Navi Mumbai Institute of Research In Mental And Neurological Handicap

Dr. Anil B. Jalan,
MD DCH MCPS

With effect from 1st April 2010 (Revised).

A brief introduction to NIRMAN and our team:-

Navi Mumbai Institute of Research In Mental And Neurological Handicap was established in Navi Mumbai on 15th Dec. 1998 under the leadership of Dr. Anil B. Jalan (MD, DCH, and MCPS). Dr. Jalan passed MD in Paediatrics from Mumbai University in 1989. He developed interest in genetics and Metabolic disorders while working in PRL of KEM hospital Mumbai during post graduation and later on received basic training in Metabolic disorders at CDFD Hyderabad. Thereafter Dr. Jalan has regularly attended specialized training programmes at various European universities. Over a period of 10 years he has diagnosed and managed more than 7,000 children with various metabolic, genetic and neurological disorders. Details of his training programme are as follows:-

	Name of the Course	Academy	
1	1st focused course on "Congenital Disorders of Glycosylation"	University of Catania, Catania, Italy	April 2003.
2	Training in Basic and advanced Laboratory. Technologies and Newborn screening	AKH – Kinder Klinik, University of Vienna, Austria	Nov. 2003
3	2nd Focused course on "Paroxysmal Disorders"	Orphan Europe Academy, AMC, Univ. of Amsterdam, Netherlands.	Aug. 2004
4	Clinical and Laboratory aspects of Neuro-metabolic disorders	Paed. Neurology Laboratory, Radboud University, Nijmegen, Netherlands	Sept. 2004
5	Hands on training on GC-MS and Tandem Mass Spectrometry	AKH – Kinder Klinik, University of Vienna, Austria	Sept. 2005
6	Hands on Training on Laboratoryoratory aspects of Lysosomal storage Disorders	LKH University, Graz, Austria	Oct 2005
7	Focused course on "Mitochondriopathies"	Orphan Europe Academy – Radboud University Nijmegen, Netherlands	June 2006
8	Molecular Diagnosis of IEM and Cystic Fibrosis	AKH – Kinder Klinik, University of Vienna, Austria	July 2006
9	Focused course on "Paediatric Movement Disorders"	Orphan Europe Academy – Barcelona, Spain	June 2007

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10	Molecular Diagnosis in IEM – Advanced Techniques	AKH – Kinder Klinik, University of Vienna, Austria	June 2007
11	Molecular Diagnosis of Wilson's disease and Hemochromatosis	AKH – Int. Medicine Dept, Dr. Peter Ferenci's Laboratory	Sept. 2008
12	Biochemical Diagnosis of MPS, Iso Electric Focusing and Real Time PCR	LKH – Medical University of Graz, Austria	May 2009
13	Molecular Diagnosis of Infectious Diseases	LKH – Medical University of Graz, Austria	May 2009
14	Metabolic Disorders in Newborns	Charles' University – Prague, Czech Republic	Oct. 2009
15	Gas Chromatography and TMS	Cento-Gene – Vienna Biocenter	May 2010

In the lab he has team of Jr and Senior research officers:-

1. Ms. Ketki Kudalkar – M.Sc. Biotechnology (PhD student) : Sr. Research Officer
2. Mr. Mahendra Parab – M.Sc. Biotechnology : Jr. Research Officer
3. Mr. Rishikesh Jalan – M.Sc. Biotech & Life Sceinces : Trainee Research officer (Trained at Graz & Vienna)
4. Mrs. Nutan Shirsat (Telawane) : Lab Supervisor (Trained in Vienna NBS Lab)

We had QC / PA (Quality Control and Performance Analysis) Certification from CDC (US Gov) for last 8 years consecutively.
We are member of following professional bodies:-

1. SSIEM : Society for Study in Inborn Error of Metabolism
2. SIMD : Society for Inborn Metabolic Defects
3. ESGH : European Society of Human genetics
4. ISGS : Indian Society for Genetic Screening
5. ISPAT : Indian Society for Prenatal Diagnosis and Therapy

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	Tests	Samples Required	Disorders	Lab. Charges & Dr. Charges	Suggested Patients Charges	Approx. Turn Around time
	Consultation and Counselling					
1	Urine MRST					24 Hrs
2	Ammonia , Lactate, BSL, Uric Acid & Cholesterol	3ml Heparin blood	Various IEMs and Genetic Dis.			24 Hrs
3	Urine MPS Screening – 3 Spot Tests + GAG Quantitation	24 Hr urine	MPS			48 Hrs
4	TLC – Oligosaccharides for LSD –	20 ml – Morning urine sample	Lysosomal Storage Disorders			72 Hrs
5	Urine HPLC – Orotic Acid	20 ml Random Urine sample	UCDs and UMPS def.			24 Hrs
6	Pl. HPLC – Methionine, Phenylalanine, Tyrosine, Tryptophan	2 ml clear plasma or Serum	PKU, Tyrosinemia, Homocystinuria			24 Hrs
7	LC-MS-MS – Aminoacids	2 ml clear plasma or Serum	Aminoacidopathies			2 – 3 weeks
8	LC-MS-MS – Aminoacids	2 ml clear plasma + Urine / CSF	Aminoacidopathies			3 – 4 wks.
9	Detailed Newborn Screening – including MS-MS (GALT / Biotinidase / T4 / TSH / G6 PD / TMS for Carnitine – Acyl Carnitine and Aminoacids) Urine GC-MS – SOS	2 ml Heparinised blood and 2 ml EDTA blood or 4 good sized spots on S & S 903 paper	40 + Screenable Disorders			2 wks.

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	Any of the following Parameters can be selected for NBS panel. Rate swill be discussed and decided according to number of assured samples per month.					
A	T 4 or TSH		Hypothyroidism			
B	17 OHP		Cong. Adrenal Hyperplasia			
C	GALT enzyme		Galactosemia			
D	Galactose		Galactosemia			
E	Biotinidase Enzyme		Biotinidase Def.			
F	Methionine		Tyrosinemia, Hcy			
G	Tyrosine		Tyrosinemia			
H	Phenylalanine		PKU			
I	Homocysteine		Homocystinuria			
J	Leucine		MSUD			
K	G 6 PD		G 6 PD Def.			
L	Carnitine / Acyl Carnitine profile		Organic Acidemias and FAODs			
M	HPLC – Hb for Sickle Cell Dis.		Sickle Cell Dis.			

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10	IGF-1	2 ml Plasma or Serum	Growth Monitoring			24 Hrs
11	Tandem Mass Spectrometry (TMS) for Carnitine/Acyl-Carnitine Profile	2 ml Heparinsied blood or DBS	Organic Acidemia and FAODs			3 Wks. ± 1 Wk.
12	Urine GC-MS for Organic acids	10 – 15 ml Urine + 1ml chloroform	Organic Acidemias and other 101 dis.			3 Wks ± 1 Wk.
13	Blood TMS + Ur. GC-MS	3 ml Heparinsied blood + 15 ml Ur	Various IEMs			3 Wks ± 1 Wk.
14	Urine HPLC for Purine and Pyrimidine metabolites by HPLC – 20 ml urine	20 ml Morning urine	Purine and Pyrimidine metabolism defects			1 Wk.
15	Transferrin Isoelectric Focusing for CDG – 2 ml Serum	2 ml Serum	Cong. Disorder of Glycosylation			6 – 8 Wks.
16	Biotinidase enzyme assay – Quantitative	2 ml Plasma or Serum	Biotinidase def & Holocarboxylase def			24 – 48 hrs.
17	ADA / PNP Enzymes	3 ml Heparinised blood + 10 ml urine	Cong. Immune Deficiency			6 – 8 Wks.
18	HPRT / APRT Enzymes	3 ml Heparinised blood + 10 ml urine	Lesch Nyhan Syndrome			6 – 8 Wks.
19	Galactosemia Screening – Urine MRST, TLC Sugar, GALT & Galactose – 20 ml urine + 3 ml Heparinised blood	3 ml Heparinised blood + 10 ml urine	Galactosemia			24 Hrs.
20	Protein Challenge – for OTC Carrier – Follow protocol	Follow the protocol	OTC Carrier status in Women			3 Days

21	Tyrosinemia Profile – Pl. Tyrosine, Phe , Meth, AFP, SA, PBG Synthase, Urine SA, dALA and SAA. [30 ml urine + 3 ml Heparinsied blood and 3 ml EDTA blood]	3 ml Heparinised blood, 3 ml EDTA blood, 2 ml Serum and 20 ml urine	Tyrosinemia Type I, II and III			4 – 6 wks.
22	Detailed metabolic profile – Critically ill newborns / MR / Autism / Detailed NB Screening with past H/S/O IEM	Same as above	Various IEMs			3 – 4 wks.
23	Homocysteine / Folic Acid / B 12 / Methionine	3 ml EDTA blood + 3 ml Serum (Fasting)	Homocystinuria – CBS / MTHFR / MS Def			3 – 4 Days
24	VLCFA Analysis	3 ml Serum and 2 ml Heparinised blood	Peroxisomal Disorders			3 – 4 wks.
25	Pipecolic Acid – Plasma	2 ml Plasma or Serum	Peroxisomal Disorders			3 – 4 wks.
26	Pipecolic Acid – CSF	2 ml CSF or Plasma	Peroxisomal Dis and Pyridoxine Dependant Seizures			3 – 4 wks.
27	Plasmalogen –Blood	3 ml Hep. Blood	Peroxisomal Disorders			3 – 4 wks.
28	7 DHC and 8 DHC for Cholesterol Synthesis defects	3 ml Serum or 10 ml Urine	Disorder of Cholesterol Synth.			4 – 6 wks.
29	Quantitative NAA estimation in Urine by Capillary Gas Chromatography	3 ml Serum or 10 ml Urine	Canavan's Dis.			4 – 6 wks.
30	Free Fatty Acids in Plasma – Quantitation	3 ml Serum and 2 ml Heparinised	Long Chain Fatty Acid Oxidation			1 – 2 Wks

		blood	Defects			

Lysosomal Storage Disorders

31 A	MPS (Mini) Profile – MPS Spot / GAG Qty. / MPS – EPP / TLC – Oligosaccharides / 4 enzymes – Aryl Sulphatase A / B, Beta Galactosidase, Beta Glucuronidase.	5 ml EDTA blood + 3 ml Heparinsied blood + 2 ml Serum + 20 ml urine from 24 Hr Ur collection.	Muco Poly-saccharoidosis			6 – 8 Weeks
31 B	MPS (Detailed) Profile – Above tests + Iduronidase and Iduronate Sulphatase	Same as above	Muco Poly-saccharoidosis			6 – 8 Weeks
32	**Enzymes for Sanfilippo Syndrome and Morquio A are done either at Netherlands or Graz University**					
33	Lysosomal enzymes	Samples Required	Disorders	Lab Ch.	Pt Ch.	
1	Iduronidase	5 ml Heparinsied blood with 5 ml EDTA blood + 15 ml Urine	MPS I			
2	Iduronate Sulphatase		MPS II			
3	Beta Galactosidase		MPS IV B			
4	Aryl Sulphatase B		MPS VI			
5	Beta Glucuronidase		MPS VII			
A	Aryl Sulphatase A		MLD			

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B	Aryl Sulphatase A + B		Multiple Sulphatase			
C	Hexosaminidase – Total and A		Sandoff & Tay Sach			
D	Beta Galactosidase		GM 1			
E	Beta Glucosidase		Gaucher's			
F	Alfa – Galactosidase		Fabry's Dis.			
G	Alfa – Glucosidase		Pompe's Dis.			
H	Alpha Mannosidase		Mannosidosis			
I	Alpha Fucosidase		Fucosidosis			
J	Sphinogolylinase		Niemann Pick's Dis.			
34	Chitotriosidase	2 ml Serum or Plasma	LSD monitoring			
35	GL - 3 and Lyso GL - 3	3 ml EDTA blood		Contact Lab before sending		
36	GB – 3	3 ml EDTA blood	Fabry's Dis. Monitoring	Contact Lab before sending		
37	14 Leukocyte enzymes	2 ml Plasma / Serum				
38	Red blood Cell – Glutathione Assay (Reduced GSH)	3 ml EDTA blood in ice or cold chain.	Glutathione Synthesis Dis.			3 – 4 weeks

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39	Pterins – In Urine / Blood / Serum / CSF	Contact Lab before sending	Pterin and Phe Met. Disorders			4 – 6 Wks
40	Sulfocysteine – Plasma / Urine	Contact Lab before sending	So / XO Def.			4 – 6 Wks
41	S – Adenosyl-homocysteine S – Adenosyl-methionine (Plasma / Sr)	Contact Lab before sending	Meth-Hcy Metabolism defects			8 – 10 Wks
42	Homogentisic Acid Quantitation	10 ml urine in ice	Alkaptonuria			2 – 3 Wks
43	Quantitation of Glycerol and Glycerol 3 – Phosphate (Ur / Plasma / Sr.)	Contact Lab before sending	FBP Def. and Glycerol met defect			4 – 6 Wks
44	Quantitative estimation of Co.Q10 (Plasma / Serum)	Contact Lab before sending	Mitochondriopathy			4 – 6 Wks
45	Urinary Acyl Glycines by GC-MS (Quantitative)	Contact Lab before sending	Organic Acidemias			8 – 10 Wks
46	CSF – 5 MTHF	Contact Lab before sending	Neonatal Seizures			4 – 6 Wks
47 A	Sulfatides (Qualitative)	10 morning Urine sample in cold		Contact Lab before sending	Contact Lab before sending	
47 B	Sulfatides (Quantitative)	10 morning Urine sample in cold		Contact Lab before sending	Contact Lab before sending	
48	TMS of Bile Salts	3 ml Serum or 10 ml urine	Bile Acid Synthesis Defects			4 – 6 wks

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49	TMS of Bile Salts (Both urine + Serum)	3 ml Serum or 10 ml urine				4 – 6 wks

	Other Important Enzymes			**Lab Charges**	**Patient Charges**	
50	Enzymes for NCL – 1 (PPT) and NCL – 2 (TTP)	2ml Serum, 3 ml Heparinised blood and 3 ml EDTA blood				
51	Following Test performed at Vienna on TMS					
	5 Leukocyte enzymes from DBS - 1. Gaucher's Disease 2. Pompe's Disease 3. Nieman pick's Disease 4. Krabbe's Disease 5. Fabry's Disease	Only 1 - 2 ml Heparinised blood is sufficient or 2 filter papers with 8 Spots.				

	Serum Drug Levels by HPLC			**Lab Charges**	**Patient Charges**	
52	Serum Sodium Benzoate Levels	2ml Serum, 3 ml Heparinised blood and 3 ml EDTA blood				Within 24 hrs

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I	**Quantitative estimation of Metabolites in Amniotic Fluid**					
a	Aminoacids (Citrulline etc.)	10 ml AF in ice	Aminoacidopathies			2 – 3 weeks
b	Homogentisic Acid	10 ml AF in ice	Alkaptonuria			2 – 3 weeks
c	Methyl Malonic Acid	10 ml AF in ice	MMA			2 – 3 weeks
d	Iso Valeryl Glycine	10 ml AF in ice	Iso Valeric Acidemia			2 – 3 weeks
e	Glutaric Acid	10 ml AF in ice	Glutaric Acidemia			2 – 3 weeks
f	Hexanoyl Glycine	10 ml AF in ice				2 – 3 weeks
g	Fumaric Acid	10 ml AF in ice	Fumaric Aciduria			2 – 3 weeks
h	7 Dehydro Cholesterol	10 ml AF in ice	SLO			2 – 3 weeks
i	N-Acetyl Aspartate	10 ml AF in ice	Canavan's Dis.			2 – 3 weeks
	Prenatal Diagnosis by Enzyme assay or uptake studies					
	Disorder	Technique	Sample			
1	MSUD	[1-^{14}C] Leucine Oxidation	Cultured Cells CVBx, AF (with cells)	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
		Mutation Analysis (BCKDHA, BCKDHB and DBT	DNA	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.

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		genes)				
2	Citrullinemia	AF – Citrulline levels	Uncultured AF			4 – 6 Wks.
3	Citrullinemia	[^{14}C] Citrulline uptake	Cultured Cells CVBx, AF (with cells)	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
		Mutation Analysis (ASS Gene)	DNA	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
4	Argininosuccinic Aciduria	[^{14}C] Citrulline uptake	Cultured Cells CVBx, AF (with cells)	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
		Mutation Analysis (ASL Gene)	DNA	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
5	Methyl Malonic Acidemia	MMA – Quantitation	Uncultured AF	31,500/-	35,000/-	2 – 3 weeks
		Propionyl and Methyl Malonyl carnitine	Uncultured AF	31,500/-	35,000/-	2 – 3 weeks
		MCM (mutase) Activity	Cultured Amniocytes	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
		[1-^{14}C] Propionate +/- OHCbl uptake	Cultured Cells CVBx, AF (with cells)	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
		Mutation analysis – MUT, MMAA, MMAB, MMADHC genes	DNA	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.

6	MMA with Homocystinuria	MMA	Uncultured AF	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
		Total Hcy	Uncultured AF	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
		Propionyl and Methyl Malonyl carnitine	Uncultured AF	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
		[1-^{14}C] Propionate +/- OHCbl uptake	Cultured Cells CVBx, AF (with cells)	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
7	Propionic Acidemia	Methyl citrate	Uncultured AF	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
		Propionyl carnitine	Uncultured AF	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
		Propionyl Co-A Carboxylase activity	Cultured Cells CVBx, AF (with cells)	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
		Mutation Analysis (PCCA & PCCB genes)	DNA	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
8	Iso Valeric Acidemia	Iso Valeryl Carnitine	Uncultured AF	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
		^{14}C-IsoValerate Uptake	Cultured Cells CVBx, AF (with cells)	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.

9	Multiple Carboxylase Deficiency	Mutation Analysis (HLCS gene)	DNA	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
10	Methyl Crotonyl glycinuria	Enzyme Analysis (MCC)	Cultured Cells CVBx, AF (with cells)	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
		Mutation Analysis (MCC-1 and MCC2 gene)	DNA	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
11	Methyl Glutaconic Aciduria Type I	Mutation Analysis (AUH gene)	DNA	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
12	Methyl Glutaconic Aciduria Type III (Costeff Syndrome)	Mutation Analysis (OPA 3 gene)	DNA	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
13	Pyruvate Carboxylase Def. (Cong. Lactic Acidosis)	Pyruvate Carboxylase Activity	Cultured Cells CVBx, AF (with cells)	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
14	Fatty Acids Oxidation Def. – CPT I and CPT II	Carnitine Palmitoyl Transferase Def. I & II enzyme activity	Cultured Cells CVBx, AF (with cells)	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
		Mutation Analysis (CPT I & II gene)	DNA	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
15	LCHAD	Mutation Analysis (HADHA gene)	DNA	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
16	MCAD	Mutation Analysis (ACADM gene)	DNA	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
17	Epilepsy Pyridoxin dependant	Mutation Analysis	DNA	Contact Lab	Contact Lab	4 – 6 Wks.

	(Antiquitin Gene defect)	(ALDH7A1 gene)		before sending	before sending	
18	Phenylketonuria	Mutation Analysis (PAH gene)	DNA	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
19	Defects of BH4 metabolism defect	Mutation Analysis (GDH1, PTPS, QDPR and SPR gene)	DNA	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
20	Adenylosuccinase Lyase Def.	Mutation Analysis (ADSL gene)	DNA	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
	There are many other disorders available for Prenatal Diagnosis					

Newer additions for Neurological disorders

Please contact before sending samples as there are specific requirements with each test

	Tests			Lab Charges	Patient Charges	Turn around time
1	CSF – INF Alfa Interferon (For Aicardi Gautier Syndrome)	2ml Clear CSF + 2 ml Clear Serum tubes kept in Ice.				1 week
2	CSF – Oligoclonal Antibodies (For Multiple Sclerosis)	2ml Clear CSF + 2 ml Clear Serum tubes				3 – 4 weeks

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		kept in Ice.				
3a	CSF – Protein 14 – 3 – 3 (For Cruetzfeldt Jacob Disease)	2ml Clear CSF + 2 ml Clear Serum tubes kept in Ice.				3 – 4 weeks
3b	CSF – Protein 14 – 3 – 3 (For Cruetzfeldt Jacob Disease) – Western blott	2ml Clear CSF + 2 ml Clear Serum tubes kept in Ice.				3 -4 weeks
4	CSF IgG Index – Measles IgG Ab (For SSPE and MS)	2ml Clear CSF + 2 ml Clear Serum tubes kept in Ice.				1 Week
5	NMDA receptor Antibodies – NR2	2ml Clear CSF + 2 ml Clear Serum tubes kept in Ice.		Contact Lab before sending	Contact Lab before sending	
6	HLA – B1502	3 ml EDTA blood		Contact Lab before sending	Contact Lab before sending	

Hepatology & Gastroenterology

1	Gluten Sensitivity Profile	Samples Required	Disease	Lab Charges	Patient Charges	
A	Antigliadin IgG	2 ml Serum	Coeliac Dis.			24 Hrs
B	Anti Gliadin IgA	2 ml Serum	Coeliac Dis.			24 Hrs
C	Anti Gliadin IgG + IgA	2 ml Serum	Coeliac Dis.			24 Hrs

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D	Anti Endomyseal Ab Ig A	2 ml Serum	Coeliac Dis.			24 Hrs
E	Anti Glia IgG + IgA + Endomy. IgA	2 ml Serum	Coeliac Dis.			24 Hrs
F	Anti Parietal cell Ab Ig A	2 ml Serum	Autoimmune Enteropathy			24 Hrs
G	Anti Intrinsic Factor Ig A	2 ml Serum	Autoimmune Enteropathy			24 Hrs
H	Anti parietal + Anti intrinsic Factor	2 ml Serum	Autoimmune Enteropathy			24 Hrs
I	Anti Gliadin Ab - All 5 Parameters	2 ml Serum	Coeliac Dis.+ Autoimmune Enteropathy			24 Hrs

For many other IEMs we can arrange Prenatal Diagnosis through our national international collaborators. Kindly send the clinical details, especially of the index case to discuss further. The estimate of expenses can be generated only after a proper Index case evaluation and carrier detection of the couple is completed.

Instructions for sending samples: -

1. **Urine for GC-MS (Organic Acids):** Atleast 20 ml of urine (liquid) should be collected in a sterile container. If only GC-MS is required, add few drops of chloroform or keep in the deep fridge. If HPLC- Ur. Orotic acid or Purine / Pyrimidines are required in addition to GC-MS for Organic acids please do not add Chloroform. After performing Orotic acid, we shall add Chloroform. Chloroform interferes with analysis of Orotic acid and Purine / Pyrimidines.
2. **Tandem Mass Spectrometry (MS-MS on blood):-** Blood should be collected in Heparinised tube or can be plotted directly from scalp vein on S & S 903 filter paper. Please keep the paper horizontal on a clean surface and put 2 – 3 drops of blood for one spot of about half inch circle. The blood must soak through and should be seen from the other side exactly like the spot

seen from front side. Allow the paper to dry in horizontal position for 3 hrs atleast and then place the papers in a plastic envelope. For one patent we must have two papers (atleast 8 spots of good size). If preparing paper seems difficult in your busy practice, please collect 2 ml blood in a heparin (green top) vaccuatainer and keep in the butter compartment of fridge till you send it to our lab (preferably dispatch the same day).

3. **HPLC – Aminoacids / Phe-Tyr-Meth etc.:-** Collect 3 ml blood in an EDTA tube and centrifuge the tune immediately (within 15 minutes). Separate the plasma and keep in deep fridge (- 20 C) or send it immediately. Additionally collect two filter papers (8 spots) in case confirmation with MS-MS is required or other tests required.
4. **For critically ill new borns:-** Send 20 ml urine, 3 ml Heparinised blood, 3 ml EDTA blood and 2 ml clear serum. If you have already done CSF, please send 2 ml of clear non hemorrhagic serum as well.

For better interpretation we request for following reports along with samples: - Ammonia, Lactate, BSL, Uric Acid and ABG with Electrolytes where ever indicated (especially in critically ill newborns).

We would strongly recommend a detailed history with clinical summary for all samples referred for metabolic work-up. In absence of history no interpretation will be / can be given. For any special tests or DNA studies please discuss with Dr. Anil B Jalan - (Mobile 098211 24578) before collecting samples.

Payment terms: - All the samples must accompany a DD in favor of “NIRMAN” payable in Mumbai. In absence of DD, the sample will not be processed. We shall not accept DD in the name of Dr. Anil Jalan or any bank transfer.

Thanking you,

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Paediatric Geneticist

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Useful contact numbers

		Mobile #	Office #
1	Dr. Anil B. Jalan – NIRMAN	098211 24578	0091-22-67910236
2	Mr. Sushil Upadhyay (Manager) – NIRMAN	098921 58501	0091-22-67910237
3	Mrs. Nutan Shirsat (Lab Supervisor) – NIRMAN	099870 73020	0091-22-67910237
4	Mr. Harshal Telawane (Accountant) – NIRMAN	098214 04448	0091-22-67910237

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