

North Dakota Newborn Screening Program List of Disorders July 2016

Amino Acid Disorders

- (ASA) Argininosuccinic Aciduria *
- (CIT) Citrullinemia, Type I or ASA Synthetase Deficiency *
- (HCY) Homocystinuria (Cystathionine Beta Synthetase) *
- (MSUD) Maple Syrup Urine Disease *
- (PKU) Classic Phenylketonuria *
- (TYR-1) Tyrosinemia, Type I *
- (ARG) Argininemia**
- (BIOPT-BS) Biopterin Defect in Cofactor Biosynthesis **
- (CIT-II) Citrullinemia, Type II **
- (BIOPT REG) Biopterin Defect in Cofactor Regeneration **
- (H-PHE) Benign Hyperphenylalaninemia **
- (MET) Hypermethioninemia **
- (TYR II) Tyrosinemia, Type II **
- (TYR III) Tyrosinemia, Type III **

Organic Acid Conditions

- (GA-1) Glutaric Acidemia, Type I *
- (HMG) 3-Hydroxy 3-Methylglutaric Aciduria *
- (IVA) Isovaleric Acidemia *
- (3-MCC) 3-Methylcrotonyl-CoA carboxylase *
- (Cbl-A,B) Methylmalonic Acidemia (Cobalamin disorders, Vitamin B12 Disorders) *
- (β KT) beta-Ketothiolase Deficiency *
- (MUT) Methylmalonic Acidemia (Methylmalonyl-CoA Mutase) *
- (PROP) Propionic Acidemia *
- (MCD) Holocarboxylase Synthase Deficiency *
- (2M3HBA) 2-Methyl-3-Hydroxybutyric Aciduria **
- (2MBG) 2-Methylbutyrylglycinuria **
- (3MGA) 3-Methylglutaconic Aciduria **
- (Cbl-C, D) Methylmalonic Acidemia with Homocystinuria **
- (MAL) Malonic Acidemia **

Fatty Acid Oxidation Disorders

- (CUD) Carnitine Uptake Defect/Carnitine Transport Defect *
- (LCHAD) Long-chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency *
- (MCAD) Medium-chain Acyl-CoA Dehydrogenase Deficiency *
- (TFP) Trifunctional Protein Deficiency *
- (VLCAD) Very Long-chain Acyl-CoA Dehydrogenase Deficiency *
- (CACT) Carnitine Acylcarnitine Translocase Deficiency **
- (CPT-Ia) Carnitine Palmitoyltransferase, Type I **
- (CPT-II) Carnitine Palmitoyltransferase, Type II **
- (GA2) Glutaric Acidemia, Type II **
- (MCKAT) Medium-chain Ketoacyl-CoA Thiolase Deficiency **
- (M/SCHAD) Medium/short-chain L-3-hydroxyacyl-CoA Dehydrogenase Deficiency **

Endocrine Disorders

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

Hemoglobin Disorders

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

Other Disorders

- (BIOT) Biotinidase Deficiency *
- (CF) Cystic Fibrosis *
- (GALT) Classic Galactosemia *
- (HEAR) Hearing Loss *
- (CCHD) Critical Congenital Heart Disease *
- (SCID) Severe Combined Immune Deficiency *

Disorders on the SACHDNC recommended panel that ND does not screen:

- (MPS I) Mucopolysaccharidosis type I*
- (GSD II) Glycogen Storage Disease Type II (Pompe) *
- (X-ALD) X-linked Adrenoleukodystrophy*
- (DE-RED) 2,4 Dienoyl-CoA Reductase Deficiency **
- (GALK) Galactokinase Deficiency **
- (GALE) Galactopimerase Deficiency **
- (IBG) Isobutyrylglycinuria **
- (SCAD) Short-chain acyl-CoA Dehydrogenase **

* Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) Recommended Uniform Screening Panel – Core Panel

** ACHDNC Recommended Uniform Screening Panel – Secondary Targets – Screening for the Core Panel of disorders may show information about secondary conditions (by-products of mandatory screening)

The possibility of a false negative or a false positive result must always be considered when screening newborns for metabolic disorder.