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Test list Oct 2017

Tests and Profiles

	Tests	Method	Sample Needed	TAT
	Genetic Consultation / Counseling	**-**	**All samples to be sent at 2 – 8 C.**	
1	Urine MRST	-	15 ml Urine	24 hrs
2	Sr. Ammonia, Lactate, Sugar	Fuji Dry Chem	2 ml Heparinised blood in cold chain	2 hrs
3	Sr. Free Fatty Acids	Colorimetry	2 ml Separated Serum in cold chain	24 hrs
4	Beta Hydroxybutyrate	Colorimetry	2 ml Separated Serum in cold chain	24 hrs
5	TMS of Blood – DBS - Carnitine / Acyl-carnitine Profile	TMS - DBS	2 ml Heparinised blood or Dried Blood Spots on S & S 903 paper	4 – 5 days
6	TMS - Carnitine / Acyl-carnitine Profile – Plasma	TMS - Plasma	2 ml Heparinised blood in cold chain	4 – 5 days
7	TMS - Carnitine / Acyl-carnitine Profile - Urine	TMS - Urine	15 ml Urine in Cold Chain	7 – 8 days
8	GC-MS of Urine (For Organic Acids)	GC-MS	15 ml Urine in cold chain	2 – 3 days
9	Biotinidase enzyme- Quantitative assay in Serum	Colorimetric	2 ml Separated Serum in cold chain	24 hrs
10	HPLC - Urine Orotic Acid	HPLC	15 ml Urine in cold chain	1 – 2 days
11	HPLC - Urine Purine / Pyrimidines	HPLC	15 ml Urine in cold chain	2 – 3 days
12	UHPLC - Plasma / Serum Aminoacids :(23AA)	UHPLC	2 ml Separated Serum in cold chain	1 – 2 days

13	UHPLC - Urine Aminoacids :(23AA)	UHPLC	15 ml Urine in cold chain	2 – 3 days
14	UHPLC - CSF Aminoacids :(23 AA)	UHPLC	2 ml Clear CSF in cold chain	1 – 2 days
	Galactosemia Profile	**Method**	**Sample Needed** **All samples to be sent at 2 – 8 C.**	**TAT**
15	GALT Enzyme	Fluorometry	2 ml Heparinised blood in cold chain	5 – 6 days
16	Total Galactose and Galactose 1, Phosphate	Colorimetric	2 ml Heparinised blood in cold chain	2 – 3 days
17	TLC Sugar	TLC	15 ml Urine in cold chain	2 – 3 days
18	Total Galactosemia Profile	-	2 ml Heparinised blood + 15 ml urine	5 – 6 days
	Homocysteine Profile			
19	Sr. Homocystine	HPLC	3 ml EDTA Blood and 2 ml Serum	5 – 6 days
20	Sr. Homocysteine, B_{12} and Folic Acid	-	3 ml EDTA Blood and 2 ml Serum	5 – 6 days
21	MTHFR Mutation Analysis – 2 common Mutations	PCR- RFLP	3 ml EDTA Blood	5 – 6 days
22	Sr. Homocysteine, Sr. Folic Acid, Sr. B_{12}, Plasma Aminoacids, MTHFR Mutations		3 ml EDTA Blood and 2 ml Serum	5 – 6 days
	Pyridoxine dependent seizures			
23	Plasma Pipecolic Acid	GC-MS/SIM	2 ml Separated Serum or Plasma	4 – 5 days
24	CSF – Pipecolic Acid	GC-MS/SIM	2 ml Clear CSF in cold chain	4 – 5 days

25	Urine AASA and Pipecolic Acid	GC-MS/SIM	10 ml morning urine sample, frozen	4 – 5 days
26	Plasma + CSF Pipecolic Acid & Urine AASA	GC-MS/SIM	2 ml Separated Plasma + 2 ml CSF + 10 ml urine – Samples must be sent frozen	8 – 10 days
27	Antiquitin Gene Studies (Performed in Europe)		3 ml EDTA blood x 2 tubes and DNA consent form signed.	
	Enzymes on DBS	**Method**	**Sample Needed All samples to be sent at 2 – 8 C.**	**TAT**
28	ADA / PNP		2 ml Heparinised blood in cold chain	4 – 6 weeks
29	HPRT / APRT		2 ml Heparinised blood in cold chain	4 – 6 weeks
	Special Quantitative Assays			
30	Plasma Argininosuccinic Acid	UHPLC	2 ml Separated Serum or Plasma	5 – 6 days
31	Urine Argininosuccinic acid	UHPLC	15 ml Urine in cold chain	5 – 6 days
	Plasma Argininosuccinic Acid	LCMS/MS	2 ml Separated Serum or Plasma	
	Urine Argininosuccinic acid	LCMS/MS	15 ml Urine in cold chain	
32	Quantitation of Succinylacetone in urine	GC-MS/SIM	15 ml Urine in cold chain	5 – 6 days
	Quantitation of Succinylacetone in urine	LCMS/MS		
33	Quantitation of Methylmalonic acid in Urine / Plasma	GC-MS/SIM	2 ml Separated Plasma + 15 ml Urine	5 – 6 days
34	Quantitation of Glutaric acid in Urine / Plasma	GC-MS/SIM	2 ml Separated Plasma + 15 ml Urine	5 – 6 days
35	Quantitation of Mevalonic acid in	GC-MS/SIM	15 ml Urine in cold chain	5 – 6 days

	urine			
36	Quantitation of Homogentisic acid in urine	GC-MS/SIM	15 ml Urine in cold chain	5 – 6 days

37	Quantitation of Oxalic acid in urine	GC-MS/SIM	15 ml Urine in cold chain	5 – 6 days
	Peroxisomal Disorders			
38	Very Long Chain Fatty Acids (VLCFA)	GC-MS/SIM	2 ml Separated Serum or Plasma	10 – 12 days
39	Plasmalogens in RBC	GC-MS/SIM	3 ml EDTA blood in cold chain	10 – 12 days
	Molybdenum Cofactor deficiency	**Method**	**Sample Needed**	**TAT**
41	Urine Sulfites	-	15 ml Urine in cold chain	2 Hrs
42	Urine Sulfocysteine	UHPLC	15 ml Urine in cold chain	5 – 6 days
43	Plasma Sulfocysteine	UHPLC	2 ml Separated Serum or Plasma	5 – 6 days
44	Urine + Plasma Sulfocysteine + Sulfites	UHPLC	2 ml Separated Plasma + 15 ml Urine	5 – 6 days
45	Gene Studies (MOCS1, MOCS2, GEPH, SUOX) Tests performed in Germany		3 ml EDTA blood x 2 tubes and DNA consent form signed	
	Pterin metabolism Studies			
46	Urine Pterins or CSF	UHPLC	15 ml urine / 2 ml CSF in cold chain	10 – 12 days
47	DBS Pterins	UHPLC	2 ml Heparinised blood / DBS	10 – 12 days
48	DBS + Urine or CSF Pterins	UHPLC	DBS / 15 ml urine/2 ml CSF in cold chain	10 – 12 days

49	DHPR Enzyme on DBS	Flurometry	2 ml Heparinised blood / DBS	5 – 6 days

	Folates and CSF – NT			
50	CSF – MTHF (Folates)	UHPLC	CSF collected in serially numbered vials 1 ml in each – 1, 2, 3 and 4. Send samples frozen. Clinical details and Plasma Aminoacid results important for interpretation.	10 – 12 days
51	CSF – MTHF + CSF Pterins + Ur Pterins	UHPLC		10 – 12 days
52	CSF Neurotransmitters (Samples sent to Germany)	UHPLC		3 – 4 weeks
	Cholesterol Synthesis Defect			
53	Plasma Sterols	GC-MS/SIM	2 ml Separated Serum or Plasma	10 – 12 days
54	SLO Gene Studies (DHCR7) Tests performed in Germany		3 ml EDTA blood x 2 tubes and DNA consent form signed	
	Lysosomal Storage Disorders	**Method**	**Sample Needed**	**TAT**
55	Urine MPS – GAG Quantitation, EPP and Oligosaccharides	TLC/EPP	15 ml Urine in cold chain	5 – 6 days
56				
a	Iduronidase	MPS I -	3 ml Heparinised blood + 3 ml EDTA blood + 2 ml Serum and 15 – 20 ml urine in Cold chain	5 – 6 days
b	Iduronate Sulphatase	MPS II		10 – 12 days
c	N-acetylgalactosamine-6-sulfatase	MPS IV – A		10 – 12 days
d	Beta Galactosidase	MPS IV – B		5 – 6 days
e	Aryl Sulphatase B	MPS VI		5 – 6 days

f	Beta Glucuronidase	MPS VII	3 ml Heparinised blood + 3 ml EDTA blood + 2 ml Serum and 15 – 20 ml urine in Cold chain	5 – 6 days
g	Aryl Sulphatase A	MLD		5 – 6 days
h	Aryl Sulphatase A + B	Multiple Sulphatase Def :		5 – 6 days
i	Hexosaminidase – Total and A	Sandhoff's Disease		10 – 12 days
j	Hexosaminidase – A	Tay Sach's Disease		10 – 12 days
k	Beta Glucosidase	Gaucher Disease		5 – 6 days
l	Alfa – Galactosidase	Fabry's Disease		10 – 12 days
m	Alfa – Glucosidase / Acid Maltase	Pompe Disease		5 – 6 days
n	Alpha Mannosidase	α Mannosidosis		5 – 6 days
o	Beta Mannosidase	β Mannosidosis		5 – 6 days
p	Alpha Fucosidosis	Fucosidosis		5 – 6 days
q	Sphingomyelinase	Niemann Pick A/B :		10 – 12 days
r	Beta- galactocerebrosidase	Krabbe Disease :		10 – 12 days
	Acid Lipase			
s	Chitotriosidase	Biomarker		5 – 6 days
t	CCL – 18	Biomarker		5 – 6 days
u	MPS III A, B, C and D Can be performed with our			

	collaborators. Cost per enzyme -			
	Miscellaneous	**Method**	**Sample Needed**	**TAT**
57	CDG Screening (Serum by IEF+ Urine) (No discount)	IEF	2 ml Serum + 15 ml Urine in Cold chain	10 – 12 days
58	CSF – IFN (Alfa Interferon) for AGS	ELISA	2 ml Serum + 2 ml CSF in Cold chain	5 – 6 days
59	Leucocyte Cysteine	UHPLC	3 ml Heparinsied or EDTA blood	5 – 6 days
60	Gluten Sensitivity profile – AGA-IgG, IgA, Anti Endomyseal Ab-IgA and TTG	ELISA	2 ml Serum	2 – 3 days
61	Total Bile Acids	Colorimetry	2 ml Serum / Plasma + 15 ml Urine in Cold Chain	3 – 4 days
62	TMS Bile Acids (Europe)	TMS	2 ml Serum / Plasma + 15 ml Urine in Cold Chain	4 – 6 weeks
63	Venous blood Karyotype		3 ml Heparinised blood	2 – 3 weeks
	Creatine Metabolism Studies			
64	GAA, Creatine and Creatinine – Urine Quantitative assay (Useful for follow-up and confirmatory diagnosis)	LC/MS/MS	24 Hr urine collected in clean bottle and 15 ml sent after total volume of urine measured and weight of the patient mentioned. Urine must be sent in Dry ice.	1 week
65	GAA, Creatine and Creatinine Plasma Quantitative assay (Not useful for followup)	LC/MS/MS	2 ml Hepoarinised plasma or Serum in Dry ice and one DBS Card (2 ml heparinised blood) Sent in a hteormocl box with coolant gel. No Dry ice required.	1 week
66	DBS GAA / Creatine	TMS	DBS – 4 Good sized spots	1 Week

	Profiles	Sample Needed All samples to be sent at 2 – 8 C.	TAT
67	**Critically ill Newborn Profile (Blood + Ur)** – Ur MRST, TLC Sugar, GALT, Galactose, Biotinidase, 17 OHP, G6PD HPLC Ur Orotic Acid. HPLC – Ur. Purine/Pyrimidines, TMS of blood, GC-MS of Urine, Plasma Aminoacids	3 ml Heparinised blood + 3 ml EDTA blood + 2 ml Serum and 15 – 20 ml urine in Cold chain	8 – 10 days
68	**Detailed Metabolic Workup (Blood + Ur)** – Ur MRST, TLC Sugar, TLC – Oligosaccharide-rides, GALT, Galactose, Biotinidase, HPLC Ur. Orotic Acid. HPLC Ur. Purine / Pyrimidines Sr. Hcy and Sr. B12 Level, TMS of blood, GC-MS of Urine, Plasma Aminoacids	3 ml Heparinised blood + 3 ml EDTA blood + 2 ml Serum and 15 – 20 ml urine in Cold chain	10 – 12 days

69	**Autism Profile –** Ur MRST, TLC – Oligosaccharide, Biotinidase, HPLC Ur Orotic Acid. HPLC – Ur. Purine / Pyrimidines Sr. Hcy and Sr. B12 Level, TMS of blood, GC-MS of Urine, Plasma Aminoacids, Antigliadin Ab Endomyseal Ab, (PCR -Fragile X and Karyotype Excluded)		3 ml Heparinised blood + 3 ml EDTA blood + 2 ml Serum and 15 – 20 ml urine in Cold chain	10 – 12 days
70	**Metabolic Liver Profile –** (Please provide detailed LFT report including GGT) Galactosemia Profile : GALT / Galactose, TLC Sugar Tyrosinemia Profile – Urine SA, Blood SA, AFP Plasma Aminoacids HLH profile – LDH, TG, Cholesterol, Ferritin and sCD25 TLC Oligosaccharides. TMS of Blood GC-MS of Urine **(CDG is not included in this profile = Cost Rs. 15,000/- extra)**		3 ml Heparinised blood + 3 ml EDTA blood + 2 ml Serum and 15 – 20 ml urine in Cold chain	10 – 12 days
71	**Detailed Newborn Screening –** TMS, Galactose, Biotinidase, T4, TSH, SA, IRT, 17 OHP, G6PD		2 ml heparinised blood or DBS with 4 good sized spots	5 – 6 days

72	**NBS + GC-MS of Urine**		2 ml heparinised blood or DBS with 4 good sized spots + 15 ml Urine	5 – 6 days
73	**Tyrosinemia Profile –** AFP, PBG Synthase, Blood & Urine Succinyl Acetone, GC-MS of Urine, Plasma Aminoacids		3 ml Heparinised blood + 3 ml EDTA blood + 2 ml Serum and 15 – 20 ml urine in Cold chain	5 – 6 days

74	**Proximal RTA Profile –** For Galactosemia, Wilson Disease, Tyrosinemia Type 1and Cystinosis GALT and Galactose Sr. and Urine Copper, Ceruloplasmin Plasma Aminoacids and Urine Succinyl Acetone Quantitation Leucocyte Cysteine		3 ml Heparinised blood + 3 ml EDTA blood + 2 ml Serum and 15 – 20 ml urine (from 24 Hr urine collection) in Cold chain	12 – 15 days
75	**HLH Screening –** Sr. Triglycerides, Sr. LDH, Sr. ferritin and soluble CD25		3 ml Heparinised blood + 3 ml EDTA blood + 2 ml Serum and 15 – 20 ml urine in Cold chain	5 – 6 days

76	**Recurrent Seizures Profile –** Sr. Lactate, CSF Lactate Urine MRST - Sulfites Biotinidase enzyme in Serum Carnitine / Acyl carnitine Profile in detail		2 ml Heparinised blood + 2 ml EDTA blood + 2 ml Serum + 2 ml CSF + 15 ml urine in cold chain	10 – 12 days

	GC/MS of Urine Plasma Aminoacids CSF Aminoacids CSF - 5 MTHF CSF Pipecolic acid and Urine AASA (If Sulfites negative) or Plasma and Urine Sulfocysteine (If Sulfites positive)			

77	**Leukodystrophy Profile –** Serum Lactate Aryl sulfatase A Enzyme (MLD) Krabbe Enzyme Urine GCMS for Canavan's Disease and Organic acidemias VLCFA and Plasmalogens for Peroxisomal Disorders Carnitine / Acyl Carnitine Profile Serum Homocysteine and Vitamin B12 levels Plasma Pipecolic acid TLC for Oligosaccharides		3 ml Heparinised blood x 2 tubes + 3 ml EDTA blood + 2 ml Serum and 15 – 20 ml urine in Cold chain	10 – 12 days

	Tests for Porphyria	**Method**	**Sample Needed**	**TAT**
78	Urinary Total Porphyrins	Column Chromato	10 – 15 ml Urine in Covered tube in cold chain (24 hr urine volume to be mentioned)	1 – 2 days
79	Urine ALA and PBG Quantitation	Column Chromato		1 – 2 days

80	Urine Total Porphyrins + ALA + PBG	Column Chromato		1 – 2 days
81	HPLC Urine Porphyrins – Uro I,III, Hepta, Hex, Penta and Copro I and III	UHPLC		5 – 6 days
82	RBC Free Porotoporphyrins and Zinc Protoporphyrins	UHPLC	3 ml EDTA blood	5 – 6 days
83	HPLC Stool Porphyrins – Uro I,III, Hepta, Hex, Penta and Copro I ,III	UHPLC	5 – 10 Gm in a sterile tube	5 – 6 days

84	PBG Synthase	Fluorometry enzyme		
85	PBG Deaminase	Fluorometry enzyme	3 ml EDTA blood	5 – 6 days
86	Uro-decarboixylase	Fluorometry enzyme	3 ml EDTA blood	5 – 6 days
87	**Full Porphyria biochemical workup –** Urine Porphyrin, PBG, ALA, Urine HPLC, Stool - HPLC, Blood Porphyrin isomers HPLC, PBG Synthase enzyme, PBG Deaminase enzyme, Uro Decarboxylase enzyme		10 – 15 ml Urine in Covered tube in cold chain (24 hr urine volume to be mentioned), 3 ml EDTA blood, 3 ml Heparinised blood and 2 ml Serum	5 – 6 days

	ELISA Tests	Method	Sample Needed	TAT
88	Antigliadin Ab – IgG and IgA	ELISA	2ml Serum at room temperature	24 hrs
89	Anti Endomyseal Ab – IgA	ELISA	2ml Serum at room temperature	24 hrs
90	Soluble CD25	ELISA	2 ml Serum	1 – 2 days
91	Serum Ferritin	ELISA	2ml Serum	1 - 2 days
92	Vitamin D	ELISA	2 ml Serum	2 - 3 days
93	Vitamin B12	ELISA	2 ml Serum	2 - 3 days
94	Folic Acid	ELISA	2 ml Serum	2 - 3 days

	Molecular Studies on Research Basis			
A	**Lysosomal Storage Diseases**			
1	Gaucher's Disease		Full clinical details Date of Birth Height, weight Metabolic workup / enzyme analysis confirming the diagnosis **3 ml heparinised blood + 3 ml EDTA blood + 2 ml Serum + 15 – 20 ml morning urine sample**	3 – 6 Months
2	Niemann Pick A/B			3 – 6 Months
3	Niemann Pick Type C			3 – 6 Months
4	GM1 Gangliosidosis			3 – 6 Months
5	GM2 Gangliosidosis – Tay Sachs and Sandhoff's Disease			3 – 6 Months
6	Krabbe Disease			3 – 6 Months
7	Metachromatic Leukodystrophy			3 – 6 Months
8	Fabry's Disease			3 – 6 Months
9	Farber's Disease			3 – 6 Months
10	Pompe Disease			3 – 6 Months
11	Wolmann Disease			3 – 6 Months

B	Mucopolysaccharidosis			
1	Hurler Disease MPS I		Full clinical details Date of Birth, Height, weight Metabolic workup / enzyme analysis confirming the diagnosis **3 ml heparinised blood + 3 ml EDTA blood + 2 ml Serum + 15 – 20 ml morning urine sample**	3 – 6 Months
2	Hunter/s Disease MPS II			3 – 6 Months
3	Sanfilippo Disease MPS III			3 – 6 Months
4	Morquio Disease MPS IV			3 – 6 Months
5	Maroteaux :Lamy Disease MPS VI			3 – 6 Months
6	Sly Disease MPS VII			3 – 6 Months
7	Mucolipidosis MLS I, II, III			3 – 6 Months
C	**Urea Cycle Defects**			
1	CPS Deficiency	*CPS1*	3 ml Fresh heparinised blood is needed.	3 – 6 Months
2	NAGS Deficiency	*NAGS*	Full clinical details Date of Birth Height / weight Metabolic workup / enzyme analysis confirming the diagnosis **3 ml heparinised blood + 3 ml EDTA blood + 2 ml Serum + 15 – 20 ml morning urine sample**	3 – 6 Months
3	Orotic Aciduria	*OTC*		3 – 6 Months
4	Argininosuccinate Synthase Def	*ASS*		3 – 6 Months
5	Argininosuccinate Lyase Deficiency	*ASL*		3 – 6 Months
6	Arginase Deficiency	*ARG1*		3 – 6 Months
7	Citrin Deficiency	*SLC25A13*		3 – 6 Months
8	Lysinuric Protein Intolerance	*SLC7A7*		3 – 6 Months

D	Glycogen Storage Diseases	Gene	Samples Needed	
1	GSD Type Ia	*G6PC*	Full clinical details Date of Birth Height / weight Metabolic workup **3 ml heparinised blood + 3 ml EDTA blood + 2 ml Serum + 15 – 20 ml morning urine sample**	3 – 6 Months
2	GSD Type Ib-d	*SLC37A4*		
3	GSD Type II	*GAA*		
4	GSD Type III	*AGL*		
5	GSD Type IV	*GBE1*		
6	GSD Type V	*PYGM*		
7	GSD Type VI	*PYGL*		
8	GSD Type VII	*PFKM*		
9	GSD Type VIII	*PHKA2*		

10	GSD Type IX	*PGK1*		
11	GSD Type X	*PGAM2*		

	Some more research Projects	**Gene**	**Samples Needed**	
1	Alport Syndrome	*COL4A5*	Full clinical details Date of Birth Height / weight Metabolic workup / enzyme analysis confirming the diagnosis **3 ml heparinised blood + 3 ml EDTA blood**	3 – 6 Months
2	Cystic Fibrosis	*CFTR*		3 – 6 Months
3	Cystinosis	*CTNS*		3 – 6 Months
4	Creatine Synthesis Defect			
5	Duchenne Muscular Dystrophy			
6	Hereditary Angioedema			
7	Gilbert Disease			