DISORDER 01/2021	ANALYTE	NORMAL RANGE
Fatty Acid Oxidation Disorders		
Carnitine Uptake Defect	C0	> 8.60 < 80.00 µmol/L
	C2	> 6.70 µmol/L
	C3	> 0.55 < 6.00 µmol/L
	C3/Met	> 0.03 < 0.80
	C3+C16	> 2.00 µmol/L
	(C0+C2+C3+C16+C18:1)/Cit	> 1.00 < 10.50
Carnitine Uptake Defect, Carnitine Palmitoyl Transferase Deficiency Type 1	C16	> 0.65 < 6.50 µmol/L
	C18	> 0.28 < 2.00 µmol/L
	C18:1	> 0.40 < 4.00 µmol/L
Carnitine Palmitoyl Transferase Deficiency Type 1	C0	> 8.60 < 80.00 µmol/L
	C0/(C16+C18)	> 2.00 < 30.00
	(C16+C18:1)/C2	> 0.06 < 1.00
	(C16+C18:1)/C3	> 0.50 < 7.50
Short Chain Acyl-CoA Dehydrogenase Deficiency, Glutaric Acidemia Type 2	C4	< 1.10 µmol/L
	C4/C2	< 0.08
Short Chain Acyl-CoA Dehydrogenase Deficiency	C4/C8	< 20.00
Medium/Short Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	C3DC+C4OH	< 1.20 µmol/L
	C5DC+C6OH	< 0.35 µmol/L
Glutaric Acidemia Type 2	C5	< 0.50 µmol/L
	C5/C2	< 0.04
	C5/C8	< 9.80
	C5DC+C6OH/C3DC+C4OH	> 0.2 < 0.84
	C5DC+C6OH/C4DC+C5OH	< 1.90
	C4	< 1.10 µmol/L
	C4/C2	< 0.08
Glutaric Acidemia Type 2, Medium Chain Acyl-CoA Dehydrogenase Deficiency	C5DC+C6OH	< 0.35 µmol/L
	C6	< 0.15 µmol/L
	C8	< 0.23 µmol/L
	C8/C2	< 0.02
	C10	< 0.45 µmol/L
Medium Chain Acyl-CoA Dehydrogenase Deficiency	C3DC+C4OH	< 0.95 µmol/L
	C8/C10	< 1.50
	C10:1	< 0.15 µmol/L
2,4 Dienoyl CoA Reductase Deficiency	C10:2	< 0.04 µmol/L
Glutaric Acidemia Type 2, Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency, Very Long Chain Acyl-CoA Dehydrogenase Deficiency, Trifunctional Protein Deficiency	C12	< 0.35 µmol/L
	C14:1	< 0.45 µmol/L
	C14:1/C2	< 0.04
	C14:1/C8	< 7.00
	C14:2	< 0.14 µmol/L
Glutaric Acidemia Type 2, Very Long Chain Acyl-CoA Dehydrogenase Deficiency	C12:1	< 0.33 µmol/L
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	< 0.50 µmol/L
Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency, Trifunctional Protein Deficiency	C14OH	< 0.20 µmol/L
	C16OH	< 0.20 µmol/L
	C16OH/C16	< 0.07
	C16:1OH	< 0.17 µmol/L
	C18OH	< 0.10 µmol/L
	C18OH/C18	< 0.13
	C18:1OH	< 0.23 µmol/L
	C18:2OH	< 0.15 µmol/L
Carnitine Palmitoyl Transferase Deficiency Type 2, Carnitine/Acylcarnitine Translocase Deficiency	C16	> 0.65 < 6.50 µmol/L
	C3/C16	> 0.17 < 3.00
	C0/(C16+C18)	> 2.00 < 30.00
	(C16+C18:1)/C2	> 0.06 < 1.00
	(C16+C18:1)/C3	> 0.50 < 7.50
	C18	> 0.28 < 2.00 µmol/L
	C18:1	> 0.40 < 4.00 µmol/L
	C18:2	< 1.20 µmol/L
Organic Acid Disorders		
Propionic Acidemia, Methylmalonic Acidemia, Multiple CoA Carboxylase Deficiency	C3	> 0.55 < 6.00 µmol/L
	C3/C2	< 0.25
	C3/C16	> 0.17 < 3.00
	(C16+C18:1)/C3	> 0.50 < 7.50
Propionic Acidemia, Methylmalonic Acidemia	C3/Met	> 0.03 < 0.80
	C16:1OH	< 0.17 µmol/L
Malonic Aciduria	C3DC+C4OH	< 1.20 µmol/L
	C3DC+C4OH/C4	< 5.50
	C3DC+C4OH/C10	< 12.00
	C3DC+C4OH/C16	< 0.65
	C4DC+C5OH/C3DC+C4OH	> 0.20 < 2.80
	C5DC+C6OH/C3DC+C4OH	> 0.20 < 0.84
Isobutyryl-CoA dehydrogenase deficiency, Ethylmalonic Encephalopathy	C4	< 1.10 µmol/L
	C4/C2	< 0.08
	C4/C8	< 20.00
Isovaleric Acidemia, Ethylmalonic Encephalopathy, 2-Methyl Butyryl-CoA Dehydrogenase Deficiency	C5	< 0.50 µmol/L
	C5/C2	< 0.04
Isovaleric Acidemia	C5/C5DC+C6OH	< 4.80
	C5/C8	< 9.80
Beta-Ketothiolase Deficiency, 2-Methyl 3 Hydroxy Butyric Aciduria	C5:1	< 0.04 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	C4DC+C5OH	< 0.90 µmol/L
	C4DC+C5OH/C3DC+C4OH	> 0.20 < 2.80
	C4DC+C5OH/C4	< 3.60
	C4DC+C5OH/C8	< 13.00
3-Hydroxy-3-Methylglutaryl CoA Lyase Deficiency	C6DC	< 0.27 µmol/L
Glutaric Acidemia Type 1	C5DC+C6OH	< 0.35 µmol/L
	C5DC+C6OH/C3DC+C4OH	> 0.20 < 0.84
	C5DC+C6OH/C4DC+C5OH	< 1.90
	C5DC+C6OH/C8	< 5.20
	C5DC+C6OH/C12	< 4.80

DISORDER 01/2021	ANALYTE	NORMAL RANGE
Amino Acid and Urea Cycle Disorders Collected ≥ 24 hours of age		
Ornithine Transcarbamylase Deficiency, Carbamoyltransferase Deficiency, Carbamoyl Phosphate Synthetase I Deficiency	Citrulline (Cit)	> 6.45 < 49.00 µmol/L
	Ala/Cit	> 6.60 < 76.00
	Glutamine (Gln)	< 4000.00 µmol/L
	Gln/Cit	> 0.25 < 345.00
	Glutamic Acid (Glu)	> 235 < 1490.00 µmol/L
	Glu/Cit	> 12.00 < 126.00
	Cit/Phe	> 0.10 < 0.67
	Met/Cit	> 0.13 < 3.40
	Orn/Cit	< 15.00
	(C0+C2+C3+C16+C18:1)/Cit	> 1.00 < 10.50
Argininemia, Arginase Deficiency	Arginine (Arg)	< 60.00 µmol/L
	Arg/Gly	< 0.15
	Arg/Orn	< 0.66
	Arg/Phe	< 0.94
	Cit/Arg	> 0.28 < 4.50
Citrullinemia, Argininosuccinic Aciduria, Pyruvate Carboxylase Deficiency	Citrulline (Cit)	> 6.45 < 49.00 µmol/L
	Cit/Arg	> 0.28 < 4.50
	Cit/Phe	> 0.10 < 0.67
	Glutamic Acid (Glu)	> 235 < 1490.00 µmol/L
	Glu/Cit	> 12.00 < 126.00
	Gln/Cit	> 0.25 < 345.00
Argininosuccinic Aciduria	Argininosuccinic Acid (ASA)	< 21.00 µmol/L
	Asa/Arg	< 2.00
Maple Syrup Urine Disease	Leucine + Isoleucine (Leu)	< 250.00 µmol/L
	Leu/Ala	< 0.60
	Leu/Phe	< 4.00
	Valine (Val)	< 210.00 µmol/L
	Val/Phe	< 3.50
Homocystinuria due to MTHFR (5,10-methylenetetrahydrofolate reductase (NADPH)) deficiency, Remethylation Defect	Methionine (Met)	> 4.00 < 40.00 µmol/L
	Met/Ala	> 0.01 < 0.15
	Met/Cit	> 0.13 < 3.40
	Met/Leu	> 0.02 < 0.47
	Met/Phe	> 0.04 < 0.700
Homocystinuria, Hypermethionemia	Methionine (Met)	> 4.00 < 40.00 µmol/L
	Met/Ala	> 0.01 < 0.15
	Met/Cit	> 0.13 < 3.40
	Met/Leu	> 0.02 < 0.47
	Met/Phe	> 0.04 < 0.700
	C3/Met	> 0.03 < 0.80
Phenylketonuria, Hyperphenylalaninemia, Defects of Biopterin Cofactor Regeneration, Defects of Biopterin Cofactor Biosynthesis	Phenylalanine (Phe)	< 100.00 µmol/L
	Phe/Tyr	< 1.40
Tyrosinemia	Succinylacetone (SUAC)	< 1.80 µmol/L
	SUAC/Met	< 0.37
	SUAC/Phe	< 0.04
	SUAC/Tyr	< 0.03
	Tyrosine (Tyr)	< 290.00 µmol/L
Endocrine Disorders		
Congenital Hypothyroidism	TSH Collected ≥ 24 hours of age:	
	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	
Other Hemoglobinopathies	Variant Hemoglobins	
SBeta-Thalasemia	Hemoglobin Sβ° Thal	
Alpha-Thalasemia (Bart's Hb, Hemoglobin H disease)	Hemoglobin Bart's / Hemoglobin H disease	F > A, FAST<10.7%
Beta-Thalasemia	Hemoglobin β° Thal	F > A, A>4.8%
Immune Disorders		
	TREC:	RNase P Control DNA:
Severe Combined Immunodeficiency (SCID)	All EGA, Ct < 34.2	All EGA, Ct < 26
T-cell related lymphocyte deficiencies		
Adenosine Deaminase Severe Combined Immunodeficiency (ADA-SCID)	Adenosine (ADO)	< 5.20 µmol/L
	Deoxyadenosine (DADO)	< 0.90 µmol/L
Peroxisomal Disorders		
X-Linked Adrenoleukodystrophy	C26:0-lysophosphatidylcholine (C26:0-LPC)	< 0.130 µmol/L
Neuromuscular Disorders		
	SMN1	RNase P Control DNA:
Spinal Muscular Atrophy	Ct < 28	Ct < 26
Lysosomal Storage Disorders		
Pompe Disease	Acid alpha-glucosidase (GAA) activity	> 22% GAA Daily Patient Median Activity (µM/h)
Mucopolysaccharidosis Type I (MPS-I)	Alpha-L-iduronidase (IDUA) activity	> 18% IDUA Daily Patient Median Activity (µM/h)