Missouri Newborn Screening Disorders Tested

Biotinidase Deficiency (BIOT)
Classic Galactosemia (GALT)
Congenital Adrenal Hyperplasia (CAH)
Primary Congenital Hypothyroidism (CH)
Cystic Fibrosis (CF)
Severe Combined Immunodeficiency (SCID)
Spinal Muscular Atrophy (SMA)

Amino Acid Disorders
- Argininemia (ARG)
- Argininosuccinic aciduria (ASA)
- Citrullinemia, type I (CIT)
- Citrullinemia, type II (CIT II)
- Biopterin defect in cofactor biosynthesis (BIOPT-BS)
- Biopterin defect in cofactor regeneration (BIOPT-REG)
- Homocystinuria (HCY)
- Hyperphenylalaninemia (H-PHE)
- Hypermethioninemia (MET)
- Maple syrup urine disease (MSUD)
- Phenylketonuria (PKU)
- Tyrosinemia, type I (TYR I)
- Tyrosinemia, type II (TYR II)
- Tyrosinemia, type III (TYR III)

Fatty Acid Oxidation Disorders
- 2,4-Dienoyl-CoA reductase deficiency (DE RED)
- Carnitine acylcarnitine translocase deficiency (CACT)
- Carnitine palmitoyl transferase type I deficiency (CPT-IA)
- Carnitine palmitoyl transferase type II deficiency (CPT-II)
- Carnitine uptake defect (CUD)*****
- Glutaric acidemia type II (GA-2)
- Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
- Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
- Medium-chain ketoacyl-CoA thiolase deficiency (MCAT)
- Medium/Short-chain hydroxyacyl-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-methylglutaryl-CoA lyase deficiency (HMG)
- 3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)
- 3-Methylglutaconic aciduria (3MGA)
- Beta-ketothiolase deficiency (BKT)
- Glutaric acidemia type I (GA-1)
- Holocarboxylase synthetase deficiency (MCD)

Organic Acid Disorders (continued)

Isobutyryl-CoA dehydrogenase deficiency (IBG)
Isovaleric acidemia (IVA)
Malonic acidemia (MAL)*
Methylmalonic acidemia (Cbl A,B)
Methylmalonic acidemia (Cbl C,D)
Methylmalonic acidemia (Methylmalonyl-CoA mutase deficiency) (MUT)
Propionic acidemia (PROP)

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
Gaucher
Krabbe
Mucopolysaccharidosis type I (MPS-I)
Mucopolysaccharidosis type II (MPS-II)
Pompe

Point of Care Screening

Critical congenital heart disease (CCHD)
Hearing loss

***** There is a lower probability of detection of this disorder during the immediate newborn period.

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.

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