North Dakota Newborn Screening List of Disorders

Core Conditions

Organic Acid Conditions (Metabolic Disorders)

- Propionic Acidemia (PROP)
- Methylmalonic Acidemia (Methylmalonyl-CoA Mutase) (MUT)
- Methylmalonic Acidemia (Cobalamin disorders, Vitamin B12 Disorders) (Cbl A, B)
- Isovaleric Acidemia (IVA)
- 3-Methylcrotonyl-CoA carboxylase (3-MCC)
- 3-Hydroxy 3-Methylglutaric Aciduria (HMG)
- Holocarboxylase Synthase Deficiency (MCD)
- βeta-Ketothiolase Deficiency (βKT)
- Glutaric Acidemia, Type I (GA1)

Fatty Acid Oxidation Disorders (Metabolic Disorders)

- Carnitine Uptake Defect/Carnitine Transport Defect (CUD)
- Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
- Very Long-chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)
- Long-chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
- Trifunctional Protein Deficiency (TFP)

Amino Acid Disorders (Metabolic Disorders)

- Argininosuccinic Aciduria (ASA)
- Citrullinemia, Type I or ASA Synthetase Deficiency (CIT)
- Maple Syrup Urine Disease (MSUD)
- Homocystinuria (Cystathionine Beta Synthetase) (HCY)
- Classic Phenylketonuria (PKU)
- Tyrosinemia, Type I (TYR I)

Endocrine Disorders

- Primary Congenital Hypothyroidism (CH)
- Congenital Adrenal Hyperplasia (CAH)

Hemoglobin Disorders

- S,S Disease (Sickle Cell Anemia) (Hb SS)
- S, βeta-Thalassemia (HB S/βTh)
- S,C Disease (Hb S/C)

Other Disorders

- Biotinidase Deficiency (Metabolic Disorder) (BIOT)
- Critical Congenital Heart Disease (CCHD)
- Cystic Fibrosis (CF)
- Classic Galactosemia (GALT)
- Hearing Loss (HEAR) (Not mandated in ND)
- Severe Combined Immune Deficiency (SCID)
- Spinal Muscular Atrophy (SMA)
- Glycogen Storage Disease Type II (Pompe) (GSD II)
- Mucopolysaccharidosis Type I (MPS I)

Secondary Conditions

Organic Acid Conditions (Metabolic Disorders)

- Methylmalonic Acidemia with Homocystinuria (Cbl C, D)
- Malonic Acidemia (MAL)
- 2-Methylbutyrylglycinuria (2MBG)
- 3-Methylglutaconic Aciduria (3MGA)
- 2-Methyl-3-Hydroxybutyric Aciduria (2M3HBA)

Fatty Acid Oxidation Disorders (Metabolic Disorders)

- Medium/short-chain L-3-hydroxyacyl-CoA Dehydrogenase Deficiency (M/SCHAD)
- Glutaric Acidemia, Type II (GA2)
- Medium-chain Ketoacyl-CoA Thiolase Deficiency (MCAT)
- Carnitine Palmitoyltransferase, Type I (CPT IA)
- Carnitine Palmitoyltransferase, Type II (CPT II)
- Carnitine Acylcarnitine Translocase Deficiency (CACT)

Amino Acid Disorders (Metabolic Disorders)

- Argininemia (ARG)
- Citrullinemia, Type II (CIT II)
- Hypermethioninemia (MET)
- Benign Hyperphenylalaninemia (H-PHE)
- Biopterin Defect in Cofactor Biosynthesis (BIOPT BS)
- Biopterin Defect in Cofactor Regeneration (BIOPT REG)
- Tyrosinemia, Type II (TYR II)
- Tyrosinemia, Type III (TYR III)

Hemoglobin Disorders

- Various other Hemoglobinopathies (Var Hb)

Other Disorders

- T-cell related lymphocyte deficiencies

ND does not screen for the following Recommended Uniform Screening Panel conditions:

Core Conditions

- Mucopolysaccharidosis Type II (MPS II)
- X-linked Adrenoleukodystrophy (X-ALD)
- Guanidinoacetate Methyltransferase Deficiency (GAMT)
- Krabbe Disease (Low Galactocerebrosidase, GALC)

Secondary Conditions

- 2,4 Dienoyl-CoA Reductase Deficiency (DE RED)

- Galactoepimerase Deficiency (GALE)
- Galactokinase Deficiency (GALK)
- Isobutyrylglycinuria (IBG)
- Short-chain acyl-CoA Dehydrogenase (SCAD)

For a complete listing of the disorders on the National Recommended Uniform Screening Panel, visit https://www.hrsa.gov/advisory-committees/heritable-disorders/rusp

The possibility of a false negative or a false positive result must always be considered when screening newborns for disorders, particularly if the infant is symptomatic.