

# Cardiovascular Pharmacogenetic Report

## Genetic Risk Assessment: 5 Cardiac Conditions

### ✓ Type III Hyperlipoproteinemia

#### Not Associated with Type III Hyperlipoproteinemia

The patient is negative for both the APOE c.388 T>C (Cys130Arg) and c.526 C>T (Arg176Cys) mutations. The patient's genotype is wild-type, which is the most common genotype in the general population (frequency: >60%).

A patient with wild-type genotype does not have a defect in the apolipoprotein E (APOE), which is an integral structure of lipoprotein particles that have critical roles in blood lipid metabolism and transport. The APOE ε3/ε3 genotype is not associated with increased risk of cardiovascular disease. No action is needed when a patient is normolipidemic.

### ✓ Platelet Hyperactivity

#### Normal Response to Aspirin

The patient is negative for the ITGB3 176T>C (Leu59Pro) mutation. The genotype for the integrin β3 gene is wild-type, which is the most common genotype in the general population.

The wild-type genotype results confers a "normal" platelet reactivity, and is not associated with a resistance to the antithrombotic effects of aspirin. However, because the variability in response to antiplatelet drugs is multifactorial and not caused by single gene mutations, testing for the ITGB3 mutation alone should not be used as a diagnostic tool.

### ✓ Hyperlipidemia/Atherosclerotic Cardiovascular Disease

#### No increased risk of cardiovascular disease

The patient is a non carrier of the risk alleles in LPA gene for both the variants (rs3798220 and rs10455872).

The patient's genotype is associated with normal lipoprotein levels. The patient has no increased risk of atherosclerosis and cardiovascular disease as compared to the general population unless other risk factors are present.

No action is needed for this patient unless other genetic and non genetic risk factors (e.g. high blood pressure, smoking, diabetes, obesity, high blood cholesterol and excessive alcohol use) are present.

### ✓ Thrombophilia

#### Normal Risk of Thrombosis

The patient does not carry the F5 c.1601G>A variant (also known as Factor V Leiden) or the F2 c.\*97G>A variant (also known as Factor II 20210G>A).

The patient's risk of thrombosis is not increased (average risk of clotting is about 1 in 1000 for anyone in a year). However, because this test cannot find all of the inherited reasons for abnormal clotting, other factors may affect this risk assessment.

Assess thrombotic risk based on other genetic and/or circumstantial risk factors such as smoking, obesity, malignancy, prolonged immobilization or surgery.

**Estrogen-containing contraceptive and hormone replacement therapy:** unless other genetic and/or circumstantial risk factors are present, consider standard prescribing and monitoring practices.

### ✓ Hyperhomocysteinemia - Thrombosis

#### No Increased Risk of Hyperhomocysteinemia

The patient carries one copy of MTHFR c.1286A>C variant (heterozygous). MTHFR enzyme activity is reduced (80% of normal activity).

The patient's slightly reduced MTHFR activity is not a risk factor for hyperhomocysteinemia. Unless other risk factors are present, the patient is not expected to have an increased risk for venous thromboembolism (VTE).

The patient's MTHFR activity is slightly reduced.