



# Disorder Panels (Apr 2016)

			Hb+FS+	FS+	FS	MS	Bio7	Bio6	Bio5	Bio4	Hb+	ACMG
			57	53	52	47	7	6	5	4	4	
<b>Disorders Screened by Tandem Mass Spectrometry</b>												
<b>A</b>	<b>Fatty Acid Oxidation Disorders</b>		11	11	11	11						
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 I <sup>1</sup>	CPT-I	•	•	•	•						•
7	2,4-Dienoyl-CoA Reductase Deficiency <sup>1</sup>	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	•	•	•	•						•
<b>B</b>	<b>Organic Acid Disorders</b>		14	14	14	14						
12	3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency	HMG	•	•	•	•						•
13	Glutaric Acidemia Type I	GA-I	•	•	•	•						•
14	Isobutyryl-CoA Dehydrogenase Deficiency	IBG	•	•	•	•						•
15	Isovaleric Acidemia	IVA	•	•	•	•						•
16	2-Methylbutyryl-CoA Dehydrogenase Deficiency	2MBG	•	•	•	•						•
17	3-Methylcrotonyl-CoA Carboxylase Deficiency	3MCC	•	•	•	•						•
18	3-Methylglutaconyl-CoA Hydratase Deficiency	3MGA	•	•	•	•						•
	<b>Methylmalonic Acidemias</b>	MMA	•	•	•	•						•
19	Methylmalonyl-CoA Mutase Deficiency	MUT	•	•	•	•						•
20	Some Adenosylcobalamin Synthesis Defects	CBL A,B/C,D	•	•	•	•						•
21	Maternal Vitamin B12 Deficiency		•	•	•	•						•
22	Mitochondrial Acetoacetyl-CoA Thiolase Deficiency	BKT	•	•	•	•						•
23	Propionic Acidemia	PROP	•	•	•	•						•
24	Multiple CoA Carboxylase Deficiency	MCD	•	•	•	•						•
25	Malonic Aciduria	MAL	•	•	•	•						•
<b>C</b>	<b>Amino Acid Disorders</b>		16	16	16	16						
26	Argininemia	ARG	•	•	•	•						•
27	Argininosuccinic Aciduria	ASA	•	•	•	•						•
28	5-Oxoprolinuria <sup>1</sup>	5-OXO	•	•	•	•						•
29	Carbamoylphosphate Synthetase Deficiency <sup>1</sup>	CPS	•	•	•	•						•
30	Citrullinemia	CIT-I	•	•	•	•						•
31	Homocystinuria	HCY	•	•	•	•						•
32	Hypermethioninemia	MET	•	•	•	•						•
33	Hyperammonemia, Hyperornithinemia, Homocitrullinuria Syndrom	HHH	•	•	•	•						•
34	Hyperornithinemia with Gyral Atrophy <sup>1</sup>	HOGA	•	•	•	•						•
35	Maple Syrup Urine Disease	MSUD	•	•	•	•						•
	<b>Phenylketonuria</b>	PKU	•	•	•	•						•
36	Classical/Hyperphenylalaninemia		•	•	•	•						•
37	Biopterin Cofactor Deficiencies		•	•	•	•						•
	<b>Tyrosinemia</b>	TYR	•	•	•	•						•
38	Transient Neonatal Tyrosinemia		•	•	•	•						•
39	Tyrosinemia Type I <sup>1</sup>	TYR I	•	•	•	•						•
40	Tyrosinemia Type II	TYR II	•	•	•	•						•
41	Tyrosinemia Type III	TYR III	•	•	•	•						•
<b>D</b>	<b>Other</b>		6	6	6	6						
42	Hyperalimentation		•	•	•	•						
43	Medium Chain Triglyceride Oil Administration	MCT	•	•	•	•						
44	Treatment with Benzoate, Pyvalic Acid, or Valproic Acid		•	•	•	•						



# Disorder Panels (Apr 2016)

		Hb+FS+	FS+	FS	MS	Bio7	Bio6	Bio5	Bio4	Hb+	ACMG
		57	53	52	47	7	6	5	4	4	
45	Liver Disease	•	•	•	•						
46	Presence of EDTA Coagulants in Blood Specimen	•	•	•	•						
47	Carnitine Uptake Deficiency										•
<b>Disorders Screened by Other Technologies</b>											
48	Congenital Hypothyroidism					•	•	•	•		•
49	Galactosemia					•	•	•	•		•
50	Congenital Adrenal Hyperplasia					•	•	•	•		•
51	Glucose-6-Phosphate Dehydrogenase Deficiency					•	•	•	•		•
52	Biotinidase Deficiency					•	•	•			•
53	Cystic Fibrosis		•			•					•
54	Phenylketonurea					•	•				
55	Sickle Cell Anemia		•							•	•
56	Sickle-C Disease		•							•	•
57	S-beta Thalassemia		•							•	•
58	Hb Variants		•							•	•

<sup>1</sup> There is a lower probability of detection of this condition during the immediate newborn period