

North Dakota Newborn Screening Program List of Disorders January 2012

AMINO ACIDEMIAS AND UREA CYCLE DISORDERS

- (ASA) Argininosuccinic aciduria*
- (CIT) Citrullinemia, type 1 or ASA synthetase deficiency*
- (HCY) Homocystinuria (cystathionine beta synthetase)*
- (MSUD) Maple syrup urine disease*
- (PKU) Classic phenylketonuria*
- (TYR-I) Tyrosinemia, type I*
- (ARG) Argininemia**
- (BIOPT-BS) Defects of biopterin cofactor biosynthesis**
- (CIT-II) Citrullinemia, type II**
- (BIOPT-REG) Defects of biopterin cofactor regeneration**
- (H-PHE) Benign hyperphenylalaninemia**
- (MET) Hypermethioninemia**
- (TYR II) Tyrosinemia, type II**
- (TYR III) Tyrosinemia, type III**

ORGANIC ACIDEMIAS

- (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) β -ketothiolase*
- (MUT) Methylmalonic acidemia (methylmalonyl-CoA mutase)*
- (PROP) Propionic acidemia*
- (MCD) Holocarboxylase synthase*
- (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*
- (MCAD) Medium chain acyl-CoA dehydrogenase*
- (TFP) Trifunctional protein deficiency*
- (VLCAD) Very long-chain acyl-CoA dehydrogenase*
- (CACT) Carnitine acylcarnitine translocase**
- (CPT-Ia) Carnitine palmitoyltransferase, type I**
- (CPT-II) Carnitine palmitoyltransferase, type II**
- (GA2) Glutaric acidemia, type II**
- (MCAT) Medium-chain ketoacyl-CoA thiolase**
- (M/SCHAD) Medium/Short chain L-3-hydroxyacyl-CoA dehydrogenase**

ENDOCRINE

- (CAH) Congenital adrenal hyperplasia*
- (CH) Primary congenital hypothyroidism*

HEMOGLOBINOPATHIES

- (Hb SS) S,S disease (Sickle cell anemia)*
- (Hb S/C) S,C disease*
- (HB S/βTh) S, β-thalassemia*
- (Var Hb) Variant hemoglobinopathies**

OTHER

- (BIOT) Biotinidase deficiency*
- (CF) Cystic fibrosis*
- (GALT) Classic galactosemia*
- (GALE) Galactose epimerase deficiency**
- (HEAR) Hearing loss*

* Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) Recommended Uniform Screening Panel – Core Panel

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

Disorders on the SACHDNC recommended panel that we do not screen:

- (SCID) Severe combined immunodeficiency*
- (CCHD) Critical congenital heart disease*
- (DE-RED) 2,4 Dienoyl-CoA reductase deficiency**
- (GALK) Galactokinase deficiency**
- (SCAD) Short-chain acyl-CoA dehydrogenase**
- (IBG) Isobutyrylglycinuria**

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