



Sonic
Genetics

Quality is in our DNA

Cardiac genetics

Information for Medical Specialists

Cardiac genetic disorders can have long-term and significant health implications for both the individual and their biological relatives. Some cardiac genetic disorders can be attributed to an abnormality in one of a number of genes. Sometimes an abnormality in one gene may cause many different disorders, including non-cardiovascular disorders. Having information about the presence of an abnormality in a gene can assist in the ongoing clinical management of your patient.

The purpose of genetic testing in cardiac disorders

A cardiac genetic disorder, or 'familial cardiovascular disorder', is a disorder that runs in the family because of an inherited genetic error. If an inherited genetic error is the major underlying cause of your patient's cardiac disorder, the abnormal gene may have been inherited by the patient, placing them at risk of developing the disorder.

If a cardiac genetic disorder is suspected, genetic testing should be considered to determine whether the underlying genetic cause can be identified. The identification of the genetic abnormality responsible for a cardiac disorder carries important implications for both the patient and their relatives. For this reason, pre-test counselling of the patient and family about the potential genetic implications of the test result is required. Sonic Genetics facilitates pre-test counselling with qualified counsellors. Post-test counselling is also available, and can be arranged by the genetic counsellor. Both pre- and post-test genetic counselling are included in the test cost.

Although there is no current cure for cardiac genetic disorders, the confirmation of a genetic cause for a patient's heart disease can provide valuable information about:

- The types of cardiac problems that could arise
- The risk and severity of life-threatening events, such as a cardiac arrest
- The most appropriate way to monitor cardiac health and risks
- The most effective medication to reduce the risk or severity of cardiac complications
- The potential value of more expensive interventions, such as implantation of a defibrillator
- Clinical screening advice for other at-risk family members
- Options for family planning, including pre-implantation genetic diagnosis



Cardiac genetic testing with Sonic Genetics

Sonic Genetics and Bioscientia, our sister laboratory in Germany, offer your patients a comprehensive cardiac genetics testing service, incorporating the latest laboratory analytics and interpretation of results. The Bioscientia laboratory is accredited to provide medical genetic testing by both German and American regulatory authorities, and has strong academic links across Europe. Genetic testing services provided by Sonic Genetics can be requested through your local Sonic Pathology Australia laboratory.

We currently offer testing of more than 130 different genes that have a known association with familial cardiovascular disease. We have grouped these genes into 11 panels that correspond to typical clinical presentations. Some genes are associated with a number of different clinical disorders and so are represented in more than one group.

If you wish to order more than one panel or test for a condition that is not listed below, please contact us on 1800 010 447.

The clinical disorders (and corresponding name of the gene panel) currently offered are as follows:

Cardiac diseases

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|---------------------|--|
| Cardiomyopathies | <ul style="list-style-type: none">➤ Dilated cardiomyopathy (DCM)➤ Hypertrophic cardiomyopathy (HCM)➤ Arrhythmogenic right ventricular cardiomyopathy (ARVC)➤ Left ventricular non-compaction (LVNC) |
| Cardiac arrhythmias | <ul style="list-style-type: none">➤ Long QT syndrome (LQTS) (including Jervell and Lange-Nielsen & Romano-Ward syndromes)➤ Brugada syndrome |

Vasculopathies

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| Vascular diseases and connective tissue diseases | <ul style="list-style-type: none">➤ Aortopathy (including Marfan syndrome)➤ Vasculopathies➤ Pulmonary hypertension |
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Hyperlipidaemias

- Familial hypercholesterolaemia (FH)
- Familial hypertriglyceridaemia (FHTG)

Access to pre- and post-test genetic counselling

Sonic Genetics provides pre- and post-test genetic counselling for patients referred for diagnostic cardiac genetic testing. The counselling service is provided by experienced genetic counsellors via phone, with the requesting clinician being kept informed. Fees associated with genetic counselling are included in the test cost and this service is provided by qualified genetic counsellors.

Arranging a test

- 1 Please complete a dedicated Sonic Genetics test request form, available on our website,* to select the appropriate test for your patient.
- 2 If the selected panel is not rebated by Medicare, the patient should be directed to contact us on 1800 010 447 to arrange pre-payment and sample collection.
- 3 If required, the patient will then be contacted by a qualified genetic counsellor to discuss testing and obtain informed consent, as appropriate.
- 4 The results of the test are reported in 4-8 weeks.
- 5 Post-test counselling by a qualified genetic counsellor is available to the patient, and can be arranged by the genetic counsellor.

*www.sonicgenetics.com.au/cardiacrf