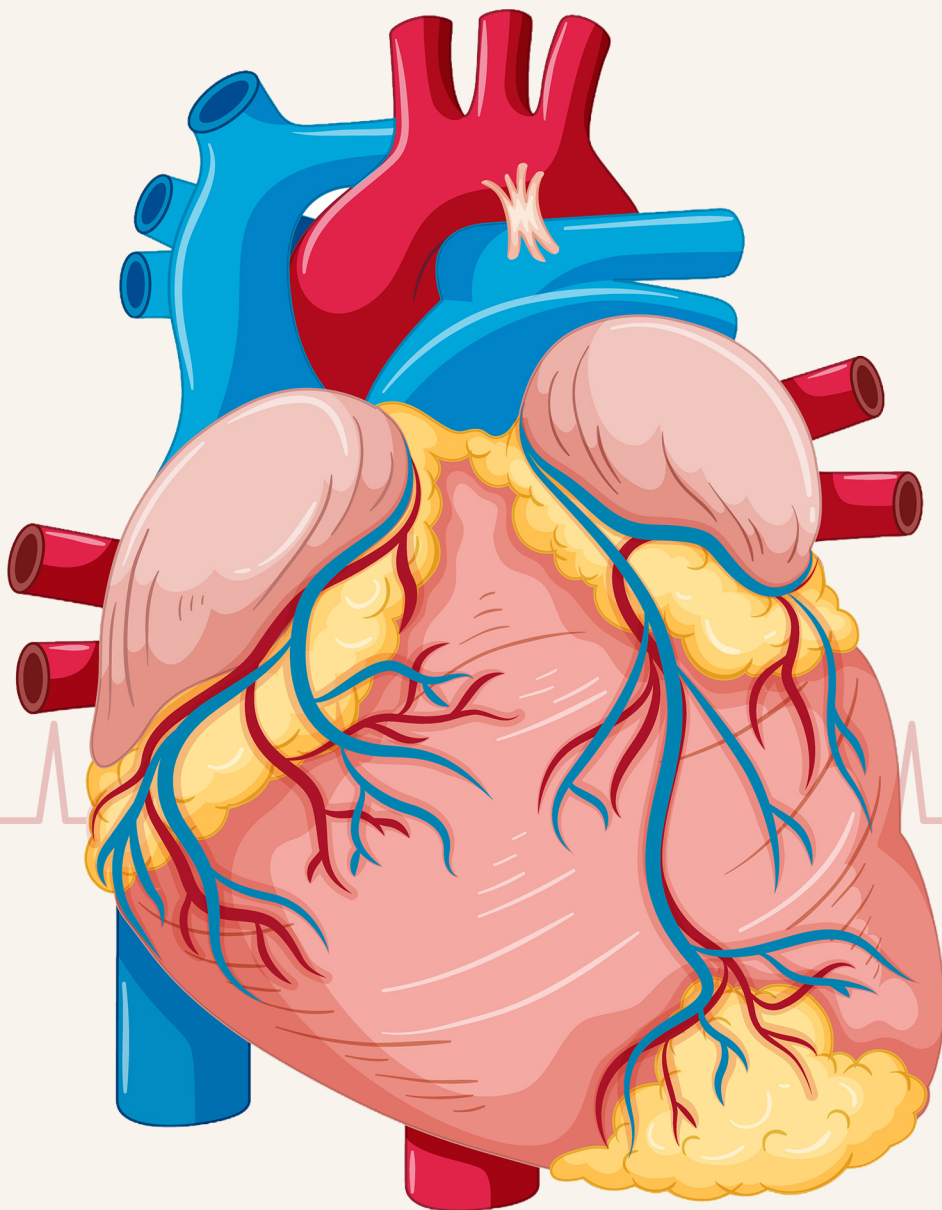




Comprehensive Cardio Genomics





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Our cardiovascular genetic test identifies inherited variations affecting heart structure, rhythm, and function, enabling early detection and personalized management. In India, congenital heart disease affects **~8 per 1,000* live births**, highlighting the importance of early genetic evaluation in at risk families.

*PMID: 27625516

Familial hypercholesterolemia (genetic high cholesterol) affects
~1 in 250 Indians (~5 million).

PMID: 38599725

List of conditions covered in WES



Cardiomyopathies

Structural and functional disorders of the myocardium associated with heart failure, and sudden cardiac death.

Conditions include: HCM | ARVC | DCM | LVNC | RCM etc.

Genes covered: MYBPC3, TNNT2, TNNI3, LMNA, TTN, MYH7, SCN5A, PKP2, DSP, DSG2, DSC2, and many more.



Inherited Arrhythmia Syndromes

Channelopathies caused by pathogenic variants affecting cardiac conduction, increasing susceptibility to life threatening arrhythmias.

Conditions include: Brugada Syndrome | LQTS | SQTS | CCD | Timothy Syndrome etc.

Genes covered: KCNQ1, KCNH2, SCN5A, RYR2, CASQ2 and many more.



Genetic Dyslipidemias

Genetic disorders of lipid metabolism that increase the risk of early atherosclerosis and premature coronary artery disease.

Conditions include: Familial Hypercholesterolemia | Dysbetalipoproteinemia | Familial Chylomicronemia | Sitosterolemia etc.

Genes covered: LDLR, APOB, PCSK9 and many more.



Heritable Aortopathies

Genetic conditions involving progressive dilation or structural weakness of the aorta, often asymptomatic until advanced.

Conditions include: FTAAD | Marfan Syndrome | Loeys-Dietz Syndrome | Ehlers-Danlos Syndrome etc.

Genes covered: FBN1, TGFB1, TGFB2, SMAD3, COL3A1 and many more.





Other Inherited Cardiovascular Conditions

Systemic genetic disorders with significant cardiovascular involvement.

Conditions include: Pulmonary Arterial Hypertension | Hereditary Hemorrhagic Telangiectasia | RASopathies | Hereditary Hemochromatosis | Noonan Syndrome | etc.

Genes covered: MPR2, ACVRL1, ENG, SMAD4, RASA1, PTPN11, SOS1, RAF1, KRAS, RIT1, TFR2, HFE, SLC40A1 and many more.

List of conditions covered in CMA or FISH



Congenital Heart Defects with Genetic Basis

These are structural abnormalities of the heart present at birth caused by inherited or de novo genetic variations affecting cardiac development.

Conditions include: Familial Atrial Septal Defect (ASD) | DiGeorge Syndrome (22q11.2 deletion) | Williams Syndrome

Genes covered: NKX2.5, GATA4, TBX1 (within deletion), ELN (elastin deletion) and many more.

Who Should Consider Cardiac Genetic Testing?

- Individuals with a personal history of early-onset heart disease.
- Unexplained cardiomyopathy, arrhythmia, or sudden cardiac arrest.
- Family history of sudden cardiac death (especially <50 years).
- First degree relatives of individuals diagnosed with a genetic heart condition.
- Individuals with persistently high cholesterol despite lifestyle modification.
- Patients with congenital heart defects.

When Should Testing Be Done?

- After a confirmed or suspected diagnosis of an inherited cardiac disorder.
- When there is a strong family history of heart disease or sudden death.
- Before initiating long-term management in genetically suspected conditions.
- During family screening once a pathogenic variant is identified.
- Early in life for at-risk individuals to enable preventive strategies.

Test Specifications



| TESTS | Platform | Variant Type | Sensitivity & Specificity | Sample Type | Turn Around Time |
|-------|------------------------------------|-----------------------------|---------------------------|-----------------------|------------------|
| WES | Illumina Novaseq 6000 | SNVs, CNVs, MtDNA Mutations | 99 - 100% | Blood in EDTA tube | 4 weeks |
| CMA | Illumina Infinium GSA -24 v3.0 | Copy Number Variants | 99 - 100% | Blood in EDTA tube | 3-4 weeks |
| FISH | Fluorescence In Situ Hybridization | Chromosomal Abberations | 100% | Blood in Heparin tube | 1 week |

Other Services



Phramaco Genomics

Genetics-guided drug selection



Preventive Genomics

Predicting health risks before symptoms appear



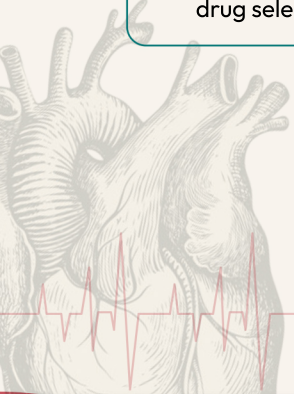
Reproductive Genomics

Pre-conception to Post-natal genetic care



Gutmicrobiome Test

Understanding gut health and balance



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SCAN TO KNOW MORE

