

DISORDERS SCREENED BY THE PROGRAM

ENDOCRINE DISORDERS:

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

CYSTIC FIBROSIS *

HEMOGLOBINOPATHIES*:

- Sickle cell disease and other hemoglobin disorders

METABOLIC DISORDERS:

- Biotinidase deficiency *
- Galactosemia *

Amino Acid Disorders - Disorders identified through tandem mass spectrometry testing, listed with abbreviations and names:

- (ASA) Argininosuccinate acidemia*
- (CIT 1) Citrullinemia or ASA Synthetase Deficiency*
- (HCY) Homocystinuria (cystathionine beta synthetase)
- (MSUD) Maple Syrup Urine Disease*
- (PKU) Phenylketonuria*
- (TYR-1) Tyrosinemia Type 1*
- (ARG) Arginemia**
- (BIOPT-BS) Defects of bipterin cofactor biosynthesis**
- (CIT-II) Citrullinemia type II**
- (BIOPT-RG) Defects of bipterin cofactor regeneration**
- (H-PHE) Benign hyperphenylalaninemia**
- (MET) Hypermethioninemia**
- (TYR II) Tyrosinemia type II**

- (TRY III) Tyrosinemia type III**

Fatty Acid Oxidation Disorders - Disorders identified through tandem mass spectrometry testing, listed with abbreviations and names:

- (CUD) Carnitine uptake defect (Carnitine transport defect)
- (LCHAD) Long-chain L-3 hydroxyacyl-CoA dehydrogenase*
- (MCAD) Medium chain acyl-CoA dehydrogenase*
- (TRP) Trifunctional protein deficiency*
- (VLCAD) Very long-chain acyl-CoA dehydrogenase*
- (CACT) Carnitine acylcarnitine translocase**
- (CPT-Ia) Carnitine palmitoyltransferase I**
- (CPT-II) Carnitine palmitoyltransferase II**

- (GA-II) Glutaric acidemia Type II**
- (MCKAT) Medium-chain ketoacyl-CoA thiolase**
- (M/SCHAD) Medium/Short chain L-3-hydroxy acyl-CoA dehydrogenase**
- (SCAD) Short-chain acyl-CoA dehydrogenase**

Organic Acid Disorders - Disorders identified through tandem mass spectrometry testing, listed with abbreviations and names:

- (GA-1) Glutaric acidemia type 1*
- (HMG) 3-Hydroxy 3-methylglutaric aciduria *
- (IVA) Isovaleric acidemia*
- (3-MCC) 3-Methylcrotonyl-CoA carboxylase*
- (Cbl-A,B) Methylmalonic acidemia (vitamin B12 disorders)*
- (BKT) Beta Ketothiolase*
- (MUT) Methylmalonic Acidemia (methylmalonyl-CoA mutase)*
- (PROP) Propionic acidemia*
- (MCD) Multiple carboxylase*
- (2M3HBA) 2-Methyl-3-hydroxybutyric aciduria**
- (2MGB) 2-Methylbutyryl-CoA dehydrogenase**
- (3MGA) 3-Methylglutaconic aciduria**
- (Cbl-C, D) Methylmalonic acidemia**
- (IBG) Isobutyryl-CoA dehydrogenase**
- (MAL) Malonic acidemia**

* American College of Medical Geneticists Recommended Disorders - Core Panel

** American College of Medical Geneticists Recommended Disorders - Secondary Targets

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