

Services

S.No.	Test	Test Code	Gene/Mutation	TAT	Price (INR)
Next Generation Sequencing					
1	Exome sequencing# (Singleton, CREV3 capture kit, this covers the mitochondrial genome)	WES001	Not applicable	8 weeks	42000
2	Exome sequencing# (Singleton, TWIST capture kit, this does not cover the mitochondrial genome)	WES002	Not applicable	8 weeks	31000
3	Duo exome sequencing (CREV3 capture kit)	DES001	Not applicable	8 weeks	75000
4	Duo exome sequencing (TWIST capture kit)	DES002	Not applicable	8 weeks	60000
5	Trio exome sequencing (CREV3 capture kit)	TES001	Not applicable	8 weeks	110000
6	Trio exome sequencing (TWIST capture kit)	TES002	Not applicable	8 weeks	80000
7	Re-analysis of exome/genome data#	REA001	Not applicable	6 weeks	20000
8	Mitochondrial genome sequencing	MTS001	Not applicable	8 weeks	20000

(*includes segregation of up to three prioritized variants by Sanger sequencing in the family. We request you to share the capture kit details for re-analysis of exome sequencing data)

Mutation analysis for specific diseases (targeted variants) by Sanger sequencing					
9	Targeted mutation analysis of Achondroplasia	ACH001	FGFR3 (c.1138G>A and c.1138G>C)	7 working days	4000
10	Targeted mutation analysis of Hypochondroplasia	HCH001	FGFR3 (c.1620C>A and c.1620C>G)	7 working days	4000
11	Targeted mutation analysis of Apert syndrome	APT001	FGFR2 (c.755_756delCGinsTT, c.755C>G, c.756_758delGCCinsCTT and c.758C>G)	7 working days	4000
12	Targeted mutation analysis of Sickle cell disease	SCD001	HBB (c.20A>C)	7 working days	4000
13	Targeted mutation analysis of Thanatophoric dysplasia type I	TDY001	FGFR3 (c.742C>T and c.1118A>G)	7 working days	4500
14	Targeted mutation analysis of Thanatophoric dysplasia type II	TDY002	FGFR3 (c.1948A>G and c.1949A>T)	7 working days	4000
15	Targeted mutation analysis of Factor V Leiden mutation	FVL001	F5 (c.1601G>A)	7 working days	4000
16	Targeted mutation analysis of any common single variant	TMA001	Not applicable	7 working days	5000
17	Targeted mutation analysis of any unique single variant	TMA002	Not applicable	3 weeks	6000
18	Segregation analysis for single variant in proband, father and mother	SEG001	Not applicable	2 weeks	12000

Mutation analysis for specific monogenic diseases (full gene analysis) by Sanger sequencing					
19	Mutation analysis of Gilbert syndrome and Crigler-Najjar syndrome	GCN001	UGT1A1 (Regulatory, promotor, exonic and flanking intronic regions of UGT1A1)	2 weeks	16000
20	Mutation analysis of beta thalassemia (sequencing the coding regions, 619 bp deletion detection and sequencing of reported variants in promoter and intronic regions as necessary)	HBB001	HBB	10 working days	10000
21	Mutation analysis of Rett syndrome	RET001	MECP2	2 weeks	12000
22	Mutation analysis of Oculocutaneous albinism type I	OCA001	OCA1	2 weeks	12000
23	Mutation analysis of Deafness, autosomal recessive 1A (Connexin 26)	CON001	GJB2	2 weeks	10000
24	Mutation analysis of arthropathy, progressive pseudorheumatoid of childhood	PPD001	WISP3	2 weeks	12000
25	Mutation analysis of campomelic dysplasia	CDY001	SOX9	2 weeks	16000
26	Mutation analysis of transcobalamin 2 deficiency	TCN001	TCN2	2 weeks	16000

Mutation analysis for specific disease by MLPA/TP-PCR					
27	Mutation analysis of Fragile X syndrome (TP-PCR)	FMR001	FMR1	2 weeks	9000
28	Mutation analysis of Huntington disease (TP-PCR)	HDI001	HTT	7 working days	9000
29	Mutation analysis of Disorders of Sex Development (MLPA)	DSD001	DMRT1, CYP17A1, SRD5A2 and HSD17B3	7 working days	11000
30	Mutation analysis of Friedreich ataxia (TP-PCR)	FDA001	FXN	2 weeks	9000
31	Mutation analysis of Myotonic dystrophy Type I (TP-PCR)	MDY001	DMPK	2 weeks	9000
32	Mutation analysis of Common microdeletions (MLPA)	MDS001	Not applicable	7 working days	7000
33	Mutation analysis of Angelman syndrome/ Prader Willi syndrome (MS-MLPA)	PWA001	MKRN3, MAGEL2, NDN, SNRPN and the snoRNA cluster; UBE3A	7 working days	10000
34	Mutation analysis of Spinal muscular atrophy by MLPA (or carrier testing)	SMA002	SMN1 and SMN2	7 working days	7000
35	Mutation analysis of Duchenne muscular dystrophy (MLPA)	DMD001	DMD	7 working days	11000
36	Mutation analysis of Hunter syndrome (MLPA)	HNT001	IDS	7 working days	11000
37	Mutation analysis of Neuronal ceroid lipofuscinosis (MLPA)	NCL001	PPT1, TPP1, CLN3, CLN6, and CLN8	7 working days	11000
38	Congenital adrenal hyperplasia (Sanger sequencing and MLPA)	CAH001	CYP21A2 (Deletion/duplication and sequence analysis of CYP21A2) MLPA and Sanger sequencing	2 weeks	26000
39	Unique MLPA on demand	UMD001	Not applicable	8 weeks	50000

Prenatal Diagnosis*					
40	Rapid aneuploidy testing for chromosomes 13, 18, 21, X and Y by QF-PCR	PND001	Not applicable	7 working days	10000
41	Prenatal diagnosis of any monogenic disease, detection of common aneuploidies and maternal cell contamination (if a diagnosis is established in the proband or if the parents are confirmed carriers)	PND002	Not applicable	10 working days	19000

* Please provide fetal samples (amniotic fluid 10 ml or sufficient chorionic villi) and 2 ml EDTA blood samples of both parents. Also provide copies of previous genetic test reports of the proband and family members.

Sample requirements: 2-3 ml EDTA blood samples or DNA or tissue of proband and parents, affected and unaffected siblings (as necessary). Turnaround time begins on the date of receipt of payment or samples, whichever is later. For patient information sheets, consent forms, test request forms and information for clinicians, please see our website: www.sumagenomics.in

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