MEDICOVER GENETICS

# CARDIAC AND AORTIC PANELS

## Know&Manage

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Patient Information

# WHAT IS CARDIAC AND AORTIC PANELS KNOW&MANAGE

## KNOW

Hereditary cardiac disorders have a prevalence of 3% in the population. Arrythmogenic 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.

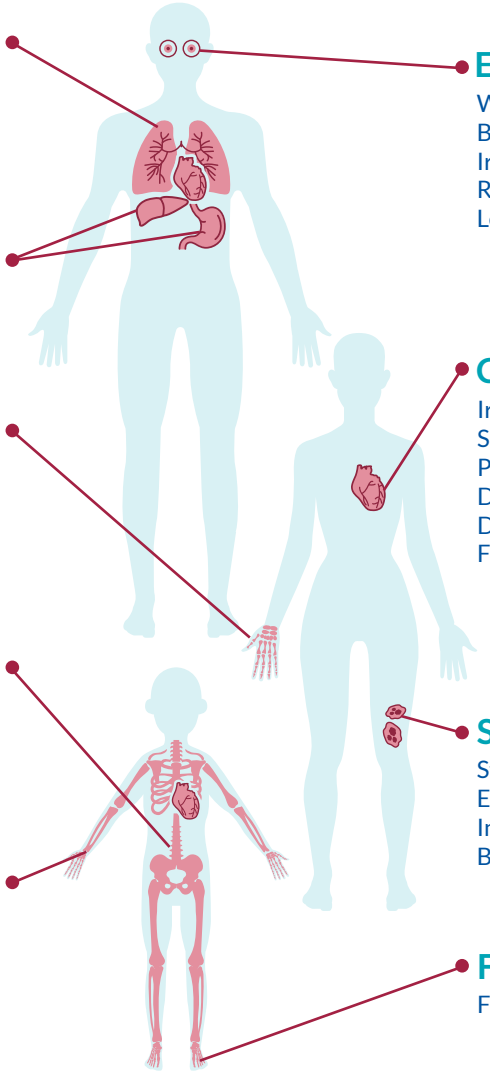
# WHO COULD BENEFIT FROM THIS TEST

- 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 who is recommended to test 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.

# COMMON SYMPTOMS



## CHEST
Pain (AD, CM)
Pigeon chest (MS)
Shortness of breath (AD, CHD, CM)
Breathing difficulty (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), Arrythmias (AR), Congenital Heart Defects (CHD), Cardiomyopathies (CM), Ehlers-Danlos Syndrome (EDS), Marfan Syndrome (MS), Marfan-Like Disorders (MLS)

https://rarediseases.info.nih.gov/diseases

# HOW TO GET THIS TEST ORDERED

**STEP 1**
Visit a physician or one of our medical geneticists to choose the right test for you

**STEP 2**
A sample is collected at your nearest blood drawing point (accepted samples: EDTA blood, buccal swab)

**STEP 3**
Sequencing is performed in our accredited laboratory in Germany

**STEP 4**
A medical report is delivered

**STEP 5**
Genetic counselling by our local medical counsellors is available upon request

Step 3-4 takes 15-25 working days.

Please note that symptoms vary in type and severity between people and that not all symptoms are listed for each disorder. Additionally each patient may have different signs and symptoms, which are assessed by your physician and genetic counsellor.

## 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 asymptomatic disorders that develop later in life. We will report pathogenic and likely pathogenic variants as well as variants of unknown significance.

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

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 may not be able to detect all variants that lead to an inherited disease.

## MEDICAL GENETIC COUNSELLING

We can provide expert medical interpretation of the results for the specialist as well as for the patient. This includes advice on monitoring procedures and clinical interventions, such as medication or surgery.

## WHY US

- A network of laboratories and medical institutions makes us a leader in genetic testing in Germany with foundations dating back to 1998

- A clinical team comprised of scientists, physicians and medical geneticists, several with >20 years of experience in genetic testing, assuring meaningful and comprehensive genetic tests

- Up-to-date diagnostic algorithms

- Expertise in gene variant analysis ensuring "no variant left behind"

- Cutting-edge technology in sequencing and laboratory methods allows for short turnaround times

- Quality assessed by several certifying bodies, including EFI,
DIN EN ISO 15189 accreditation for medical laboratories,
DIN EN ISO/IEC 17025 accreditation for testing and calibration laboratories

- Data privacy is your right and our priority

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Contact info

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