

DISORDER 01/2020	ANALYTE	NORMAL RANGE
Fatty Acid Oxidation Disorders		
Carnitine Uptake Defect	C0	> 10.50 < 95.0 µmol/L
	C2	> 9.50 µmol/L
	C3	> 0.52 < 6.50 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
	C4/C8	< 30.0
Short Chain Acyl-CoA Dehydrogenase Deficiency	C4OH	< 0.65 µmol/L
Medium/Short Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	C6OH	< 0.22 µmol/L
	C5	< 0.68 µmol/L
Glutaric Acidemia Type 2	C5/C0	< 0.017
	C5/C2	< 0.0167
	C5/C3	> 0.008 < 0.28
	C5DC/C5OH	< 4.00
	C5DC/C0	< 0.011
	C5DC	< 0.3 µmol/L
Glutaric Acidemia Type 2, Medium Chain Acyl-CoA Dehydrogenase Deficiency	C6	< 0.33 µmol/L
	C8	< 0.39 µmol/L
	C8/C2	< 0.015
	C10	< 0.40 µmol/L
	C3DC	< 0.20 µmol/L
	C8/C10	< 7.50
Medium Chain Acyl-CoA Dehydrogenase Deficiency	C10:1	< 0.30 µmol/L
	C10:2	< 0.20 µmol/L
2,4 Dienoyl CoA Reductase Deficiency	C12	< 0.64 µmol/L
Glutaric Acidemia Type 2, Very Long Chain Acyl-CoA Dehydrogenase Deficiency, Carnitine Palmitoyl Transferase Deficiency Type 2, Carnitine/Acylcarnitine Translocase Deficiency	C14:1	< 0.65 µmol/L
	C14:1/C2	< 0.019
Glutaric Acidemia Type 2, Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency, Very Long Chain Acyl-CoA Dehydrogenase Deficiency, Trifunctional Protein Deficiency	C12:1	< 0.50 µmol/L
	C14:1/C12:1	< 15.00
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	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
	C18OH	< 0.12 µmol/L
Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency, Trifunctional Protein Deficiency, 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
	C18:2	< 0.98 µmol/L
Organic Acid Disorders		
Propionic Acidemia, Methylmalonic Acidemia, Multiple CoA Carboxylase Deficiency	C3	> 0.52 < 6.50 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	< 0.80 µmol/L
	C5OH/C0	< 0.025
	C5OH/C8	< 16.00
3-Hydroxy-3-Methylglutaryl CoA Lyase Deficiency	C6DC	< 0.29 µmol/L
	C5DC	< 0.30 µmol/L
Glutaric Acidemia Type 1	C5DC/C0	< 0.011
	C5DC/C5OH	< 4.00
	C5DC/C8	< 9.00
	C5DC/C16	< 0.12
	C5DC/C16	< 0.12

DISORDER 01/2020	ANALYTE		NORMAL RANGE	
Amino Acid and Urea Cycle Disorders				
Ornithine Transcarbamylase Deficiency, Carbamoyltransferase Deficiency, Carbamoyl Phosphate Synthetase I Deficiency	Citrulline (Cit)	> 5.50 < 40.00 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)	> 5.50 < 40.00 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				
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			
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:		RNase P Control DNA:	
	All EGA	Ct < 34.2	All EGA	Ct < 26
T-cell related lymphocyte deficiencies				
Peroxisomal Disorders				
X-Linked Adrenoleukodystrophy	C24:0-lysophosphatidylcholine (C24:0-LPC)	< 0.192 µmol/L		
	C26:0-lysophosphatidylcholine (C26:0-LPC)	< 0.130 µmol/L		
Neuromuscular Disorders				
Spinal Muscular Atrophy	SMN1		RNase P Control DNA:	
	Ct < 28		Ct < 26	