

DISORDER 01/2021	ANALYTE	NORMAL RANGE
Fatty Acid Oxidation Disorders		
Camitine 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
Camitine Uptake Defect, Camitine 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
Camitine 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
	C5DC+C6OH	< 0.35 µmol/L
Glutaric Acidemia Type 2, Medium Chain Acyl-CoA Dehydrogenase Deficiency	C6	< 0.15 µmol/L
	C8	< 0.23 µmol/L
	C8/C2	< 0.02
	C10	< 0.45 µmol/L
	C3DC+C4OH	< 0.95 µmol/L
Medium Chain Acyl-CoA Dehydrogenase Deficiency	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, Camitine Palmitoyl Transferase Deficiency Type 2, Camitine/Acylcamitine Translocase Deficiency	C14	< 0.50 µmol/L
	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
Camitine Palmitoyl Transferase Deficiency Type 2, Camitine/Acylcamitine Translocase Deficiency	C18:2OH	< 0.15 µmol/L
	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
Malonic Aciduria	C16:1OH	< 0.17 µmol/L
	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
	C4	< 1.10 µmol/L
Isobutyryl-CoA dehydrogenase deficiency, Ethylmalonic Encephalopathy	C4/C2	< 0.08
	C4/C8	< 20.00
	C5	< 0.50 µmol/L
Isovaleric Acidemia, Ethylmalonic Encephalopathy, 2-Methyl Butyryl-CoA Dehydrogenase Deficiency	C5/C2	< 0.04
	C5/C5DC+C6OH	< 4.80
Isovaleric Acidemia	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			
Alpha-Thalasemia (Bart's Hb)	Hemoglobin Bart's			
Beta-Thalasemia	Hemoglobin Sβ ⁰ Thal			
Other Hemoglobinopathies	Variant Hemoglobins			
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)	