Delaware Division of Public Health Newborn Screening Program

Disorder Name & Abbreviation	Analyte / Marker		Normal Reference Range		
Endocrine Disorder			**Initial ****	**2nd *****	**Units**
• Congenital Hypothyroidism (CH)	Thyroid Stimulating Hormone (TSH) *		< 25.0	< 25.0	µU/mL serum
	Thyroxine (T4) – used as confirmatory test		> 5.0	> 5.0	µg/dL serum
• Congenital Adrenal Hyperplasia (CAH)	17-αHydroxyprogesterone	Birth Weight			ng/mL serum
		>/= 2200 gm	< 25	< 25	
		1300-2199 gm	< 70	< 70	
		< 1300 gm	< 120	< 120	
Amino Acid / Urea Cycle Disorders			**Initial ****	**2nd *****	**Units**
• Phenylketonuria (PKU)	Phenylalanine *		< 99.0	< 108.0	µmol/L
• Hyperphenylalanemia (HPHE)	Phenylalanine/Tyrosine ratio		< 1.70	< 1.50	N/A
• Maple Syrup Urine Disease (MSUD)	Leucine *		< 222.0	< 375.0	µmol/L
	Leucine/Phenylalanine ratio		< 4.90	< 7.75	N/A
• Homocystinuria (HCYS)	Methionine *		< 48.0	< 80.0	µmol/L
• Hypermethioninemia (HMET)	Methionine/Phenylalanine ratio		< 0.95	< 1.40	N/A
• Tyrosinemia, types I, II, or III (TYR)	Tyrosine *		< 217.0	< 295.0	µmol/L
	Tyrosine/Phenylalanine ratio		< 5.20	< 6.00	N/A
• Argininemia (ARG)	Arginine		< 33.0	< 58.0	µmol/L
• Citrullinemia (Argininosuccinate Synthetase Def. (CIT)	Citrulline *		< 37.0	< 65.0	µmol/L
• Argininosuccinate Lyase Deficiency (ASL)	Citrulline/Arginine ratio		< 8.50	< 6.90	N/A
Organic Acid Disorders			**Initial ****	**2nd *****	**Units**
• Glutaric Acidemia, type I (GA I)	Glutarylcarnitine (AC5DC)		< 0.20	< 0.15	µmol/L
• Propionic Acidemia (PA)	Propionylcarnitine (AC3) *		< 5.60	< 5.60	µmol/L
• Methylmalonic Acidemia (MMA)	Hydroxyisovalerylcarnitine (AC5-OH)		< 0.76	< 0.91	µmol/L
• Multiple Carboxylase Deficiency (MCD)					
• Isovaleric Acidemia (IVA)	Isovalerylcarnitine (AC5)		< 0.38	< 0.90	µmol/L
• 2-Methylbutyryl-CoA Dehydrogenase Def. (2-MBCD)					
• 3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)	Hydroxyisovalerylcarnitine (AC5-OH)		< 0.76	< 0.91	µmol/L
• 3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency (HMG)					
• Beta-Ketothiolase Deficiency (BKT)	Tiglylcarnitine (AC5:1) *		< 0.08	< 0.10	µmol/L
	Hydroxyisovalerylcarnitine (AC5-OH)		< 0.76	< 0.91	µmol/L
• Isobutyryl-CoA Dehydrogenase Def. (IBCD)	Butyrylcarnitine (AC4)		< 1.00	< 1.00	µmol/L
Fatty Acid Oxidation Disorders			**Initial ****	**2nd *****	**Units**
• Medium Chain Acyl-CoA Dehydrogenase Def. (MCAD)	Octanoylcarnitine (AC8) *		< 0.26	< 0.46	µmol/L
	Decenoylcarnitine (AC10:1)		< 0.23	< 0.54	µmol/L
	Hexanoylcarnitine (AC6)		< 0.22	< 0.25	µmol/L
• Carnitine Palmioyltransferase II Deficiency (CPT II)	Hexadecanoylcaritine (AC16) *		< 6.60	< 5.20	µmol/L
• Carnitine/Acylcarnitine Translocase Deficiency (CAT)	Octadecenoylcarnitine (AC18:1)		< 2.80	< 3.90	µmol/L
• Glutaric Acidemia, Type II (GA II)	Butyrylcarnitine (AC4)		< 1.00	< 1.00	µmol/L
• Multiple Acyl-CoA Dehydrogenase Deficiency (MADD)	Isovalerylcarnitine (AC5)		< 0.38	< 0.90	µmol/L
	Octanoylcarnitine (AC8)		< 0.26	< 0.46	µmol/L
	Decanoylcarnitine (AC10) *		< 0.48	< 0.42	µmol/L
	Hexadecanoylcaritine (AC16)		< 6.60	< 5.20	µmol/L
	Glutarylcarnitine (AC5DC)		< 0.20	< 0.15	µmol/L
• Short-Chain Acyl-CoA Dehydrogenase Deficiency (SCAD)	Butyrylcarnitine (AC4) *		< 1.00	< 1.00	µmol/L
	Isovalerylcarnitine (AC5)		< 0.38	< 0.90	µmol/L
• Long-Chain Hydroxyacyl-CoA Dehydrogenase Def. (LCHAD)	Hydroxyhexadecanoylarnitine (AC16-OH)		< 0.14	< 0.10	µmol/L
• Trifunctional Protein Deficiency (TFP)					
• Very Long-Chain Acyl-CoA Dehydrogenase Def. (VLCAD)	Tetradecenoylcarnitine (AC14:1) *		< 0.65	< 0.45	µmol/L
	Hydroxyhexadecanoylarnitine (AC16-OH)		< 0.14	< 0.10	µmol/L
• Carnitine Uptake Deficiency (CUD)	Free Carnitine (C0)		> 8.00	> 7.60	µmol/L
Other Genetic Disorders			**Initial**	**2nd**	**Units**
• Galactosemia	Galactose-1-Phosphate UridylTransferase (GALT)		> 3.1	> 2.0	U/gm Hb
	Total Galactose (TG)		< 10.0	< 10.0	mg/dL
• Hemoglobinopathies	Hemoglobin S,C,E,O,D,G, Bart's		FA	FA	---
	Hemoglobin X (unknown variant)				
• Biotinidase Deficiency	Biotinidase		> 36.0	> 52.0	MRU
• 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 blood
	CF-DNA Mutation Analysis		No Mutations Found		
• Severe Combined Immunodeficiancy (SCID)	T-cell Receptor Excision Circles (TREC)		>= 27	>=27	TREC copies

* Indicates Primary Marker ** Also 2nd specimen < 7 days old *** Also Initial spec > 7 days Doc. #35-05-20/05/05/15 Version-N- 07/18/14