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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DNA studies performed in our Lab.

	DNA studies			Lab Charges	Patient Charges	
A	DNA – MTHFR (2 Mutations)					2 weeks
B	Fragile X –by Methylation Specific PCR					3 – 4 weeks
C	Prader Willi Syndrome / Angelman Syndrome – Methylation Specific PCR					3 – 4 weeks
D	Hemochromatosis – Adult type					3 – 4 weeks
E	MECP2 Gene Sequencing					6 – 8 wks.

	There are more than 1000 different DNA studies available through our international collaborators. Charges for the test :- 1. Actual Charges + 2. 20 % TDS + 3. Foreign Exchange remittance charges (Bank Charges) 4. DNA Extraction Charges + 5. Courier Charges + 6. Service charges – Rs. 5,000/- per person.			
	DNA Extraction Charges – Per vial (300 uL of blood by Pure gene kit) or 1.5 ml Blood – By Chemical method			

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	DNA Chip – Microarray				Lab Charges	Pt Charges	
	DNA Chip – Molecular Cytogenetics – CGH Array						
A	Neurological Cases with Dysmorphic Features – Screens 1500 Gene s						
B	Autism Chip						
C	Prenatal Diagnosis – CVBx or Abortus material						
D	Other chips available are – Ashkenazi jew, Retinitis Pigmentosa (AR), RP (AD), USHER Syndrome, Congenital Deafness etc.					Please see the details on our website	

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	DNA Tests for Various Disorders available through our International Collaborations	
1	**Metabolic**	**Gene**
1	2,4 Dienoyl-CoA Reductase Deficiency	DECR1
2	3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency (HMG)	HMGCL
3	3-Hydroxyacyl CoA Dehydrogenase Deficiency	HADH
4	3-Methylcrontonyl CoA Carboxylase Deficiency (3-MMC)	MCCC1
5	Adrenogenital Syndrom (CYP21A2) (CAH)	CYP21A2
6	Alpha-1-Antitrypsin-Deficiency (SERPINA 1)	SERPINA1
7	Alpha-Mannosidosis (α-Mannosidase Def. - MAN2B1)	MAN2B1
8	Apolipoprotein-E- Deficiency (APOE)	APOE
9	Arginine:Glycin-Amidinotransferase- Deficiency - (AGAT)	AGAT
10	Argininosuccinic Aciduria (ASL) (OMIM 608310)	ASL
11	Beta-Mannosidose (β-Mannosidase Def. - MANBA) MANBA	
12	Biotinidase- Deficiency (BTD)	BTD
13	Carnitine-Palmitoyl-Transferase- Deficiency, Type II (CPT2)	CPT2
14	CCL 18 [PARC] (Monitoring: Gaucher, Disease)	
15	Ceroid-Lipofuszinosen, neuronal, Type 2 (NCL2) - (TPP1)	TPP1
16	Ceroid-Lipofuszinosen, neuronal, Type 5 (NCL5) -(CLN5)	CLN5
17	Ceroid-Lipofuszinosen, neuronal, Type 6 (NCL6) -(CLN6)	CLN6
18	Ceroid-Lipofuszinosen, neuronal, Type 7 (NCL7) -(MFSD8)	MFSD8
19	Ceroid-Lipofuszinosen, neuronal, Type 8 (NCL8) -(CLN8)	CLN8
20	Creatine Transporter Deficiency (OMIM 300036)	SLC6AB
21	Chitotriosidase (Monitoring: Gaucher, Morbus / genetics: Null Mutant)	
22	Cystinosis (CTNS)	CTNS
23	Cystic Fibrosis (Mucoviscidosis) (CFTR)	CFTR
24	Diabetes insipidus, nephrogenic, X-chromosomal - (AVPR2)	AVPR2

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25	Diabetes insipidus, nephrogenic, autosomal -(AQP2)	AQP2
26	Epilepsy, Pyridoxin- Dependency (EPD) (ALDH7A1)	ALDH7A1
27	Fabry Disease (OMIM 301500) (α- Galactosidase Def.) - (OMIM 300644)	GLA
28	Farber's Disease (Saure Ceramidase Def. - ASAH)	ASAH
29	Fucosidosis (α-L-Fucosidase Def. - FUCA1)	FUCA1
30	Galactokinase -Deficiency (GALK1)	GALK1
31	Galactosemia (OMIM 230400)	GALT
32	Galactose Epimerase - Deficiency (GALE)	GALE
33	Gaucher Disease, type I (OMIM 230800) GBA (OMIM 606463)	GBA
34	Gaucher Disease, type II (OMIM 230900) GBA (OMIM 606463)	GBA
35	Gaucher Disease, type III (OMIM 231000) GBA (OMIM 606463)	GBA
36	Gaucher Disease, type IIIC (OMIM 231005) GBA (OMIM 606463)	GBA
37	Gaucher Disease, perinatal lethal (OMIM 608013) GBA (OMIM 606463)	GBA
38	Glucose-6-Phosphat-Dehydrogenase - Deficiency (without Spherocytosis)	G6PD
39	Glutaryl-Coenzyme A (CoA)-Dehydrogenase- Deficiency (GDD) (GCDH)	GCDH
40	GSD Type 1A (von-Gierke-Krankheit) - (G6PC)	G6PC
41	GSD Type 1B (von-Gierke-Krankheit) - (G6PT1)	G6PT1
42	GSD Type 1C (von-Gierke-Krankheit) - (G6PT1)	G6PT1
43	GSD Type 2 (Pompe, Morbus) (α- Glukosidase Def. - GAA)	GAA
44	GSD Type 3 (Cori- oder Forbes- Krankheit) (AGL)	AGL
45	GSD Type 4 (Andersen-Krankheit) (GBE1)	GBE1
46	GSD Type 5 (McArdle, Morbus) (PYGM)	PYGM
47	GSD Type 6A, auf Grund von Phosphorylase-Kinase Defizienz (PHKA2)	PHKA2
48	GSD Type 6B, auf Grund von hepatischer Phosphorylase-Defizienz (PYGL)	PYGL
49	GSD Type 7 (Tarui-Krankheit) (PFKM)	PFKM
50	Congenital disorder of glycosylation, type Ia (CDG1A) (OMIM 212065) (OMIM 601785)	PMM2

51	Congenital disorder of glycosylation, type Ib (CDG1B) (OMIM 602579) (OMIM 154550)	MPI
52	Congenital disorder of glycosylation, Typ 1C (CDG1C) (ALG6)	ALG6
53	Congenital disorder of glycosylation, Typ 1D (CDG1D) (ALG3)	ALG3
54	Congenital disorder of glycosylation, Typ 1E (CDG1E) (DPM1)	DPM1
55	Congenital disorder of glycosylation, Typ 1F (CDG1F) (MPDU1)	MPDU1
56	Congenital disorder of glycosylation, Typ 1G (CDG1G) (ALG12)	ALG12
57	Congenital disorder of glycosylation, Typ 1H (CDG1H) (ALG8)	ALG8
58	Congenital disorder of glycosylation, Typ 1I (CDG1I) (ALG2)	ALG2
59	Congenital disorder of glycosylation, Typ 1J (CDG1J) (DPAGT1)	DPAGT1
60	Congenital disorder of glycosylation, Typ 1K (CDG1K) (ALG1)	ALG1
61	Congenital disorder of glycosylation, Typ 1L (CDG1L) (ALG9)	ALG9
62	Congenital disorder of glycosylation, Typ 1M (CDG1M) (TMEM15)	TMEM15
63	Congenital disorder of glycosylation, Typ 1N (CDG1N) (RFT1)	RFT1
64	Congenital disorder of glycosylation, Typ 2A (CDG2A) (MGAT2)	MGAT2
65	Congenital disorder of glycosylation, Typ 2C (CDG2C) (SLC35C1)	SLC35C1
66	Congenital disorder of glycosylation, Typ 2D (CDG2D) (B4GALT1)	B4GALT1
67	Congenital disorder of glycosylation, Typ 2E (CDG2E) (COG7)	COG7
68	Congenital disorder of glycosylation, Typ 2F (CDG2F) (SLC35A1)	SLC35A1
69	Congenital disorder of glycosylation, Typ 2G (CDG2G) (COG1)	COG1
70	Congenital disorder of glycosylation, Typ 2H (CDG2H) (COG8)	COG8
71	GM1-Gangliosidosis, type I (OMIM 230500) (β-Galaktosidase Def. - GLB1) (OMIM 611458)	GLB1
72	GM1-Gangliosidosis, type II (OMIM 230600) (OMIM 611458)	GLB1
73	GM1-Gangliosidosis, type III (OMIM 230650) (OMIM 611458)	GLB1
74	Guanidinoacetate Methyltransferase Defizienz (GAMT)	GAMT
75	Hemochromatosis (HFE)	HFE
76	Hemochromatosis, juvenile, Typ 2A (AR, HFE2A) (HJV)	HJV
77	Hemochromatosis, juvenile, Typ 2B (AR, HFE2B) (HAMP)	HAMP

78	Hemochromatosis, Typ 3 (AR, HFE3) (TFR2)	TFR2
79	Hemochromatosis, Typ 4 (AD, HFE4) (SLC40A1)	SLC40A1
80	Hartnup-Syndrom (SLC6A19)	SLC6A19
81	Hypercholesterolemia, familiär, auf Grund eines Defektes im LDL-Rezeptor (LDL-R)	LDLR
82	Hypercholesterolemia, AD, Typ 3 (HCHOLA3) (PCSK9)	PCSK9
83	Hypercholesterolemia, AD, Typ B (APOB)	APOB
84	Hypercholesterolemia, AR (ARH)	ARH
85	Hyperoxaluria Typ 1 (AGXT)	AGXT
86	Hyperoxaluria Typ 2 (GRHPR)	GRHPR
87	Hyper-phenylalaninemia (PKU) PAH (OMIM 261600)	PAH
88	Hyper-phenylalaninemia (PKU) PAH (OMIM 261600) Exon deletion studies by MLPA	PAH
89	Hyper-phenylalaninemia – GTP Cyclohydrolase 1 Def. (OMIM 600225)	GCH1
90	Hyper-phenylalaninemia, 6 - @Piruvonyl-tetrahydropterin Synthase Def. (PTS) (OMIM 261640)	PTPS
91	Hyper-phenylalaninemia – Dihydropteridine Reductase Deficiency (DHPR) (OMIM 261630)	QDPR
92	Hyper-phenylalaninemia – Sepiapterin Reductase deficiency (SPR) (OMIM 182125)	SPR
93	Iso Valeric Acidemia (IVA) (IVD)	IVD
94	Ketoacidosis due to beta-Ketothiolase-Dezienz (ACAT1)	ACAT1
95	Krabbe Disease (OMIM 245200) (Galactocerebrosidase Def.) (OMIM 606890)	GALC
96	Kreatin (Creatine)Transporter-Defect (SLC6A8)	SLC6A8
97	Kurzketten (Short Chain) - Acyl-CoA-Dehydrogenase-Deficiency (SCAD) (ACADS)	ACADS
98	Lipodystrophy, congenital generalised, Typ 1 (Berardinelli-Seip; CGL1) (AGPAT2)	AGPAT2
99	Lipodystrophy, congenital generalised, type 2 (CGL2) (OMIM 269700) (OMIM 606158)	BSCL2
100	Mannose-Binding Protein Dezienz (MBL2)	MBL2
101	Maple Syrup urine Disease (MSUD) BCKDHA (OMIM 608348)	BCKDHA
102	Maple Syrup urine Disease (MSUD) BCKDHB (OMIM 608348)	BCKDHB
103	Maple Syrup urine Disease (MSUD) DBT (OMIM 248610)	DBT
104	Maple Syrup urine Disease (MSUD) DLD (OMIM 238331)	DLD

105	McArdle Dis – Glycogen Storage Dis Typ V (PYGM)	PYGM
106	Metachromatic Leukodystrophy (MLD) (Arylsulphatase A Def. - ARSA)	ARSA
107	Methylmalonic acidemia, Vitamin-B12-sensible Form, Typ cbl A (MMAA) (OMIM 607481)	MMAA
108	Methylmalonic acidemia, Type cbl B (MMAB) (OMIM 605768)	MMAB
109	Methylmalonic aciduria - Homocystinuria, Typ cbl C (MMACHC)	MMACHC
110	Methylmalonic aciduria - Homocystinuria, Typ cbl D (C2ORF25)	C2ORF25
111	Methylmalonic aciduria, Methylmalonyl-CoA-Mutase-Deficiency (MUT) (OMIM 609058)	MUT
112	3 – Methylglutaconic Aciduria Type I (OMIM 600529)	AUH
113	3 – Methylglutaconic Aciduria Type III (OMIM 606580)	OPA3
114	Mittelketten (Medium Chain)-Acyl-CoA-Dehydrogenase-Dezienz (MCAD) (ACADM)	ACADM
115	Mediterranean Fever - familial (MEFV)	MEFV
116	MODY Syndrom Typ 1 (HNF4A)	HNF4A
117	MODY Syndrom Typ 2 (GCK)	GCK
118	MODY Syndrom Typ 3 (HNF1A)	HNF1A
119	MODY Syndrom Typ 5 (HNF1B)	HNF1B
120	Mucolipidosis Typ 2, I-Zellkrankheit (GNPTAB)	GNPTAB
121	Mucolipidosis Typ 3, Alpha/Beta (GNPTAB)	GNPTAB
122	Mucolipidosis Typ 4 (MCOLN1)	MCOLN1
123	Mucopolysaccharidose Typ 1 (MPS I, Hurler/Scheie-Krankheit) (α-L-Iduronidase Def.-IDUA)	IDUA
124	Mucopolysaccharidose Typ 2 (MPS II, Hunter-Krankheit) (Iduronate 2-Sulphatase Def. - IDS)	IDS
125	Mucopolysaccharidose Typ 3a (MPS IIIa, Sanlippo Syndrom A) (N-SulphoglukosamineSulphohydrolase Def. - SGSH)	SGSH
126	Mucopolysaccharidose Typ 3b (MPS IIIb,Sanlippo Syndrom B) (α-Nacetylglukosaminidase Def. - NAGLU)	NAGLU
127	Mucopolysaccharidose Typ 3c (MPS IIIc, Sanlippo Syndrom C) (Heparan-α-Glukosaminide-N-Acetyltransferase Def. -HGSNAT)	HGSNAT
128	Mucopolysaccharidose Typ 3d (MPS IIId, Sanlippo Syndrom D) (N-Acetylglukosamine-6-Sulphatase	GNA

	Def. – GNA)	
129	Mucopolysaccharidose Typ 4a (MPS IVa, Morquio Syndrom A) (Galaktosamine-6-Sulphate Sulphatase Def. - GALNS)	GALNS
130	Morquio syndrome B (OMIM 253010) (β-Galaktosidase-Def. - GLB1) (OMIM 611458)	GLB1
131	Mucopolysaccharidose Typ 6 (MPS VI, Maroteaux-Lamy Syndrom) (Arylsulphatase B Def. - ARSB)	ARSB
132	Mucopolysaccharidose Typ 7 (MPS VII, Sly Syndrom) (β-Glukuronidase Def. - GUSB)	GUSB
133	Mucopolysaccharidose Typ 9 (MPS IX) (HYAL1)	HYAL1
134	Multiple Acyl-CoA-Dehydrogenase-Deficiency (MADD) (ETFA)	ETFA
135	Multiple-Sulfatase-Deficiency (MSD) (SUMF1)	SUMF1
136	Niemann-Pick Disease, type A (OMIM 257200),type B (OMIM 607616) (Sphingomyelinase Def.SMPD1) (OMIM 607608)	SMPD1
137	Niemann-Pick Disease, type C1 (NPC1) (OMIM 257220) (Filipin Test only in Fibroblasts) (OMIM 607623)	NPC1
138	Niemann-Pick Disease, type C2 (OMIM 607625) (Filipin Test only in Fibroblasts) (OMIM 601015)	NPC2
139	Non Ketotic Hyperglycinemia (HGNK) GLDC (OMIM 238300)	GLDC
140	Non Ketotic Hyperglycinemia (HGNK) AMT (OMIM 238310)	AMT
141	Ornithine Transcarbamylase Deficiency (OTC) (OMIM 300461)	OTC
142	Pompe's Disease, GSD Type II (α- Glukosidase Def. - GAA)	GAA
143	Porphyria, acute intermittent (AIP) (HMBS)	HMBS
144	Propionic Acidemia (PA) (PCCA, PCCB)	PCCA + PCCB
145	Propionic Acidemia (PA) (PCCA)	PCCA
146	Propionic Acidemia (PA) (PCCB)	PCCB
147	Propionic Acidemia (PCCA) Identification of Exonic deletion by MLPA	PCCA
148	Prosaposin-Defect (PSAP)	PSAP
149	Pyruvate kinase deficiency with hemolytic anemia (OMIM 266200) (OMIM 609712)	PKLR
150	Refsum Syndrom (PEX7, PHYH) PEX7,	PHYH
151	Refsum Syndrom (PEX7)	PEX7

152	Refsum Syndrom (PHYH)	PHYH
153	Sandhoff's Disease GM2-Gangliosidose (Hexosaminidase-A+B-Def. - HEXB)	HEXB
154	Schindler Disease (α -Nacetylgalaktosaminidase Def. - NAGA)	NAGA
155	Sehr-langkettige (Very Long Chain) -Acyl-CoA-Dehydrogenase - Dezienz (VLCAD) (ACADVL)	ACADVL
156	Tay-Sachs, Morbus, AB Variante (GM2A)	GM2A
157	Tay-Sachs Disease (TSD) (OMIM 272800)(Hexosaminidase-A-Def. - HEXA)(OMIM 606869)	HEXA
158	Tyrosinemia Type I (FAH) (OMIM 276700)	FAH
159	Wilson's Disease Morbus (ATP7B)	ATP7B
160	Wolman Disease (LIPA)	LIPA
2	**Neurological disease**	
2.1	**Hereditary spastic paraplegia (HSP)**	
2.1.1.	**X-chromosomale SPG`s**	
1	SPG1 (MASA-Syndrom) (L1CAM)	L1CAM
2	SPG2 (PLP1)	PLP1
2.1.2.	**Autosomal dominate SPG`s**	
1	SPG3A (Atlastin SPG3A)	Atlastin SPG3A
2	Spastic paraplegia 4, autosomal dominant (SPG4) (OMIM 182601)(OMIM 604277)	SPG4
3	SPG6 (NIPA1)	NIPA1
4	SPG8 (KIAA0196)	KIAA0196
5	SPG10 (KIF5A)	KIF5A
6	Spastic paraplegia 13, autosomal dominant (SPG13) (OMIM 605280)(OMIM 118190)	HSPD1
7	Leukodystrophy, hypomyelinating, 4 (OMIM 612233)(OMIM 118190)	HSPD1
8	Spastic paraplegia 17 (SPG17) (OMIM 270685) (OMIM 606158)	BSCL2
9	SPG20 (Spartin SPG20)	Spartin SPG20
10	SPG21 (MAST Syndrom) (ACP33)	ACP33
2.2.	**Hereditary polyneuropathy (CMT)**	
1	CMT1-Screening PMP22 (OMIM 601097), MPZ (OMIM 159440), LITAF (OMIM 603795), EGR2	PMP22, MPZ, LITAF,

	(OMIM 129010), NEFL (OMIM 162280)	EGR2,NEFL
2	Charcot-Marie-Tooth Disease, type 1A (CMT1A) (OMIM 118220) (**Seq**. + MLPA) (OMIM 601097)	PMP22
3	Charcot-Marie-Tooth Disease, type 1A (CMT1A) (OMIM 118220) (MLPA) (OMIM 601097)	PMP22
4	Charcot-Marie-Tooth Disease, type 1A (CMT1A) (OMIM 118220) (Seq.)(OMIM 601097)	PMP22
5	Charcot-Marie-Tooth Disease, type 1B (CMT1B) (OMIM 118200) (OMIM 159440)	MPZ
6	Charcot-Marie-Tooth Disease, type 1C (CMT1C) (OMIM 601098) (OMIM 603795)	LITAF
7	Charcot-Marie-Tooth Disease, type 1D (CMT1D) (OMIM 607678) (OMIM 129010)	EGR2
8	Charcot-Marie-Tooth Disease, type 1F (OMIM 607734) (OMIM 162280)	NEFL
9	Neuropathy, hereditary, with liability to pressure palsies (HNPP) (OMIM 162500) (OMIM 601097)	
10	CMT2-Screening (KIF1B, MFN2, RAB7, LMNA,GARS, NEFL, HSPB1, MPZ, GDAP1, HSPB8,GJB1)KIF1B, MFN2 (OMIM 608507)	
11	CMT2A1 (KIF1B) RAB7,LMNA, GARS (OMIM 600287), NEFL (OMIM 162280), HSPB1 (OMIM 602195), MPZ (OMIM 159440), GDAP1, HSPB8, GJB1	KIF1B
12	Charcot-Marie-Tooth Disease, type 2A2 (CMT2A2) (OMIM 609260) (Seq. + MLPA)MFN2 (OMIM 608507)	
13	Charcot-Marie-Tooth Disease, type 2A2 (CMT2A2) (OMIM 609260) (MLPA) (OMIM 608507)	MFN2
14	Charcot-Marie-Tooth Disease, type 2A2 (CMT2A2) (OMIM 609260) (Seq.) (OMIM 608507)	MFN2
15	Charcot-Marie-Tooth Disease, type 6 (CMT6) (OMIM 601152) (Seq.) (OMIM 608507)	MFN2
16	CMT2B (RAB7)	RAB7
17	CMT2B1 (LMNA)	LMNA
18	Charcot-Marie-Tooth Disease, type 2D (CMT2D) (OMIM 601472) (OMIM 600287)	GARS
19	Charcot-Marie-Tooth Disease, type 2E (OMIM 607684) (OMIM 162280)	NEFL
20	Charcot-Marie-Tooth Disease, type 2F (CMT2F)(OMIM 606595) (OMIM 602195)	HSPB1
21	Charcot-Marie-Tooth Disease, type 2I (CMT2I)(OMIM 607677), type 2J (CMT2J) (OMIM 607736) (Seq. + MLPA) MPZ (OMIM 159440)	
22	Charcot-Marie-Tooth Disease, type 2I (CMT2I)(OMIM 607677), type 2J (CMT2J) (OMIM 607736)	

	(MLPA) MPZ (OMIM 159440)	
23	Charcot-Marie-Tooth Disease, type 2I (CMT2I)(OMIM 607677), type 2J (CMT2J) (OMIM 607736) (Seq.) MPZ (OMIM 159440)	
24	CMT2K (GDAP1)	GDAP1
25	CMT2L (HSPB8)	HSPB8
26	Dejerine-Sottas Syndrome (DSS) (OMIM 145900)CMT3/CMT4F: MPZ (OMIM 159440),PMP22 (OMIM 601097), PRX, EGR2 (OMIM 129010),GJB1	
27	CMT4 CMT4A (GDAP1)	GDAP1
28	CMT4B1 (MTMR2)	MTMR2
29	CMT4B2 (SBF2)	SBF2
30	CMT4C (SH3TC2)	SH3TC2
31	CMT4C1 (LMNA)	LMNA
32	CMT4C4 (GDAP1)	GDAP1
33	CMT4D (NDRG1)	NDRG1
34	Charcot-Marie-Tooth Disease, type 4E (CMT4E)(OMIM 605253)EGR2 (OMIM 129010)	
35	Charcot-Marie-Tooth Disease, type 4E (CMT4E)(OMIM 605253) (Seq. + MLPA) MPZ (OMIM 159440)	
36	CMT4F (PRX)	PRX
37	CMT4H (FGD4)	FGD4
38	CMT4J (FIG4)	FIG4
	Autonomic Neuropathies	
1	HSAN1 (SPTLC1)	SPTLC1
2	HSAN2 (HSN2)	HSN2
3	HSAN3 (IKBKAP)	IKBKAP
4	HSAN4 (NTRK1)	NTRK1
5	HSAN5 (NGFB; NTRK1)	NGFB; NTRK1
6	HSAN5 (NGFB)	NGFB

7	HSAN5 (NTRK1)	NTRK1
8	Minifaszikuläre Neuropathie mit 46, XY partieller Gonadendysgenesie (DHH)	DHH
	Rein motorische Neuropathie	
1	Neuropathie, distal kongenital (AR, DSMA4) (PLEKHG5)	PLEKHG5
2	Neuropathie, distal betont, rein motorisch, Typ 2A (AD, HMN2A) (HSPB8)	HSPB8
3	Neuronopathy, distal hereditary motor, type IIB (HMN2B) (OMIM 608634)(OMIM 602195)	HSPB1
4	Neuronopathy, distal hereditary motor, type V (HMN5) (OMIM 600794) (OMIM 600287)	GARS
5	Neuropathie, distal betont, rein motorisch, Typ 6 (HMN6) (IGHMBP2)	IGHMBP2
6	Neuropathie, distal betont, rein motorisch, Typ 7B (HMN7B) (DCTN1)	DCTN1
7	Sonstiges Andermann Syndrom (ACCPN) (SLC12A6)	SLC12A6
8	CMTX1 (GJB1)	GJB1
9	CMTX5 (PRPS1)	PRPS1
10	Charcot-Marie-Tooth Disease, dominant intermediate D (OMIM 607791) (OMIM 159440)	MPZ
11	DI-CMTB (DNM2)	DNM2
12	DI-CMTC (YARS)	YARS
13	Giant Axonal Neuropathie (GAN)	GAN
14	Hereditäre neuralgische Amyotrophie (HNA) (SEPT9)	Sep-09
15	Katarakte, kongenitale - facial Dysmorphien - Neuropathie (CCFDN) (CTDP1)	CTDP1
16	Refsum Syndrom (PEX7, PHYH)	PEX7, PHYH
17	Refsum Syndrom (PEX7)	PEX7
18	Refsum Syndrom (PHYH)	PHYH
19	Slowed Nerve Conduction Velocity (slow NCV; AD) (ARHGEF10)	ARHGEF10
	2.3. Vascular diseases	
1	Antithrombin-Defizienz, kongenital (AT3)	AT3
2	CADASIL (OMIM 125310) (OMIM 600276)	NOTCH3
3	Fabry Disease (OMIM 301500) (Genetic) * (α- Galactosidase A) (OMIM 300644)	GLA
4	Faktor II-Defizienz, kongenital (F2)	F2

5	Faktor V-Defizienz [R506Q] (F5)	F5
6	Glycoprotein 1A-Defizienz (ITGA2)	ITGA2
7	Methylcobalamine deficiency, cblG Type (OMIM 250940)(OMIM 156570)	MTR
8	Homocystinuria (Cystathionine beta-Synthase, CBS)	CBS
9	Homocystinuria due to Methylen- Tetrahydrofolat-Reductase-Defizienz (MTHFR)	MTHFR
10	Mitochondrial Myopathie - Lactic acidosis (MELAS)MTND1, MTND5,MTND6, MTTL1,MTTQ, MTTS1,MTTK, MTTS2, MTTH	
11	Plasminogen-Activator-Inhibitor-1-Deficiency, Congenital (PAI1)	PAI1
12	Protein-C- Deficiency, Congenital (PROC)	PROC
13	Protein-S- Deficiency, Congenital (PROS1)	PROS1
	2.4. non-ataxic movement disorder	
	2.4.1. Dystonia	
1	Dystonia 1, Torsion, autosomal dominant (DYT1) (OMIM 128100), (OMIM 605204)	Exon 5 TOR1A
2	Dystonia 1, Torsion, autosomal dominant (DYT1) (OMIM 128100), (OMIM 605204)	Complete TOR1A
3	Dopa-responsive Dystonia, autosomal dominant (DYT5A) (OMIM 128230) (OMIM 600225)	GCH1
4	Dopa-responsive Dystonia, autosomal recessive (DYT5B) (OMIM 191290)	TH
5	Dystonia 6, Torsion (Dyt6) (OMIM 602629) (OMIM 609520)	THAP1
6	DYT8 - Dystonie 8 (MR1)	MR1
7	Dystonia 11 (DYT11) (OMIM 159900) (OMIM 604149)	SGCE
8	DYT12 (ATP1A3)	ATP1A3
9	Dystonia 16 (DYT16) (OMIM 612067)(OMIM 603424)	PRKRA
10	Dystonia 18 (DYT18) (OMIM 612126)(OMIM 138140)	SLC2A1
	2.4.2. Parkinson	
1	Parkinson Disease, familial, type 1 (PARK1)(OMIM 168601)(OMIM 163890)	SNCA
2	Parkinson Disease 4, autosomal dominant Lewy Body (PARK4) (OMIM 605543)(OMIM 163890)	SNCA
3	Parkinson Disease 2, autosomal recessive juvenile (PARK2) (OMIM 600116)(OMIM 602544)	PARK2
4	Parkinson Disease 6, autosomal recessive earlyonset (PARK6) (OMIM 605909)(OMIM 608309)	PINK1

5	Parkinson Disease 7, autosomal recessive earlyonset (PARK7) (OMIM 606324) (OMIM 602533)	DJ1
6	Parkinson PARK8 (LRRK2)	LRRK2
7	Parkinson PARK9 (ATP13A2)	ATP13A2
8	Parkinson PARK14 (PLA2G6)	PLA2G6
9	Parkinson PARK15 (FBXO7)	FBXO7
	2.4.3. Sonstiges	
1	Glucose transport defect, blood-brain barrier (OMIM 606777)(OMIM 138140)	SLC2A1
2	Hallervorden-Spatz-Krankheit (PKAN) (PANK2)	PANK2
3	Tourette-Syndrom (SLITRK1, DRD2, SLC6A4, HTR2A, TH) SLITRK1, DRD2, SLC6A4, HTR2A, TH	
4	Tourette syndrome (TS) (OMIM 137580) (OMIM 609678)	SLITRK1
5	Tourette-Syndrom (DRD2)	DRD2
6	Tourette-Syndrom (SLC6A4)	SLC6A4
7	Tourette-Syndrom (HTR2A)	HTR2A
8	Tourette-Syndrom (TH)	TH
9	Wilson, Morbus (ATP7B)	ATP7B
	2.5. Ataxia and trinucleotide disease	
1	Arts-Syndrom (PRPS1)	PRPS1
2	Ataxia with oculomotor Apraxia 1 (AOA1)(APTX)	APTX
3	Ataxie mit okulomotorischer Apraxie 2 (AOA2)(SETX)	SETX
4	Ataxia-Teleangiectasia (ATM)	ATM
5	Chorea Huntington (HD)	HD
6	Chorea Huntington like-1 (PRNP)	PRNP
7	Chorea Huntington like-2 (JPH3)	JPH3
8	Denato-rubro-pallido-luysiale Atrophie (DRPLA)(ATN1)	ATN1
9	Episodic Ataxia, Type 2 (EA2) (OMIM 108500)(OMIM 601011)	CACNA1A
10	Familial Hemiplegic Migraine 1 (FHM1) (OMIM 141500)(OMIM 601011)	CACNA1A
11	Friedreich'sche Ataxie (FRDA) (FXN) (FL)	FXN

12	Friedreich'sche Ataxie (FRDA) (FXN) **(Seq.)**	FXN
13	Kennedy, Morbus (SBMA) (AR) (FL)	AR
14	Myotone Dystrophie 1 (DM1) (DMPK) (FL)	DMPK
15	Myotone Dystrophie 2 (PROMM / DM2) (ZNF9)(FL)	ZNF9
16	Myotone Dystrophie 2 (PROMM / DM2) (ZNF9)(Southern Blot)	ZNF9
17	Spinozerebelläre Ataxie Typ 1 (SCA1) (ATXN1)(FL)	ATXN1
18	Spinozerebelläre Ataxie Typ 2 (SCA2) (ATXN2)(FL)	ATXN2
19	Spinozerebelläre Ataxie Typ 3 (SCA3) (ATXN3)(FL)	ATXN3
20	Spinozerebelläre Ataxie Typ 4 (SCA4) (PLEKHG4)	PLEKHG4
21	Spinozerebelläre Ataxie Typ 5 (SCA5) (SPTBN2)	SPTBN2
22	Spinocerebellar Ataxia 6 (SCA6) (CACNA1A)(FL) (OMIM 183086) (OMIM 601011)	CACNA1A
23	Spinozerebelläre Ataxie Typ 7 (SCA7) (ATXN7)(FL)	ATXN7
24	Spinozerebelläre Ataxie Typ 8 (SCA8) (SCA8)(FL)	SCA8
25	Spinozerebelläre Ataxie Typ 10 (SCA10)(ATXN10) (FL)	ATXN10
26	Spinozerebelläre Ataxie Typ 11 (SCA11)(TTBK2)	TTBK2
27	Spinozerebelläre Ataxie Typ 12 (SCA12)(PPP2R2B) (FL)	PPP2R2B
28	Spinozerebelläre Ataxie Typ 13 (SCA13)(KCNC3) (Seq.)	KCNC3
29	Spinozerebelläre Ataxie Typ 14 (SCA14)(PRKCG)	PRKCG
30	Spinozerebelläre Ataxie Typ 17 (SCA17) (TBP)(FL)	TBP
31	Spinozerebelläre Ataxie Typ 27 (SCA27)(FGF14)	FGF14
32	All SCA by sequencing	
	2.6. Leukodystrophy and other CNS - disorders	
1	Acyl-CoA-Oxydase-Defizienz (ACOX1)	ACOX1
2	Adrenoleukodystrophie (X-ALD) (ABCD1)	ABCD1
3	Aicardi-Goutière-Syndrom 1 (AGS1) (TREX1)	TREX1
4	Aicardi-Goutière-Syndrom 2 (AGS2) (RNASEH2B)	RNASEH2B
5	Aicardi-Goutière-Syndrom 3 (AGS3) (RNASEH2C)	RNASEH2C

6	Aicardi-Goutière-Syndrom 4 (AGS4) (RNASEH2A)	RNASEH2A
7	Aicardi-Goutière-Syndrom 5 (AGS5) (TREX1)	TREX1
8	Alexander, Morbus (GFAP)	GFAP
9	CADASIL (OMIM 125310)(OMIM 600276)	NOTCH3
10	Canavan, Morbus (ASPA)	ASPA
11	Externe progressive Ophthalmoplegie 1 (POLG)	POLG
12	Externe progressive Ophthalmoplegie 3 (C10ORF2)	C10ORF2
13	Fukosidose* (α-L-Fukosidase Def. - FUCA1)	FUCA1
14	Giant Axonal Neuropathie (GAN)	GAN
15	GM1-Gangliosidosis, type I (OMIM 230500)* (β- Galaktosidase Def. - GLB1)(OMIM 611458)	GLB1
16	Congenital disorder of glycosylation, type Ia (CDG1A) (OMIM 212065) * (OMIM 601785)	PMM2
17	Congenital disorder of glycosylation, type Ib (CDG1B) (OMIM 602579) (OMIM 154550)	* MPI
18	Glykosylierungsdefekt, Typ 1C (CDG1C) (ALG6)	* ALG6
19	Glykosylierungsdefekt, Typ 1D (CDG1D) (ALG3)	* ALG3
20	Glykosylierungsdefekt, Typ 1E (CDG1E) (DPM1)	* DPM1
21	Glykosylierungsdefekt, Typ 1F (CDG1F)(MPDU1)	* MPDU1
22	Glykosylierungsdefekt, Typ 1G (CDG1G)(ALG12)	* ALG12
23	Glykosylierungsdefekt, Typ 1H (CDG1H) (ALG8)	* ALG8
24	Glykosylierungsdefekt, Typ 1I (CDG1I) (ALG2)	* ALG2
25	Glykosylierungsdefekt, Typ 1J (CDG1J) (DPAGT1)	* DPAGT1
26	Glykosylierungsdefekt, Typ 1K (CDG1K) (ALG1)	* ALG1
27	Glykosylierungsdefekt, Typ 1L (CDG1L) (ALG9)	* ALG9
28	Glykosylierungsdefekt, Typ 1M (CDG1M)(TMEM15)	* TMEM15
29	Glykosylierungsdefekt, Typ 1N (CDG1N) (RFT1)	* RFT1
30	Glykosylierungsdefekt, Typ 2A (CDG2A)(MGAT2)	* MGAT2
31	Glykosylierungsdefekt, Typ 2C (CDG2C)(SLC35C1)	* SLC35C1
32	Glykosylierungsdefekt, Typ 2D (CDG2D)(B4GALT1)	* B4GALT1

33	Glykosylierungsdefekt, Typ 2E (CDG2E)(COG7)	* COG7
34	Glykosylierungsdefekt, Typ 2F (CDG2F)(SLC35A1)	* SLC35A1
35	Glykosylierungsdefekt, Typ 2G (CDG2G) (COG1)	* COG1
36	Glykosylierungsdefekt, Typ 2H (CDG2H) (COG8)	* COG8
37	Krabbe Disease (OMIM 245200) (Genetic)*(OMIM 606890)	GALC
38	Leukoencephalopathies mit Beteiligung von Hirnstamm/Rückenmark und Laktaterhöhung (LBSL) (DARS2)	DARS2
39	Leucoencephalopathies Vanishing White Matter (VWM) (EIF2B1, EIF2B2, EIF2B3, EIF2B4,EIF2B5)EIF2B1, EIF2B2,EIF2B3, EIF2B4,EIF2B5	
40	Megalenzephale Cystic Leukoencephalopathies (MLC1) MLC1	
41	MERRF (Mitochondrial, MTTK, MTTL1, MTTH,MTTS1, MTTS2, MTTF)Mitochondrial, MTTK,MTTL1, MTTH,MTTS1, MTTS2,MTTF	
42	Metachromatic Leukodystrophy (MLD) *(Arylsulphatase A Def. - ARSA)	ARSA
43	Metachromatic Leukodystrophy, Saposin B Defizienz * (PSAP)	PSAP
44	Multiple-Sulfatase-Defizienz (MSD) (SUMF1)	SUMF1
45	Niemann-Pick Disease, type C1 (NPC1) (OMIM 257220) * (NPC1)(OMIM 607623)	NPC1
46	Osteodysplasie, lipomembranöse polyzystischesklerosierende Leukoencephalopathies (PLOSL)(TYROBP; TREM2)	TYROBP; TREM2
47	Osteodysplasie, lipomembranöse polyzystischesklerosierende Leukoencephalopathies (PLOSL)(TYROBP; TREM2)	TYROBP
48	Osteodysplasie, lipomembranöse polyzystischesklerosierende Leukoencephalopathies (PLOSL)(TYROBP; TREM2)	TREM2
49	Pelizaeus-Merzbacher-Krankheit (PMD) (PLP1)	PLP1
50	Pelizaeus-Merzbacher-like-Krankheit (PMLD1)(GJA12)	GJA12
51	Phosphoglycerate Dehydrogenase Defizienz (PHGDH)	PHGDH
52	Sialinsäure-Speicherkrankheit (SLC17A5)	SLC17A5
53	Sjogren-Larsson Syndrom (SLS) (ALDH3A2)	ALDH3A2

54	Tuberöse Hirnsklerose (TSC1, TSC2)	TSC1, TSC2
55	Tuberöse Hirnsklerose (TSC1)	TSC1
56	Tuberous Sclerosis-1 (OMIM 191100)(OMIM 191092)	TSC2
57	Waardenburg Syndrom / Hirschsprung Krankheit (PCWH) (SOX10)	SOX10
58	Zellweger Syndrom (ZS) (PEX1, PEX2, PEX3,PEX5, PEX6, PEX12, PEX14, PEX26)PEX1, PEX2, PEX3,PEX5, PEX6, PEX12,PEX14, PEX26	
59	Zerebrotendinöse Xanthomatose (CYP27A1)	CYP27A1
	2.7. Muscle disorders and familiar ALSdiseases	
1	Amyotrophe Lateralsklerose 2 (ALS2) (Alsin)	Alsin
2	Amyotrophe Lateralsklerose 4 (ALS4) (SETX)	SETX
3	Amyotrophe Lateralsklerose 8 (ALS8) (VAPB)	VAPB
4	Amyotrophe Lateralsklerose 10 (ALS10)(TARDBP)	TARDBP
5	Limb-Girdle Muscular Dystrophy, Type 1A, AD (LGMD1A) (OMIM 159000) (OMIM 604103)	MYOT
6	Spheroid Body Myopathy (OMIM 182920) (OMIM 604103)	MYOT
7	Myotilinopathy (OMIM 609200) (OMIM 604103)	MYOT
8	Gliedergürtelmuskeldystrophie, AD, Typ 1B (LGMD1B) (Lamin A/C, LMNA)	Lamin A/C, LMNA
9	Gliedergürtelmuskeldystrophie, AD, Typ 1C (LGMD1C) (Caveolin 3, CAV3)	Caveolin 3, CAV3
10	Gliedergürtelmuskeldystrophie, AR, Typ 2A (LGMD2A) (Calpain 3, CAPN3)	Calpain 3, CAPN3
11	Gliedergürtelmuskeldystrophie, AR, Typ 2B (LGMD2B) (Dysferlin, DYSF)	Dysferlin, DYSF
12	Gliedergürtelmuskeldystrophie, AR, Typ 2C (LGMD2C) (Sarcoglycan, Gamma, SGCG)	Sarcoglycan, Gamma,SGCG
13	Gliedergürtelmuskeldystrophie, AR, Typ 2D (LGMD2D) (Sarcoglycan, Alpha, SGCA)	Sarcoglycan, Alpha,SGCA
14	Gliedergürtelmuskeldystrophie, AR, Typ 2E (LGMD2E) (Sarcoglycan, Beta, SGCB)	Sarcoglycan, Beta, SGCB
15	Gliedergürtelmuskeldystrophie, AR, Typ 2F (LGMD2F) (Sarcoglycan, Delta, SGCD)	Sarcoglycan, Delta,SGCD

16	Gliedergürtelmuskeldystrophie, AR, Typ 2G (LGMD2G) (Titin-Cap, TCAP)	TitinCap, TCAP
17	Muscular dystrophy, limb-girdle, type 2H (LGMD2H) (OMIM 254110)(OMIM 602290)	TRIM32
18	Gliedergürtelmuskeldystrophie, AR, Typ 2I (LGMD2I) (Fukutin-Related Protein, FKRP)	Fukutin Related Protein, FKRP
19	Gliedergürtelmuskeldystrophie, AR, Typ 2J (LGMD2J) (Titin, TTN)	Titin, TTN
20	Gliedergürtelmuskeldystrophie, AR, Typ 2K (LGMD2K) (Protein O-Mannosyltransferase 1, POMT1)Protein Omannosyltransferas e 1,	POMT1
21	Muskeldystrophie, fazio-skapulo-humeral (FSHD1A) (FRG1) FRG1 Muskeldystrophie, kongenital, merosin-positiv (FKRP)	FKRP
22	Muskeldystrophie, okulopharyngeal (OPMD)(PABPN1) (FL)	PABPN1
23	Muskelatrophie, proximal, spinal, Typ 1, 2, 3, 4 (SMA I, II, III, IV) (Werdnig-Hofmann SMN1,BIRC1 [NAIP])WerdnigHofmann	SMN1, BIRC1 [NAIP]
24	Muskeldystrophie, Typ Fukuyama (FCMD)(FKTN)	FKTN
	2.8. Dementia	
1	Alzheimer-Krankheit, familiäre (FAD1) (APP)	APP
2	Alzheimer-Krankheit, familiäre (FAD3) (PSEN1)	PSEN1
3	Alzheimer-Krankheit, familiäre (FAD4) (PSEN2)	PSEN2
4	Creutzfeldt-Jakob-Krankheit (CJD) (PRNP)	PRNP
5	Gerstmann-Sträussler-Scheinker-Syndrom (GSD) (PRNP)	PRNP
6	Fatale familiäre Insomnie (FFI) (PRNP)	PRNP
7	Frontotemporale Demenz (GRN, MAPT)	GRN, MAPT
8	Frontotemporal lobar degeneration with ubiquitin-positive inclusions (FTLDU) (OMIM 607485) (OMIM 138945)	GRN
9	Frontotemporal Dementia (FTD) (OMIM 600274) (OMIM 157140)	MAPT
10	Prader-Willi-Syndrom / Angelmann-Syndrom (Chr.region 15q11-q13, Methylationtest;UBE3A)Chr.region15q11q13,	Methylationtest;UBE3A
	3. Ophthalmological disorders	

1	Alström-Syndrom (ALMS1)	ALMS1
2	Aniridie, Typ 2 (AN2) (PAX6)	PAX6
3	Best-Krankheit (VMD) (BEST1)	BEST1
4	Fundus Favimaculatus (FFM) (ABCA4, PRPH2)	ABCA4, PRPH2
5	Fundus Favimaculatus (FFM) (ABCA4)	ABCA4
6	Fundus Favimaculatus (FFM) (PRPH2)	PRPH2
7	Glaukom, hereditäres (CYP1B1)	CYP1B1
8	Hornhautdystrophie, Avellino Typ (CDA)(TGFB1)	TGFB1
9	Hornhautdystrophie, Bowman Layer, Typ 1 (CDB1) (TGFB1)	TGFB1
10	Lowe Syndrom (OCRL)	OCRL
11	Mikrophthalmie, isoliert, Typ 3 (MCOP3) (RAX)	RAX
12	Mikrophthalmie, syndromal, Typ 3 (MCOPS3)(SOX2)	SOX2
13	Netzhautdystrophie Typ Bothnia (RLBP1)	RLBP1
14	Netzhautdystrophie Typ Bothnia-1A (GLC1A)(MYOC)	MYOC
15	Netzhautdystrophie Typ Bothnia-1E (GLC1E)(OPTN)	OPTN
16	Netzhautdystrophie Typ Bothnia-1G (GLC1G)(WDR36)	WDR36
17	Optikus-Neuropathie Typ Leber (LOHN)(Mitochondrial, MTND4, MTND4L, MTND1,MTND, MTCO3, MTND5, MTND2, MTCO1,MTATP6)	
18	Retinale Dystrophie (AD) – Screening (RHO,RGR, PDE6B, GUCY2D, HPRP3, PRPF31, PRPF8)	
19	Retinale Dystrophie (AR) – Screening (RPE65,LRAT, ABCA4, RHO, PDE6A, PDE6B, CNGA1,CNGB1, SAG, GUCY2D, MERTK, CRB1, USH2A)	
20	Rieger-Syndrom (PITX2)	PITX2
21	Stargardt-Krankheit 1 (STGD1) (ABCA4; CNGB3)	ABCA4; CNGB3
22	Stargardt-Krankheit 1 (STGD1) (ABCA4)	ABCA4
23	Stargardt-Krankheit 1 (STGD1) (ABCA4)	ABCA4
24	Stargardt-Krankheit 1 (STGD1) (CNGB3)	CNGB3
25	Stargardt-Krankheit 3 (STGD3) (ELOVL4)	ELOVL4

26	Stickler-Syndrom, AR (COL9A1)	COL9A1
27	Stickler-Syndrom 1 (STL1) (COL2A1)	COL2A1
28	Stickler-Syndrom 2 (STL2) (COL11A1)	COL11A1
29	Stickler-Syndrom 3 (STL3) (COL11A2)	COL11A2
30	Wagner-Krankheit (WGN1) (CSPG2)	CSPG2
31	Weill-Marchesani-Syndrom, AD (FBN1)	FBN1
32	Weill-Marchesani-Syndrom, AR (ADAMTS10)	ADAMTS10
33	Wolfram-Syndrom 1 (WFS1)	WFS1
34	Wolfram-Syndrom 2 (ZCD2)	ZCD2
	4. ENS-diseases	
1	Alport-Syndrom, AR (COL4A3, COL4A4)	COL4A3, COL4A4
2	Alport-Syndrom, X-chromosomal (COL4A5)	COL4A5
3	Taubheit - Dystonie - Optikusatrophie (TIMM8A)	TIMM8A
4	Wolfram-Syndrom 1 (WFS1)	WFS1
5	Wolfram-Syndrom 2 (ZCD2)	ZCD2
	Nicht-syndromale, autosomal recessive Schwerhörigkeit (AR) Deafness	
1	Schwerhörigkeit, Typ 1 (DFNB1) (GJB2; GJB6)	GJB2; GJB6
2	Schwerhörigkeit, Typ 1 (DFNB1)	(GJB2) GJB2
3	Deafness, autosomal recessive 1B (DFNB1B)(OMIM 612645) (OMIM 604418)	GJB6
4	Schwerhörigkeit, Typ 3 (DFNB3) (MYO15A)	MYO15A
5	Schwerhörigkeit, Typ 6 (DFNB6) (TMIE)	TMIE
6	Schwerhörigkeit, Typ 7 (DFNB7, DFNB11)(TMC1)	TMC1
7	Schwerhörigkeit, Typ 9 (DFNB9) (OTOF)	OTOF
8	Schwerhörigkeit, Typ 16 (DFNB16) (STRC)	STRC
9	Schwerhörigkeit, Typ 18 (DFNB18) (USH1C)	USH1C
10	Schwerhörigkeit, Typ 28 (DFNB28) (TRIOBP)	TRIOBP

11	Schwerhörigkeit, Typ 30 (DFNB30) (MYO3A)	MYO3A
	Nicht-syndromale, autosomal dominant Deafness - Schwerhörigkeit (AD)	
1	Schwerhörigkeit, Typ 1 (DFNA1) (DIAPH1)	DIAPH1
2	Schwerhörigkeit, Typ 2 (DFNA2) (GJB3, KCNQ4)	GJB3, KCNQ4
3	Schwerhörigkeit, Typ 2 (DFNA2) (GJB3)	GJB3
4	Schwerhörigkeit, Typ 2 (DFNA2) (KCNQ4)	KCNQ4
5	Schwerhörigkeit, Typ 3 (DFNA3) (GJB2, GJB6)	GJB2, GJB6
6	Schwerhörigkeit, Typ 3 (DFNA3) (GJB2)	GJB2
7	Deafness, autosomal dominant 3B (DFNA3B)(OMIM 612643) (OMIM 604418)	GJB6
8	Ectodermal dysplasia, hidrotic, autosomaldominant (OMIM 129500)(OMIM 604418)	GJB6
9	Schwerhörigkeit, Typ 4 (DFNA4) (MYH14)	MYH14
10	Schwerhörigkeit, Typ 5 (DFNA5) (DFNA5)	DFNA5
11	Schwerhörigkeit, Typ 6 (DFNA6, DFNA14,DFNA38) (WFS1)	WFS1
12	Schwerhörigkeit, Typ 9 (DFNA9) (COCH)	COCH
13	Schwerhörigkeit, Typ 10 (DFNA10) (EYA4)	EYA4
14	Schwerhörigkeit, Typ 11 (DFNA11) (MYO7A)	MYO7A
15	Schwerhörigkeit, Typ 12 (DFNA12, DFNA8)(TECTA)	TECTA
16	Schwerhörigkeit, Typ 13 (DFNA13) (COL11A2)	COL11A2
17	Schwerhörigkeit, Typ 15 (DFNA15) (POU4F3)	POU4F3
18	Schwerhörigkeit, Typ 17 (DFNA17) (MYH9)	MYH9
19	Schwerhörigkeit, Typ 22 (DFNA22) (MYO6)	MYO6
20	Schwerhörigkeit, Typ 23 (DFNA23) (SIX1)	SIX1
21	Schwerhörigkeit, Typ 36 (DFNA36) (TMC1)	TMC1
22	Schwerhörigkeit, Typ 44 (DFNA44) (CCDC50)	CCDC50
	5. Bone diseases	
1	Arthrogrypose, distal, Typ 1 (TPM2)	TPM2
2	Arthrogrypose, distal, Typ 2A (MYH3)	MYH3

3	Arthrogrypose, distal, Typ 2B (TNNT3, TNNI2,MYH3)	TNNT3, TNNI2, MYH3
4	Arthrogrypose, distal, Typ 2B (TNNT3)	TNNT3
5	Arthrogrypose, distal, Typ 2B (TNNI2)	TNNI2
6	Arthrogrypose, distal, Typ 2B (MYH3)	MYH3
7	MPS-Screening* (IDUA, IDS, SGSH, NAGLU,HGSNAT, GNA, GALNS, GLB1, ARSB, GUSB,HYAL1)	
8	Mukopolysaccharidose Typ 4a (MPS IVa, Morquio Syndrom A)* (GALNS)	GALNS
9	Osteogenesis imperfecta, type I (OMIM 166200)(OMIM 120150)	COL1A1
10	Caffey Disease (OMIM 114000)(OMIM 120150)	COL1A1
11	Osteogenesis imperfecta, type IIA (OMIM 166210)(OMIM 120150)	COL1A1
12	Osteogenesis imperfecta, type III (OMIM 259420)	COL1A1
13	Osteogenesis imperfecta, type IV (OMIM 166220)	COL1A1
14	Ehlers-Danlos Syndrome, type VII, autosomal dominant (OMIM 130060) (OMIM 120150)	COL1A1
15	Osteogenesis imperfecta, type IV (OMIM 166220) (OMIM 120150)	COL1A1
16	Ehlers-Danlos Syndrome, type VII, autosomal dominant (OMIM 130060)(OMIM 120150)	COL1A1
17	Osteogenesis imperfecta, Typ 2 A (COL1A2)	COL1A2
	6. Cardiololgical diseases	
1	Danon-Disorder, X-chromosomal Cardiomyopathy and Myopathy (LAMP2)	LAMP2
2	Cardiomyopathy, dilated (CMD) (MYBPC3)	MYBPC3
3	Cardiomyopathy, dilated, 1A (CMD1A)(LMNA)	LMNA
4	Cardiomyopathy, dilated, 1D (CMD1D)(TNNT2)	TNNT2
5	Cardiomyopathy, dilated, 1G (CMD1G) (TTN)	TTN
6	Cardiomyopathy, dilated, 1R (CMD1R)(ACTC1)	ACTC1
7	Cardiomyopathy, dilated, 1S (CMD1S)(MYH7)	MYH7
8	Cardiomyopathy, dilated, 1Y (CMD1Y) (TPM1)	TPM1
9	Cardiomyopathy, dilated, 2A (CMD2A) (OMIM 611880) (OMIM 191044)	TNNI3
10	Cardiomyopathy, familial hypertrophic, 1 (CMH1) (MYH7)	MYH7
11	Cardiomyopathy, familial hypertrophic, 2 (CMH2) (TNNT2)	TNNT2

12	Cardiomyopathy, familial hypertrophic, 3 (CMH3) (TPM1)	TPM1
13	Cardiomyopathy, familial hypertrophic, 4 (CMH4) (MYBPC3)	MYBPC3
14	Cardiomyopathy, familial hypertrophic, 7, included (CMH7, included) (OMIM 191044) (OMIM 191044)	TNNI3
15	Cardiomyopathy, familial restrictive, 1 (RCM1)(OMIM 115210) (OMIM 191044)	TNNI3
16	Cardiomyopathy, familial hypertrophic, 8 (CMH8) (MYL3)	MYL3
17	Cardiomyopathy, familial hypertrophic , 9 (CMH9) (TTN)	TTN
18	Cardiomyopathy, familial hypertrophic, 10 (CMH10) (MYL2)	MYL2
19	Cardiomyopathy, familial hypertrophic, 11 (CMH11) (ACTC1)	ACTC1
20	Cardiomyopathy, familial hypertrophic, 12 (CMH12) (CSRP3)	CSRP3
21	Cardiomyopathy, familial hypertrophic, mit Wolf-Parkinson-White Syndrom (PRKAG2)	PRKAG2
	7. Nephrological disorders	
1	Alport Syndrom, AR (COL4A3, COL4A4)	COL4A3, COL4A4
2	Alport Syndrom, AR (COL4A3)	COL4A3
3	Alport Syndrom, AR (COL4A4)	COL4A4
4	Alport Syndrom X-chromosomal (COL4A5)	COL4A5
5	Bardet-Biedl-Syndrom (BBS1)	BBS1
6	Cystinose (CTNS)	CTNS
7	Diabetes insipidus, nephrogen, X-chromosomal (AVPR2)	AVPR2
8	Diabetes insipidus, nephrogen, autosomal (AQP2)	AQP2
9	Epstein-Syndrom (MYH9)	MYH9
10	Gitelman-Syndrom (SLC12A3)	SLC12A3
11	Kallmann-Syndrom (KAL1)	KAL1
12	Liddle Syndrom (SCNN1B, SCNN1G)	SCNN1B, SCNN1G
13	Liddle Syndrom (SCNN1B)	SCNN1B
14	Liddle Syndrom (SCNN1G)	SCNN1G
15	Lowe Syndrom (OCRL)	OCRL

16	Meckel-Syndrom Typ 1 (MKS1)	MKS1
17	Meckel-Syndrom Typ 3 (TMEM67)	TMEM67
18	Meckel-Syndrom Typ 4 (CEP290)	CEP290
19	Nephronophthise, AD (UMOD)	UMOD
20	Nephronophthise, AR, Typ 1 (NPHP1)	NPHP1
21	Nephronophthise, AR, Typ 2 (INVS)	INVS
22	Nephrotic Syndrom, familiär, idiopathic, steroid-resistant, AD (ACTN4)	ACTN4
23	Nephrotic Syndrom, congenital, Finnish Type (NPHS1)	NPHS1
24	Nephrotic Syndrom, familiär, idiopathic, steroid-resistant, AR (NPHS2)	NPHS2
25	Nierenkrankheit (Kidney Dis.), polycystic, AD (PKD1, PKD2)	PKD1, PKD2
26	Nierenkrankheit,(Kidney Dis.) polycystic, AD (PKD1)	PKD1
27	Nierenkrankheit,(Kidney Dis.) polycystic, AD (PKD2)	PKD2
28	Nierenkrankheit,(Kidney Dis.) polycystic, AR (PKHD1)	PKDH1
29	Nierenzellkarzinom (Renal Carcinoma), familial papillary (MET)	MET
30	Rickets, Vitamin D-resistant, AD (FGF23)	FGF23
31	Rickets, Vitamin D-resistant, AR (DMP1)	DMP1
32	Rickets, Vitamin D-resistant, X-chromosomal (PHEX)	PHEX
33	Vesikoureterale Re%ux 2 (ROBO)	ROBO2
34	Cystinuria (SLC3A1, SLC7A9)	SLC3A1, SLC7A9
35	Cystinuria (SLC3A1)	SLC3A1
36	Cystinuria (SLC7A9)	SLC7A9
	8. Oncological disorders	
1	Alagille-Syndrom 1 (JAG1)	JAG1
2	Alagille-Syndrom 2 (NOTCH2)	NOTCH2
3	Alpha-1-Antitrypsin-Defi zienz (SERPINA 1)	SERPINA
4	APC-assoziierte Polyposis (FAP) (APC)	APC
5	Beckwith-Wiedemann-Syndrom (CDKN1C,NSD1)(OMIM 606681)	CDKN1C, NSD1

6	Beckwith-Wiedemann-Syndrom (CDKN1C)	CDKN1C
7	Beckwith-Wiedemann Syndrome (BWS) (OMIM 130650)(OMIM 606681)	NSD1
8	Weaver Syndrome (WSS) (OMIM 277590) (OMIM 606681)	NSD1
9	Breast Cancer, familial (BRCA1, BRCA2)	BRCA1, BRCA2
10	Breast Cancer, familial (BRCA1)	BRCA1
11	Breast Cancer, familial (BRCA2)	BRCA2
12	Ki-RAS Gen Sequenzierung (KRAS)	KRAS
13	Kolonkarzinom, Typ 1, hereditär, nicht-polypös (HNPCC1) (MSH2)	MSH2
14	Kolonkarzinom, Typ 2, hereditär, nicht-polypös (HNPCC2) (MLH1)	MLH1
15	Kolonkarzinom, Typ 3, hereditär, nicht-polypös (HNPCC3) (PMS1)	PMS1
16	Kolonkarzinom, Typ 4, hereditär, nicht-polypös (HNPCC4) (PMS2)	PMS2
17	Kolonkarzinom, Typ 5, hereditär, nicht-polypös (HNPCC5) (MSH6)	MSH6
18	Lymphoproliferative Erkrankung, X chromosomal (SH2D1A)	SH2D1A
19	Magenkarzinom, familiär (CDH1)	CDH1
20	Multiple Endocrine Neoplasie Typ 1 (MEN1)	MEN1
21	Multiple Endocrine Neoplasie Typ 2 (RET)	RET
22	MYH-assoziierte Polyposis (MUTYH)	MUTYH
23	Nierenzellkarzinom, familiär, papillär (MET)	MET
24	Pancreatitis, hereditary chronic (HCP) (PRSS1,SPINK1, CFTR, CTRC)	PRSS1, SPINK1, CFTR,CTRC
25	Pancreatitis, hereditary chronic (HCP) (PRSS1)	PRSS1
26	Pancreatitis, hereditary chronic (HCP)(SPINK1)	SPINK1
27	Pancreatitis, hereditary chronic (HCP) (CFTR,CTRC)	CFTR
28	Pancreatitis, hereditary chronic (HCP) (CTRC)	CTRC
29	Peutz-Jeghers Syndrom (STK11)	STK11
30	Prostate Cancer, hereditary (ELAC2, RNASEL,BRCA2, SRD5A2)	ELAC2, RNASEL,BRCA2,

		SRD5A2
31	Prostate Cancer, hereditary (ELAC2)	ELAC2
32	Prostate Cancer, hereditary (RNASEL)	RNASEL
33	Prostate Cancer, hereditary (BRCA2)	BRCA2
34	Prostate Cancer, hereditary (SRD5A2)	SRD5A2
35	Shwachman-Diamond-Syndrom (SDS) (SBDS)	SBDS
36	Sotos Syndrome (OMIM 117550)(OMIM 606681)	NSD1
37	Stromatumor, gastrointestinal (GIST) (KIT)	KIT
38	Stromatumor, gastrointestinal (GIST) (PDGFRA)	PDGFRA
	9. others	
1	Nemaline Myopathy 3 (NEM3) (ACTA1)	ACTA1
2	Brugada Syndrom 1 SCN5A	SCN5A
3	DUCHENNE MUSCULAR DYSTROPHY and BECKER MUSCULAR DYSTROPHY DMD	
4	Neuraminidase Deficiency	NEU1
5	Congenital stationary Nightblindness Typ 2A (CACNA1F)	CACNA1F
6	Kallmann-Syndrom 3 (PROKR2)	PROKR2
7	Kallmann-Syndrom 4 (PROK2)	PROK2
8	Hypogonadotropic isolated Hypogonadism (GPR54)	GPR54
9	McKusick-Kaufman-Syndrom (MKKS)	MKKS
10	DIARRHEA 2, WITH MICROVILLOUS ATROPHY;	DIAR2 MYO5B
11	**Retinal Dystrophy Screen (AR)**	
a	RPE65	RPE65
b	LRAT	LRAT
c	ABCA4	ABCA4
d	RHO	RHO
e	PDE6A	PDE6A
f	PDE6B	PDE6B

g	CNGA1	CNGA1
h	CNGAB1	CNGAB1
i	SAG	SAG
j	GUCY2D	GUCY2D
k	MERTK	MERTK
l	CRB1	CRB1
m	USH2A	USH2A
12	**Retinal Dystrophy Screen (AD)**	
a	RHO	RHO
b	RGR	RGR
c	PDE6B	PDE6B
d	GUCY2D	GUCY2D
e	HPRP3	HPRP3
f	PRPF31	PRPF31
g	PRPF8	PRPF8
13	**CMT autosomal recessive**	
a	CMT2B1 (LMNA)	LMNA
b	CMT2K / CMT4A (GDAP1)	GDAP1
c	CMT4B1 (MTMR2)	MTMR2
d	CMT4B2 (SBF2)	SBF2
e	CMT4C (SH3TC2)	SH3TC2
f	CMT4D (NDRG1)	NDRG1
g	CMT4F (PRX)	PRX

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).

For more information please log on to www.metabolicerror.com

Navi Mumbai Institute of Research In Mental And Neurological Handicap

Dr. Anil B. Jalan,
MD DCH MCPS
With effect from 1st April 2010 (Revised).

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

For more information please log on to www.metabolicerror.com

Navi Mumbai Institute of Research In Mental And Neurological Handicap

Dr. Anil B. Jalan,
MD DCH MCPS

With effect from 1st April 2010 (Revised).

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