

Genome-wide Sequencing Tests

GENOME-WIDE SEQUENCING TESTS	GENES ANALYZED	SAMPLE TYPE	TECHNOLOGY	TAT
NIPT Test- Ion Torrent	NA	2-3 ml EDTA blood	NGS	4-5 weeks
NIPT Test- Illumina	NA	2-3 ml EDTA blood	NGS	4-5 weeks
Clinical exome sequencing	NA	2-3 ml EDTA blood	NGS	4-5 weeks
Whole exome sequencing	NA	2-3 ml EDTA blood	NGS	4-5 weeks
Mitochondrial Sequencing	NA	2-3 ml EDTA blood	NGS	4-5 weeks
Epilepsy Panel	NA	2-3 ml EDTA blood	NGS	4-5 weeks
BRACA1 & BRACA 2	NA	2-3 ml EDTA blood	NGS	4-5 weeks
Hemophilia Panel	NA	2-3 ml EDTA blood	NGS	4-5 weeks
Skeletal Dysphasia Panel	NA	2-3 ml EDTA blood	NGS	4-5 weeks
CF Panel	NA	2-3 ml EDTA blood	NGS	4-5 weeks
Achondroplasia (FGFR3 Full gene sequencing)	NA	2-3 ml EDTA blood	NGS	4-5 weeks
TSC1 & TSC2 Gene Analysis	NA	2-3 ml EDTA blood	NGS	4-5 weeks
Couple Carrier Genetic testing (CGT)	NA	2-3 ml EDTA blood from both	NGS	4-5 weeks

GENE PANELS for specific diseases (Cardiac/ Cancer/ Nephrology/ Neurology/ Endocrinology/ Gastroenterology)				
TEST NAME	GENES ANALYSED	REQUIRED SAMPLE	TECHNOLOGY USED	TAT
Clinical Exome for Cardiac Diseases	NA	2-3 ml EDTA blood	NGS	4-5 weeks
Organ specific cancer (oncology) panel	NA	2-3 ml EDTA blood	NGS	4-5 weeks
Nephrology panel	NA	2-3 ml EDTA blood	NGS	4-5 weeks
Neurology panel	NA	2-3 ml EDTA blood	NGS	4-5 weeks
Endocrinology panel	NA	2-3 ml EDTA blood	NGS	4-5 weeks
Gastroenterology & hepatology panel	NA	2-3 ml EDTA blood	NGS	4-5 weeks
Liver transplant & hepatobiliary panel	NA	2-3 ml EDTA blood	NGS	4-5 weeks
Thrombophilia panel	(Factor V Leiden, G1691A/R506Q, Factor V R2, H1299R, Prothrombin/Factor II, G20210A, 5,10-Methylenetetrahydrofolate Reductase (MTHFR), C677T, MTHFR, A1298C)	2-3 ml EDTA blood	Microarray	2-3 weeks
Complete Pharmacogenetic panel	100+ markers	2-3 ml EDTA blood	NGS	4- 5 weeks