Tests performed through our international collaboration

Molecular Studies for Inborn Errors of Metabolism

A	Inborn Errors of Metabolism	Defective Gene	
1	Acyl - Co A oxidase deficiency	ACOX 1	
2	Argininosuccinate lyase deficiency	ASL	
3	Aromatic L – amino acid decarboxylase (AADC) Deficiency	DDC	
4	Carnitine – acylcarnitine translocase (CACT) Deficiency	CACT	
5	Primary (systemic) carnitine deficiency	OCTN2	
6	Carnitine palmitoyltransferase I (CPT1)	CPT1A	
7	Carnitine palmitoyltransferase 2 (CPT2)	CPT2	
8	CHILD Syndrome	NSDHL	
9	Conradi – Hunermann – Happle syndrome (CDPX2)	EBP	
10	D – Bifunctional protein (DBP) Deficiency	DBP, MFE2	
11	Desmosterolosis	DHCR24	
12	Dihydropyrimidinase (DHP) Deficiency	DPYS	
13	Dihydropyrimidine dehydrogenase (DPD) Deficiency	DPYD	
14	Ethylmalonaciduria (Ethylmalonic encephalopathy)	ETHE1	
15	Fructose intolerance, hereditary	ALDOB	
16	Galactosemia, classic	GALT	
17	Galactokinase deficiency	GALK1	
18	Glutaric aciduria type I (Glutaryl – Co A dehydrogenase deficiency	GCDH	
19	Glycogen storage disease 0	GYS2	

20	Greenberg skeletal dysplasia (Sterol – delta 14 reductase deficiency)	LBR	
21	GTP cyclohydrolase I deficiency	GCH1	
22	Hydroxyacyl – Co A dehydrogenase deficiency (2 – Methyl – 3 – hydroxybutyryl – CoA dehydrogenase deficiency)	HADH2	
23	Hyper Ig D Syndrome (Mevalonate Kinase deficiency)	MVK	
24	Hyperoxaluria type I	AGXT	
25	Isovaleric acidemia	IVD	
26	Lathosterolosis	SC5DL	
27	3 – methylglutaconicaciduria type I (3 – methylglutaconyl – CoA hydratase deficiency)	AUH	
28	Medium – chain – acyl – Co A dehydrogenase (MCAD) Deficiency	ACADM	
29	Mevalonaciduria (Mevalonate Kinase deficiency)	MVK	
30	Mitochondrial DNA Depletion Syndrome, Myopathic Form	SUCLA2	
31	Mitochondrial trifunctional protein (MTP) Deficiency	HADHA	
32	Mitochondrial trifunction protein (MPT) Deficiency	HADHB	
33	Multiple acyl – Co A dehydrogenase deficiency	ETFB	
34	Multiple acyl – Co A dehydrogenase deficiency	ETFDH	
35	Refsum Disease	PHYH	
36	Rhizomelic chondrodysplasia punctata (RCDP) Type 1	PEX7	
37	Rhizomelic chondrodysplasia punctata (RCDP) type 2 (DHAPAT Deficiency)	DHAPAT	
38	Rhizomelic chondrodysplasia punctata (RCDP) Type 3 (alkyl – DHAP synthase deficiency)	AGPS	
39	Short – chain acyl – CoA dehydrogenase (SCAD) Deficiency	ACADS	
40	Sjogren – Larsson syndrome	ALDH10	
41	Smith – Lemli – Opitz Syndrome	DHCR7	

42	Tyrosine hydroxylase deficiency	TH	
43	Very long – chain acyl – Co A dehydrogenase (VLCAD) Deficiency	ACADVL	
44	X – linked adrenoleukodystrophy	X –ALD	
45	Zellweger Syndrome Neonatal adrenoleukodystrophy Infantile Refsum disease	PEX1	
46	Zellweger Syndrome Neonatal adrenoleukodystrophy Infantile Refsum disease	PEX2	
47	Zellweger Syndrome Neonatal adrenoleukodystrophy Infantile Refsum disease	PEX3	
48	Zellweger Syndrome Neonatal adrenoleukodystrophy Infantile Refsum disease	PEX5	
49	Zellweger Syndrome Neonatal adrenoleukodystrophy Infantile Refsum disease	PEX6	
50	Zellweger Syndrome Neonatal adrenoleukodystrophy Infantile Refsum disease	PEX10	
51	Zellweger Syndrome Neonatal adrenoleukodystrophy Infantile Refsum disease	PEX12	
52	Zellweger Syndrome Neonatal adrenoleukodystrophy Infantile Refsum disease	PEX13	
53	Zellweger Syndrome Neonatal adrenoleukodystrophy Infantile Refsum disease	PEX14	
54	Zellweger Syndrome Neonatal adrenoleukodystrophy Infantile Refsum disease	PEX16	
55	Zellweger Syndrome Neonatal adrenoleukodystrophy Infantile Refsum disease	PEX19	
56	Zellweger Syndrome Neonatal adrenoleukodystrophy Infantile Refsum disease	PEX26	
57	Zellweger Syndrome Neonatal adrenoleukodystrophy Infantile Refsum disease	Complementation testing – Skin fibroblasts	