Disorder Panels (Apr 2021)

			Hb+FS+	FS+	FS	MS	BioX*	Hb+	ACMG
			65	61	60	55	7	4	
	Disorders Screened by Tandem Mass Spectrometry								
A	**Fatty Acid Oxidation Disorders**		**12**	**12**	**12**	**12**			
1	Carnitine / Acylcarnitine Translocase Deficiency	CACT	•	•	•	•			•
2	3-Hydroxy Long Chain Acyl-CoA Dehydrogenase Deficiency	LCHAD	•	•	•	•			•
3	Medium Chain Acyl-CoA Dehydrogenase Deficiency	MCAD	•	•	•	•			•
4	Neonatal Carnitine Palmitoyl Transferase Deficiency Type II	CPT-II	•	•	•	•			•
5	Very Long Chain Acyl-CoA Dehydrogenase Deficiency	VLCAD	•	•	•	•			•
6	Carnitine Palmitoyl Transferase Deficiency Type IA[1]	CPT-IA	•	•	•	•			•
7	2,4-Dienoyl-CoA Reductase Deficiency1	DE-RED	•	•	•	•			•
8	Multiple Acyl-CoA Dehydrogenase Deficiency	GA-II	•	•	•	•			•
9	Short-chain Acyl-CoA Dehydrogenase Deficiency	SCAD	•	•	•	•			•
10	Trifunctional Protein Deficiency	TFP	•	•	•	•			•
11	Short chain Hydroxy Acyl-CoA Dehydrogenase Deficiency	SCHAD	•	•	•	•			•
12	Medium Chain Ketoacyl-CoA Thiolase Deficiency	MCKAT	•	•	•	•			
B	**Organic Acid Disorders**		**16**	**16**	**16**	**16**			
13	3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency	HMG	•	•	•	•			•
14	Glutaric Acidemia Type I	GA-I	•	•	•	•			•
15	Isobutyryl-CoA Dehydrogenase Deficiency	IBG	•	•	•	•			•
16	Isovaleric Acidemia	IVA	•	•	•	•			•
17	2-Methylbutyryl-CoA Dehydrogenase Deficiency	2MBG	•	•	•	•			•
18	3-Methylcrotonyl-CoA Carboxylase Deficiency	3MCC	•	•	•	•			•
19	3-Methylglutaconyl-CoA Hydratase Deficiency	3MGA	•	•	•	•			•
20	2-Methyl-3-Hydroxybutyric Aciduria	2M3HBA	•	•	•	•			
	Methylmalonic Acidemias	MMA	•	•	•	•			•
21	Methymalonyl-CoA Mutase Deficiency	MUT	•	•	•	•			•
22	Methylmalonic Acidemia (Cobalamin disorders)	CBL A,B							
23	Methylmalonic Acidemia with Homocystinuria	CBL C,D	•	•	•	•			•
24	Maternal Vitamin B12 Deficiency		•	•	•	•			
25	Mitochondrial Acetoacetyl-CoA Thiolase Deficiency	BKT	•	•	•	•			•
26	Propionic Acidemia	PROP	•	•	•	•			•
27	Multiple CoA Carboxylase Deficiency	MCD	•	•	•	•			•
28	Malonic Aciduria	MAL	•	•	•	•			•
C	**Amino Acid Disorders**		**21**	**21**	**21**	**21**			
29	Argininemia	ARG	•	•	•	•			•
30	Argininosuccinic Aciduria	ASA Lyase	•	•	•	•			•
31	5-Oxoprolinuria1	5-OXO	•	•	•	•			•
32	Carbamoylphosphate Synthetase Deficiency[1]	CPS	•	•	•	•			•
33	Ornithine Transcarbamylase Deficiency[1]	OTC	•	•	•	•			•
34	Citrullinemia	CIT-I	•	•	•	•			
35	Citrullinemia Type II	CIT-II	•	•	•	•			
36	Homocystinuria	HCY	•	•	•	•			•
37	Hypermethioninemia	MET	•	•	•	•			•
38	Hyperammonemia, Hyperornithinemia, Homocitrullinuria Syndrome1	HHH	•	•	•	•			•
39	Hyperornithinemia with Gyral Atrophy1	HOGA	•	•	•	•			•
40	Maple Syrup Urine Disease	MSUD	•	•	•	•			•
	Phenylketonuria								•
41	Classical/Hyperphenylalaninemia	PKU	•	•	•	•			•
42	Benign Hyperphenylalaninemia	H-PHE	•	•	•	•			
43	Defects of Biopterin Cofactor Biosynthesis	BIOPT BS	•	•	•	•			
44	Defects of Biopterin Cofactor Regeneration	BIOPT REG	•	•	•	•			•
	Tyrosinemia								•
45	Transient Neonatal Tyrosinemia	TTN	•	•	•	•			
46	Tyrosinemia Type I1	TYR I	•	•	•	•			•
47	Tyrosinemia Type II	TYR II	•	•	•	•			•
48	Tyrosinemia Type III	TYR III	•	•	•	•			•
49	Nonketotic Hyperglycinemia[1]	NKHG	•	•	•	•			
D	**Other**		**6**	**6**	**6**	**6**			
50	Hyperalimentation		•	•	•	•			
51	Medium Chain Triglyceride Oil Administration	MCT	•	•	•	•			
52	Treatment with Benzoate, Pyvalic Acid, or Valproic Acid		•	•	•	•			
53	Liver Disease		•	•	•	•			
54	Presence of EDTA Coagulants in Blood Specimen		•	•	•	•			
55	Carnitine Uptake Deficiency	CUD	•	•	•	•			•
56	Congenital Hypothyroidism	CH	•	•	•		•		•
57	Galactosemia	TGAL	•	•	•		•		•
58	Congenital Adrenal Hyperplasia	CAH	•	•	•		•		•
59	Glucose-6-Phosphate Dehydrogenase Deficiency	G6PD	•	•	•		•		
60	Biotinidase Deficiency	BIO	•	•	•		•		•
61	Cystic Fibrosis	CF	•	•			•		•
	Phenylketonurea	PKU					•		•
62	Sickle Cell Anemia	Hb S/S	•					•	•
63	Sickle-C Disease	Hb S/C	•					•	•
64	S-βeta Thalassemia	Hb S/βTh	•					•	•
65	Hb Variants	Var Hb	•					•	•

1 There is a lower probability of detection of this condition during the immediate newborn period