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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	BOH Cases	**All Cytogenetic Tests are performed by our Collaborative Lab in Mumbai.**		**Lab Charges**	**Patient Charges**	
A	Karyotype – Single	3 ml Heparinised blood	Chromosomal Disorders	Contact Lab before sending	Contact Lab before sending	
B	High resolution karyotype	3 ml Heparinised blood				
C	Karyotype – Couple	3 ml Heparinised blood				
D	High resolution karyotype	3 ml Heparinised blood				
E	Abortus Karyotype / Products of Conception – D & C	Sample in sterile Saline				
F	Karyotype of CVBx – For prenatal diagnosis	CVBx Sample in sterile Saline				
G	Karyotype of AF – For prenatal diagnosis	AF in sterile container – 15 ml with cells				
H	Amniotic Fluid – Aminoacids, GC-MS for Organic Acids and Orotic Acids	15 ml of clear AF in a sterile tube, kept at 2 – 8 C.	Various IEMs			
I	CVBx or Amniotic Fluid – Prenatal Screening for Lysosomal Storage Disease (Known Index Case)	30 mgm of clear C. V. Bx sample in 15 ml RPMI medium		Depends on enzyme and DNA		
J	Couple's Screening for LSD	3 ml Heparinised blood, 3 ml EDTA blood, 2 ml Serum and 20 ml urine				

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k	Couple's Screening for IEM (Other then LSD)	3 ml Heparinised blood, 3 ml EDTA blood, 2 ml Serum and 20 ml urine				
L	MLPA (Multiple Ligand Probe Amplification) – for Cytogenetic abnormalities	3 ml EDTA and 3 ml heparinised blood	At the moment as a research project	Not done as a stand alone test		Cost of DNA extraction applicable
M	Molecular Cytogenetic – DNA chip					

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

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	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 genes)	DNA	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
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

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		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 (Antiquitin Gene defect)	Mutation Analysis (ALDH7A1 gene)	DNA	Contact Lab before sending	Contact Lab before sending	4 – 6 Wks.
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					

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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).

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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,

Dr. Anil B. Jalan,
MD DCH MCPS
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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