7. HEPATOLOGY

	MCNJS	Crigler Najjar Syndrome (Sequencing)	4 ml blood EDTA	6-8 wks
	GALT	Galactosemia enzyme (GALT Quantitative)	**6 ml Heparin/ DBS**	3-4 days
	BGALEM	Galactosemia Panel: Galt+ Epimerase+ Galactose+ Gal-1-PO4	**6 ml Heparin blood**	3-4 days
	GALTSEQ	Galactosemia Gene sequencing (GALT)	4 ml blood EDTA	6-8 wks
	MGDP	Gilbert's disease (UGT1A1 Promoter polymorphism)	4 ml blood EDTA	3-4 wks
	MGDS	Gilbert's Disease Sequencing	4 ml blood EDTA	6-8 wks
	MGS	Glycogen storage 1A (common Indian mutation)	4 ml blood EDTA	3-4 wks
	MGSDS	Glycogen Storage 1A Gene Sequencing	4 ml blood EDTA	6-8 wks
	GLYRBC	Glycogen content in RBC (GSD Type III) - Fasting	**6 ml Heparin blood**	one wks
	MGSD3	Glycogen Storage disease III- known mutation AGL gene	4 ml blood EDTA	6-8 wks
	MGSD	GSD III PND by Linkage (AGL gene on 1p21)	4 ml blood EDTA	1-2 wks
	MHACM	Haemochromatosis (2 mutations)	4 ml blood EDTA	3-4 wks
	MALDOB1	Hereditary Fructose intolerance (HFI)-ALDOB gene common mutations	4 ml EDTA	2 to 3 wks
	MOTC	OTC gene sequencing	4 ml blood EDTA	3-4 wks
	MATP8B1	PFIC1-Progressive familial intrahepatic cholestasis type 1 - ATP8B1 gene sequencing	4 ml EDTA	6 to 8 wks
	MABCB11	PFIC2-Progressive familial intrahepatic cholestasis type 2 -ABCB11 gene Sequencing	4 ml EDTA	6 to 8 wks
	MABCB4	PFIC3-Progressive familial intrahepatic cholestasis type 3 - ABCB4 gene Sequencin	4 ml EDTA	6 to 8 wks
	MHEXBT	Sandhoff disease- HEX B gene targeted mutations analysis	4 ml blood EDTA	3-4 wks
	BSA	Succinyl acetone	Blood spots on filter paper/ 2 ml Heparin Blood / urine	2-3 days
	MTYS	Tyrosinemia - FAH Gene sequencing	4 ml blood EDTA	3-4 wks
	BCPL	Ceruloplasmin (Serum)	**4 ml blood in Plain tube**	3-4 days
	MWDC	Wilsons Disease- 5 common mutations	4 ml EDTA	2 to 3 wks
	MWDS	Wilson Disease - ATP7B Gene Sequencing	4 ml blood EDTA	3-4 wks
	MWD	Wilson disease - prenatal diagnosis by linkage	4 ml blood EDTA	3-4 wks
	MWLP	Wilson linkage presymptomatic	4 ml blood EDTA	2-3 wks
	BSC	Sugar chromatography	5 ml morning Urine	one wks