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.
****** 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.

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