

CARDIOLOGY

Know&Manage

Comprehensive cardiac health with genetic insights

MEDICOVER GENETICS

CARDIOLOGY KNOW&MANAGE

KNOW THE CAUSE

Inherited cardiovascular disorders, including cardiomyopathies, inherited arrhythmia syndromes, aortopathies, familial lipid disorders, congenital heart defects, and metabolic conditions with cardiac involvement, are major causes of heart failure, life-threatening arrhythmias, and sudden cardiac death[1].

MANAGE THE RISK

Genetic testing provides essential diagnostic and prognostic information, enabling earlier and more accurate diagnosis, personalised risk stratification, and implementation of guideline-directed surveillance and management[1]. A confirmed molecular diagnosis can inform treatment decisions, identify patients eligible for genotype-specific therapies, and support cascade testing of at-risk relatives, facilitating early intervention[1].

Genetic testing can distinguish overlapping conditions, identify alternative causes, and shorten the path to a definitive diagnosis.

WHY **RECOMMEND** GENETIC TESTING

- Precise molecular diagnosis
- Reduced diagnostic delay and patient burden
- Genotype-guided therapy and surveillance
- Risk stratification and prognosis assessment
- Predictive family screening and early intervention in asymptomatic relatives
- Pharmacogenetic optimisation of cardiovascular therapy

WHEN TO **CONSIDER** GENETIC TESTING

Patient-related findings

- Unexplained ventricular arrhythmias, recurrent syncope or aborted sudden cardiac death (SCD)
- Abnormal ECG findings suggestive of inherited arrhythmia syndrome
- Imaging findings suggestive of cardiomyopathy, particularly in children and young adults
- Heritable thoracic aortic aneurysm or dissection, or bicuspid aortic valve
- Heart defects where a genetic aetiology is suspected, particularly syndromic or familial forms
- Premature coronary artery disease
- Patients receiving cardiovascular polypharmacy where metaboliser status may affect drug efficacy or safety

Family history–related findings

- Positive family history of an inherited cardiovascular disease or characteristic clinical phenotype
- SCD in a first- or second-degree relative
- First-degree relative of an index case once a causal genetic variant has been identified (predictive testing)

This selection highlights common cardiovascular genetic findings and does not replace comprehensive clinical evaluation. For detailed diagnostic pathways and management recommendations, please refer to current (ESC) Guidelines and ESC cardiovascular genomics consensus statements[2].

1. Elliott P, et al. Eur Heart J. 2025;46(4):344-353. doi:10.1093/eurheartj/ehae747
2. European Society of Cardiology. ESC Clinical Practice Guidelines. Available from: https://www.escardio.org/guidelines/clinical-practice-guidelines/all-esc-practice-guidelines

OUR **TESTS**

Our cardiogenomics portfolio delivers comprehensive molecular support across the full spectrum of inherited cardiovascular disease, helping clinicians optimise patient care and improve long-term outcomes.

CARDIOLOGY - COMPREHENSIVE PANEL

ARRHYTHMIAS / ION CHANNEL DISORDERS

Arrhythmia – comprehensive panel

- Atrial fibrillation
- Brugada syndrome
- Calcium release deficiency syndrome
- Catecholaminergic polymorphic ventricular tachycardia
- Early repolarisation syndrome
- Long QT syndrome
- Progressive cardiac conduction disease
- Short QT syndrome
- Sinus node disease

CARDIOMYOPATHIES

Cardiomyopathies – comprehensive panel
(also recommended in cases of recurrent myocarditis)

- Arrhythmogenic cardiomyopathy
- Dilated cardiomyopathy
- Hypertrophic cardiomyopathy
- Left ventricular non-compaction cardiomyopathy (LVNC)
- Restrictive cardiomyopathy

CONGENITAL HEART DEFECTS[3]

Congenital heart defects – comprehensive panel

- Alagille syndrome
- Heterotaxy
- RASopathies (incl. Noonan syndrome)

CONNECTIVE TISSUE AND AORTIC DISORDERS

- Bicuspid aortic valve with risk of aortic valve stenosis and dilatation
- Vascular Ehlers-Danlos syndrome
- Loeys-Dietz syndrome
- Marfan syndrome
- Thoracic aortic aneurysm and dissection

ADDITIONAL ANALYSES

LIPID METABOLISM DISORDERS

- Cholesteryl ester storage disease / Wolman disease
- Familial combined hyperlipidaemia (dysbetalipoproteinaemia)
- Familial hypercholesterolaemia
- Hyperlipidaemia
- Hypobetalipoproteinaemia
- LCAT deficiency / Norum disease / Fish eye disease
- Mixed hyperlipoproteinaemia
- Primary hypertriglyceridaemia / Familial chylomicronaemia syndrome
- Sitosterolaemia
- Tangier disease

STORAGE DISEASES

- Fabry disease
- Hereditary ATTR amyloidosis
- Glycogen storage disease II (Pompe disease)

PHARMACOGENETICS[4]

- PGx Cardiovascular *(CYP2C19, CYP2C9, CYP2D6, CYP4F2, SLCO1B1, VKORC1)*
- PGx Mavacamten *(CYP2C19)*

CYTOGENOMICS[4]

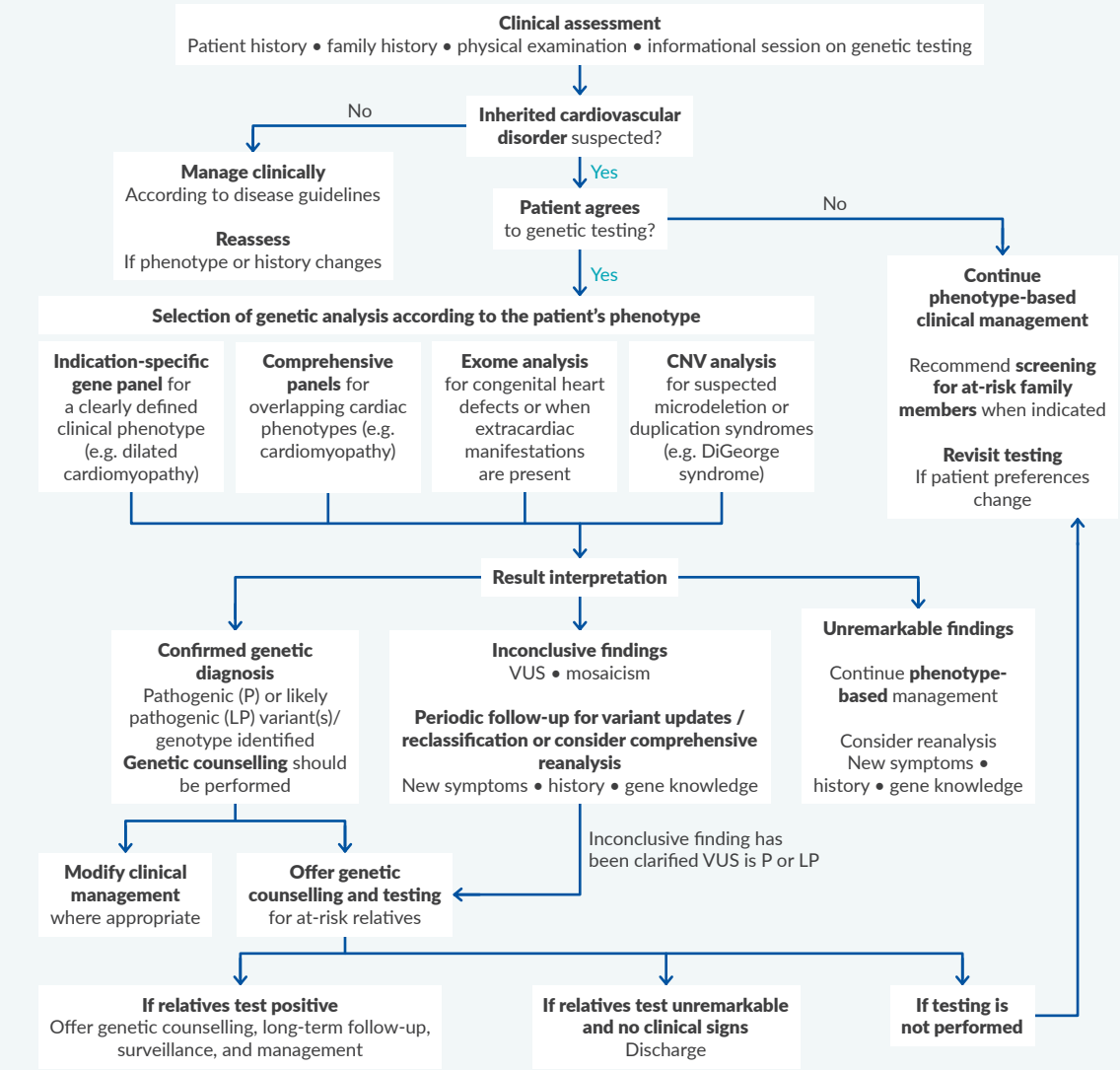
- CNV analysis (CMA or low-coverage WGS)

TRINUCLEOTIDE REPEAT DISORDERS[4]

- Myotonic dystrophy type 1 (DMPK)
- Friedreich ataxia (FXN)

3. In selected cases of suspected genetically determined congenital heart defects, comprehensive whole-exome analysis may be considered in line with European Society of Cardiology (ESC) guidance on genetic testing in congenital heart disease.
4. Not included in Cardiology - comprehensive panel

DIAGNOSTIC **ALGORITHM**

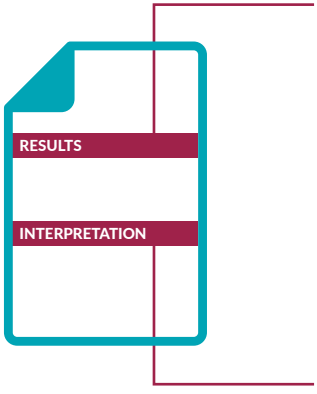


Workflow based on current cardiovascular genetics guidelines from the German Cardiac Society (DGK), the Society for Human Genetics (GfH) and the German Society for Pediatric Cardiology (DGPK) and European Society of Cardiology (ESC).

POSSIBLE **OUTCOMES** OF TESTING

Interpretation of genetic findings requires correlation with the patient's clinical phenotype, family history, and other relevant clinical information. Variants are classified according to the five-tier ACMG/AMP framework, namely pathogenic, likely pathogenic, variant of uncertain significance, likely benign or benign. Where appropriate, gene- and disease-specific criteria are also considered, in line with ESC cardiovascular genomics recommendations[2].

Results are reported according to clinically relevant variant categories:



PATHOGENIC OR LIKELY PATHOGENIC VARIANT

One or more genetic variants known or likely to be disease-causing may be identified. This can confirm or support the molecular diagnosis and inform risk stratification, clinical management, and family testing.

VARIANT OF UNCERTAIN SIGNIFICANCE (VUS)

One or more genetic variants with currently insufficient evidence to determine whether they are disease-causing or benign may be identified. Clinical management should remain guided by the patient's phenotype and family history. Re-evaluation over time may support variant reclassification. Predictive testing in relatives based solely on a VUS is not recommended.

NO CLINICALLY RELEVANT VARIANT IDENTIFIED

No disease-associated variants are detected in the analysed genes. This does not exclude a genetic cause. Clinical management should remain guided by the patient's phenotype and family history, and reanalysis may be considered over time.

TESTING **METHODOLOGY AND QUALITY**

Our cardiology panels, whole-exome sequencing (WES), and low-coverage whole-genome sequencing (lcWGS) are based on next-generation sequencing (NGS), enabling comprehensive detection of sequence variants and copy number variations. Additional methods may be applied when clinically indicated.

Variant interpretation follows established professional standards and is performed in the context of the patient's clinical presentation and family history.

HOW TO **ORDER**



Select the appropriate genetic test based on patient's phenotype; inform and obtain consent



Collect the sample(s) (preferably EDTA blood)



Send the sample(s) to **Medicover Genetics**



The sample(s) will be analysed at **Medicover Genetics** laboratories



Results will be sent to you

CONTACT US

If you have additional questions or concerns, please contact us at **info.genetics@medicover.com**

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