Missouri Newborn Screening Disorders Tested

Biotinidase Deficiency (BIOT)
Classical Galactosemia (GALT)
Congenital Adrenal Hyperplasia (CAH)
Congenital Primary Hypothyroidism (CH)
Cystic Fibrosis (CF)
Severe Combined Immunodeficiency (SCID) **

Amino Acid Disorders
- Argininemia (ARG, arginase deficiency)
- Argininosuccinate acidemia (ASA, argininosuccinase)
- Citrullinemia type I (CIT-I, argininosuccinate synthetase)
- Citrullinemia type II (CIT-II, citrin deficiency)
- Defects of biopterin cofactor biosynthesis (BIOPT-BS)
- Defects of biopterin cofactor regeneration (BIOPT-RG)
- Homocystinuria (HCY, cystathionine beta synthase)
- Hyperphenylalaninemia (H-PHE)
- Hypermethioninemia (MET)
- Maple syrup urine disease (MSUD, branched-chain ketoacid dehydrogenase)
- Phenylketonuria (PKU, phenylalanine hydroxylase)
- Tyrosinemia type I (TYR-1, fumarylacetoacetate hydrolase) **
- Tyrosinemia type II (TYR-II, tyrosine aminotransferase)
- Tyrosinemia type III (TYR-III, hydroxyphenylpyruvate dioxygenase)

Fatty Acid Disorders
- Carnitine acylcarnitine translocase deficiency (CACT)
- Carnitine uptake defect (CUD, carnitine transport defect) **
- Carnitine palmitoyl transferase deficiency I (CPT-1a)
- Carnitine palmitoyl transferase deficiency II (CPT-II)
- Dienoyl-CoA reductase deficiency (DE-RED)
- Glutaric acidemia type II (GA-II, multiple acyl-CoA dehydrogenase deficiency)
- Long-chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
- Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
- Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)
- Medium/Short chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (M/SCHAD)
- Short-chain acyl-CoA dehydrogenase deficiency (SCAD)
- Trifunctional protein deficiency (TFP)
- Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Organic Acid Disorders
- 2-Methyl-3-hydroxybutyric aciduria (2M3HBA)
- 2-Methylbutyryl-CoA dehydrogenase deficiency (2MBG)
- 3-Hydroxy 3-methylglutaric aciduria (HMG, 3-Hydrox 3-methylglutaryl-CoA lyase)
- 3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)
- 3-Methylglutaconic aciduria (3MGA, Type I hydratase deficiency)
- Beta ketothiolase (BKT, mitochondrial acetoacetyl-CoA thiolase, short-chain ketoacyl thiolase)
- Glutaric acidemia type I (GA-1, glutaryl-CoA dehydrogenase)
- Isobutyryl-CoA dehydrogenase deficiency (IBG)

Organic Acid Disorders (continued)

Isovaleric acidemia (IVA, Isovaleryl-CoA dehydrogenase)
Malonic acidemia (MAL, malonyl-CoA decarboxylase)
Methylmalonic acidemia (CBL A,B; vitamin B12 disorders)
Methylmalonic acidemia (CBL C,D)
Methylmalonic acidemia (MUT, methylmalonyl-CoA mutase)
Multiple carboxylase deficiency (MCD, holocarboxylase synthetase)
Propionic acidemia (PROP, propionyl-CoA carboxylase)

Hemoglobinopathies

Sickle cell disease (Hb S/S)
Sickle hemoglobin-C disease (Hb S/C)
Sickle beta zero thalassemia disease
Sickle beta plus thalassemia disease
Sickle hemoglobin-D disease
Sickle hemoglobin-E disease
Sickle hemoglobin-O-Arab disease
Sickle hemoglobin Lepore Boston disease
Sickle HPFH disorder
Sickle "Unidentified"
Hemoglobin-C beta zero thalassemia disease
Hemoglobin-C beta plus thalassemia disease
Hemoglobin-E beta zero thalassemia disease
Hemoglobin-E beta plus thalassemia disease
Hemoglobin-H disease
Homozygous beta zero thalassemia disease
Homozygous-C disease
Homozygous-E disorder
Double heterozygous beta thalassemia disease

Lysosomal Storage Disorders

Fabry (GLA)
Gaucher (GBA)
Hurler/MPS-I (IDUA)
Krabbe (GALC) **
Pompe (GAA)

Point of Care Screening

Critical Congenital Heart Defects (CCHD)
Hearing

***** There is a lower probability of detection of this disorder during the immediate newborn period.
****** Currently conducting statewide pilot/implementation testing.

The Missouri Newborn Screening Laboratory's goal is to identify infants at risk and in need of diagnostic testing for the above disorders. A normal screening result does **NOT** rule out the possibility of an underlying metabolic/genetic disease.

Revised: 1/3/17