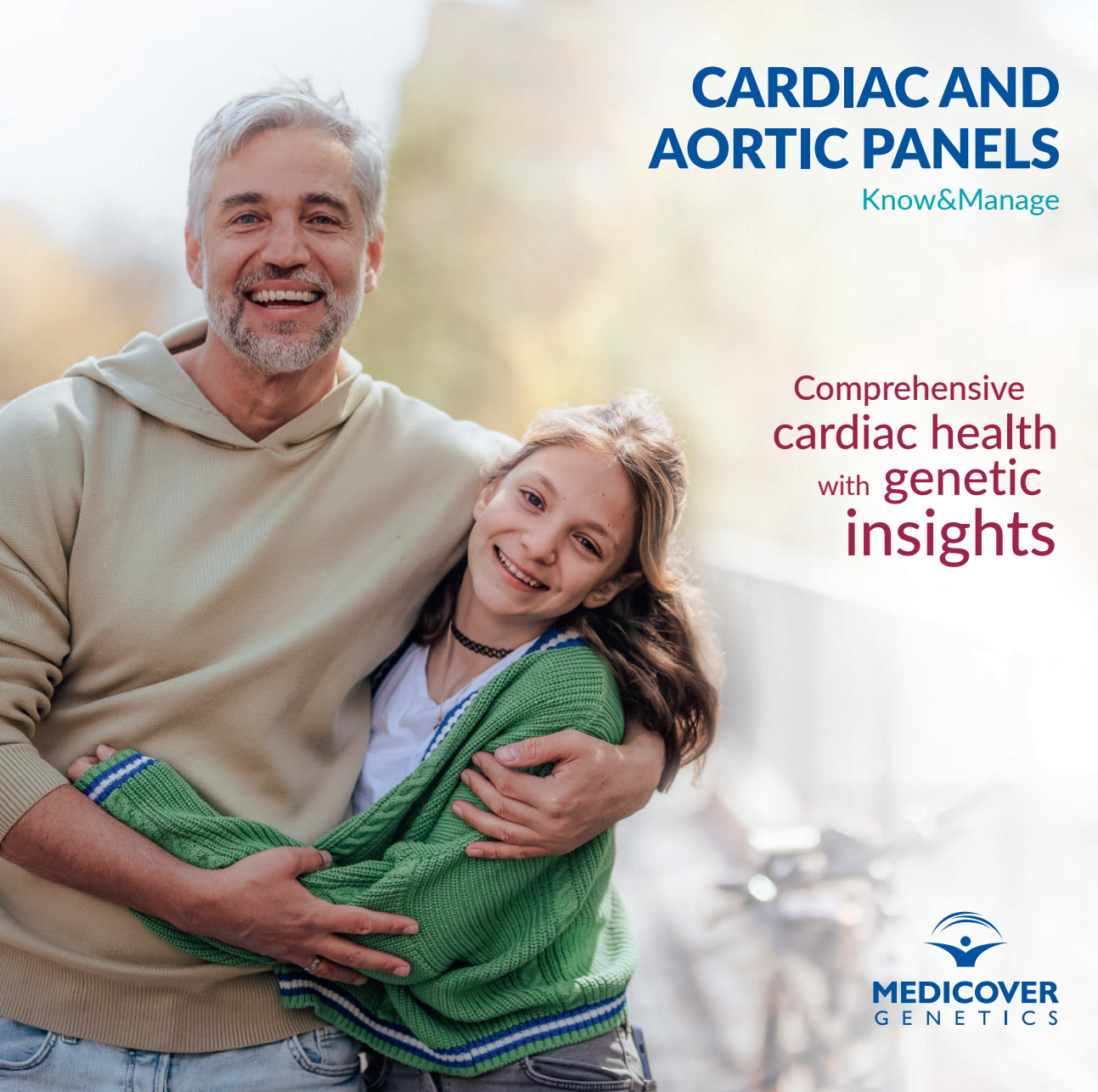


CARDIAC AND AORTIC PANELS

Know&Manage

Comprehensive cardiac health with genetic insights

MEDICOVER GENETICS

WHAT IS **CARDIAC AND AORTIC PANELS Know&Manage**?

KNOW

Hereditary cardiac disorders have a prevalence of 3% in the population. Arrhythmogenic diseases are responsible for most cardiac mortality in the young and congenital heart defects are the most common type of birth defect (1% of all live births). Owing to improved treatment and management options, there are more adults living with congenital heart defects than children. Importantly, deaths from aortic aneurysms may be prevented if individuals at risk are identified and managed.

We offer comprehensive and syndrome-specific panels testing for cardiac and aortic disorders.

MANAGE

Genetic information can improve clinical management by determining the right treatment and follow-up plan. It can be used to determine the prognosis, predict therapy response, and in some cases it can identify gene therapy options.

Knowing about a cardiac or aortic disorder allows you to be proactive about your health with management strategies, such as medication, lifestyle changes or surgery.

WHO COULD **BENEFIT** FROM THIS TEST?

- You have a family history of unexplained cardiac arrest, cardiac death or sudden aortic events
- You have a child born with heart defects
- You have a clinical diagnosis of a heart defect and need a differential diagnosis
- You have a child with developmental delay and it is recommended to test them for heart disorders
- You need to determine the right timing for prophylactic aortic surgery
- You have a hereditary heart disorder and want to estimate the risk of a pregnancy

IMPORTANCE OF GETTING TESTED

If you or a family member has a risk of heart disease, identifying the cause can guide actions to improve the outcome of the disorder. Additionally, family members can be informed and encouraged to also get tested. Our genetic counsellors can provide medical advice.

OUR TESTS

ARRHYTHMOGENIC HEART DISORDERS[1]

Arrhythmia (Channelopathies)[1]

- Atrial fibrillation
- Brugada syndrome
- Catecholaminergic polymorphic ventricular tachycardia
- Early repolarization syndrome
- Long QT syndrome
- Progressive cardiac conduction disorder
- Short QT syndrome
- Sinus node dysfunction

Cardiomyopathies[1]

- Arrhythmogenic cardiomyopathy
- Dilated cardiomyopathy
- Hypertrophic cardiomyopathy
- Noncompaction cardiomyopathy
- Restrictive cardiomyopathy

Syndromic forms of HCM with extracardiac manifestation

- Amyloidosis
- Endocardial fibroelastosis
- Friedreich's ataxia
- Hemochromatosis
- Cardiofaciocutaneous syndrome
- Laing syndrome
- Lysosomal storage disorders (Danon disease, Fabry disease, Gaucher disease, Pompe disease, Mucopolysaccharidoses type I and II)
- Mitochondrial syndromes (Leigh syndrome, MELAS, MERFF)
- Myofibrillar myopathy
- PRKAG2 cardiomyopathy
- RASopathies (Costello syndrome, Noonan syndrome)

CONGENITAL HEART DEFECTS[1]

- Alagille syndrome
- DiGeorge syndrome
- Heterotaxy
- Isolated & syndromic congenital heart defects
- RASopathies
- Williams Beuren syndrome

AORTIC DISORDERS[1]

- Ehlers Danlos syndrome
- Loeys Dietz syndrome
- Marfan syndrome
- Thoracic aortic aneurysm (+/- dissection)

LIPID METABOLISM DISORDERS

- Familial hypercholesterolemia
- Hyperlipidemia / Hyperlipoproteinemia
- Hypolipidemia / Hypolipoproteinemia

PHARMACOGENETICS

- ß-Blocker *(CYP2D6)*
- Clopidogrel *(CYP2C19)*
- Mavacamten *(CYP2C19)*
- Statine *(SLCO1B1)*
- Sartane *(CYP2C9)*

[1] comprehensive panel available

Whole exome sequencing (WES)

WES is recommended in cases of undiagnosed, rare, or atypical presentations of cardiac and aortic conditions. It is particularly useful when symptoms indicate multiple genetic causes or when initial panel results are inconclusive.

Three testing options:

Trio WES *highest diagnostic yield*	Duo WES	Single WES
patient and 2 biological parents	patient and 1 biological parent	patient only

COMMON SYMPTOMS

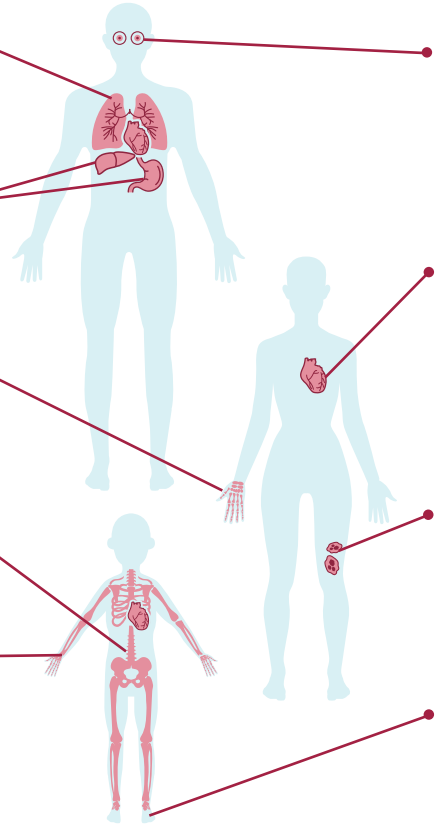
CHEST
Pain (AD, CM)
Pigeon chest (MS)
Shortness of breath (AD, CHD, CM)
Breathing difficulties (CHD)

METABOLISM
Enlarged liver (CHD, CM)
Gastrointestinal problems (CHD)
Indigestion (CHD)

FINGERS
Long slender fingers (AD, CHD, MS, MLS)
Short fingers (CHD)
Abnormal fingernails (CHD)

SPINE
Skeletal anomalies (AD, CHD, EDS, MS)
Growth retardation (AD, CHD)

JOINTS
Hypermobility (AD, EDS, CHD)
Dislocation (EDS, MLS)
Inflammation (EDS, MS)



EYES
Widely spaced (AD, CHD)
Blurred vision (CHD)
Iris abnormalities (AD)
Retinal abnormalities (AD)
Lens luxation (MLS)

CARDIOVASCULAR
Irregular heartbeat (AR, CM)
Sudden cardiac arrest (AR, CM)
Palpitations (AR, CHD, CM, EDS)
Dizziness/fainting (AD, CM)
Difficulty exercising (CHD, CM)
Fatigue (CHD, CM, MS)

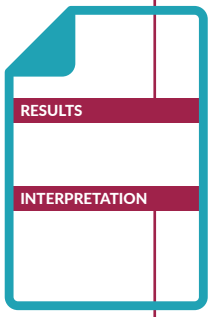
SKIN
Stretchy (EDS)
Easily bruised (AD, CHD, EDS)
Inelastic, wrinkled (AD)
Bluish color (CHD)

FEET
Flat (AD, CHD, MS)

Aortic disorders (AD), Arrhythmia (AR), Congenital heart defects (CHD), Cardiomyopathies (CM), Ehlers Danlos syndrome (EDS), Marfan syndrome (MS), Marfan-like disorders (MLS)
https://rarediseases.info.nih.gov/diseases

WHAT ARE THE POSSIBLE **OUTCOMES** OF THE TEST?

A molecular genetic diagnostic report outlining the results of the sequencing analysis is provided. Changes in DNA sequences (variants) can be detrimental and lead to the development of a cardiac or aortic disorder, including disorders that manifest later in life. We will report on the following types of variants:



PATHOGENIC AND LIKELY PATHOGENIC VARIANTS

The genetic cause of the observed symptoms has been identified and may help determine the right treatment and management plan.

VARIANTS OF UNKNOWN SIGNIFICANCE (VUS)

There was not enough evidence to classify the variant as either pathogenic or benign. Annual variant reclassification and testing family members is recommended.

Please note that we do not report every VUS, only those with a posterior probability to be pathogenic of at least 50% and variants with less than 50% only when the main gene(s) are affected.

It is important to note that a **negative result** does not guarantee the absence of a disorder or that the disorder does not have a genetic cause. Genetic testing is a rapidly advancing field, yet it may still miss certain variants associated with inherited diseases due to methodological limitations an scientific knowledge, as not all relevant genes or genetic changes have been identified.

Summary of the results and recommendations

Interpretation of the molecular genetic results relies on an accurate clinical picture of the patients

MEDICAL **GENETIC COUNSELLING**

Medical genetic counselling is an essential part of a genetic testing journey that we offer before and after testing. Genetic counsellors will obtain a detailed family history, explain the method of testing that will be used, its risks and benefits, the limitations of the diagnosis, and advise you on the consequences of the results including management options and recurrence risk. The goal of counselling is to provide you with a greater understanding of the results and the ability to make more informed choices.

Availability of genetic counselling services may vary by country. Please contact us to check for more information on access in your region.

HOW TO **ORDER**?

Visit a physician or one of our medical geneticists to choose the right test for you

The sample is sent to **Medicover Genetics**

Discuss the medical report with your physician

Arrange sample collection at your nearest blood drawing point (accepted samples: EDTA blood, buccal swab)

Sequencing is performed in our accredited laboratory in Germany

MORE **QUESTIONS**?

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

MEDICOVER GENETICS

Medicover Genetics GmbH
www.medicover-genetics.com

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