Genome.One offers a portfolio of genomic sequencing based panel tests for patients with suspected genetic cardiac conditions. The portfolio includes both disease-specific panels and broader category panels for cardiomyopathies, arrhythmias, aortopathies and RASopathies.

Disease-specific panels are designed to be flexible, offering analysis of either the complete panel or the core set of most commonly implicated genes.

All Genome.One cardiac panels can be based on either whole genome sequencing (WGS) or whole exome sequencing (WES), and use a targeted analysis approach.

### Genome.One cardiac panels

<table>
<thead>
<tr>
<th>Condition</th>
<th>Core Panel</th>
<th>Complete Panel</th>
</tr>
</thead>
<tbody>
<tr>
<td>All cardiomyopathies</td>
<td></td>
<td>91</td>
</tr>
<tr>
<td>Dilated cardiomyopathy</td>
<td>29</td>
<td>48</td>
</tr>
<tr>
<td>Hypertrophic cardiomyopathy</td>
<td>14</td>
<td>54</td>
</tr>
<tr>
<td>Arrhythmogenic right ventricular cardiomyopathy</td>
<td>4</td>
<td>9</td>
</tr>
<tr>
<td>Left ventricular noncompaction</td>
<td>9</td>
<td>10</td>
</tr>
<tr>
<td>All arrhythmias</td>
<td></td>
<td>43</td>
</tr>
<tr>
<td>Long QT syndrome</td>
<td>10</td>
<td>15</td>
</tr>
<tr>
<td>Short QT syndrome</td>
<td>4</td>
<td></td>
</tr>
<tr>
<td>Catecholaminergic polymorphic ventricular tachycardia</td>
<td>4</td>
<td></td>
</tr>
<tr>
<td>Atrial fibrillation</td>
<td>8</td>
<td>10</td>
</tr>
<tr>
<td>Brugada syndrome</td>
<td>1</td>
<td>25</td>
</tr>
<tr>
<td>Aortopathies</td>
<td></td>
<td>20</td>
</tr>
<tr>
<td>RASopathies</td>
<td></td>
<td>15</td>
</tr>
<tr>
<td>Congenital heart disease</td>
<td></td>
<td>27</td>
</tr>
<tr>
<td>Familial hypercholesterolaemia</td>
<td></td>
<td>4</td>
</tr>
</tbody>
</table>

The requesting clinician is responsible for selecting the panel to be tested.
**Indications for genetic testing**

Genetic testing using Genome.One cardiac panels is recommended in patients:

- With a clinical or suspected diagnosis of a genetic cardiac condition, with or without a family history
- Where no prior genetic testing has been performed
- When a genetic diagnosis of a genetic cardiac condition has not yet been made in an accredited laboratory
How to arrange a test

Discuss the testing options with the patient and complete the patient consent form. Alternatively, contact us to arrange a pre-test consultation and consent with a Genome.One genetic counsellor. Contact details can be found on the back of this sheet.

Determine whether the costs of the test will be covered by your institution or privately paid for by the patient. Private patients will be sent an invoice by Genome.One for payment.

Complete either our online or paper test request form.

Give a copy of the completed request form to the patient for a whole blood sample collection (5 mL for adults, 2 mL for paediatrics).

You will receive a report detailing the results 8-12 weeks after the specimen reaches Genome.One.
Benefits of genetic testing for cardiac conditions

- The potential for earlier, more accurate diagnosis
- Identification of an existing genetic cause to support a suspected diagnosis
- More informed management and treatment options
- Predictive testing or pre-symptomatic screening for at-risk relatives, with ongoing management for people who are affected or at-risk
- Guidance for family planning (Genome.One does not offer preimplantation genetic testing)

What Genome.One offers

- Whole genome sequencing and whole exome sequencing-based panel tests with gene lists regularly maintained based on current evidence
- The ability to start with the most commonly implicated genes (core panel) and expand to a larger extended panel if no variants are found
- Expert pathology and genetic counselling support, with in-house genetic counselling services available to patients both before and after testing, at no additional cost
- Optional reanalysis of data in the future to answer clinical questions that may arise*
- Streamlined ordering process through www.genome.one

*additional fees may apply

Genetic counselling for cardiac conditions

The Human Genetics Society of Australasia recommends genetic counselling for all patients considering or undergoing genetic testing for cardiac conditions.

Genetic counselling aims to provide individuals and their families with information and support when there is a suspected genetic condition in the family. Genetic counselling is usually performed pre- and post-genetic testing to help patients understand the implications of genetic information for themselves and their family.

Genome.One offers genetic counselling to individuals undergoing genetic testing with our laboratory at no additional cost. You can refer your patient to our genetic counselling services using the contact details on the back.

Alternatively, a list of Australian genetic counselling services can be found on the Centre for Genetics Education website at: www.genetics.edu.au/genetics-services/genetic-counselling-services

Insurance information

Under current Australian law, a person’s private health insurance will not be affected by their decision to have genetic testing, or by the result of genetic testing. However, genetic testing can have implications for a person’s ability to obtain risk-rated insurance policies, including life insurance, disability insurance and income protection insurance. This could include denial of cover, exclusion from cover or increased premiums.

Patients outside of Australia should obtain advice about the implications of genetic
Genome.One cardiac panels

**Dilated cardiomyopathy**
- **Core**
  - ACTC1, BAG3, CRYAB, CSRP3, DE2, DMD, DSG2, DSP, FLNC, HFE, ILK, LAMA4, LAMP2, LB3, LMNA, MYBPC3, MYH7, NEXN, NXX2-5, PLN, RRM20, SCNA5, TAZ, TNNC1, TNN3, TNNT2, TPM1, TTN, VCL
- **Extended**
  - ACTN2, ALMS1, ANKRD1, DNAJC19, DOLK, EMD, EYA4, FKTN, GATAD1, JUP, MYH6, MYPN, OBSCN, SDHA, SGCB, SGCD, SYNE1, SYNE2, TCAP

**Long QT syndrome**
- **Core**
  - ACTN2, CACNA1C, CALM1, CALM2, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, SCN5A
- **Extended**
  - AKAP9, CAV3, KCNJ5, SCN4B, SNTA1

**Short QT syndrome**
- **Core**
  - CACNA2D1, KCNH2, KCNJ2, KCNQ1

**Catecholaminergic polymorphic ventricular tachycardia**
- **Core**
  - CALM1, CASQ2, RYR2, TRDN
- **Extended**
  - SCN1B, SCN2B

**Atrial fibrillation**
- **Core**
  - EMD, GJA5, KCNA5, NPPA, NUP155, SCN3B, SCN4B, SCN5A
- **Extended**
  - SCN1B, SCN2B

**Brugada syndrome**
- **Core**
  - SCNA5
- **Extended**
  - ABCC9, AKAP9, ANK2, CACNA1C, CACNA2D1, CACNB2, FGFT2, GPD1L, HCN4, KCN2D, KCND3, KCNE1L, KCNE3, KCNH2, KCNJ8, PKP2, RANGRF, SCN10A, SCN1A, SCN2B, SCN3B, SEMA3A, SLMAP, TRPM4

**Dilated cardiomyopathy**
- **Core**
  - ACTC1, BAG3, CRYAB, CSRP3, DE2, DMD, DSG2, DSP, FLNC, HFE, ILK, LAMA4, LAMP2, LB3, LMNA, MYBPC3, MYH7, NEXN, NXX2-5, PLN, RRM20, SCNA5, TAZ, TNNC1, TNN3, TNNT2, TPM1
- **Extended**
  - ACTN2, ALMS1, ANKRD1, DNAJC19, DOLK, EMD, EYA4, FKTN, GATAD1, JUP, MYH6, MYPN, OBSCN, SDHA, SGCB, SGCD, SYNE1, SYNE2, TCAP

**Hypertrophic cardiomyopathy**
- **Core**
  - ACTC1, ACTN2, CSRP3, GLA, LAMP2, MYBPC3, MYH7, MYL2, MYL3, PLN, PRKG2, TNM1
- **Extended**
  - ACTA1, ATP5E, BRF, CACNA1C, CALR3, CAV3, CBL, CRYAB, FHI1, FLNC, FXN, GAA, HRAS, JPH2, KRAS, LDB3, LMNA, MAP2K1, MAP2K2, MYH6, MYLK2, MYOZ2, MYVN, NEXN, NF1, NRAS, PTNP11, RAF1, RASA1, RIT1, SHOC2, SLC2A4, SOS1, SPRED1, TCAP, TNNC1, TSPM, TTR, VCL

**Arrhythmogenic right ventricular cardiomyopathy**
- **Core**
  - DSC2, DSG2, DSP, PKP2
- **Extended**
  - CTNNA3, JUP, RYR2, TGB3, TMEM43

**Left ventricular noncompaction**
- **Core**
  - ACTC1, DTNA, LB3, MYBPC3, MYH7, PRDM16, TAZ, TNNT2, TPM1
- **Extended**
  - MIB1

**All cardiomyopathies**
- **Complete**
  - ALL GENES AS ABOVE

**All arrhythmias**
- **Complete**
  - ALL GENES AS ABOVE

Please visit [www.genome.one](http://www.genome.one) for any updates to the gene compositions of our cardiac panels.
Privacy

Blood or saliva samples are held securely in the Genome.One laboratory in accordance with applicable law and standard laboratory practice.

Patient personal information will be securely stored in paper-based and electronic records in the Genome.One laboratory in accordance with the Genome.One privacy policy and applicable law. Genomic data is held on servers in the United States of America for the purposes of storage and highly specialised data processing and analysis. However, before any genomic data is transmitted to the USA, information identifying the patient is removed and replaced with a secure code. Only staff at Genome.One have the ability to link the code with the patient’s personal information.

Information from WGS screening will not be shared with other people without the patient’s permission, except where this is required or permitted by law. Information from WGS is important for medical research about the genetic causes of disease. When providing consent, the patient has a choice whether their deidentified genomic information can also be shared with researchers or included in research data banks.

The full Genome.One privacy policy is available at www.genome.one/privacy-policy

Contact us for an example report or for any further information about the cardiac testing process.

Contact details

Genome.One

370 Victoria St Darlinghurst NSW 2010 Australia

P 1300 G-NOME-1 (1300 466631)

E clinical@genome.one

Genome.One mission statement

Genome.One is a pioneering health information company providing genetic answers to health questions, enhancing the lives of patients, families and communities across the world.

As a wholly-owned subsidiary of the Garvan Institute of Medical Research, we’re backed by pioneering clinical discovery and translational medicine research excellence. Our laboratory is accredited by NATA for Medical Testing and meets ISO 15189 requirements.