardiac genetics improves. The genes tested are listed on our website. This list will be updated as medical knowledge improves.

## **Before you have a cardiac genetic test**

A genetic test for cardiovascular disorders may be significant for both you and your family.

A discussion with your medical specialist or a genetic counsellor, together with a signed consent form, are required prior to the test.

The pre-test discussion will address the potential medical, social, psychological and financial impact of your genetic test result on you and your family.

Genetic counselling can also be provided by Sonic Genetics, usually at no additional cost to you, with no obligation to proceed with testing.

If a genetic error causing a cardiovascular disorder is found, your doctor will explain how this information will guide further investigations and therapy. It is also important that your close relatives are informed of your result and that genetic testing is available for them.

### **How you will receive your test results**

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.

You can also speak to a genetic counsellor about what the result means for you and your family.

**For further information, please refer to our website, [www.sonicgenetics.com.au/cardiac](http://www.sonicgenetics.com.au/cardiac), and at the Australian Genetic Heart Disease Registry, [www.heartregistry.org.au](http://www.heartregistry.org.au)**



**SONIC PATHOLOGY  
AUSTRALIA**

Sonic Genetics is part of Sonic Pathology Australia, the largest pathology provider in the country. Our state-of-the-art laboratories and extensive network of collection centres serve each state and territory capital, as well as regional and rural Australia.

Douglass Hanly Moir Pathology

Sullivan Nicolaidis Pathology

Melbourne Pathology

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Clinipath Pathology

Hobart Pathology

Launceston Pathology

North West Pathology

Southern.IML Pathology

For further information, please refer to our website,  
[www.sonicgenetics.com.au](http://www.sonicgenetics.com.au) or call us on 1800 010 447

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**Sonic Pathology Australia**

Level 3, 14 Giffnock Avenue, Macquarie Park, NSW 2113, Australia  
[www.sonicpathology.com.au](http://www.sonicpathology.com.au)