

## Molecular Diagnostic Services

MOLECULAR DIAGNOSTICS	GENES ANALYZED	SAMPLE TYPE	TECHNOLOGY	TAT
Beta-Thalassemia	HBB - 3 exons	2-3 ml EDTA blood	Sanger sequencing	3 weeks
Sickle cell anaemia	HBB - exon1	2-3 ml EDTA blood	Sanger sequencing	3 weeks
Mal de Meleda syndrome	SLURP- 3 exons	2-3 ml EDTA blood	Sanger sequencing	3 weeks
LHON disease (3 common mutations)	G3460A,G11778A,T14484C	2-3 ml EDTA blood	Sanger sequencing	3 weeks
MTHFR gene polymorphisms	677C>T and 1298A>C mutations	2-3 ml EDTA blood	Sanger sequencing	3 weeks
DMD (MLPA Based 79 exons)	DMD - 79 exon	2-3 ml EDTA blood	MLPA	3 weeks
SMA (MLPA)	SMA	2-3 ml EDTA blood	MLPA	3 weeks
G6PD	G6PD	2-3 ml EDTA blood	Sanger sequencing	3 weeks
Fragile X	-	2-3 ml EDTA blood	Sanger sequencing	15 days
Spino Cerebral Ataxia (SCA) - Single form	SCA	2-3 ml EDTA blood	Sanger sequencing	18 days
Spino Cerebral Ataxia (SCA) - Panel (1,2,3,6,7,10@12)	0	2-3 ml EDTA blood	Sanger sequencing	18 days
Rett Syndrome	-	2-3 ml EDTA blood	Sanger sequencing	12 days
DNA isolation and storage up to two years	NA	2-3 ml EDTA blood	NA	NA
Customised Monogenic Disorder tests	As per test	2-3 ml EDTA blood	Sanger sequencing	3-4 weeks