HEART & GENETICS

Cholesterol Genetic Screen

Familial hypercholesterolemia (FH) is a hereditary genetic condition whereby the body is unable to remove harmful cholesterol from the bloodstream. In such a case, the bad cholesterol LDL is extremely high and this causes atherosclerotic plaque formation in the arteries thereby increasing risk for heart attacks and strokes.

Abnormally functioning LDL-receptors cause deposition of cholesterol in different parts of the body eg xanthelasma (skin), xanthomas (tendons), and coronary arteries (atherosclerosis).

When to Suspect FH

	LDL CHOLESTEROL
People 20 years of age or older	≥190 mg/dL
People under 20 years of age	≥160 mg/dL

Diagnostic clinical criteria have been published for FH and are based on extreme hypercholesterolemia, premature coronary artery disease (CAD), physical examination findings (e.g. xanthoma or corneal arcus), family history of hypercholesterolemia or CAD, and presence of a disease-causing mutation in an FH-related gene.

Benefits of Genetic testing for FH

-Confirm a diagnosis, particularly when clinical criteria are unclear or borderline in an individual
-Tailor medical treatment
-Clarify risks to family members

How is genetic testing done?

The test is done using a blood sample. The sample will be kept in a special kit, then sent to an approved genetic testing lab in US whereby the genes are analysed for mutations.

Genetic Tests Available	**Price Inclusive of Consultation & GST**
Cholesterol Genetic Panel (4 genes) *APOB, LDLR, PCSK9*, and *LDLRAP1*	$5400

Interpretations of genetic test results

The results will be ready in 3 weeks. There are 3 possible results: positive, negative, variant of unknown significance (inconclusive).

Positive test result = Mutation found in a gene that causes Familial Hypercholesterolemia. Medications and regular cardiac followup is recommended.

Negative test result = No mutations are detected.

Variant of Unknown Significance / Inconclusive test result = A genetic change is found, but it is not yet known if associated with FH.