9. METABOLIC AND STORAGE DISORDERS

	a	Small Molecule Disorders		
	BAMT	Urine Metabolic Test	10 ml urine	1-2 days
	BAAM	TLC/Thin Layer Chromatography - (Plasma/urine) + Urine Metabolic screening tests	10 ml urine	one wks
	BHPLC	HPLC/High Performance Liquid Chromatography -Quantitative amino acid assay	**3 ml heparin blood/ Urine/ CSF**	one wks
	MASL	Arginosuccinic aciduria -ASL gene sequencing	4 ml blood EDTA	6-8wks
	BALK	Alkaptonuria- Qualitative	10 ml urine	2-3 days
	MALK	Alkaptonuria, Linkage studies (2 affected required)	4 ml blood EDTA	3-4 wks
	MAPD	Alkaptonuria, Prenatal Diagnosis	CVS + 4 ml EDTA blood couple/ Affected subject	1-2 wks
	MTAZ	Barth synd. (3-methylglutaconic aciduria type II) TAZ gene	4 ml blood EDTA	6-8 wks
	MBKT	Beta Ketothiolase sequencing	4 ml EDT	4-6 wks
	BIOTINEA	Biotinidase/BTD -enzyme assay	**4 ml Heparin blood/ DBS**	3-4 days
	MBTD	Biotinidase Deficiency- BTD gene sequencing	4 ml blood EDTA	6-8 wk
	MSLC19A3	Biotin-responsive basal ganglia disease (BBGD)-SLC19A3 gene sequencing	4 ml EDTA	6 to 8 wks
	MCIT	Citrullinemia R157H (G390R)	4 ml EDTA	2-3 wks
	MASS1	Citrullinemia type 1-ASS1 gene sequencing	4 ml blood EDTA	3-4 wks
	MFBP1	Fructose 1,6 Biphosphatase Deficiency-FBP1 gene sequencing	4 ml EDTA	3 to 4 wks
	GALT	Galactosemia - GALT enzyme assay	**6 ml Heparin blood/ DBS**	3-4 days
	BGK	Galactokinase -GALK enzyme assay	**6 ml Heparin blood**	3-4 days
	BGALEM	Galactosemia Panel: Galt+ Epimerase+ Galactose+ Gal-1-PO4	**6 ml Heparin blood**	3-4 days
	MGALT6	GALT - 6 Common Mutations	4 ml EDTA	1 to 2 wks
	GALTSEQ	Galactosemia- GALT Gene sequencing	4 ml blood EDTA	6-8 wks
	MHFIA	Hereditary Fructose intolerance (HFI)-ALDOB gene common mutations	4 ml EDTA	2 to 3 wks
	MALDOB	Hereditary Fructose intolerance (HFI)-ALDOB gene sequencing	4 ml EDTA	2 to 3 wks
	MMSUDA	Maple Syrup Urine Disease- BCKDHA gene sequencing	4 ml blood EDTA	6-8 wks

	MMSUDB	Maple Syrup Urine Disease- BCKDHB gene sequencing	4 ml blood EDTA	6-8 wks
	MMSUDDBT	Maple Syrup Urine Disease- DBT gene sequencing	4 ml blood EDTA	6-8 wks
	MCAD	MCAD mutation (Medium chain Acyl-coA dehydrogenase)	4 ml blood EDTA	3-4 wks
	MACADM	MCAD gene sequencing (ACADM gene)	4 ml EDTA	4 to 6 wks
	BMAT	Methyl malonic Acid Test	10 ml urine-morning	2-3 days
	MMMAA	Methyl Malonic Aciduria- MMAA gene sequencing	4 ml blood EDTA	6-8 wks
	MMMMA	Methyl Malonic Aciduria- MMAB gene sequencing	4 ml blood EDTA	6-8 wks
	MMUT	Methylmalonic acedimia- MUT gene sequencing	4 ml blood EDTA	6-8 wks
	MOTC	OTC gene sequencing	4 ml blood EDTA	3-4 wks
	MPCCA	Propionic aciduria- PCCA gene sequencing	4 ml blood EDTA	3-4 wks
	MPCCB	Propionic aciduria- PCCB gene sequencing	4 ml blood EDTA	3-4 wks
	BSC	Sugar chromatography	5 ml morning Urine	one 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
	MACADVL	VLCADD, Very Long Chain Fatty Acid Dehyd Def.-ACADVL gene	4 ml blood EDTA	3-4 wks
	b	**Glycogen Strorage Disorders**		
	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
	BPOM	Pompe disease - a glucosidase enzyme assay	**6 ml Heparin blood/ DBS**	3-4 days
	MGAAT	Pompe- GAA gene targeted mutation analysis	4 ml blood EDTA	3-4 wks
	MGAA	Pompe -GAA gene sequencing	4 ml blood EDTA	6-8 wks
	MGSD3	Glycogen Storage disease III- known mutation AGL gene	4 ml blood EDTA	6-8 wks
	SR49	McArdle- common mutation (R49 X mutation)	4 ml blood EDTA	2-3 wks
	MPYGM	McArdle disease gene sequencing - PYGM gene	4 ml EDTA	8 to 10 wks
	c	**Mucoploysaccharidosis**		
	BURN	MPS screen in urine (Biochemical genetics)	5 ml morning Urine	next day
	BQMPS	MPS estimation urine - Quantitative	5 ml morning Urine	one wks

	BUME	MPS Electrophoresis for GAGs	5 ml morning Urine	one wks
	BHUR	MPS I/Hurler syndrome - Iduronidase enzyme	**6 ml Heparin blood/DBS**	3-4 days
	IDSUEA	MPS 2/Hunter Syndrome - Iduronate 6 sulphate enzyme	**6 ml Heparin blood**	3-4 days
	MHUNTD	MPS II/Hunter syndrome (IDS gene deletions)	4 ml blood EDTA	6-8 wks
	MHUNTS	Hunter syndrome - IDS gene sequencing	4 ml EDTA	8 to 10 wks
	MMPSII	MPS II Gene Sequencing	4 ml EDTA	6 to 8 wks
	BMP3A	MPS III A/Sanfillipo Type A - Heparan sulphamidase	**6 ml Heparin blood**	one wks
	MMPS3A	MPS IIIA/ Sanfillipo Disease-SGSH gene sequencing	4 ml blood EDTA	6-8 wks
	BMP3B	MPS III B/Sanfilipo Type B- alpha-N acetyl glucosaminidase	**6 ml Heparin blood**	one wks
	MMPS3B	MPS IIIB Sanfillipo Disease-NAGLU gene sequencing	4 ml EDTA	4 to 6 wks
	BMP3C	MPS III C/Sanfillipo Type C -N- acetyl transferase	**6 ml Heparin blood**	one wks
	BMP3D	MPS III D-Sanfillipo Type D- α-N acetyl glucosaminide 6 - sulfate sulfatase	**6 ml Heparin blood**	one wks
	BMP3 (A+B)	MPS III/Sanfillippo (A+B) or(C+D)	**6 ml Heparin blood**	one wks
	G6S	MPS IVA/Morquio IVA- Galactose -6-Sulphatase	**6 ml Heparin blood**	3-4 days
	MGALNS	MPS IVA (Morquio A) - GALNS gene targeted mutations	4 ml EDTA Blood	2 to 3 wks
	BMP7G	MPS IV B/Morquio IVB- β-galactosidase enzyme assay	**6 ml Heparin blood/ DBS**	3-4 days
	BMP6	MPS VI/Aryl sulphatase - B enzyme assay	**6 ml Heparin blood/ DBS**	3-4 days
	MASRB	MPS VI -ASRB gene sequencing	4 ml EDTA	6 to 8 wks
	BGAC	MPS VII/Sly syndrome-β-glucuronidase enzyme assay	**6 ml Heparin blood/ DBS**	3-4 days
	MMS	Multiple Sulphatase Deficiency/MSD-SUMF1 gene sequencing	4 ml blood EDTA	6-8 wks
	BNAGA	Schindler disease - alpha N Acetyl Galactosaminidase	4 ml blood Heparin	3-4 days
	d	**Lysosomal Storage Disorders**		
	BEPK	Enzyme Package - (organomegaly) - Gaucher+NP+GM1	**8 ml Heparin blood/DBS**	3-4 days
	BFD	Fabry disease- α-Galactosidase enzyme	**6 ml Heparin blood / DBS**	3-4 days
	MGLA	Fabry disease - GLA gene sequencing	4 ml EDTA	4 to 6 wks
	BFUC	Fucosidase enzyme assay (Fucosidosis)	**6 ml Heparin blood**	3-4 days

	BMP7	GM1 gangliosidosis -β- galactosidase enzyme assay	**6 ml Heparin blood/ DBS**	3-4 days
	MGLB1	GM1 Gangliosidosis-GLB1 gene sequencing	4 ml blood EDTA	6-8 wks
	BTAYSACH	GM2 Gangliosidosis/Tay Sach's disease- - β- Hexosaminidase A enzyme	**6 ml Heparin blood/ DBS**	3-4 days
	MHEXA	GM2 Gangliosidosis/Tay Sach's disease-HEXA gene sequencing	4 ml blood EDTA	6-8 wks
	BSD	GM2 Gangliosidosis -Sandoff disease-Hexosaminidase (A+B) enzyme	**6 ml Heparin blood**	3- 4 days
	MHEXBT	GM2 Gangliosidosis- Sandhoff disease-HEX B gene targeted mutations analysis	4 ml blood EDTA	3-4 wks
	MHEXB	GM2 Gangliosidosis -Sandhoff disease-HEXB gene sequencing	4 ml EDTA	6 to 8 wks
	BCHT	Gaucher Disease-Chitotriosidase enzyme	**3 ml Heparin blood/ DBS**	3-4 days
	BGM1	Gaucher Disease-β- glucosidase enzyme assay	**6 ml Heparin blood/ DBS**	3-4 days
	MGDM	Gaucher's disease (4 common mutations)	4 ml EDTA	3 to 4 wks
	MGDSG	Gaucher disease - GBA gene sequencing	4 ml EDTA	4 to 6 wks
	BIC	I-Cell dis. (Plasma enzymes: b-Galactosidase+ Aryl sulfatase A+ β-Hex A)	**6 ml Heparin blood**	3-4 days
	BMANA	Mannosidosis (Mannosidase A enzyme)	**6 ml Heparin blood**	3-4 days
	BMANB	Mannosidosis (Mannosidase B enzyme)	**6 ml Heparin blood**	3-4 days
	BMAN	Mannosidase enzyme (Mannosidosis A & B)	**6 ml Heparin blood**	3-4 days
	BARA	Metachromatic Leukodystrophy-Aryl sulphatase - A enzyme	**6 ml Heparin blood**	3-4 days
	MMLD	Metachromatic Leukodystrophy gene sequencing	4 ml blood EDTA	6-8 wks
	BNPD	Niemann Picks disease-Sphingomyelinase enzyme assay	**6 ml Heparin blood**	3-4 days
	MNP	Niemann Pick disease gene sequencing	4 ml blood EDTA	6-8 wks
	BVLC	Peroxisomal Disorders-VLCFA Analysis	4 ml blood EDTA	3-4 wks
	BWM	Wolman disease, Acid esterase	**6 ml Heparin blood**	3-4 days
	MXALD	X linked adrenoleukodystrophy - ABCD1 gene sequencing	4 ml blood EDTA	6-8 wks