Ventrilia®

cardiovascular test

MEDICOVER GENETICS

WHAT IS **Ventrilia**?

Ventrilia is a genetic test that analyzes up to **292 genes** to detect mutations that cause multiple cardiovascular conditions with complex phenotypes. Ventrilia can provide you with comprehensive genetic insight about your patient's cardiovascular health and guide towards an optimal clinical management plan.

CLINICAL UTILITY

Cardiovascular diseases are the leading cause of illness and death worldwide, responsible for **31% of all global deaths**[1]. Previously, assessment of disease risk was based on an individual's lifestyle and their family medical history. The role of genetic testing is now becoming increasingly important by **refining the understanding** behind many hereditary cardiovascular diseases and leading towards a more personalized clinical management based on the identified genetic mutations.

Identification of a disease-causing genetic mutation through Ventrilia can lead to:

- effective clinical management and treatment for many cardiovascular conditions
- improved prognosis and quality of life of your patient
- prevention of potential complications, such as sudden cardiac arrest, through prophylactic measures

RECOMMENDATIONS FROM INTERNATIONAL ORGANISATIONS:

Genetic testing for cardiovascular diseases is recommended by professional societies such as the American College of Cardiology (ACC), American Heart Association (AHA), and the European Society of Cardiology (ESC), based on an individual's family and clinical history. The recommendations include disease-specific considerations, testing for specified genes and applicable clinical care depending on the genetic testing results[2].

WHO IS **Ventrilia** FOR?

- Symptomatic patients with an unidentified cardiovascular genetic disorder
- Presymptomatic patients with family history of inherited cardiovascular disease or sudden cardiac death
- Patients experiencing fainting or unexplained seizures
- Patients with clinical diagnosis of channelopathies (irregular heart rhythm)
- Patients suspected of having a cardiovascular-associated genetic condition due to clinical symptoms (shortness of breath, excessive sweating, heart pain, weakness)
- Asymptomatic patients in high-risk groups (eg. high cholesterol) with non-specific phenotype

1. World Health Organization: WHO. "Cardiovascular Diseases (CVDs)." www.who.int, June 2021, www.who.int/news-room/fact-sheets/detail/cardiovascular-diseases-(cvds).

2. Musunuru, Kiran, et al. "Genetic Testing for Inherited Cardiovascular Diseases: A Scientific Statement From the American Heart Association." Circulation, vol. 13, no. 4, Wiley, July 2020.

WHAT DOES **Ventrilia** TEST FOR?

Ventrilia tests for autosomal recessive, autosomal dominant, and X-linked cardiovascular diseases. Ventrilia is available in 7 disease-category panels and a comprehensive panel that includes all genes tested in the disease-category panels.

48 genes

AORTOPATHY PANEL

Aortopathy refers to a group of diseases that affect the aorta, causing enlargement, dissection or aortic aneurysm.
Aortopathy related diseases include: Marfan Syndrome, Ehlers-Danlos Syndrome, Loeys-Dietz Syndrome, etc.

42 genes

ARRHYTHMIA PANEL

Arrhythmia refers to irregular, too fast, or too slow heartbeat caused by the improper working of the electrical impulses that coordinate the heartbeat.
Arrhythmia related diseases include: Atrial Fibrillation, Brugada Syndrome, Long QT Syndrome, Short QT Syndrome, etc.

98 genes

CARDIOMYOPATHY PANEL

Cardiomyopathy is a group of diseases of the heart muscle (myocardium) which reduce the efficiency of the heart to pump blood.
Cardiomyopathy related diseases include: Dilated Cardiomyopathy, Hypertrophic Cardiomyopathy, etc.

80 genes

CONGENITAL HEART DEFECTS (CHD) PANEL

CHD are present from birth and affect the heart's structure and efficiency to function.
CHD related diseases include: Atrioventricular Septal defect, Ventricular Septal defect, Tetralogy of Fallot, etc.

11 genes

FAMILIAL HYPERCHOLESTEROLEMIA (FH) PANEL

FH is a common inherited genetic disorder that causes high levels of LDL and could lead to heart disease and heart attacks, if untreated.

11 genes

PULMONARY HYPERTENSION (PH) PANEL

PH refers to the high blood pressure in the arteries of the lungs and the right side of the heart.

30 genes

RASOPATHIES PANEL

RASopathies are a group of genetic conditions that affect the RAS-MAPK pathways and lead to developmental syndromes.
RASopathies related diseases include: Cardio-Facio-Cutaneous Syndrome, Neurofibromatosis Type 1, Noonan Syndrome, etc.

292 genes

COMPREHENSIVE PANEL

The comprehensive panel includes all **292 genes** tested in the disease-category panels.

WHY RECOMMEND **Ventrilia** TO MY PATIENTS?

Ventrilia can help you:

IDENTIFY
- complex cardiovascular diseases
- the specific disease by differentiating between diseases with similar phenotype

EVALUATE
- patient's level of risk
- the most beneficial treatment options
- at-risk family members

MANAGE
- decision-making before an invasive treatment
- specific therapies and predict their response

Ventrilia can help you take informed, accurate, and early decisions on the best clinical management for your patient through a simple, non-invasive procedure.

CASE STUDY

5-year-old female whose mother and maternal aunt have prolonged corrected QT interval (QTc). Patient's mother was treated with Implantable Cardioverter Defibrillator (ICD), while her maternal aunt has passed away.

After Ventrilia Cardiovascular testing results

A **heterozygous variant** in the **KCNH2** gene was detected. This is associated with Romano-Ward syndrome, an autosomal dominant disorder which is the most common form of Long QT Syndrome (LQTS). If left untreated, LQTS could lead to arrhythmia and sudden death.

Ventrilia facilitated the patient's early diagnosis, which can initiate the start of appropriate clinical care, leading to improved prognosis. If the condition were to remain undiagnosed, and thus untreated, the prognosis would be poor as up to 1 in 5 **untreated** LQTS patients die within a year of symptoms manifestation.

TECHNOLOGICAL ADVANTAGES

TARGETED TECHNOLOGY

Ventrilia is based on a novel, **Target Capture Enrichment Technology** which utilizes Next Generation Sequencing, and has been thoroughly validated for its accuracy and precision.

NOVEL BIOINFORMATICS

Innovative bioinformatic pipelines analyze the sequencing data produced from each sample, increasing the sensitivity and specificity of Ventrilia.

FULL COVERAGE

Ventrilia screens for **all coding regions*** on the genes tested, and targets adjacent non-coding sequence, increasing the chances of identifying any pathogenic or likely pathogenic mutations:
- single nucleotide variants (SNVs)
- small insertions and deletions (INDELs)
- copy number variants (CNVs)

**Exceptions on regions containing repeats, sequences of high homology (pseudogenes and segmental duplications) or high GC-content.*

BENEFITS OF **Ventrilia**

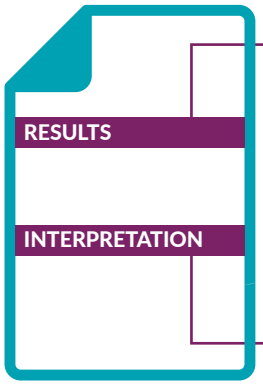
SAFE
Non-invasive sample collection

VALUABLE
Guides informed clinical management

INFORMATIVE
Early identification of asymptomatic patients

THOROUGH
Full exonic coverage*

WHAT WILL THE **REPORT** TELL ME?



CLINICALLY SIGNIFICANT VARIANT DETECTED
A pathogenic or likely pathogenic variant has been identified in a gene associated with a cardiovascular disease.

NO CLINICALLY SIGNIFICANT VARIANT DETECTED
No disease-causing genetic variant has been identified.

VARIANT OF UNCERTAIN SIGNIFICANCE
A genetic change has been detected, but it is currently unknown whether that change is associated with a genetic disorder.

Summary of the results and recommendations

Carrier status will not be reported for recessive conditions.
Variant re-evaluation can be requested by healthcare providers at defined timeframes.

WHAT CAN I DO AFTER **Ventrilia**?

Ventrilia can detect cardiovascular disease-causing mutations in symptomatic, presymptomatic and asymptomatic individuals. This will help you to:

- Provide the most optimal clinical management plan to reduce your patient's risk of sudden cardiovascular events
- Recommend appropriate examinations at key time intervals for high-risk, asymptomatic patients identified with a cardiovascular genetic mutation
- Recommend testing and genetic counseling to family members, if applicable

**Exceptions apply*

HOW TO ADMINISTER **Ventrilia**?

Recommend the ideal **Ventrilia** panel to your patient

Collect a buccal swab from your patient

Send the sample to **Medicover Genetics**

The sample will be analyzed at **Medicover Genetics** laboratories

Results will be sent to you within 2-4 weeks from sample receipt

MORE **QUESTIONS**?

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

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