suma genomics
simplifying rare diseases

91 94494 98467
office@sumagenomics.in
www.sumagenomics.in

# 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 | 48000 |
| 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 |
| 9 | Whole genome sequencing# | WGS001 | Not applicable | 8 weeks | 98000 |
| 10 | Trio genome sequencing | TGS001 | Not applicable | 8 weeks | 190000 |

(#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** | | | | | |
| 11 | Targeted mutation analysis of Achondroplasia | ACH001 | FGFR3 (c.1138G>A and c.1138G>C) | 7 working days | 4000 |
| 12 | Targeted mutation analysis of Hypochondroplasia | HCH001 | FGFR3 (c.1620C>A and c.1620C>G) | 7 working days | 4000 |
| 13 | 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 |
| 14 | Targeted mutation analysis of Sickle cell disease | SCD001 | HBB (c.20A>C) | 7 working days | 4000 |
| 15 | Targeted mutation analysis of Thanatophoric dysplasia type I | TDY001 | FGFR3 (c.742C>T and c.1118A>G) | 7 working days | 4500 |
| 16 | Targeted mutation analysis of Thanatophoric dysplasia type II | TDY002 | FGFR3 (c.1948A>G and c.1949A>T) | 7 working days | 4000 |
| 17 | Targeted mutation analysis of Factor V Leiden mutation | FVL001 | F5 (c.1601G>A) | 7 working days | 4000 |
| 18 | Targeted mutation analysis of any common single variant | TMA001 | Not applicable | 7 working days | 5000 |
| 19 | Targeted mutation analysis of any unique single variant | TMA002 | Not applicable | 3 weeks | 6000 |
| 20 | 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** | | | | | |
| 21 | Mutation analysis of Gilbert syndrome and Crigler-Najjar syndrome | GCN001 | UGT1A1 (Regulatory, promotor, exonic and flanking intronic regions of UGT1A1) | 2 weeks | 16000 |
| 22 | 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 |
| 23 | Mutation analysis of Rett syndrome | RET001 | MECP2 | 2 weeks | 12000 |
| 24 | Mutation analysis of Oculocutaneous albinism type I | OCA001 | OCA1 | 2 weeks | 12000 |
| 25 | Mutation analysis of Deafness, autosomal recessive 1A (Connexin 26) | CON001 | GJB2 | 2 weeks | 10000 |
| 26 | Mutation analysis of arthropathy, progressive pseudorheumatoid of childhood | PPD001 | WISP3 | 2 weeks | 12000 |
| 27 | Mutation analysis of campomelic dysplasia | CDY001 | SOX9 | 2 weeks | 16000 |
| 28 | Mutation analysis of transcobalamin 2 deficiency | TCN001 | TCN2 | 2 weeks | 16000 |
| **Mutation analysis for specific disease by MLPA/TP-PCR** | | | | | |
| 29 | Mutation analysis of Huntington disease (TP-PCR) | HDI001 | HTT | 7 working days | 9000 |
| 30 | Mutation analysis of Disorders of Sex Development (MLPA) | DSD001 | DMRT1, CYP17A1, SRD5A2 and HSD17B3 | 7 working days | 11000 |
| 31 | Mutation analysis of Common microdeletions (MLPA) | MDS001 | Not applicable | 7 working days | 7000 |
| 32 | Mutation analysis of Angelman syndrome/ Prader Willi syndrome (MS-MLPA) | PWA001 | MKRN3, MAGEL2, NDN, SNRPN and the snoRNA cluster; UBE3A | 7 working days | 10000 |
| 33 | Mutation analysis of Spinal muscular atrophy by MLPA (or carrier testing) | SMA002 | SMN1 and SMN2 | 7 working days | 7000 |
| 34 | Mutation analysis of Duchenne muscular dystrophy (MLPA) | DMD001 | DMD | 7 working days | 11000 |
| 35 | Mutation analysis of Hunter syndrome (MLPA) | HNT001 | IDS | 7 working days | 11000 |
| 36 | Mutation analysis of Neuronal ceroid lipofuscinosis (MLPA) | NCL001 | PPT1, TPP1, CLN3, CLN6, and CLN8 | 7 working days | 11000 |
| 37 | Congenital adrenal hyperplasia (Sanger sequencing and MLPA) | CAH001 | CYP21A2 (Deletion/duplication and sequence analysis of CYP21A2) MLPA and Sanger sequencing | 2 weeks | 26000 |
| 38 | Unique MLPA on demand | UMD001 | Not applicable | 8 weeks | 50000 |
| **Prenatal Diagnosis*** | | | | | |
| 39 | Rapid aneuploidy testing for chromosomes 13, 18, 21, X and Y by QF-PCR | PND001 | Not applicable | 7 working days | 10000 |
| 40 | 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 Storage** | | | | | |
| 41 | DNA storage of precious sample for one year | DNA001 | Not applicable | Not applicable | 5000 |

**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

**Shipping address:** **Suma Genomics Private Limited,** MCBR (Annexe), Manipal Center for Biotherapeutics Research
MIT campus, Manipal - 576 104, Karnataka, India | **E-mail:** **office@sumagenomics.in** | **Phone:** **+91 94494 98467**

Version 1.2, 2023