DISORDER 07/2016	ANALYTE	NORMAL RANGE
Fatty Acid Oxidation Disorders		
Carnitine Uptake Defect	C0	> 12.00 < 95.0 µmol/L
	C2	> 7.50 µmol/L
	C3	> 0.42 < 5.50 µmol/L
	C3/Met	> 0.016 < 0.23
	C3+C16	> 2.00 µmol/L
	(C0+C2+C3+C16+C18:1)/Cit	> 1.60 < 11.8
Carnitine Uptake Defect, Carnitine Palmitoyl Transferase Deficiency Type 1	C16	> 0.40 < 7.50 µmol/L
	C18	> 0.20 < 2.20 µmol/L
	C18:1	> 0.23 < 2.75 µmol/L
Carnitine Palmitoyl Transferase Deficiency Type 1	C18:2	> 0.03 < 1.10 µmol/L
	C0	> 12.00 < 95.0 µmol/L
	C0/(C16+C18)	> 4.00 < 50.00
	(C16+C18:1)/C2	> 0.062 < 0.33
Short Chain Acyl-CoA Dehydrogenase Deficiency, Glutaric Acidemia Type 2	C4	< 1.08 µmol/L
	C4/C2	< 0.047
	C4/C3	> 0.038 < 0.78
Short Chain Acyl-CoA Dehydrogenase Deficiency	C4/C8	< 20.0
Medium/Short Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	C4OH	< 0.80 µmol/L
	C6OH	< 0.25 µmol/L
Glutaric Acidemia Type 2	C5	< 0.55 µmol/L
	C5/C0	< 0.018
	C5/C2	< 0.032
	C5/C3	>0.022 < 0.56
	C5DC/C5OH	< 3.20
	C5DC/C0	< 0.01
Glutaric Acidemia Type 2, Medium Chain Acyl-CoA Dehydrogenase Deficiency	C5DC	< 0.30 µmol/L
	C6	< 0.31 µmol/L
	C8	< 0.32 µmol/L
	C8/C2	< 0.017
	C10	< 0.40 µmol/L
Medium Chain Acyl-CoA Dehydrogenase Deficiency	C3DC	< 0.20 µmol/L
	C8/C10	< 5.00
	C10:1	< 0.32 µmol/L
2,4 Dienoyl CoA Reductase Deficiency	C10:2	< 0.15 µmol/L
Glutaric Acidemia Type 2, Very Long Chain Acyl-CoA Dehydrogenase Deficiency, Carnitine Palmitoyl Transferase Deficiency Type 2, Carnitine/Acylcarnitine Translocase Deficiency	C12	< 0.70 µmol/L
Glutaric Acidemia Type 2, Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency, Very Long Chain Acyl-CoA Dehydrogenase Deficiency, Trifunctional Protein Deficiency	C14:1	< 0.55 µmol/L
	C14:1/C2	< 0.026
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	C12:1	< 0.43 µmol/L
	C14:1/C12:1	< 9.00
Glutaric Acidemia Type 2, Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency Very Long Chain Acyl-CoA Dehydrogenase Deficiency, Trifunctional Protein Deficiency, Carnitine Palmitoyl Transferase Deficiency Type 2, Carnitine/Acylcarnitine Translocase Deficiency	C14	< 0.72 µmol/L
Glutaric Acidemia Type 2, Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency, Very Long Chain Acyl-CoA Dehydrogenase Deficiency, Trifunctional Protein Deficiency	C14:1/C16	< 0.22
	C14:2	< 0.17 µmol/L
Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency, Trifunctional Protein Deficiency	C16OH	< 0.18 µmol/L
	C16OH/C16	< 0.09
	C16:1OH	< 0.23 µmol/L
	C18OH/C18	< 0.17
	C18:1OH	< 0.15 µmol/L
	C14OH	< 0.16 µmol/L
Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency, Trifunctional Protein Deficiency, Carnitine/Acylcarnitine Translocase Deficiency	C18OH	< 0.14 µmol/L
Carnitine Palmitoyl Transferase Deficiency Type 2, Carnitine/Acylcarnitine Translocase Deficiency	C16	> 0.40 < 7.50 µmol/L
	C3/C16	> 0.180 < 2.60
	C0/(C16+C18)	> 4.00 < 50.00
	(C16+C18:1)/C2	> 0.062 < 0.33
	C18	> 0.20 < 2.20 µmol/L
	C18:1	> 0.23 < 2.75 µmol/L
	C18:2	> 0.03 < 1.10 µmol/L
Organic Acid Disorders		
Propionic Acidemia, Methylmalonic Acidemia, Multiple CoA Carboxylase Deficiency	C3	> 0.42 < 5.50 µmol/L
	C3/C2	< 0.19
	C3/C16	> 0.180 < 2.60
	C4/C3	> 0.038 < 0.78
	C5/C3	>0.022 < 0.56
Propionic Acidemia, Methylmalonic Acidemia	C3/Met	>0.016 < 0.23
Methylmalonic Acidemia	C4DC	< 1.60 µmol/L
Malonic Aciduria	C3DC	< 0.20 µmol/L
	C3DC/C10	< 5.00
Isobutyryl-CoA dehydrogenase deficiency, Ethylmalonic Encephalopathy, Formiminoglutamic Acidemia, Glutamate Formiminotransferase Deficiency	C4	< 1.08 µmol/L
	C4/C2	< 0.047
	C4/C3	> 0.038 < 0.78
	C4/C8	< 20.00
Isovaleric Acidemia, Ethylmalonic Encephalopathy, 2-Methyl Butyryl-CoA Dehydrogenase Deficiency	C5	< 0.55 µmol/L
	C5/C0	< 0.018
	C5/C2	< 0.032
	C5/C3	>0.022 < 0.56
Beta-Ketothiolase Deficiency, 2-Methyl 3 Hydroxy Butyric Aciduria	C5:1	< 0.16 µmol/L
3-Hydroxy-3-Methylglutaryl CoA Lyase Deficiency, 3-Methylcrotonyl CoA Carboxylase Deficiency, Multiple CoA Carboxylase Deficiency, Beta-Ketothiolase Deficiency, 2-Methyl 3 Hydroxy Butyric Aciduria, 3-Methylglutaconic Aciduria	C5OH	< 0.65 µmol/L
	C5OH/C0	< 0.024
	C5OH/C8	< 15.00
3-Hydroxy-3-Methylglutaryl CoA Lyase Deficiency	C6DC	< 0.22 µmol/L
Glutaric Acidemia Type 1	C5DC	< 0.30 µmol/L
	C5DC/C0	<0.01
	C5DC/C5OH	< 3.20
	C5DC/C8	< 8.50
	C5DC/C16	< 0.17

DISORDER 07/2016	ANALYTE	NORMAL RANGE
Amino Acid and Urea Cycle Disorders		
Ornithine Transcarbamylase Deficiency, Carbamoyltransferase Deficiency, Carbamoyl Phosphate Synthetase I Deficiency	Alanine	< 700 µmol/L
	Arginine	> 2.60 µmol/L
	Citrulline	> 6.00 < 43.00 µmol/L
	Cit/Phe	> 0.095 < 0.80
	(C0+C2+C3+C16+C18:1)/Cit	> 1.60 < 11.8
	Orn/ Cit	< 19.00
Argininemia, Arginase Deficiency	Arginine	< 55.00 µmol/L
	Arg/Ala	< 0.19
	Arg/Orn	< 0.70
	Arg/Phe	< 1.00
Argininemia, Arginase Deficiency, Ornithine Transcarbamylase Deficiency, Carbamoyltransferase Deficiency, Carbamoyl Phosphate Synthetase I Deficiency	Cit/Arg	> 0.32 < 5.95
Citrullinemia, Arginosuccinic Aciduria, Pyruvate Carboxylase Deficiency	Citrulline (Cit)	> 6.00 < 43.00 µmol/L
	Cit/Arg	> 0.32 < 5.95
	Cit/Phe	> 0.095 < 0.80
Nonketotic Hyperglycinemia	Glycine	< 1210 µmol/L
Maple Syrup Urine Disease	Leucine + Isoleucine	< 265 µmol/L
	Leu/Ala	< 1.05
	Leu/Phe	< 4.70
	Valine	< 280 µmol/L
	Val/Phe	< 5.20
Homocystinuria due to MTHFR (5,10-methylenetetrahydrofolate reductase (NADPH)) deficiency, Remethylation Defect	Methionine	> 9.10 < 68 µmol/L
	Met/Phe	> 0.17 < 1.20
	Met/Cit	> 0.68 < 5.50
	Met/Leu	> 0.09 < 0.65
	Met/Tyr	> 0.09 < 1.50
Homocystinuria, Hypermethionemia	Methionine	> 9.10 < 68 µmol/L
	Met/Phe	> 0.17 < 1.20
	Met/Leu	> 0.09 < 0.65
	Met/Tyr	> 0.09 < 1.50
	C3/Met	> 0.016 < 0.23
Homocystinuria, Hypermethionemia, Ornithine Transcarbamylase Deficiency, Carbamoyltransferase Deficiency, Carbamoyl Phosphate Synthetase I Deficiency	Met/Cit	> 0.68 < 5.50
Hyperornithinemia-Hyperammonemia-Homocitrullinuria, Ornithine Transcarbamylase Deficiency, Carbamoyltransferase Deficiency, Carbamoyl Phosphate Synthetase I Deficiency	Ornithine	< 280 µmol/L
Phenylketonuria, Hyperphenylalaninemia, Defects of Biopterin Cofactor Regeneration, Defects of Biopterin Cofactor Biosynthesis	Phenylalanine	< 120 µmol/L
	Phe/Tyr	< 1.90
Tyrosinemia	Tyrosine	< 290 µmol/L
Endocrine Disorders		
	TSH Collected ≥ 24 hours of age:	
Congenital Hypothyroidism	TSH	< 30.0 uIU/mL
Congenital Adrenal Hyperplasia	17-OHP by birthweight, Collected ≥ 24 hours of age:	
	> 2500g	< 38.3 ng/mL
	1500g < 2500g	< 75.0 ng/mL
	< 1500g	< 100 ng/mL
Metabolic Disorders		
Galactosemia	Galactose-1-phosphate Uridyltransferase (UT-GALT) activity	> 3.1 U/g Hb
	Total Galactose	< 9.1 mg/dL
Biotinidase Deficiency	Biotinidase	> 49.5 MRU
Hemoglobin Carriers/Traits		
Carrier A/S	Hemoglobin AS	F > A, no other bands present
Carrier A/C	Hemoglobin AC	
Carrier A/D	Hemoglobin AD	
Carrier A/E	Hemoglobin AE	
Carrier A/Other	Hemoglobin AOther	
Hemoglobin Disorders		
Disease S/S	Hemoglobin S	F > A, no other bands present
Disease S/C	Hemoglobin SC	
Disease C/C	Hemoglobin C	
Disease S/D	Hemoglobin SD	
Disease D/D	Hemoglobin D	
Disease S/E	Hemoglobin SE	
Disease E/E	Hemoglobin E	
Alpha-Thalasemia (Bart's Hb)	Hemoglobin Bart's	
Beta-Thalasemia	Hemoglobin Sβ° Thal	
Other Hemoglobinopathies	Variant Hemoglobins	
Immune Disorders		
Severe Combined Immunodeficiency Syndrome (SCID)	TREC by EGA:	
	< 37 weeks EGA	> 25 copies/µL
	≥ 37 weeks EGA	> 30 copies/µL
	RNase P Control DNA:	
	All EGA	Ct <28
Peroxisomal Disorders		
X-Linked Adrenoleukodystrophy	C24:0-lysophosphatidylcholine (C24:0-LPC)	< 0.157 µmol/L
	C26:0-lysophosphatidylcholine (C26:0-LPC)	< 0.157 µmol/L