

# 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 bipterin cofactor biosynthesis (BIOPT-BS)  
Defects of bipterin 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-I, 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.

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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