North Carolina State Laboratory of Public Health
Newborn Screening Normal Reference Ranges
Last Updated: February 8, 2022

| DISORDER | ABBREVIATION | ANALYTE | | REFERENCE RANGE |
|---|---|---|---|---|
| **FATTY ACID OXIDATION DISORDERS** | | | | |
| Carnitine Uptake Deficiency | CUD | Free carnitine (C0) | | >8.0 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 |
| 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-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 |
| Beta-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) | | <130 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 |

**The values listed above are normal reference ranges. Some condition interpretations are based on the combination of primary markers with their corresponding ratios and/or additional analytes.**