

DISORDER	ANALYTE / PRIMARY MARKER	REFERENCE RANGE
Amino Acid Disorders (by MS/MS*)		
HCY \ HMet	Methionine (Met)	< 35 μ mole/L
MSUD	Met/Phe	< 1.00
PKU \ HyperPhe	Leucine (Leu)	< 305 μ mole/L
TYR-II, III	Phenylalanine (Phe)	< 151 μ mole/L
TYR-I	Tyrosine (Tyr)	< 414 μ mole/L
Endocrine Disorders (by Time-Resolved Fluoroimmunoassay)		
CAH	17alpha-hydroxyprogesterone	< 35 ng/mL, Birthweight of \geq 1751 gms
CH	Thyroid Stimulating Hormone	< 26 uU/mL Age collected \leq 36 hours from Nov 1 to May 31 and < 25 uU/mL from June 1 to Oct 31
Fatty Acid Oxidation Disorders (by MS/MS*)		
CUD	Free Carnitine (C0)	> 10.0 μ mole/L (or > 8.0 & $C3 \geq 1.0$ μ mole/L)
CPT-I	Total Acylcarnitines (SUM AC)	> 7.50 μ mole/L
CAT \ CPT-II	C0/(C16 + C18)	< 50.0, Based of age of \leq 7 days
2,4Di	Hexadecanoylcarnitine (C16)	< 7.00 μ mole/L or C0/(C16+C18) > 3.00
LCHAD \ TFP	Octadecenoylcarnitine (C18:1)	< 2.50 μ mole/L or C0/(C16+C18) > 3.00
MADD (GA-II) \ MCAD \ MCKAT	Decadienoylcarnitine (C10:2)	< 0.10 μ mole/L
SCAD	Hydroxyhexadecanoylcarnitine (C16OH)	< 0.10 μ mole/L
VLCAD	Hydroxyoctadecenoylcarnitine (C18:1OH)	< 0.10 μ mole/L
M/SCHAD	Hexanoylcarnitine (C6)	< 0.30 μ mole/L
	Octanoylcarnitine (C8)	< 0.35 μ mole/L
	Butyrylcarnitine (C4)	< 1.20 μ mole/L
	Tetradecanoylcarnitine (C14)	< 0.65 μ mole/L
	Tetradecenoylcarnitine (C14:1)	< 0.60 μ mole/L
	Hydroxybutyrylcarnitine (C4OH)	< 0.80 μ mole/L
	Hydroxyhexanoylcarnitine (C6OH)	< 0.15 μ mole/L
Hemoglobin Disorders (by HPLC)		
Disease SS, SC, CC, Other Hemoglobinopathies	Hemoglobin S, SC, C	Hemoglobin F>A, No other Hemoglobin Variants Present
Carrier AS, AC, A/Other	Hemoglobin AS, AC, A/Other	Hemoglobin F>A, No other Hemoglobin Variants Present
Infectious Diseases		
HIV-1 (by Immunoassay)*	HIV-1 Antibodies	Non-reactive
Organic Acid Disorders (by MS/MS*)		
BKT \ MHBD	Tiglylcarnitine (C5:1)	< 0.07 μ mole/L
GA-I	Glutarylcarnitine (C5DC)	< 0.22 μ mole/L
IBCD	Butyrylcarnitine (C4)	< 1.20 μ mole/L
IVA \ 2-MBCD	Isovalerylcarnitine (C5)	< 0.70 μ mole/L
HMG \ 3-MCC \ 3-MGA	Hydroxyisovalerylcarnitine (C5OH)	< 0.65 or \geq 0.65 and < 0.80 μ mole/L and C5OH/C8 < 10.00
MA	Malonylcarnitine (C3DC)	< 0.22 μ mole/L
MCD \ MUT \ Cbl A,B \ Cbl C,D \ MMA \ PA	Propionylcarnitine (C3)	< 7.0 μ mole/L
Other Genetic Conditions		
BIOT (by Qualitative Colorimetric Method*)	Biotinidase	Activity present
CF (by Time-Resolved Fluoroimmunoassay)	IRT	< Top 5%
GALT (by Qualitative Fluorescence Method*)	Galactose Transferase	Activity present
Krabbe (by MS/MS*)	Galactocerebrosidase	Activity present
Pompe (by MS/MS*)	Alpha-glucosidase deficiency	Enzyme activity present
SCID (by Quantitative Real-Time PCR*)	T-Cell Receptor Excision Circles	TRECs present, Based on Multiple of Median < 1.079
X-Linked Adrenoleukodystrophy (X-ALD) (by LC-MS/MS*)	C26:0 Lysophosphatidylcholine (C26:0 LPC)	< 0.24 μ mole/L
Metachromatic Leukodystrophy (MLD) (by LC-MS/MS*)	C16:0 Sulfatide	< 0.16 μ mole/L
	C16:1-OH Sulfatide	< 0.24 μ mole/L
	ARSA Enzyme Activity (Mayo Clinic Laboratories, Rochester, MN)	\geq 0.1 nmole/mL/h
GAMT (by MS/MS*)	Guanidinoacetate (GUAC)	< 2.80 μ mole/L or (GUAC*1000)/Creatine < 12.00
Mucopolysaccharidosis Type 1 (MS/MS*)	Alpha-L-iduronidase	Activity present
Spinal Muscular Atrophy (qPCR*)	SMN1 Exon 7	SMN1 gene present
Urea Cycle Disorders (by MS/MS*)		
ASA \ CIT	Citrulline (Cit)	< 55 μ mole/L
	Argininosuccinic Acid (ASA)	< 1.00 μ mole/L
ARG	Arginine (Arg)	< 50 μ mole/L
	Arg/Orn	< 0.80

Attention Health Care Provider. Newborn screening tests are intended to provide an early opportunity to detect disorders before symptoms appear. These tests are not diagnostic. Regardless of screening test results, a physician should immediately evaluate any infant who exhibits findings consistent with the targeted disorders noted above. This information has been disclosed to you from confidential records which are protected by state law. State law prohibits any further disclosure of this information without the specific written consent of the person to whom it pertains, or as otherwise permitted by law.

*This test was developed and its performance characteristics determined by the Newborn Screening Program - Wadsworth Center - David Axelrod Institute, 120 New Scotland Ave, Albany, NY. It has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary.

Specimens will be screened for Metachromatic Leukodystrophy (MLD) beginning on 9/12/2025.