

DISORDER	ABBREVIATION	ANALYTE	REFERENCE RANGE	
FATTY ACID OXIDATION DISORDERS				
Carnitine Uptake Deficiency	CUD	Free carnitine (C0)	>6.0 µM	
Short chain acyl-CoA dehydrogenase deficiency	SCAD	Butyrylcarnitine (C4)	<1.20 µM	
Glutaric Acidemia, type II	GA-II	Butyrylcarnitine (C4)	<1.20 µM	
		Isovalerylcarnitine (C5)	<0.83 µM	
Medium chain acyl-CoA dehydrogenase deficiency	MCAD	Octanoylcarnitine (C8)	<0.40 µM	
Medium chain keto acyl-CoA thiolase deficiency	MCKAT	Octanoylcarnitine/Decanoylcarnitine (C8/C10)	<2.50	
Long chain acyl-CoA dehydrogenase deficiency	LCAD	Tetradecenoylcarnitine (C14:1)	<0.50 µM	
Very long chain acyl-CoA dehydrogenase deficiency	VLCAD	Tetradecenoylcarnitine/Dodecenoylcarnitine Ratio (C14:1/C12:1)	<3.50	
Long chain 3-Hydroxy acyl-CoA dehydrogenase deficiency	LCHAD	3-Hydroxy-hexadecanoylcarnitine (C16OH)	<0.12 µM	
Trifunctional Protein deficiency	TFP	3-Hydroxy-octadecanoylcarnitine (C18OH)	<0.06 µM	
Carnitine palmitoyltransferase II deficiency	CPT-II	Hexadecanoylcarnitine (C16)	<9.50 µM	
Carnitine/acylcarnitine translocase deficiency	CACT	Octadecenoylcarnitine (Oleylcarnitine) (C18:1)	<2.50 µM	
Carnitine palmitoyltransferase I deficiency	CPT-I	C0/(C16+C18) by age	0-7 days	<68.00
			>7 days	<132.00
Medium/Short chain 3-Hydroxy acyl-CoA dehydrogenase deficiency	M/SCHAD	Malonylcarnitine\3-Hydroxy-butrylcarnitine (C3DC+C4OH)	<0.60 µM	

ORGANIC ACIDEMIA DISORDERS			
Propionic acidemia	PA	Propionylcarnitine (C3)	<7.65 µM
Methylmalonic acidemia – (methylmalonyl-CoA mutase)	MMA-MUT	Propionylcarnitine/Acetylcarnitine Ratio (C3/C2)	<0.35
Methylmalonic acidemia – (cobalamin disorders)	MMA-CBL		
Isobutyrylglycinuria	IBG	Butyrylcarnitine (C4)	<1.20 µM
Isovaleric acidemia	IVA	Isovalerylcarnitine (C5)	<0.83 µM
2-Methylbutyrylglycinuria	2MBG		
3-Hydroxy-3-methylglutaryl-CoA lyase deficiency	HMG		
3-methylcrotonyl-CoA carboxylase deficiency	3MCC	Methylmalonyl\3-Hydroxy-isovalerylcarnitine (C4DC+C5OH)	<0.78 µM
Multiple carboxylase deficiency	MCD		
3-Methylglutaconic aciduria	3-MGA		
Glutaric acidemia type I	GA-I	Glutaryl\3-Hydroxy-hexanoylcarnitine (C5DC+C6OH)	<0.44 µM
Beta-Ketothiolase deficiency	BKT	Tiglylcarnitine (C5:1)	<0.08 µM
2-Methyl-3-hydroxybutyric aciduria	2M3HBA		
Malonic acidemia	MAL	Malonylcarnitine\3-Hydroxy-butrylcarnitine (C3DC+C4OH)	<0.60 µM

AMINO ACID & UREA CYCLE DISORDERS			
Phenylketonuria	PKU	Phenylalanine (Phe)	<130 µM
Hyperphenylalaninemia		Phenylalanine/Tyrosine (Phe/Tyr)	<2.00
Bioppterin cofactor defect of biosynthesis			
Bioppterin cofactor defect of regeneration			
Maple syrup urine disease	MSUD	Leucine (Leu+Ile+Pro-OH)	<380 µM
		Valine (Val)	<300 µM
		Leucine/Phenylalanine (Leu+Ile+Pro-OH/Phe)	<5.00 µM
Tyrosinemia type I	TYR I	Succinylacetone (Suac)	<1.00 µM
Tyrosinemia type II/III	TYR II/III	Tyrosine (Tyr)	<450 µM
Homocystinuria	HCY/MET	Methionine (Met)	<105 µM
Hypermethionemia			
Citrullinemia, type I	CIT	Citrulline (Cit)	<55.0 µM
Citrullinemia, type II			
Argininosuccinic acidemia	ASA	Argininosuccinic Acid (Asa)	<0.93 µM
Argininemia	ARG	Arginine (Arg)	<120 µM

ENDOCRINE DISORDERS				
Congenital Hypothyroidism	CH	Thyroxine (T4) Thyroid-stimulating Hormone (TSH)	≥5 µg/dL <20 UIU/mL	
Congenital Adrenal Hyperplasia	CAH	17-Hydroxyprogesterone (17-OHP) by Birthweight	≥2300 g	<30 ng/mL
			1400-2299g	<50 ng/mL
			≤1399g	<73 ng/mL

METABOLIC DISORDERS			
Classic galactosemia	GAL	Galactose-1-Phosphate Uridyltransferase Activity (GALT)	≥2.2 U/gHb
		Total Galactose (Tgal)	<7.3 mg/dL
		GALT Mutational Analysis	Mutations Not Detected
Biotinidase Deficiency	BIOT	Biotinidase Activity (Biot)	≥83.6 U/dL

OTHER DISORDERS				
Hemoglobinopathies	S/S Disease	HGB	Hemoglobin	FA
	Hgb C/D/E Disease			
Cystic Fibrosis	CF	Immunoreactive Trypsinogen (IRT) Cystic Fibrosis Transmembrane Conductance Regulator (CFTR)	<96th percentile CFTR Mutations Not Detected	
Severe Combined Immunodeficiency	SCID	T-cell Receptor Excision Circles (TRECs)	TRECs Detected	
Spinal Muscular Atrophy	SMA	Homozygous Deletion of Exon 7 in SMN1	Exon 7 Detected in SMN1	
X-Linked Adrenoleukodystrophy	X-ALD	C26:0-lysophosphatidylcholine (C26:0-LPC)	<0.15 µM	
Mucopolysaccharidosis Type I	MPS I	α-L-iduronidase (IDUA) Enzyme Activity	>8% of daily median	
Pompe Disease	POMPE	Acid-α-glucosidases (GAA) Enzyme Activity	>15% of daily median	

KNOWN PKU DIET MONITORING			
Known Phenylketonuria	PKU	Phenylalanine (Phe)	120-360 µM
		Tyrosine (Tyr)	>30 µM