

SL.NO	TEST CODE	TEST NAME	TECHNIQUE USED	SAMPLE TYPE	PRICE (In rupees)	TAT
<u>MOLECULAR TESTING</u>						
1	MD 001	SRY DELETION ANALYSIS	PCR/AGE	3mL EDTA blood	1400	1 Week
2	MD 002	Y-CHROMOSOME MICRODELETION ASSAY	PCR/AGE	3mL EDTA blood	4600	1 Week

<u>HAEMOLYTIC ANAEMIAS</u>						
3	MD 003	BETA-THALASSEMIA-619 bps Deletion Analysis	PCR/AGE	3mL EDTA blood	2000	1 Week
4	MD 004	BETA-THALASSEMIA-Any single mutation (Specify mutation)	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
5	MD 005	BETA-THALASSEMIA-Common Mutations Panel	Sanger Sequencing	3mL EDTA blood	3000	2 Weeks
6	MD 006	BETA-THALASSEMIA-Complete (<i>HBB</i>) Gene Sequencing	Sanger Sequencing	3mL EDTA blood	4000	2 Weeks
7	MD 007	HAEMOGLOBIN-D DISEASE-Mutation Analysis	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
8	MD 008	HAEMOGLOBIN-E DISEASE-Mutation Analysis	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
9	MD 009	SICKLE CELL ANAEMIA-Mutation Analysis	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
10	MD 010	GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY- <i>G6PD</i> Mutation Analysis (Kerala-Kalyan, Mediterranean & Orissa Variants)	Sanger Sequencing	3mL EDTA blood	3500	2 Weeks

<u>NEUROMUSCULAR DISEASES</u>						
11	MD 011	BECKER MUSCULAR DYSTROPHY (BMD)-Mutation Analysis /Carrier Screening	MLPA	3mL EDTA blood	4000	2 Weeks
12	MD 012	DUCHENNE MUSCULAR DYSTROPHY (DMD)-Mutation Analysis /Carrier Screening	MLPA	3mL EDTA blood	4000	2 Weeks
13	OGM 002	FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY TYPE-1 (FSHD1)- D4Z4- ARRAY CONTRACTION TESTING	Optical Genome Mapping	3mL EDTA blood	70000	4 Weeks
14	MD 013	HEREDITARY SENSORY AND MOTOR NEUROPATHY (HSMN/CMT1A)- PMP22 Mutation Analysis	MLPA	3mL EDTA blood	4000	2 Weeks
15	MD 014	LIMB GIRDLE MUSCULAR DYSTROPHY TYPE 2A (LGMD2A) - <i>CAPN3</i> -Mutation Analysis [2051-1G>T, 2338G>C]	Sanger Sequencing	3mL EDTA blood	2500	2 Weeks
16	MD 015	MYOTONIC DYSTROPHY TYPE 1 (MD-1) -Mutation Analysis	TP-PCR	3mL EDTA blood	2500	2 Weeks

17	MD 016	OCULOPHARYNGEAL MUSCULAR DYSTROPHY (OPMD)-Mutation Analysis	Sanger Sequencing	3mL EDTA blood	3000	2 Weeks
18	MD 017	SPINAL AND BULBAR MUSCULAR ATROPHY (SBMA)-Mutation Analysis	Fragment Analysis	3mL EDTA blood	2500	2 Weeks
19	MD 018	SPINAL MUSCULAR ATROPHY (SMA)- Mutation Analysis /Carrier Screening	MLPA	3mL EDTA blood	3300	2 Weeks
<u>HEREDITARY ATAXIAS AND OTHER NEURODEGENERATIVE DISORDERS</u>						
20	MD 019	DENTATORUBROPALLIDOLUYSIAN ATROPHY (DRPLA) -Mutation Analysis	Fragment Analysis	3mL EDTA blood	2500	2 Weeks
21	MD 020	FRIEDREICH ATAXIA (FA)-Mutation Analysis	TP-PCR	3mL EDTA blood	2500	2 Weeks
22	MD 021	HUNTINGTON DISEASE (HD)-Mutation Analysis	Fragment Analysis	3mL EDTA blood	2500	2 Weeks
23	MD 022	SPINOCEREBELLAR ATAXIA (SCA) -any one type (Specify type)	Fragment Analysis	3mL EDTA blood	2500	2 Weeks
24	MD 023	SPINOCEREBELLAR ATAXIA (SCA) TYPES-1,2,3-Mutation Analysis	Fragment Analysis	3mL EDTA blood	5500	2 Weeks
25	MD 024	SPINOCEREBELLAR ATAXIA (SCA) TYPES-1,2,3,6-Mutation Analysis	Fragment Analysis	3mL EDTA blood	7500	2 Weeks
26	MD 025	SPINOCEREBELLAR ATAXIA (SCA) TYPES-7,17-Mutation Analysis	Fragment Analysis	3mL EDTA blood	5000	2 Weeks
27	MD 026	SPINOCEREBELLAR ATAXIA (SCA) TYPES-1,2,3,6,12-Mutation Analysis	Fragment Analysis	3mL EDTA blood	9500	3 Weeks
28	MD 027	SPINOCEREBELLAR ATAXIA TYPES (SCA) -1,2,3,6,7,12,17-Mutation Analysis	Fragment Analysis	3mL EDTA blood	13500	3 Weeks
<u>DEVELOPMENTAL DELAY / DYSMORPHISM & INTELLECTUAL DISABILITY SYNDROMES</u>						
29	MD 028	FRAGILE X SYNDROME (FRX/FXS)- Mutation Analysis/Carrier Screening	TP-PCR	3mL EDTA blood	6000	2 Weeks
30	MD 029	MICRODELETIONS/MICRODUPLICATIONS SYNDROMES-Mutation Analysis [1p36 DELETION SYNDROME, 2p16.1-P15 MICRODELETION SYNDROME, 2q23.1 MICRODELETION/MICRODUPLICATION SYNDROME, GLASS SYNDROME, 3q29 MICRODELETION/MICRO DUPLICATION SYNDROME, WOLF-HIRSCHHORN SYNDROME, CRI-DU-CHAT SYNDROME, SOTOS SYNDROME, WILLIAMSBEUREN SYNDROME, WILLIAMS-BEUREN DUPLICATION SYNDROME, LANGER-GIEDION SYNDROME, 9q22.3 MICRODELETION SYNDROME, DI-GEORGE SYNDROME-2, PRADER-WILLI SYNDROME, ANGLEMAN SYNDROME, WITTEVEEN-KOLK SYNDROME, RUBINSTEIN-TAYBI SYNDROME, MILLER-DIEKER SYNDROME/LISSENCEPHALY-1, SMITH-MAGENIS SYNDROME (POTOCKI-LUPSKI SYNDROME), NF1 MICRODELETION SYNDROME, KOOLEN-DE VRIES SYNDROME, 17q21.31 MICRODUPLICATION SYNDROME, DI-GEORGE SYNDROME, 22q11.2 MICRODUPLICATION SYNDROME, DISTAL 22q11.2 DELETION SYNDROME, PHELANMCDERMID SYNDROME, RETT SYNDROME(MECP2 DUPLICATION SYNDROME)]	MLPA	3mL EDTA blood	4000	2 Weeks
31	MD 030	ANGELMAN / PRADER-WILLI SYNDROME (AS/PWS)	MS-MLPA	3mL EDTA blood	7000	2 Weeks
32	MD 031	BECKWITH-WIEDEMANN/SILVER-RUSSELL SYNDROME (BWS/SRS)	MS-MLPA	3mL EDTA blood	7000	2 Weeks

<u>CYSTIC FIBROSIS/HEREDITARY PANCREATITIS PANEL</u>						
33	MD 032	HEREDITARY PANCREATITIS PANEL [<i>SPINK1</i> (N34S); <i>PRSS1</i> (N29I, R122H)]	Sanger Sequencing	3mL EDTA blood	3000	2 Weeks
34	MD 033	<i>CFTR</i> -Any single mutation (Specify mutation)	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
35	MD 034	<i>CFTR</i> -4 Mutation Panel [DF508, G542X, G551D, R553X]	Sanger Sequencing	3mL EDTA blood	2800	2 Weeks
36	MD 035	<i>CFTR</i> -5T/7T/9T Polymorphism Screening	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
37	MD 036	<i>CFTR</i> -36 Mutation Panel	Allele-Specific PCR/Fragment Analysis	3mL EDTA blood	7500	2 Weeks
38	MD 037	<i>SPINK1</i> - Mutation analysis (N34S)	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
<u>MITOCHONDRIOPATHIES</u>						
39	MD 038	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)- Any single mutation (Specify mutation)	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
40	MD 039	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)- Mutation Analysis (G3460A, G11778A, T14484C)	Sanger Sequencing	3mL EDTA blood	3500	2 Weeks
41	MD 040	LEIGH SYNDROME-Any single mutation (Specify mutation)	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
42	MD 041	LEIGH SYNDROME-Mutation Analysis (T12706C, A13084T, G13513A)	Sanger Sequencing	3mL EDTA blood	3500	2 Weeks
43	MD 042	MITOCHONDRIAL ENCEPHALOMYOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES (MELAS)-Mutation Analysis (A3243G)	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
44	MD 043	MITOCHONDRIAL GENOME SEQUENCING	Next Generation Sequencing	3mL EDTA blood	7500	6 Weeks
<u>COAGULOPATHIES</u>						
45	MD 044	<i>FACTOR II</i> (PROTHROMBIN)- Mutation Analysis (G20210A)	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
46	MD 045	<i>FACTOR V</i> LEIDEN-Mutation Analysis (G1601A)	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
47	MD 046	HAEMOPHILIA A- <i>Factor VIII</i> -Intron 1 inversion analysis	PCR/AGE	3mL EDTA blood	2000	2 Weeks
48	MD 047	HAEMOPHILIA A- <i>Factor VIII</i> -Intron 22 inversion analysis	PCR/AGE	3mL EDTA blood	2000	2 Weeks
49	MD 048	HAEMOPHILIA A- <i>Factor VIII</i> -Introns 1 and 22 inversion analysis	PCR/AGE	3mL EDTA blood	4000	2 Weeks
<u>HEREDITARY METABOLIC DISORDERS</u>						
50	MD 049	BIOTINIDASE DEFICIENCY-Any single mutation (Specify mutation)	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks

51	MD 050	BIOTINIDASE DEFICIENCY- <i>BTD</i> Mutation analysis	Sanger Sequencing	3mL EDTA blood	5000	2 Weeks
52	MD 051	HEREDITARY HAEMOCHROMATOSIS (HH)- Mutation Analysis (H63D, C282Y)	Sanger Sequencing	3mL EDTA blood	2500	2 Weeks
53	MD 052	<i>METHYLENETETRAHYDROFOLATE REDUCTASE (MTHFR)</i> - Mutation Analysis (A1298C, C677T)	Sanger Sequencing	3mL EDTA blood	2500	2 Weeks

MISCELLANEOUS

54	MD 053	ACHONDROPLASIA- <i>FGFR3</i> Mutation Analysis [G1138A, G1138C]	Sanger Sequencing	3mL EDTA blood	2500	2 Weeks
55	MD 054	CADASIL- <i>NOTCH3</i> Mutation Analysis [C421T]	Sanger Sequencing	3mL EDTA blood	2500	2 Weeks
56	MD 055	CONGENITAL ADRENAL HYPERPLASIA (CAH) - <i>CYP21A2</i> Deletion analysis	MLPA	3mL EDTA blood	4000	3 Weeks
57	MD 056	GILBERT SYNDROME- <i>UGT1A1</i> Promoter Mutation Analysis	Sanger Sequencing	3mL EDTA blood	2500	2 Weeks
58	MD 057	ATYPICAL HAEMOLYTIC UREMIC SYNDROME (aHUS)-CNV analysis in the <i>CFH</i> region	MLPA	3mL EDTA blood	4000	3 Weeks
59	MD 058	<i>JAK2</i> -V617F (Exon 14) Mutation Analysis	Sanger Sequencing	3mL EDTA blood	2500	2 Weeks
60	MD 059	<i>JAK2</i> - (Exon 12) Mutation Analysis	Sanger Sequencing	3mL EDTA blood	2500	2 Weeks
61	MD 060	NON-SYNDROMIC HEARING LOSS- <i>GJB2</i> Mutation Analysis	Sanger Sequencing	3mL EDTA blood	2500	2 Weeks

PRENATAL DIAGNOSIS

62	MD 061	MATERNAL CELL CONTAMINATION (MCC)-by STR Analysis	Fragment Analysis	3mL Maternal EDTA blood and Chorionic Villus Sample/20-30mL Amniotic Fluid	2500	1 Week
63	MD 062	PRENATAL DIAGNOSIS WITH MCC (In CVS/AF)	Based on Proband's diagnosis		8500	1 Week
64	MD 063	PRENATAL DIAGNOSIS WITHOUT MCC (In CVS/AF)	Based on Proband's diagnosis		5500	1 Week
65	MD 064	QUANTITATIVE FLUORESCENT PCR (QF-PCR) FOR ANEUPLOIDIES	Fragment Analysis		3500	1 Week

NEXT GENERATION SEQUENCING

66	MD 065	WHOLE EXOME SEQUENCING (WES) -Analysis	Next Generation Sequencing	3mL EDTA blood	15000	~6 Weeks
67	MD 066	WHOLE EXOME SEQUENCING (WES) -Raw Data Only	Next Generation Sequencing	3mL EDTA blood	12500	~6 Weeks
68	MD 067	WHOLE GENOME SEQUENCING (WGS) - Analysis	Next Generation Sequencing	3mL EDTA blood	75000	~6 Weeks

69	MD 068	WHOLE GENOME SEQUENCING (WGS) - Raw Data Only	Next Generation Sequencing	3mL EDTA blood	70000	~6 Weeks
<i>TARGETED MUTATION ANALYSIS-for new variants</i>						
70	MD 069	TARGETED MUTATION ANALYSIS (TMA) + PRIMER DESIGNING (1 VARIANT IN A PERSON)	Sanger Sequencing	3mL EDTA blood	5500	4 Weeks
71	MD 070	TARGETED MUTATION ANALYSIS (TMA) + PRIMER DESIGNING (1 VARIANT IN EVERY ADDITIONAL PERSON)	Sanger Sequencing	3mL EDTA blood	2500	4 Weeks