MEDICOVER GENETICS

CARDIAC AND AORTIC PANELS

Know&Manage

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Physician 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

- People with a family history of unexplained cardiac arrest, cardiac death or sudden aortic events
- A child born with heart defects, such as structural abnormalities of the heart muscle
- Patients with a clinical diagnosis of a heart defect who need a differential diagnosis
- A child with neurodevelopmental delay who might have a hereditary heart disorder
- Patients who need to determine the right timing for prophylactic aortic surgery
- Patients with a hereditary heart disorder and want to estimate the risk of a pregnancy

IMPORTANCE OF GETTING TESTED

Genetic testing can identify the cause of a family history of heart disease and may help improve clinical management. Additionally, family members can be informed and encouraged to also get tested. Our genetic counsellors can provide medical advice.

CARDIAC DISORDERS COMMON SYMPTOMS

GASTROINTESTINAL PROBLEMS
HEPATOMEGALY
ANGINA PECTORIS
PECTUS CARINATUM
ARACHNODACTYLY
DYSPNEA ONYCHOSIS
BRACHYDACTYLY CYANOSIS
EXERCISE INTOLERANCE
HYPERELASTIC SKIN
PES PLANUS
ORBITAL HYPERTELORISM
IRIS ABNORMALITIES
CARDIOPULMONARY ARREST
DYSPEPSIA
CLEFT PALATE
ECTOPIA LENTIS
ECCHYMOSIS ELASTOSIS
VENTRICULAR TACHYCARDIA
SYNCOPE ARRYTHMIA
JOINT LUXATION
JOINT HYPERMOBILITY
JOINT INFLAMMATION
FATIGUE SCOLIOSIS
GROWTH RESTRICTION
HYPERPLASIA
SKELETAL DYSPLASIA

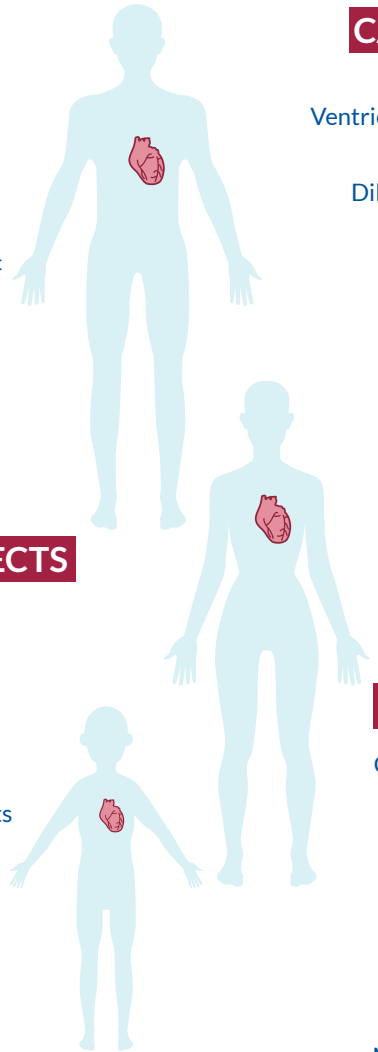
OUR PANELS

ARRYTHMIAS

- 80 genes — Comprehensive Arrythmias & Cardiomyopathies
- 9 genes — Brugada Syndrome
- 8 genes — Catecholaminergic Polymorphic Ventricular Tachycardia
- 6 genes — Short QT Syndrome
- 11 genes — Long QT Syndrome

CONGENITAL HEART DEFECTS

- 91 genes — Comprehensive Congenital Heart Defects
- 2 genes — Alagille Syndrome
- 16 genes — RASopathies With Heart Defects
- 37 genes — Syndromic Congenital Heart Defects
- 22 genes — Isolated Congenital Heart Defects
- 5 genes — Heterotaxy



CARDIOMYOPATHIES

- Arrythmogenic Right Ventricular Cardiomyopathy — 8 genes
- Dilated Cardiomyopathy — 66 genes
- Hypertrophic Cardiomyopathy — 60 genes
- Restrictive Cardiomyopathy — 6 genes
- Left Ventricular/ Noncompaction Cardiomyopathy — 10 genes

AORTIC DISORDERS

- Comprehensive Aortic Disorders — 37 genes
- Ehlers-Danlos Syndrome — 29 genes
- Marfan Syndrome — 3 genes
- Marfan-Like Disorders — 12 genes

HOW TO ORDER THE TEST FOR YOUR PATIENT

STEP 1
Complete Cardiac and Aortic Panels Test Order Form (can be downloaded from our website)

STEP 2
A sample is collected at the 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.

Most genes included in our panels have been selected for highly correlating with the specific listed syndromes. Therefore, many genes are clinically actionable and can guide management measures.

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 is an evolving field and may not detect all variants or there may not currently be enough evidence to classify all variants that lead to an inherited disease.

TECHNICAL DETAILS

DNA is isolated and next generation sequencing is performed on all coding exons and conserved intronic regions. Single base pair changes, small deletions and duplications, and copy number variants (CNV) are identified. Sequencing runs result in a Quality Score of >30 (accuracy >99.9%) in at least 75% of all bases with a coverage of >20-fold. CNV detection sensitivity is 76.99% and precision is 62.59% (with GC limitation between 0.4 and 0.6 per target, sensitivity is 77.04% and precision is 84.10%). Variant classification is performed following ACMG guidelines (Richards et al. 2015, Genet Med 17:405; Kearney et al. 2011, Genet Med 13:680).

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 preventative measures and monitoring procedures where applicable.

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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