DISORDER 08/2017	ANALYTE	NORMAL RANGE
Fatty Acid Oxidation Disorders		
Carnitine Uptake Defect	C0	> 8.00 < 95.0 µmol/L
	C2	> 8.50 µmol/L
	C3	> 0.52 < 7.00 µmol/L
	C3/Met	> 0.029 < 0.53
	C3+C16	> 2.00 µmol/L
	(C0+C2+C3+C16+C18:1)/Cit	> 1.65 < 18.00
Carnitine Uptake Defect, Carnitine Palmitoyl Transferase Deficiency Type 1	C16	> 0.45 < 8.00 µmol/L
	C18	> 0.20 < 2.60 µmol/L
	C18:1	> 0.33 < 3.15 µmol/L
Carnitine Palmitoyl Transferase Deficiency Type 1	C0	> 8.00 < 95.0 µmol/L
	C0/(C16+C18)	> 3.20 < 35.00
	(C16+C18:1)/C2	> 0.047 < 0.29
Short Chain Acyl-CoA Dehydrogenase Deficiency, Glutaric Acidemia Type 2	C4	< 1.30 µmol/L
	C4/C2	< 0.043
	C4/C3	> 0.02 < 0.66
Short Chain Acyl-CoA Dehydrogenase Deficiency	C4/C8	< 30.0
Medium/Short Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	C4OH	< 0.65 µmol/L
	C6OH	< 0.22 µmol/L
Glutaric Acidemia Type 2	C5	< 0.68 µmol/L
	C5/C0	< 0.017
	C5/C2	< 0.0167
	C5/C3	> 0.008 < 0.28
	C5DC/C5OH	< 4.00
	C5DC/C0	< 0.011
Glutaric Acidemia Type 2, Medium Chain Acyl-CoA Dehydrogenase Deficiency	C5DC	< 0.3 µmol/L
	C6	< 0.33 µmol/L
	C8	< 0.39 µmol/L
	C8/C2	< 0.015
	C10	< 0.40 µmol/L
Medium Chain Acyl-CoA Dehydrogenase Deficiency	C3DC	< 0.20 µmol/L
	C8/C10	< 7.50
	C10:1	< 0.30 µmol/L
2,4 Dienoyl CoA Reductase Deficiency	C10:2	< 0.20 µmol/L
Glutaric Acidemia Type 2, Very Long Chain Acyl-CoA Dehydrogenase Deficiency, Carnitine Palmitoyl Transferase Deficiency Type 2, Carnitine/Acylcarnitine Translocase Deficiency	C12	< 0.64 µmol/L
Glutaric Acidemia Type 2, Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency, Very Long Chain Acyl-CoA Dehydrogenase Deficiency, Trifunctional Protein Deficiency	C14:1	< 0.65 µmol/L
	C14:1/C2	< 0.019
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	C12:1	< 0.50 µmol/L
	C14:1/C12:1	< 15.00
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	C14	< 0.92 µmol/L
	C16:1	< 0.68 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	< 0.21
	C14:2	< 0.17 µmol/L
Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency, Trifunctional Protein Deficiency	C16OH	< 0.20 µmol/L
	C16OH/C16	< 0.08
	C16:1OH	< 0.24 µmol/L
	C18OH/C18	< 0.19
	C18:1OH	< 0.14 µmol/L
	C18:2OH	< 0.11 µmol/L
	C14OH	< 0.23 µmol/L
Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency, Trifunctional Protein Deficiency, Carnitine/Acylcarnitine Translocase Deficiency	C18OH	< 0.12 µmol/L
Carnitine Palmitoyl Transferase Deficiency Type 2, Carnitine/Acylcarnitine Translocase Deficiency	C16	> 0.45 < 8.00 µmol/L
	C3/C16	> 0.24 < 4.20
	C0/(C16+C18)	> 3.20 < 35.00
	(C16+C18:1)/C2	> 0.047 < 0.29
	C18	> 0.20 < 2.60 µmol/L
	C18:1	> 0.33 < 3.15 µmol/L
	C18:2	< 0.98 µmol/L
Organic Acid Disorders		
Propionic Acidemia, Methylmalonic Acidemia, Multiple CoA Carboxylase Deficiency	C3	> 0.52 < 7.00 µmol/L
	C3/C2	< 0.17
	C3/C16	> 0.24 < 4.20
	C4/C3	> 0.02 < 0.66
	C5/C3	> 0.008 < 0.28
Propionic Acidemia, Methylmalonic Acidemia	C3/Met	> 0.029 < 0.53
Methylmalonic Acidemia	C4DC	< 1.50 µmol/L
Malonic Aciduria	C3DC	< 0.20 µmol/L
	C3DC/C10	< 6.50
Isobutyryl-CoA dehydrogenase deficiency, Ethylmalonic Encephalopathy, Formiminoglutamic Acidemia, Glutamate Formiminotransferase Deficiency	C4	< 1.30 µmol/L
	C4/C2	< 0.043
	C4/C3	> 0.02 < 0.66
	C4/C8	< 30.0
Isovaleric Acidemia, Ethylmalonic Encephalopathy, 2-Methyl Butyryl-CoA Dehydrogenase Deficiency	C5	< 0.68 µmol/L
	C5/C0	< 0.017
	C5/C2	< 0.0167
	C5/C3	> 0.008 < 0.28
Beta-Ketothiolase Deficiency, 2-Methyl 3 Hydroxy Butyric Aciduria	C5:1	< 0.15 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	< 0.68 µmol/L
	C5OH/C0	< 0.025
	C5OH/C8	< 16.00
3-Hydroxy-3-Methylglutaryl CoA Lyase Deficiency	C6DC	< 0.29 µmol/L
Glutaric Acidemia Type 1	C5DC	< 0.30 µmol/L
	C5DC/C0	<0.011
	C5DC/C5OH	< 4.00
	C5DC/C8	< 9.00
	C5DC/C16	< 0.12

DISORDER 08/2017	ANALYTE	NORMAL RANGE
Amino Acid and Urea Cycle Disorders		
Ornithine Transcarbamylase Deficiency, Carbamoyltransferase Deficiency, Carbamoyl Phosphate Synthetase I Deficiency	Citrulline (Cit)	> 4.20 < 45.00 µmol/L
	Cit/Phe	> 0.11 < 0.86
	Glutamine (Gln)	< 1170.00 µmol/L
	Gln/Cit	> 2.80 < 95.00
	Glu/Cit	> 11.2 < 105.00
	Ala/Cit	> 5.50 < 100.00
	(C0+C2+C3+C16+C18:1)/Cit	> 1.65 < 18.00
	Orn/ Cit	< 23.50
Argininemia, Arginase Deficiency	Arginine (Arg)	< 66.00 µmol/L
	Arg/Ala	< 0.30
	Arg/Orn	< 0.59
	Arg/Phe	< 1.05
Argininemia, Arginase Deficiency, Ornithine Transcarbamylase Deficiency, Carbamoyltransferase Deficiency, Carbamoyl Phosphate Synthetase I Deficiency	Cit/Arg	> 0.23 < 3.40
Citrullinemia, Argininosuccinic Aciduria, Pyruvate Carboxylase Deficiency	Citrulline (Cit)	> 4.20 < 45.00 µmol/L
	Cit/Arg	> 0.23 < 3.40
	Cit/Phe	> 0.11 < 0.86
	Glutamine (Gln)	< 1170.00 µmol/L
	Gln/Cit	> 2.80 < 95.00
	Glu/Cit	> 11.2 < 105.00
	Ala/Cit	> 5.50 < 100.00
Argininosuccinic Aciduria	Argininosuccinic Acid (ASA)	< 12.50 µmol/L
	Asa/Arg	< 0.95
Maple Syrup Urine Disease	Leucine + Isoleucine (Leu)	< 235 µmol/L
	Leu/Ala	< 1.25
	Leu/Phe	< 4.60
	Valine (Val)	< 280 µmol/L
	Val/Phe	< 6.20
Homocystinuria due to MTHFR (5,10-methylenetetrahydrofolate reductase (NADPH)) deficiency, Remethylation Defect	Methionine (Met)	> 6.80 < 50.00 µmol/L
	Met/Phe	> 0.135 < 1.20
	Met/Cit	> 0.43 < 5.60
	Met/Leu	> 0.066 < 0.58
	Met/Tyr	> 0.06 < 1.00
Homocystinuria, Hypermethionemia	Methionine (Met)	> 6.80 < 50.00 µmol/L
	Met/Phe	> 0.135 < 1.20
	Met/Leu	> 0.066 < 0.58
	Met/Tyr	> 0.06 < 1.00
	C3/Met	> 0.029 < 0.53
Homocystinuria, Hypermethionemia, Ornithine Transcarbamylase Deficiency, Carbamoyltransferase Deficiency, Carbamoyl Phosphate Synthetase I Deficiency	Met/Cit	> 0.43 < 5.60
Phenylketonuria, Hyperphenylalaninemia, Defects of Biopterin Cofactor Regeneration, Defects of Biopterin Cofactor Biosynthesis	Phenylalanine (Phe)	< 100 µmol/L
	Phe/Tyr	< 1.90
Tyrosinemia	Succinylacetone (SUAC)	< 1.60 µmol/L
	SUAC/Met	< 0.13
	SUAC/Phe	< 0.037
	SUAC/Tyr	< 0.034
	Tyrosine (Tyr)	< 242 µmol/L
Endocrine Disorders		
	TSH Collected ≥ 24 hours of age:	
Congenital Hypothyroidism	TSH	< 25.5 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