VCGS
Leaders in Genetic Health

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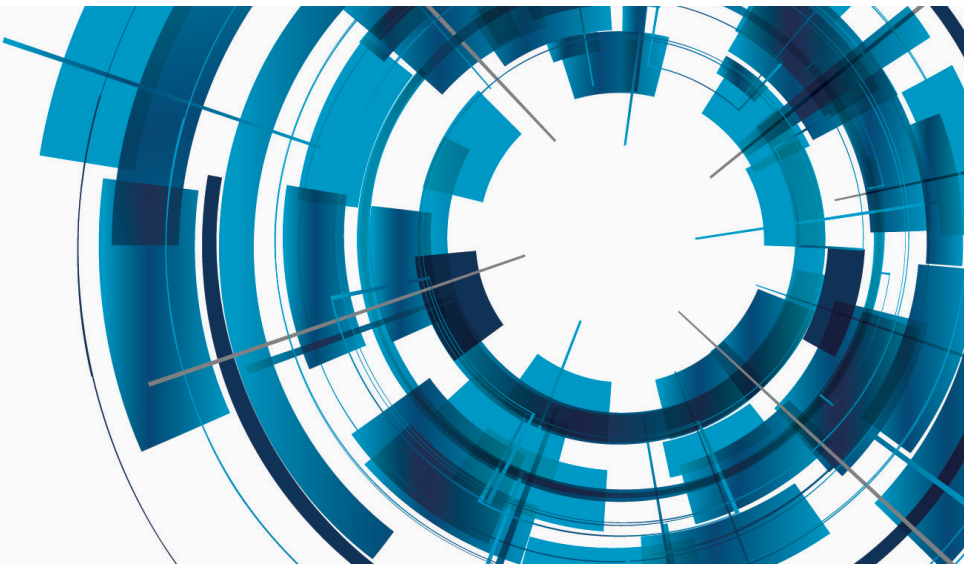
cardiac
gene panels



VCGS Cardiac Panels

VCGS offers a comprehensive range of Next Generation Sequencing based panels for the testing of hereditary cardiac conditions.

Depending on the clinical need of your patient, disease specific or broader panels are available for cardiomyopathies, arrhythmias and aortopathies. If required, VCGS offers genetic counselling support for requesting health professionals.

Benefits of VCGS Cardiac Panels

- VCGS is a leader in cardiac disease gene testing, being the first and longest standing provider of comprehensive clinical gene testing for genetic cardiac diseases in Australasia.
- VCGS has a deep knowledge base of genetic variation, built through many years of experience in cardiac disease testing.
- Carefully designed panels cover all disease relevant regions without gaps including splice sites.

With the ability to tailor panels and availability of segregation companion assays, our cardiac panels provide highly cost effective screening options.

- Panel designs are regularly reviewed to incorporate new disease associated genes.
- To complement our cardiac panels, we offer a comprehensive range of companion assays, including predictive and prenatal testing as well as customised panel design and data re-analysis options.

Our Cardiac Panels

Arrhythmia panels:	Full arrhythmia panel (33 genes)
	Long QT syndrome panel (13 genes)
	Brugada syndrome panel (12 genes)
	Catecholaminergic Polymorphic Ventricular Tachycardia panel (7 genes)
	Short QT syndrome panel (3 genes)
	Familial Atrial Fibrillation panel (4 genes)
Cardiomyopathy panels:	Full cardiomyopathy panel (63 genes)
	Dilated cardiomyopathy panel (39 genes)
	Hypertrophic cardiomyopathy panel (34 genes)
	Arrhythmogenic right ventricular cardiomyopathy panel (8 genes)
	Left ventricular non-compaction cardiomyopathy panel (8 genes)
Other panels:	Full aortopathy panel (17 genes)
	Structural congenital cardiac anomalies panel (18 genes)
Comprehensive genetic cardiac disorders panel:	All genes from the arrhythmia, cardiomyopathy & aortopathy panels (111 genes)
Customised panels (on request):	Custom selection of up to 5 genes

Cardiac Panel Report: What to expect

- VCGS uses analysis and interpretation pipelines that provide a detailed investigation of all variants found in each gene contained within the requested panel.
- Only pathogenic variants, likely pathogenic variants and variants of unknown significance (VUS) are reported.
- The laboratory will also report ACTIONABLE pathogenic/likely pathogenic variants in non-requested cardiac genes (only those previously observed in the laboratory's cohort) for consideration by the clinical team or health professional managing the patient and family.
- We also offer a re-analysis service where alternative gene panels can be screened if no significant variants have been detected in the selected panels.

VCGS: Sharing Knowledge and Supporting Choice.

Variant classification

Class 5: Pathogenic variant: Pathogenic variants are considered disease-causing.	• At-risk unaffected relatives can be offered predictive gene testing • Other affected relatives can be offered confirmatory testing • Prenatal diagnosis for the pathogenic variant is possible
Class 4: Likely pathogenic variant: The level of evidence that likely pathogenic variants are disease-causing is very high.	• At-risk unaffected relatives can be offered gene testing in conjunction with clinical screening • Other affected relatives can be offered confirmatory testing • The variant may be considered for use in prenatal diagnosis after detailed discussion with a clinical geneticist or genetic counsellor
Class 3A: Variant(s) of unknown significance with high clinical significance: VUS with high clinical significance are variants that have evidence highly suggestive of a likely pathogenic variant but there is not enough information to classify them as class 4*.	• Class 3A variants cannot be used for predictive testing or prenatal diagnosis • Co-segregation studies in affected relatives, or testing to determine if the variant is de-novo is strongly recommended as these studies may provide additional evidence to clarify the pathogenicity of class 3A variants *These variants may be re-classified based on new information; for example, family and/or functional studies (if performed).
Class 3B: Variant(s) of unknown significance: Class 3B VUS are variants for which there is insufficient evidence to classify the variant as either disease causing or likely benign.	• Class 3 variants cannot be used for predictive testing or prenatal diagnosis • In selected families, co-segregation studies in affected relatives may help to clarify pathogenicity of a class 3 VUS
Class 3C: Variant(s) of unknown significance with low clinical significance: Class 3C VUS are variant(s) of unknown significance with low clinical significance.	• Class 3C variants cannot be used for predictive testing or prenatal diagnosis
No variant of significance was found.	• Reanalysis options may be considered if the family history strongly indicates a genetic cause

Arranging cardiac panel testing

Cardiac panel testing is arranged through Specialist Cardiac Genetic Clinics and any additional queries could be sent to: molgen.general@vcgs.org.au.

SPECIMEN REQUIREMENTS:

Whole blood: 2 x 4 ml EDTA whole blood. Shipped at room temperature and must arrive within 3 days of collection. Do not freeze.

DNA: DNA will be accepted but will be assessed for quality before being processed. Contact the laboratory for further details.

When are results available?

For new cases to identify family-specific mutations:	3-4 months
For predictive testing of family members:	3-4 weeks
Prenatal testing:	2-3 weeks

VCGS expertise

Integrated Service:

VCGS offers a unique, fully integrated service that provides a range of childhood and adult pathology testing and clinical genetics services. We are scientists, genetic counsellors and clinical geneticists that work together to provide the most comprehensive service for patients and health professionals.

Expertise:

VCGS has been offering testing and genetic counselling services for 30 years. Combined with the Murdoch Childrens Research Institute, we have the largest genetic expertise in the Southern Hemisphere, with well-established pipelines for translating clinical research into diagnostics and genetic support services.

Contact details

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For more information and details about our panels and gene lists please visit: www.vcgs.org,au

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