SL.NO	TEST CODE	TEST NAME	TECHNIQUE USED	SAMPLE TYPE	PRICE (In rupees)	TAT
CYTOGENETIC TESTING						
1	CYTO 001	FISH FOR ANEUPLOIDIES (from Uncultured Chorionic Villi and Amniotic Fluid)	Fluorescence In-Situ Hybridization	Chorionic Villus Sample/20-30mL Amniotic Fluid	5000	1 Week
2	CYTO 002	FISH FOR BCR-ABL TRANSLOCATIONS	Fluorescence In-Situ Hybridization	3mL Heparin blood	5000	3 Weeks
3	CYTO 003	FISH FOR DIGEORGE SYNDROME (22q11.2 deletion syndrome)	Fluorescence In-Situ Hybridization	3mL Heparin blood	5000	3 Weeks
4	CYTO 004	FISH FOR KALLMAN SYNDROME	Fluorescence In-Situ Hybridization	3mL Heparin blood	5000	3 Weeks
5	CYTO 005	FISH FOR MILLER DIEKER SYNDROME	Fluorescence In-Situ Hybridization	3mL Heparin blood	5000	3 Weeks
6	CYTO 006	FISH FOR PRADER-WILLI SYNDROME	Fluorescence In-Situ Hybridization	3mL Heparin blood	5000	3 Weeks
7	CYTO 007	FISH FOR WILLIAM-BEUREN SYNDROME	Fluorescence In-Situ Hybridization	3mL Heparin blood	5000	3 Weeks
8	CYTO 008	KARYOTYPING FROM AMNIOTIC FLUID	AF Culture/GTG Banding	20-30mL Amniotic Fluid	5000	4 Weeks
9	CYTO 009	KARYOTYPING FROM PERIPHERAL BLOOD	Lymphocyte culturing/ GTG Banding	3mL Heparin blood	2000	3 Weeks
10	CYTO 010	SILVER NITRATE STAINING OF NUCLEOLUS ORGANIZER REGIONS	Lymphocyte culturing/Silver Nitrate staining	3mL Heparin blood	5000	3 Weeks
11	OGM 001	OGM FOR CONSTITUTIONAL CHROMOSOMAL ABNORMALITIES	Optical Genome Mapping	3-4mL EDTA blood	53000	4 Weeks
MOLECULAR TESTING						
12	MD 001	*SRY* DELETION ANALYSIS	PCR/AGE	3mL EDTA blood	1400	1 Week
13	MD 002	Y-CHROMOSOME MICRODELETION ASSAY	PCR/AGE	3mL EDTA blood	4600	1 Week
HAEMOLYTIC ANAEMIAS						
14	MD 003	BETA-THALASSEMIA-619 bps Deletion Analysis	PCR/AGE	3mL EDTA blood	2000	1 Week
15	MD 004	BETA-THALASSEMIA-Any single mutation (Specify mutation)	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
16	MD 005	BETA-THALASSEMIA-Common Mutations Panel	Sanger Sequencing	3mL EDTA blood	3000	2 Weeks
17	MD 006	BETA-THALASSEMIA-Complete *(HBB)* Gene Sequencing	Sanger Sequencing	3mL EDTA blood	4000	2 Weeks
18	MD 007	HAEMOGLOBIN-D DISEASE-Mutation Analysis	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
19	MD 008	HAEMOGLOBIN-E DISEASE-Mutation Analysis	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
20	MD 009	SICKLE CELL ANAEMIA-Mutation Analysis	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
21	MD 010	GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY-*G6PD* Mutation Analysis (Kerala-Kalyan, Mediterranean & Orissa Variants)	Sanger Sequencing	3mL EDTA blood	3500	2 Weeks

NEUROMUSCULAR DISEASES						
22	MD 011	BECKER MUSCULAR DYSTROPHY (BMD)-Mutation Analysis /Carrier Screening	MLPA	3mL EDTA blood	4000	2 Weeks
23	MD 012	DUCHENNE MUSCULAR DYSTROPHY (DMD)-Mutation Analysis /Carrier Screening	MLPA	3mL EDTA blood	4000	2 Weeks
24	OGM 002	FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY TYPE-1 (FSHD1)-D4Z4-ARRAY CONTRACTION TESTING	Optical Genome Mapping	3mL EDTA blood	46500	4 Weeks
25	MD 013	HEREDITARY SENSORY AND MOTOR NEUROPATHY (HSMN/CMT1A)- PMP22 Mutation Analysis	MLPA	3mL EDTA blood	4000	2 Weeks
26	MD 014	LIMB GIRDLE MUSCULAR DYSTROPHY TYPE 2A (LGMD2A) -*CAPN3*-Mutation Analysis [2051-1G>T, 2338G>C]	Sanger Sequencing	3mL EDTA blood	2500	2 Weeks
27	MD 015	MYOTONIC DYSTROPHY TYPE 1 (MD-1) -Mutation Analysis	TP-PCR	3mL EDTA blood	2500	2 Weeks
28	MD 016	OCULOPHARYNGEAL MUSCULAR DYSTROPHY (OPMD)-Mutation Analysis	Sanger Sequencing	3mL EDTA blood	3000	2 Weeks
29	MD 017	SPINAL AND BULBAR MUSCULAR ATROPHY (SBMA)-Mutation Analysis	Fragment Analysis	3mL EDTA blood	2500	2 Weeks
30	MD 018	SPINAL MUSCULAR ATROPHY (SMA)- Mutation Analysis /Carrier Screening	MLPA	3mL EDTA blood	3300	2 Weeks
HEREDITARY ATAXIAS AND OTHER NEURODEGENERATIVE DISORDERS						
31	MD 019	DENTATORUBROPALLIDOLUYSIAN ATROPHY (DRPLA) -Mutation Analysis	Fragment Analysis	3mL EDTA blood	2500	2 Weeks
32	MD 020	FRIEDREICH ATAXIA (FA)-Mutation Analysis	TP-PCR	3mL EDTA blood	2500	2 Weeks
33	MD 021	HUNTINGTON DISEASE (HD)-Mutation Analysis	Fragment Analysis	3mL EDTA blood	2500	2 Weeks
34	MD 022	SPINOCEREBELLAR ATAXIA (SCA) -any one type (Specify type)	Fragment Analysis	3mL EDTA blood	2500	2 Weeks
35	MD 023	SPINOCEREBELLAR ATAXIA (SCA) TYPES-1,2,3-Mutation Analysis	Fragment Analysis	3mL EDTA blood	5500	2 Weeks
36	MD 024	SPINOCEREBELLAR ATAXIA (SCA) TYPES-1,2,3,6-Mutation Analysis	Fragment Analysis	3mL EDTA blood	7500	2 Weeks
37	MD 025	SPINOCEREBELLAR ATAXIA (SCA) TYPES-7,17-Mutation Analysis	Fragment Analysis	3mL EDTA blood	5000	2 Weeks
38	MD 026	SPINOCEREBELLAR ATAXIA (SCA) TYPES-1,2,3,6,12-Mutation Analysis	Fragment Analysis	3mL EDTA blood	9500	3 Weeks
39	MD 027	SPINOCEREBELLAR ATAXIA TYPES (SCA) -1,2,3,6,7,12,17-Mutation Analysis	Fragment Analysis	3mL EDTA blood	13500	3 Weeks
DEVELOPMENTAL DELAY / DYSMORPHISM & INTELLECTUAL DISABILITY SYNDROMES						
40	MD 028	FRAGILE X SYNDROME (FRX/FXS)- Mutation Analysis/Carrier Screening	TP-PCR	3mL EDTA blood	6000	2 Weeks
41	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, WILLAMS-BEUREN SYNDROME, WILLAMS-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, PHELAN-MCDERMID SYNDROME, RETT SYNDROME(MECP2 DUPLICATION SYNDROME)]	MLPA	3mL EDTA blood	4000	2 Weeks

42	MD 030	ANGELMAN / PRADER-WILLI SYNDROME	MS-MLPA	3mL EDTA blood	7000	2 Weeks
43	MD 031	BECKWITH-WIEDEMANN/SILVER-RUSSELL SYNDROME	MS-MLPA	3mL EDTA blood	7000	2 Weeks
CYSTIC FIBROSIS/HEREDITARY PANCREATITIS PANEL						
44	MD 032	CHRONIC PANCREATITIS PANEL [*SPINK1* (N34S); *PRSS1* (N29I, R122H)]	Sanger Sequencing	3mL EDTA blood	3000	2 Weeks
45	MD 033	*CFTR*-Any single mutation (Specify mutation)	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
46	MD 034	*CFTR*-4 Mutation Panel [DF508, G542X, G551D, R553X]	Sanger Sequencing	3mL EDTA blood	2800	2 Weeks
47	MD 035	*CFTR*-5T/7T/9T Polymorphism Screening	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
48	MD 036	*CFTR*-36 Mutation Panel	Allele-Specific PCR/Fragment Analysis	3mL EDTA blood	7500	2 Weeks
49	MD 037	*SPINK1*- Mutation analysis (N34S)	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
MITOCHONDRIOPATHIES						
50	MD 038	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)- Any single mutation (Specify mutation)	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
51	MD 039	LEBER HEREDITARY OPTIC NEUROPATHY (LHON)- Mutation Analysis (G3460A, G11778A, T14484C)	Sanger Sequencing	3mL EDTA blood	3500	2 Weeks
52	MD 040	LEIGH SYNDROME-Any single mutation (Specify mutation)	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
53	MD 041	LEIGH SYNDROME-Mutation Analysis (T12706C, A13084T, G13513A)	Sanger Sequencing	3mL EDTA blood	3500	2 Weeks
54	MD 042	MITOCHONDRIAL ENCEPHALOMYOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES (MELAS)-Mutation Analysis (A3243G)	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
55	MD 043	MITOCHONDRIAL GENOME SEQUENCING	Next Generation Sequencing	3mL EDTA blood	7500	6 Weeks
COAGULOPATHIES						
56	MD 044	*FACTOR II* (PROTHROMBIN)- Mutation Analysis (G20210A)	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
57	MD 045	*FACTOR V* LEIDEN-Mutation Analysis (G1601A)	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
58	MD 046	HAEMOPHILIA A- *Factor VIII*-Intron 1 inversion analysis	PCR/AGE	3mL EDTA blood	2000	2 Weeks
59	MD 047	HAEMOPHILIA A- *Factor VIII*-Intron 22 inversion analysis	PCR/AGE	3mL EDTA blood	2000	2 Weeks
60	MD 048	HAEMOPHILIA A- *Factor VIII*-Introns 1 and 22 inversion analysis	PCR/AGE	3mL EDTA blood	4000	2 Weeks
HEREDITARY METABOLIC DISORDERS						
61	MD 049	BIOTINIDASE DEFICIENCY-Any single mutation (Specify mutation)	Sanger Sequencing	3mL EDTA blood	2000	2 Weeks
62	MD 050	BIOTINIDASE DEFICIENCY-*BTD* Mutation analysis	Sanger Sequencing	3mL EDTA blood	5000	2 Weeks
63	MD 051	HEREDITARY HAEMOCHROMATOSIS (HFE)- Mutation Analysis (H63D, C282Y)	Sanger Sequencing	3mL EDTA blood	2500	2 Weeks
64	MD 052	*METHYLENETETRAHYDROFOLATE REDUCTASE (MTHFR)*- Mutation Analysis (A1298C, C677T)	Sanger Sequencing	3mL EDTA blood	2500	2 Weeks

MISCELLANEOUS						
65	MD 053	ACHONDROPLASIA-*FGFR3* Mutation Analysis [G1138A, G1138C]	Sanger Sequencing	3mL EDTA blood	2500	2 Weeks
66	MD 054	CADASIL-*NOTCH3* Mutation Analysis [C421T]	Sanger Sequencing	3mL EDTA blood	2500	2 Weeks
67	MD 055	CONGENITAL ADRENAL HYPERPLASIA (CAH) -*CYP21A2* Deletion analysis	MLPA	3mL EDTA blood	4000	3 Weeks
68	MD 056	GILBERT SYNDROME- *UGT1A1* Promoter Mutation Analysis	Sanger Sequencing	3mL EDTA blood	2500	2 Weeks
69	MD 057	ATYPICAL HAEMOLYTIC UREMIC SYNDROME (aHUS)-CNV analysis in the *CFH* region	MLPA	3mL EDTA blood	4000	3 Weeks
70	MD 058	*JAK2*-V617F Mutation Analysis	Sanger Sequencing	3mL EDTA blood	2500	2 Weeks
71	MD 059	NON-SYNDROMIC HEARING LOSS-*GJB2* Mutation Analysis	Sanger Sequencing	3mL EDTA blood	2500	2 Weeks
PRENATAL DIAGNOSIS						
72	MD 060	MATERNAL CELL CONTAMINATION (MCC)-by STR Analysis	Fragment Analysis	3mL EDTA blood	2500	1 Week
73	MD 061	PRENATAL DIAGNOSIS WITH MCC (In CVS/AF)	Based on Proband's diagnosis	Chorionic Villus Sample/20-30mL Amniotic Fluid	8500	1 Week
74	MD 062	PRENATAL DIAGNOSIS WITHOUT MCC (In CVS/AF)	Based on Proband's diagnosis		5500	1 Week
NEXT GENERATION SEQUENCING						
75	MD 063	WHOLE EXOME SEQUENCING-Analysis	Next Generation Sequencing	3mL EDTA blood	16500	6 Weeks
76	MD 064	WHOLE EXOME SEQUENCING-Raw Data Only	Next Generation Sequencing	3mL EDTA blood	15000	4 Weeks
77	MD 065	WHOLE GENOME SEQUENCING- Analysis	Next Generation Sequencing	3mL EDTA blood	70000	6 Weeks
78	MD 066	WHOLE GENOME SEQUENCING- Raw Data Only	Next Generation Sequencing	3mL EDTA blood	60000	4 Weeks
TARGETED MUTATION ANALYSIS-for new variants						
79	MD 067	TARGETED MUTATION ANALYSIS + PRIMER DESIGNING (1 VARIANT IN A PERSON)	Sanger Sequencing	3mL EDTA blood	5500	4 Weeks
80	MD 068	TARGETED MUTATION ANALYSIS + PRIMER DESIGNING (1 VARIANT IN EVERY ADDITIONAL PERSON)	Sanger Sequencing	3mL EDTA blood	2500	4 Weeks