

WHY RECOMMEND **Ventria** TO MY PATIENTS?

Cardiovascular diseases are the **leading cause** of illness and death worldwide, responsible for 31% of all global deaths*. Previously, assessment of the disease risk was based on the lifestyle of an individual. **The role of genetic testing is now becoming increasingly important.** Genetic testing can detect the underlying condition, provide a prognosis and identify at-risk family members, who might be predisposed to the same cardiovascular disease. Identification of a genetic mutation that causes a disease, can lead to an **improved prognosis** as well as **effective clinical management and treatment** for many cardiovascular conditions†.

Cardiovascular genetic testing is highly recommended by professional societies such as the American College of Cardiology (ACC), American Heart Association (AHA), and the European Society of Cardiology (ESC).

WHAT IS THE ROLE OF CARDIOVASCULAR GENETIC TESTING?



IDENTIFY

- ◆ Genetic mutations involved in cardiovascular conditions
- ◆ Complex cardiovascular diseases
- ◆ The correct disease by differentiating between diseases with similar phenotype



EVALUATE

- ◆ Patient's level of risk
- ◆ Treatment options
- ◆ At-risk family members



MANAGE

- ◆ Clinical care
- ◆ Decision-making before an invasive treatment
- ◆ Specific therapies and predict their response

HOW TO ADMINISTER **Ventria**?



RECOMMEND THE IDEAL PANEL TO YOUR PATIENT



COLLECT A BUCCAL SWAB FROM YOUR PATIENT



SEND THE SAMPLE TO NIPD Genetics



THE SAMPLE WILL BE ANALYZED IN OUR LABORATORY



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:



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NIPD GENETICS

Ventria

cardiovascular test



Genetic insight about
your cardiovascular health

SAFE | SENSITIVE | RELIABLE

* World Health Organization (Cardiovascular Diseases fact sheets) and World Heart Federation (2017)
† Clinical Appropriateness Guidelines - Genetic Testing for Hereditary Cardiac Disease - developed by Informed Medical Decisions, Inc. (2019)

WHAT IS **Ventrilia**?

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

Ventrilia can be used to:



- ◆ Identify the genetic variant in **symptomatic individuals** with a clinically diagnosed cardiovascular condition
- ◆ Identify the genetic variant in **presymptomatic individuals** with a clinically diagnosed cardiovascular condition
- ◆ Identify the genetic variant in **asymptomatic individuals** that belong in high-risk groups

CARDIOVASCULAR DISEASE PANELS

Ventrilia tests for autosomal recessive, autosomal dominant, and X-linked cardiovascular diseases.

AORTOPATHY PANEL **48 genes**

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, Familial Thoracic Aortic Aneurysms and Dissections, Bicuspid Aortic Valve Disease

ARRHYTHMIA PANEL **42 genes**

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, Catecholaminergic Polymorphic Ventricular Tachycardia, Long QT Syndrome, Short QT Syndrome

CARDIOMYOPATHY PANEL **98 genes**

Cardiomyopathy is a group of diseases of the heart muscle (myocardium) which reduces the efficiency of the heart to pump blood. Cardiomyopathy related diseases include: Arrhythmogenic Cardiomyopathy, Arrhythmogenic Right Ventricular Cardiomyopathy, Cardiomyopathy with onset in neonatal period, infancy or childhood, Dilated Cardiomyopathy, Hypertrophic Cardiomyopathy, Left Ventricular Non-Compaction Cardiomyopathy, Restrictive Cardiomyopathy

CONGENITAL HEART DEFECTS (CHD) PANEL **80 genes**

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

FAMILIAL HYPERCHOLESTEROLEMIA (FH) PANEL **11 genes**

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

PULMONARY HYPERTENSION (PH) PANEL **11 genes**

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

RASOPATHIES PANEL **30 genes**

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, Costello Syndrome, Legius Syndrome, Neurofibromatosis Type 1, Noonan Syndrome, Noonan with multiple lentiginos

COMPREHENSIVE PANEL **292 genes**

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



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 aortic defect, or irregular cardiovascular anatomy



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*)



Patients in high-risk groups (*eg. high cholesterol*) with non-specific phenotype

WHAT ARE THE BENEFITS OF **Ventrilia** FOR MY PATIENTS?

- ✓ Faster identification of complex cardiovascular conditions
- ✓ Accurate detection of multiple cardiovascular genetic mutations
- ✓ Early identification in asymptomatic patients
- ✓ Improved prognosis
- ✓ More effective treatment
- ✓ Better clinical care
- ✓ Prevention of potential sudden onset of cardiovascular conditions, including sudden death in all patients

TECHNOLOGY YOU TRUST

Ventrilia uses a **targeted genomic analysis** and a robust, validated multiengine analysis platform to screen for clinically actionable, **highly penetrant** gene mutations with **high sensitivity and specificity**.



TARGETED TECHNOLOGY

Ventrilia is based on a novel, target capture enrichment technology that has been thoroughly validated for its accuracy and precision.



FULL COVERAGE

Ventrilia screens for **all coding regions** on the genes of interest, increasing the chances of identifying any pathogenic or likely pathogenic mutations: single nucleotide variants, small insertions and deletions, and copy number variants.



DNA SAMPLE



TARGETED REGION



SEQUENCING



ANALYSIS



REPORTING OF RESULTS

NOVEL BIOINFORMATICS

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

WHAT WILL THE REPORT TELL ME?

- ◆ Result on mutations tested
- ◆ Thorough interpretation and clinical significance of variants



Carrier status will not be reported for recessive conditions. Re-interpretation of variants of uncertain significance will be performed at routine intervals.

ADVANTAGES OF **Ventrilia** INCLUDE:

- ✓ Assessment of clinically actionable mutations
- ✓ Proven technology
- ✓ Superior targeted genomic analysis
- ✓ Easy to interpret results
- ✓ Competitive turn-around time
- ✓ Informed clinical management
- ✓ Non-Invasive sample collection

HOW DOES **Ventrilia** HELP ME?

Overlapping symptoms make it very **challenging** to clearly identify the underlying cardiovascular condition. There are many types of congenital cardiovascular diseases, ranging from simple to complex defects with severe, life-threatening symptoms.

Ventrilia can detect mutations that cause cardiovascular diseases and help you choose an ideal treatment based on the specific mutation, develop a better clinical management plan and reduce your patient's risk of sudden cardiovascular events such as stroke or heart attack.

For high-risk, asymptomatic individuals with a cardiovascular genetic mutation, you can recommend appropriate examinations at key time intervals. Additionally, you can inform any family members who might also have the same genetic mutation. Early intervention can potentially be lifesaving for symptomatic, presymptomatic and asymptomatic individuals.