

6. HAEMATOLGY AND IMMUNOLOGY

MATH	Alpha Thalassemia - Deletions	4 ml blood EDTA	3 to 4 wks
MALPHAGS	Alpha Thalassemia gene sequencing	4 ml blood EDTA	3 to 4 wks
MTH5	Beta Thalassemia mutation study	4 ml blood EDTA	2 to 3 wks
MTBMS	Beta Thalassemia mutation study - confirmation of reported mutation	4 ml blood EDTA	2 to 3 wks
MBETAGS	Beta Thalassemia gene sequencing	4 ml blood EDTA	3 to 4 wks
MBGLD	Beta Thalassemia large deletions (HPFH)	4 ml blood EDTA	2 to 3 wks
MXM1	Beta Thalassemia-XMN polymorphism	4ml blood EDTA	3 to 4 wks
MXM2	Beta Thalassemia - Bcl1 and HBSIL-MYB polymorphism	4 ml blood EDTA	2 to 3 wks
MXM3	Beta Thalassemia - XMN, Bcl1 and HBSIL- MYB polymorphism	4 ml blood EDTA	2 to 3 wks
MHBD	Hb D Iran	4 ml blood EDTA	3-4 wks
MHBP	Hb D Punjab	4 ml blood EDTA	3-4 wks
MHBE	Hb E	4 ml blood EDTA	3-4 wks
MHBQI	Hb Q India	4 ml blood EDTA	3-4 wks
MHBSI	Hb S/ Sickle Cell Anemia	4 ml EDTA	2 to 3 wks
MHPFH	Hereditary persistence of fetal hemoglobin HPFH- 5 deletions	4ml blood EDTA	4-6 wks
MHAI	Hemophilia A Inversions 1/22	4 ml blood EDTA	3-4 wks
MHAS	Hemophilia A - Gene sequencing	4 ml blood EDTA	6-8 wks
MHBS	Hemophilia B - Gene sequencing	4 ml blood EDTA	6-8 wks
MHMC	Haemophilia A/B carrier screening (Hemophilia)	4 ml blood EDTA	3-4 wks
MHMP	Haemophilia A/B prenatal diagnosis (Hemophilia)	4 ml blood EDTA	1-2 wks
BCHT	Gaucher disease-Chitotriosidase enzyme	2 ml blood heparin	3-4 days
BGM1	Gaucher's disease- Beta glucosidase enzyme assay	6 ml blood heparin	one wks
MGDM	Gaucher's disease (4 common mutations)	4 ml blood EDTA	3-4 wks
MGDSG	Gaucher disease - GBA gene sequencing	4 ml blood EDTA	6-8 wks
BG6PD	G6PD (Quantitative)	3 ml blood EDTA	3-4 days
MGP1	G-6- PD, One mutation	4 ml blood EDTA	3-4 wks
MG6PD	G-6-PD (Mediterranean & Orissa types) - G6PD	4 ml blood EDTA	3-4 wks
MJACK2	JAK-2 mutation (Molecular Genetics) Polycythemiavera	4 ml EDTA	2 to 3 wks
BPOR	Porphobilinogen (Qualitative)	10 ml morning urine	3-4 days
BPORQ	Porphobilinogen (Quantitative)	10 ml morning urine	3-4 days
BPOR BL	Porphyria (Qualitative) in blood	4 ml Heparin blood	3-4 days

	PCM	Porphyria common mutation	4 ml EDTA	2 to 3 wks
	MPSQ	Porphyria gene sequencing	4 ml EDTA	4 to 6 wks
		<i>Primary Immunodeficiency</i>		
	MBTK	Agammaglobulinemia (XLA)/ Bruton Agammaglobulinemia- BTK gene sequencing	4 ml blood EDTA	6-8 wks
	NBT	CGD/Chronic Granulomatous Disease- Nitro blue tetrazolium dye test - Slide (NBT)	4 ml Fresh Heparin blood	next day
	MXCYB B	CGD/Chronic Granulomatous Disease X linked-CYBB Gene sequencing	4 ml blood EDTA	3-4 wks
	MELA2	Cyclical Neutropenia-ELA2 gene sequencing	4 ml blood EDTA	6-8 wks
	MRAG1	SCID- RAG1 deficiency	4 ml blood EDTA	3-4 wks
	MADA	SCID-Adenosine Deaminase Deficiency ADA gene sequencing	4 ml EDTA	3 to4 wks
	MXSCID	SCID X -linked-IL2RG gene sequencing	4 ml blood EDTA	3-4 wks
	MBTK	X linked Agammaglobulinemia (XLA)/ Bruton type- BTK gene sequencing	4 ml blood EDTA	6-8 wks
	MWAS	Wiskott Aldrich gene sequencing- WAS gene	4 ml blood EDTA	3-4 wks
	MWA	Wiskott Aldrich (Prenatal Dx)	CVS + 4 ml EDTA blood couple/ Affected subject	3-4 wks