North Carolina State Laboratory of Public Health
Newborn Screening Normal Reference Ranges
Last Updated: August 29, 2025

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	Butyrlcarnitine (C4)		<1.20 µM
Glutaric Acidemia, type II	GA-II	Butyrlcarnitine (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
Very long chain acyl-CoA dehydrogenase deficiency	VLCAD	Tetradecenoylcarnitine (C14:1)		<0.50 µM
		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-butyrylcarnitine (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	Butyrlcarnitine (C4)		<1.20 µM
Isovaleric acidemia	IVA	Isovalerylcarnitine (C5)		<0.83 µM
2-Methylbutyrylglycinuria	2MBG			
3-Hydroxy-3-methylglutaryl-CoA lyase deficiency	HMG	Methylmalonyl\3-Hydroxy-isovalerylcarnitine (C4DC+C5OH)		<0.78 µM
3-methylcrotonyl-CoA carboxylase deficiency	3MCC			
Multiple carboxylase deficiency	MCD			
3-Methylglutaconic aciduria	3-MGA			
Glutaric acidemia type I	GA-I	Glutarylcarnitine\3-Hydroxy-hexanoylcarnitine (C5DC+C6OH)		<0.44 µM
β-Ketothiolase deficiency	BKT	Tiglylcarnitine (C5:1)		<0.08 µM
2-Methyl-3-hydroxybutyric aciduria	2M3HBA			
Malonic acidemia	MAL	Malonylcarnitine\3-Hydroxy-butyrylcarnitine (C3DC+C4OH)		<0.60 µM
AMINO ACID & UREA CYCLE DISORDERS				
Phenylketonuria	PKU	Phenylalanine (Phe)		<130 µM
Hyperphenylalaninemia		Phenylalanine/Tyrosine (Phe/Tyr)		<2.00
Biopterin cofactor defect of biosynthesis				
Biopterin 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)		≥5 µg/dL
		Thyroid-stimulating Hormore (TSH)		<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)		<96th percentile
		Cystic Fibrosis Transmembrane Conductance Regulator (CFTR)		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
Mucopolysaccharidosis Type II	MPS II	iduronate 2-sulfatase (I2S) Enzyme Activity		>10% of the 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