



Sonic
Genetics

Douglass Hanly Moir Pathology

Cardiogenetics

Information for Medical Specialists

Laboratory diagnosis of
genetically determined
heart diseases with
associated pre- and post-
test genetic counselling



For each cardiovascular disorder, an abnormality in any one of a number of genes can make a major contribution to that disorder, that is, one disorder can be caused by many different genes. On the other hand, an abnormality in one gene can potentially cause a number of different disorders, that is, one gene can cause many different disorders, including non-cardiovascular disorders.



The purpose of genetic testing in cardiac disorders

A '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's relatives. Relatives who have inherited the abnormal gene may also be 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 error 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 arranges pre-test counselling with HGSA-qualified counsellors. Post-test counselling is also available, and can be arranged by the genetic counsellor. Both pre- and post-test genetic counselling is included in the test cost.

Although there is no current cure for genetic heart disease, 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 through Sonic Genetics

Through partnership with Bioscientia, our sister laboratory in Germany, Sonic Genetics offers 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 Douglass Hanly Moir Pathology.

We currently offer testing of 135 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, please contact us on 1800 010 447.

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

Non-syndromic heart diseases

Cardiomyopathies

- › Dilated cardiomyopathy (DCM)
- › Hypertrophic cardiomyopathy (HCM)
- › Arrhythmogenic right ventricular cardiomyopathy (ARVC)
- › Left ventricular non-compaction (LVNC)

Cardiac arrhythmias

- › Long QT syndrome (including Jervell and Lange-Nielsen & Romano-Ward syndromes)
- › Brugada syndrome

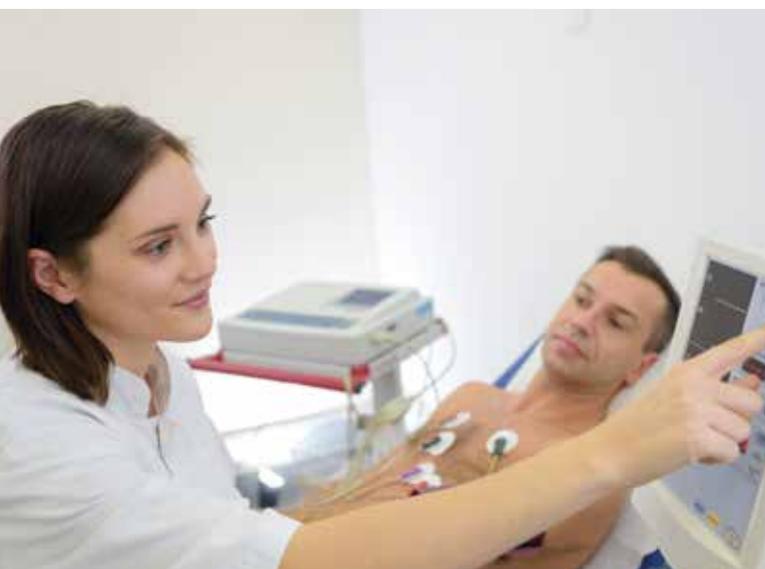
Syndromic heart diseases

Vascular diseases and connective tissue diseases

- › Aortopathy (including Marfan syndrome)
- › Vasculopathies
- › Pulmonary hypertension

Hyperlipidaemia

- › Familial hypercholesterolaemia
- › Familial hypertriglyceridaemia



Access to pre- and post-test genetic counselling

Sonic Genetics provides pre- and post-test genetic counselling for affected patients who have 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 independent genetic counsellors, who are not employees of Sonic Healthcare.

Arranging a test

- 1 Please complete our Cardiac Genetics Diagnostic Test Request Form, available on our website*, to select the appropriate test for your patient.
- 2 The patient will be asked to contact us on 1800 010 447 to arrange pre-payment and sample collection.
- 3 The patient will then be contacted by an HGSA-qualified genetic counsellor to discuss testing and obtain informed consent.
- 4 The results of the test are reported to both the 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 the patient, and can be arranged by the counsellor.

*www.sonicgenetics.com.au/cardiacrif



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PATHOLOGY

How to Access

Sonic Genetics is a member of the Sonic Healthcare group of companies.

Please contact your local Sonic Healthcare laboratory listed below:

- ACT** Capital Pathology
- NSW** Douglass Hanly Moir Pathology
Barratt & Smith Pathology
Southern.IML Pathology
- QLD** Sullivan Nicolaides Pathology
- SA** Clinpath Laboratories
- TAS** Hobart Pathology
Launceston Pathology
North West Pathology
- VIC** Melbourne Pathology
- WA** Clinipath Pathology

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

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