

Disorders Detected By Mississippi Genetic Newborn Screening

Secondary Conditions

Methylmalonic acidemia with homocystinuria
Malonic acidemia
Isobutyrylglycinuria
2-Methylbutyrylglycinuria
3-Methylglutaconic aciduria
2-Methyl-3-hydroxybutyric aciduria
Short-chain acyl-CoA dehydrogenase deficiency
Medium/short-chain L-3-hydroxyacyl-CoA dehydrogenase deficiency
Glutaric acidemia type II
Medium-chain ketoacyl-CoA thiolase deficiency
2,4 Dienoyl-CoA reductase deficiency
Carnitine palmitoyltransferase type I deficiency
Carnitine palmitoyltransferase type II deficiency
Carnitine acylcarnitine translocase deficiency
Argininemia
Citrullinemia, type II
Hypermethioninemia
Benign hyperphenylalaninemia
Bioterin defect in cofactor biosynthesis
Bioterin defect in cofactor regeneration
Tyrosinemia, type II
Tyrosinemia, type III
Various other hemoglobinopathies
Galactose epimerase deficiency
Galactokinase deficiency
T-cell related lymphocyte deficiencies

Newborn screening might not detect all genetic disorders.
Your physician may request additional testing as necessary.

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Newborn Screening



What is newborn screening?

Mississippi law requires that your baby be tested for a number of genetic disorders. All babies born in Mississippi must be tested before they are six months old.

When will my baby be tested?

The best time to collect the blood sample is while your baby is still in the hospital, and at least 24 hours after delivery.

How are the tests performed?

A small sample of blood is taken by pricking your baby's heel. Several drops of blood are dried on a piece of special absorbent paper which is sent to the laboratory. The lab will run tests on the blood to check for different genetic disorders.

Will I be told of the results?

The test results will be sent to the hospital to be filed with your baby's medical record. You and your baby's doctor will be notified if a problem is detected. Please tell the doctor as soon as possible if you move or change your phone number after your baby is born.

If an abnormal result is detected, it is important to contact you immediately. You may be contacted by someone from the Department of Health or the hospital, or by the doctor. Occasionally a re-test or an additional test may be requested by the laboratory. A common reason for re-testing a baby is that the first sample contained too little blood for adequate testing.

Are genetic disorders serious?

Many genetic disorders are serious; some are even life-threatening. Others slow down physical development or cause mental retardation. Unfortunately, most infants with these disorders show no obvious signs of disease in the first few months. The good news is that proper screening at birth can detect these problems early. With early diagnosis and treatment, some of these genetic disorders can often be managed effectively.

Can these disorders be cured?

At the present time there is no known cure for these disorders. However, the serious effects of many genetic disorders can be lessened if a special diet, medical treatment, or other intervention is started early.

If my baby has one of these disorders, does it mean my future children will also be affected?

Your doctor can discuss this with you or refer you to a specialist. These specialists have information about specific disorders and how they are inherited. A specialist can help you understand any risks for your future children.

Can my older child be tested?

Screening can be performed on children of all ages. However, the Mississippi State Department of Health only provides screening if the child is less than six months old. Some laboratories will provide testing for older children for a fee. Talk to your doctor if you want to test other children in your family.

Disorders Detected By Mississippi Genetic Newborn Screening

Core Conditions

- Propionic acidemia
- Methylmalonic acidemia (methylmalonyl-CoA mutase)
- Methylmalonic acidemia (cobalamin disorders)
- Isovaleric acidemia
- 3-Methylcrotonyl-CoA carboxylase deficiency
- 3-Hydroxy-3-methylglutaric aciduria
- Holocarboxylase synthase deficiency
- β -Ketothiolase deficiency
- Glutaric acidemia type I
- Carnitine uptake defect/carnitine transport defect
- Medium-chain acyl-CoA dehydrogenase deficiency
- Very long-chain acyl-CoA dehydrogenase deficiency
- Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency
- Trifunctional protein deficiency
- Argininosuccinic aciduria
- Citrullinemia, type I
- Maple syrup urine disease
- Homocystinuria
- Classic phenylketonuria
- Tyrosinemia, type I
- Primary congenital hypothyroidism
- Congenital adrenal hyperplasia
- S,S disease (Sickle cell anemia)
- S,Beta-thalassemia
- S,C disease
- Biotinidase deficiency
- Cystic fibrosis
- Classic galactosemia
- Severe Combined Immunodeficiencies

