ANNEXURE 3

DNA DIAGNOSTIC TESTS

DNA Test	Sample Requirement	Technique Applied	Approximate Turnaround Time	Cost(Rs.)
Hematological Disorders				
Hemoglobinopathies				
Characterization of beta thalassemia mutations	5 mL blood in EDTA	ARMS PCR/ HBB Gene Sequencing	3-4 weeks	2000/-
Characterization of other HBB variants	5 mL blood in EDTA	➢ Xmn PCR ➢ Beta-delta PCR ➢ Gene Sequencing	3-4 weeks	1000/- 2000/- 2000/-
Prenatal diagnosis of beta thalassemia	20-30 mg of Chorionic villi/Amniotic fluid 20mL	➢ ARMS PCR ➢ HBB Gene Sequencing	2 weeks	2000/-
Neuromuscular & Neurodevelopmental Disorders				
Deletion/duplication testing of DMD (dystrophin gene)	5 mL blood in EDTA	Multiplex PCR MLPA	3-4 weeks	2000/- 4000/-
Carrier testing for DMD	5 mL blood in EDTA	MLPA	3-4 weeks	2000/- per kit
Spinal Muscular Atrophy (SMN)	5 mL blood in EDTA	PCR/RFLP MLPA	3-4 weeks	1500/- 2000/-
RETT syndrome (MECP2)	5 mL blood in EDTA	Sanger sequencing	4-6 weeks	700/- per exon
Fragile X syndrome (FMR1)	5 mL blood in EDTA	TP-PCR	4 weeks	1500/-
Pantothenate kinase-associated neurodegeneration (PANK2)	5 mL blood in EDTA	Sanger sequencing	8-12 weeks	700/- per exon
Megalencephalic Leukoencephalopathy (MLC) Common Mutation	5 mL blood in EDTA	PCR / RFLP/sequencing	3-4 weeks	1000/-
Prenatal diagnosis	20-30 mg of Chorionic villi/Amniotic fluid 20mL	Multiplex PCR/ MLPA/ sequencing	2 weeks	Disorder specific

Skeletal Dysplasia				
Achondroplasia (FGFR3)(COMMON MUTATION)	5 mL blood in EDTA	PCR / RFLP	3-4 weeks	1000/-
Fibrodysplasia ossifcans (FOP) hot spot	5 mL blood in EDTA	Sanger sequencing	4-5 weeks	700/- per exon
Apert syndrome (FGFR2) hot spot	5 mL blood in EDTA	Sanger sequencing	4-5 weeks	700/-
Czech Dysplasia- (COL2A1) hotspot exon 13	5 mL blood in EDTA	Sanger sequencing	3-4 weeks?	700/- per exon
BLAU syndrome (NOD-2) hot spot exon 4	5 mL blood in EDTA	Sanger sequencing	3-4 weeks?	700/- per exon
Diastrophic Dysplasia Sulfate Transporter (SLC26A2)	5 mL blood in EDTA	Sanger sequencing	3-4 weeks?	700/- per exon
Osteogenesis Imperfecta type XV (WNTI)	5 mL blood in EDTA	Sanger sequencing	3-4 weeks?	700/- per exon
Acrodysostosis with Hormonal Resistance – (PRKARIA) hot spot exon 11	5 mL blood in EDTA	Sanger sequencing	3-4 weeks?	700/- per exon
Cartilage-hair hypoplasia (RMRP) -Hot Spot Mutation	5 mL blood in EDTA	Sanger sequencing	3-4 weeks?	700/-
Brachytelephalangic chondrodysplasia punctata (ARSE)	5 mL blood in EDTA	Sanger sequencing	3-4 weeks	700/- per exon
Progressive pseudorheumatoid dysplasia (WISP 3)	5 mL blood in EDTA	Sanger sequencing	?	700/- per exon
Methylation Disorders				
Prader–Willi syndrome	5 mL blood in EDTA	MS-MLPA	4 weeks	3000/-
Angelman syndrome	5 mL blood in EDTA	MS-MLPA	4 weeks	3000/-
Beckwith–Wiedemann syndrome	5 mL blood in EDTA	MS-MLPA	4 weeks	3000/-
Russell-Silver syndrome	5 mL blood in EDTA	MS-MLPA	4 weeks	3000/-

Inborn Error of Metabolism				
Metachromatic leukodystrophy (MLD)	5 mL blood in EDTA	Sanger sequencing	4-5 weeks	700/- per exon
Gaucher disease (GBA)	5 mL blood in EDTA	Sanger sequencing	4-5 weeks	700/- per exon
Methylmalonic acidemia (MMAA, MMAB, MMACHC)	5 mL blood in EDTA	Sanger sequencing	7-8 weeks	700/- per exon
Mucopolysaccharidosis (MPS) Type I- IDUA, Type II -IDS,	5 mL blood in EDTA	Sanger sequencing	7-8 weeks	700/- per exon
MPS III -Sanfilippo syndrome Type B (NAGLU)	5 mL blood in EDTA	Sanger sequencing	7-8 weeks	700/- per exon
Mucopolysaccharidosis (MPS) Type IV (GALNS) exon 1, 7 & 8	5 mL blood in EDTA	Sanger sequencing	7-8 weeks	700/- per exon
Glutaric aciduria type I (GA1)	5 mL blood in EDTA	Sanger sequencing	4-5 weeks	700/- per exon
Glycogen storage disease (GSD) Type I,III	5 mL blood in EDTA	Sanger sequencing	4-5 weeks	700/- per exon
Krabbe disease (common deletion)	5 mL blood in EDTA	PCR	3-4 weeks	700/-
Biotinidase deficiency (BTD)	5 mL blood in EDTA	Sanger sequencing	4-5 weeks	700/- per exon
Galactosemia (GALT)	5 mL blood in EDTA	Sanger sequencing	4-5 weeks	700/- per exon
Prenatal diagnosis	20-30 mg of Chorionic villi/Amniotic fluid 20mL	Multiplex PCR/ MLPA/ sequencing	2 weeks	Disorder specific

Miscellaneous				
Blepharophimosis, Epicanthus Inversus Syndrome (FOXL2)	5 mL blood in EDTA	Sanger sequencing	?	700/- per exon
Incontinentia pigmenti (NEMO)	5 mL blood in EDTA	PCR	3-4 weeks	700/-
Von Hippel-Lindau syndrome (VHL)	5 mL blood in EDTA	Sanger sequencing	?	700/- per exon
Ectodermal Dysplasia (*EDA & EDAR)*	5 mL blood in EDTA	Sanger sequencing	5-6 weeks	700/- per exon
Cystic Fibrosis	5 mL blood in EDTA	PCR/RFLP/ sanger sequencing	3-4 weeks	700/- per exon
Sexing PCR	5 mL blood in EDTA	PCR based	3-4 weeks	500/-
Progeria (LMNA) Common Mutations	5 mL blood in EDTA	Sanger sequencing	3-4 weeks	
Oculocutaneous Albinism Type 1 (TYR) Exon 2 Hotspot Mutation	5 mL blood in EDTA	Sanger sequencing	3-4 weeks	700/- per exon
Non-syndromic deafness (GJB2)	5 mL blood in EDTA	Sanger sequencing	3-4 weeks	700/- per exon
Multiple Endocrine Neoplasia (MEN2A)	5 mL blood in EDTA	Sanger sequencing	3-4 weeks	700/- per exon
Primary hyperoxaluria Type I- (AGXT) Hotspot exon2	5 mL blood in EDTA	Sanger sequencing	3-4 weeks	700/-
Central Hypoventilation Syndrome (PHOX2B)	5 mL blood in EDTA	Sanger sequencing	4-5 weeks	700/- per exon

RESEARCH BASIS*		
DNA Test	**Sample Requirement**	**Technique Applied**
Classical Homocystinuria (CBS)	5 mL blood in EDTA	Sanger sequencing
Duane -radial ray syndrome (SALL4) Exons 1-3	5 mL blood in EDTA	Sanger sequencing
Thrombocytopenia- absent radii (TAR) syndrome (RBM8A) Exons 1-6	5 mL blood in EDTA	Sanger sequencing
Holt-Oram Syndrome (TBX5) Exons 2-9	5 mL blood in EDTA	Sanger sequencing
Brachydactyly-Syndactyly Syndrome (HOXD13) exon 1	5 mL blood in EDTA	Sanger sequencing

**turnaround time varies*