MEDGENOME

CAP ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS

Clinical Whole Genome Sequencing
(SNVs, InDels, Repeats and CNVs)

+

Structural Variants
(Optical Genome Mapping)

Decode. Diagnose. Discover.

Key Features

- Single nucleotide variants and small insertions and deletions are detected with a high accuracy
- Uniform coverage of the whole genome allows better identification of Copy Number Variants (CNVs)
- End-to-end validation pipeline based on NA12878 reference sample, sensitivity with ~99% for SNPs and ~97% for InDels
- Detection of disease causing variants in both coding and non-coding genes of the genome
- Comprehensive evaluation of mitochondrial genome variants
- Clear detailed reports that support informed decision-making
- Unbiased genome-wide detection structural rearrangements (SVs) such as inversions and balanced translocations
- Detection of variants that could have been missed by targeted sequencing and microarray techniques
- Possibility to revisit in the later years for new and evolving clinical presentations

Test Details

Test Code	Test Name	TAT
MGM3294	ExomeMAX Reflex Whole Genome Sequencing	40 Working days
MGM275	Whole Genome Sequencing (30x) [SNVs, InDels, Repeats and CNVs]	35 Working days
MGM3296	Whole Genome Sequencing (30x) [SNVs, InDels, Repeats and CNVs] Reflex SVs by OGM	35 Working days
MGM2844	KaryoSeq (Low Pass Whole Genome Sequencing)	14 Working days

Sample Type: Peripheral Blood DNA, Direct DNA, Amniotic Fluid, Chorionic Villus Biopsy, Product of Conception/DNA

Why MedGenome

- Unmatched experience of 3,50,000+ genomes/ exomes
- South Asia's largest database with 50,00,000+ variants
- Recommended by 4,000+ hospitals & trusted by 10,000+ clinicians
- Consistent proficiency testing through "External Quality Monitoring Programs"
- Two levels of analysis and review by molecular and clinical geneticists

FREE pre & post test genetic counselling with our expert and certified genetic counsellors

Talk to the Experts

1800 103 3691 diagnostics@medgenome.com www.medgenome.com

Follow us on LinkedIn