

Delaware Division of Public Health Newborn Screening Program

Disorder Name & Abbreviation	Analyte / Marker	Normal Reference Range		
		Initial **	2nd ***	Units
Endocrine Disorder				
• Congenital Hypothyroidism (CH)	Thyroid Stimulating Hormone (TSH) *	< 20.0	< 20.0	μIU/mL
	Thyroxine (T4) – used as confirmatory test	> 3.5	> 3.5	μg/dL
• Congenital Adrenal Hyperplasia (CAH)	17-αHydroxyprogesterone	Birth Weight ≥/ 2200 gm	< 35	< 35
		1300-2199 gm	< 60	< 60
		< 1300 gm	< 75	< 75
				ng/mL
Amino Acid / Urea Cycle Disorders				
• Phenylketonuria (PKU)	Phenylalanine *	< 97.0	< 115.0	μmol/L
	Phenylalanine/Tyrosine ratio	< 2.20	< 1.84	N/A
• Hyperphenylalanemia (HPHE)	Leucine *	< 300.0	< 420.0	μmol/L
	Leucine/Phenylalanine ratio	< 7.00	< 10.00	N/A
• Maple Syrup Urine Disease (MSUD)	Methionine *	< 45.0	< 75.0	μmol/L
	Methionine/Phenylalanine ratio	< 1.07	< 1.60	N/A
• Homocystinuria (HCYS)	Tyrosine *	< 201.0	< 245.0	μmol/L
	Tyrosine/Phenylalanine ratio	< 4.94	< 5.67	N/A
• Tyrosinemia, types I, II, or III (TYR)	Arginine	< 30.0	< 55.0	μmol/L
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• Argininemia (ARG)	Citrulline *	< 27.0	< 65.0	μmol/L
	Citrulline/Arginine ratio	< 7.00	< 11.80	N/A
• Citrullinemia (Argininosuccinate Synthetase Def. (CIT))				
• Argininosuccinate Lyase Deficiency (ASL)				
Organic Acid Disorders				
• Glutaric Acidemia, type I (GA I)	Glutaryl carnitine (AC5DC)	< 0.13	< 0.12	μmol/L
• Propionic Acidemia (PA)	Propionyl carnitine (AC3) *	< 5.50	< 5.42	μmol/L
• Methylmalonic Acidemia (MMA)	Methylmalonyl carnitine (AC3-2M-DC)	< 0.60	< 1.04	μmol/L
• Multiple Carboxylase Deficiency (MCD)	Hydroxyisovaleryl carnitine (AC5-OH)	< 0.57	< 0.70	μmol/L
• Isovaleric Acidemia (IVA)	Isovaleryl carnitine (AC5)	< 0.42	< 0.69	μmol/L
• 2-Methylbutyryl-CoA Dehydrogenase Def. (2-MBCD)				
• 3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)	Hydroxyisovaleryl carnitine (AC5-OH) *	< 0.57	< 0.70	μmol/L
• 3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency (HMG)	3-Methylglutaryl carnitine (AC5-3M-DC)	< 0.11	< 0.12	μmol/L
• Beta-Ketothiolase Deficiency (BKT)	Tiglyl carnitine (AC5:1) *	< 0.11	< 0.22	μmol/L
	Hydroxyisovaleryl carnitine (AC5-OH)	< 0.57	< 0.70	μmol/L
• Isobutyryl-CoA Dehydrogenase Def. (IBCD)	Butyryl carnitine (AC4)	< 1.00	< 1.00	μmol/L
Fatty Acid Oxidation Disorders				
• Medium Chain Acyl-CoA Dehydrogenase Def. (MCAD)	Octanoyl carnitine (AC8) *	< 0.40	< 0.44	μmol/L
	Decenoyl carnitine (AC10:1)	< 0.25	< 0.42	μmol/L
	Hexanoyl carnitine (AC6)	< 0.25	< 0.32	μmol/L
• Carnitine Palmityltransferase II Deficiency (CPT II)	Hexadecanoyl carnitine (AC16) *	< 7.00	< 5.63	μmol/L
• Carnitine/Acylcarnitine Translocase Deficiency (CAT)	Octadecenoyl carnitine (AC18:1)	< 2.50	< 3.50	μmol/L
• Glutaric Acidemia, Type II (GA II)	Butyryl carnitine (AC4)	< 1.00	< 1.00	μmol/L
	Isovaleryl carnitine (AC5)	< 0.42	< 0.69	μmol/L
	Octanoyl carnitine (AC8)	< 0.40	< 0.44	μmol/L
	Decanoyl carnitine (AC10) *	< 0.30	< 0.35	μmol/L
	Hexadecanoyl carnitine (AC16)	< 7.00	< 5.63	μmol/L
• Multiple Acyl-CoA Dehydrogenase Deficiency (MADD)	Glutaryl carnitine (AC5DC)	< 0.13	< 0.12	μmol/L
• Short-Chain Acyl-CoA Dehydrogenase Deficiency (SCAD)	Butyryl carnitine (AC4) *	< 1.00	< 1.00	μmol/L
	Isovaleryl carnitine (AC5)	< 0.42	< 0.69	μmol/L
• Long-Chain Hydroxyacyl-CoA Dehydrogenase Def. (LCHAD)	Hydroxyhexadecanoyl carnitine (AC16-OH)	< 0.09	< 0.11	μmol/L
• Trifunctional Protein Deficiency (TFP)				
• Very Long-Chain Acyl-CoA Dehydrogenase Def. (VLCAD)	Tetradecenoyl carnitine (AC14:1) *	< 0.63	< 0.38	μmol/L
	Hydroxyhexadecanoyl carnitine (AC16-OH)	< 0.09	< 0.11	μmol/L
• Carnitine Uptake Deficiency (CUD)	Free Carnitine (C0)	> 7.00	> 7.00	μmol/L
Other Genetic Disorders				
• Galactosemia	Galactose-1-Phosphate Uridyl Transferase (GALT)	> 2.3	> 2.3	U/gm Hb
	Total Galactose (TG)	< 12.0	< 12.0	mg/dL
• Hemoglobinopathies	Hemoglobin S,C,E,O,D,G, Bart's Hemoglobin X (unknown variant)	FA	FA	---
• Biotinidase Deficiency	Biotinidase	≥/ 17.5	≥/ 19.0	ERU
• Cystic Fibrosis – For a complete list of mutations, refer to web site: http://www.dhss.delaware.gov/dhss/dph/lab/nbs.html	Immunoreactive Trypsin (IRT)	< 70	< 70	ng/mL
	CF-DNA Mutation Analysis	No Mutations Found		

* Indicates Primary Marker

** Also 2nd specimen < 7 days old

*** Also Initial spec > 7 days

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