NeoGen Labs

	Disorders Screened by Tandem Mass Spectrometry (TMS)		Hb+FS+	FS+	FS	MS	BioX*	Hb+	SCID	LSD	RUSP**
A	**Fatty Acid Oxidation Disorders**										
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#	CPT-Ia	•	•	•	•					•
7	2,4-Dienoyl-CoA Reductase Deficiency	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**										
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	3M3HBA	•	•	•	•					•
	Methylmalonic Acidemias										
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**										
29	Argininemia	ARG	•	•	•	•					•
30	Argininosuccinic Aciduria	ASA Lyase	•	•	•	•					•
31	5-Oxoprolinuria	5-OXO	•	•	•	•					
32	Carbamoylphosphate Synthetase Deficiency#	CPS	•	•	•	•					
33	Ornithine Transcarbamylase Deficiency#	OTC	•	•	•	•					
34	Citrullinemia	CIT-I	•	•	•	•					•
35	Citrullinemia Type II#	CIT-II	•	•	•	•					•
36	Homocystinuria	HCY	•	•	•	•					•
37	Hypermethioninemia	MET	•	•	•	•					•
38	Hyperammonemia, Hyperornithinemia, Homocitrullinuria Syndrome#	HHH	•	•	•	•					
39	Hyperornithinemia with Gyral Atrophy#	HOGA	•	•	•	•					•
40	Maple Syrup Urine Disease	MSUD	•	•	•	•					•
	Phenylketonuria										
41	*Classic Phenylketonuria*	PKU	•	•	•	•					•
42	*Benign Hyperphenylalaninemia*	H-PHE	•	•	•	•					•
43	*Defects of Biopterin Cofactor Biosynthesis*	BIOT-BS	•	•	•	•					•
44	*Defects of Biopterin Cofactor Regeneration*	BIOT-REG	•	•	•	•					•
	Tyrosinemia										
45	*Transient Neonatal Tyrosinemia*	TYR	•	•	•	•					•
46	*Tyrosinemia Type I#*	TYR I	•	•	•	•					•
47	*Tyrosinemia Type II*	TYR II	•	•	•	•					•
48	*Tyrosinemia Type III*	TYR III	•	•	•	•					•
49	Nonketonic Hyperglycemia#	NKHG	•	•	•	•					
D	**Other Disorders**										
50	Liver Disease		•	•	•	•					
51	Carnitine Uptake Deficiency	CUD	•	•	•	•					•

			Hb+FS+	FS+	FS	MS	BioX*	Hb+	SCID	LSD	RUSP**
	Disorders Screened by Tandem Mass Spectrometry (TMS)										
E	**Other Observations**										
52	Hyperalimentation		•	•	•	•					
53	Medium Chain Triglyceride Oil Administration	MCT	•	•	•	•					
54	Treatment with Benzoate, Pyvalic Acid, or Valproic Acid		•	•	•	•					
55	Presence of EDTA Coagulants in Blood Specimen		•	•	•	•					
	Disorders Screened by Other Technologies										
56	Congenital Hypothyroidism	CH	•	•	•		•				•
57	Galactosemia	GAL	•	•	•		•				•
58	Congenital Adrenal Hyperplasia	CAH	•	•	•		•				•
59	Glucose-6-Phosphate Dehydrogenase Deficiency	G6PD	•	•	•		•				•
60	Biotinidase Deficiency	BIO	•	•	•		•				•
	Phenylketonurea	PKU					•				•
61	Cystic Fibrosis	CF	•	•			•				•
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	•					•			•
	Additional Disorders Screened by TMS										
	Lysosomal Storage Disorders##	LSD									
66	*Gaucher*									•	
67	*Niemann Pick (A/B)*									•	
68	*Pompe*									•	•
69	*Krabbe*									•	
70	*Fabry*									•	
71	*Hurler (MPS-I)*									•	•
	Additional Disorders Screened by Other Technologies										
72	**Severe Combined Immunodeficiency##**	SCID							•		•

* BIOX is a customized combination of disorders from 56 to 61. X represents the number of disorders (1 to 7) in the panel.

** RUSP Recommended Uniform Screening Panel.

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

SCID and LSD screening are performed at the laboratory of our technology partner in the USA.