Heart Health Panel

What are they testing for?

Gene	Cardiomyopathies	Arrhythmias	Arteriopathies	Familial Hypercholesterolemia
ACTA2			✓	
ACTC1	✓			
APOB*				✓
COL3A1			✓	
DSC2	✓			
DSG2	✓			
DSP	✓			
FBN1			✓	
GLA	✓			
KCNH2*		✓		
KCNQ1*		✓		
LDLR**				✓
LMNA	✓			
MYBPC3	✓			
MYH7	✓			
MYH11			✓	
MYL2	✓			
MYL3	✓			
PCSK9				✓
PKP2	✓			
PRKAG2	✓			
RYR2		✓		
SCN5A		✓		
SMAD3			✓	
TGFBR1*			✓	
TGFBR2			✓	
TMEM43	✓			
TNNI3	✓			
TNNT2	✓			
TPM1	✓			

CARDIOMYOPATHY

An abnormality of the heart muscle making it harder for the heart to pump blood to the body. Inherited cardiomyopathies include: Hypertrophic cardiomyopathy, Dilated cardiomyopathy, and Arrhythmogenic right ventricular cardiomyopathy

WHAT HAPPENS WHEN YOU HAVE A CARDIOMYOPATHY?

- **Hypertrophic cardiomyopathy (HCM)** is associated with an abnormal thickening of the heart muscle, which can make it hard for the heart to pump blood.
- **Dilated cardiomyopathy (DCM)** is associated with an enlargement of the heart, which can make it hard for the heart to pump blood.
- **Arrhythmogenic cardiomyopathy (AC)** is associated with a replacement of heart tissue with fat and/or fibrous tissue, which can make it hard for the heart to pump blood

ARRHYTHMIA

An abnormality of the heart rhythm causing the heart to beat too fast, too slow or irregularly. Hereditary Arrhythmias can affect as many as 1 in 5000 people

WHAT HAPPENS WHEN YOU HAVE AN ARRHYTHMIA?

- **Long QT Syndrome (LQTS)** is associated torsade de pointes.

- Symptoms of LQTS may include fainting, or sudden cardiac arrest
- **Short QT Syndrome (SQTS)** can cause one of two rhythm problems:
  - Atrial fibrillation: this can cause shortness of breath, dizziness, chest tightness, fatigue, and fainting.
  - Ventricular tachycardia or fibrillation: This can cause fainting and sudden cardiac death
- **Catecholaminergic polymorphic ventricular tachycardia (CPVT)** can cause dangerously fast heartbeat in the ventricles.
  - Sudden cardiac death can occur, even in individuals who have no other symptoms.
- **Brugada syndrome** is associated with a dangerously fast and chaotic heartbeat (VFib).
  - Symptoms may include fainting or sudden cardiac arrest.

ARTERIOPATHY

Problems with the structure of the arteries in many parts of the body, including the heart. Aortic dissection is one of the most common causes of death in arteriopathies.

WHAT HAPPENS WHEN YOU HAVE AN ARTERIOPATHY?

- **Familial thoracic aortic aneurysm and dissection (FTAAD):** Individuals with FTAAD commonly have problems with the thoracic aorta (dilation, aneurysm, and dissection).
- **Vascular Ehlers-Danlos syndrome (vEDS):** problems with connective tissue in many parts of the body, which can causing weakness of the blood vessel walls and other organs, fragile skin, and easy bruising and bleeding, dissections commonly occur in the aorta.
- **Marfan syndrome:** problems with connective tissue in many parts of the body, causing weakness of the blood vessel walls, ectopia lentis, and increased flexibility in the joints. Problems with the heart and the surrounding blood vessels, especially the aorta (dilation, aneurysm, and dissection) are common.
- **Loeys-Dietz syndrome (LDS):** problems connective tissue in many parts of the body, which can cause weakness of the blood vessel walls, skeletal problems, and abnormal bruising and scarring of the skin. Aneurysms or dissections in arteries throughout the body and can have arterial tortuosity.

FAMILIAL HYPERCHOLESTEROLEMIA (FH)

A hereditary disorder that causes very high cholesterol levels from an early age
About 1 in 50 people with high cholesterol are born with Familial Hypercholesterolemia (FH)

WHAT HAPPENS WHEN YOU HAVE FH?

- Your liver is unable to remove enough LDL (bad cholesterol) from your blood
- This means your LDL level remains high, despite positive lifestyle choices
- You are 22x more likely to develop coronary heart disease than are those with normal cholesterol and no FH

Knowledge of inherited high cholesterol can also lead to improved compliance and reduced risk of heart disease

NEGATIVE RESULTS

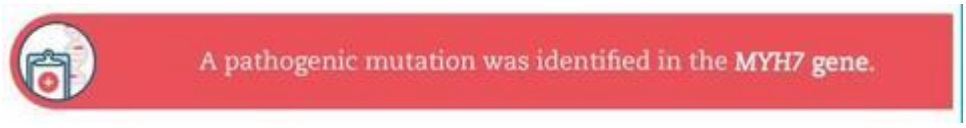
No mutations associated with an increased risk of hereditary arrhythmia, arteriopathy, cardiomyopathy, or familial hypercholesterolemia were identified in any of the 30 genes analysed.

It's still important to follow a prevention plan - While negative results can be reassuring, it's still important to follow the screening and prevention plan recommended by your healthcare provider.

When you receive your results,

1. What if I have a negative result? - If test is negative, this does not mean there is no risk as there may be other complications that this test does not cover. We recommend you always contact your GP if you have any questions or concerns.

POSITIVE RESULTS



A mutation that causes an inherited heart condition was identified in the genes analysed.

Taking preventive action - It's important to discuss results with your healthcare provider to design a personalised screening and prevention plan to manage the risk.