

**\*\*NOTE:** Newborn screening started with testing for PKU 1964. This is why newborn screening has been referred to as the "PKU test".

### **FATTY ACID OXIDATION DISORDERS**

Fatty Acids are stored energy sources in the body. Children with these disorders cannot properly change fatty acids to energy when the body is running out of its main source of energy, glucose. If this problem is not found or treated early, it can cause brain damage and death. These disorders are treated with diet, medication, plus regular medical care. The disorders are:

(CACT) Carnitine Acylcarnitine Translocase Deficiency  
(CPT-1) Carnitine Palmitoyl Transferase Deficiency-Type1  
(CPT-2) Carnitine Palmitoyl Transferase Deficiency-Type2  
(CTD) Carnitine Transport Defect  
(GA-2) Glutaric Acidemia-type2  
(LCHAD) Long Chain 3 Hydroxylacyl-CoA Dehydrogenase Deficiency  
(MCAD) Medium Chain Acyl-CoA Dehydrogenase Deficiency  
(SCAD) Short Chain Acyl-CoA Dehydrogenase Deficiency  
(VLCAD) Very Long Chain Acyl-CoA Dehydrogenase Deficiency

### **ORGANIC ACID DISORDERS**

Amino acids are the building blocks of proteins (found in large quantity in meat, fish, eggs, milk) and have many functions in the body. Children with these disorders cannot breakdown amino acids in the body properly, and in the process, they form abnormal or harmful amounts of organic acids in the body. If this problem is not found or treated early, brain damage and death can occur. These disorders are treated with special formula, other special diets, medication, plus regular medical care. The disorders are:

(2MBCD) 2-Methylbutyryl-CoA Dehydrogenase Deficiency  
(MBHD) 2-Methyl-3-Hydroxybutyryl-CoA Dehydrogenase Deficiency  
(3MCC) 3-Methylcrotonyl-CoA Carboxylase Deficiency  
(BKD) Mitochondrial Acetoacetyl-CoA Thiolase Deficiency or  $\beta$ -Ketothiolase Deficiency  
(GA-1) Glutaric Acidemia Type-1  
(HMG) 3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency  
(IBD) Isobutyryl-CoA Dehydrogenase Deficiency  
(IVA) Isovaleric Acidemia  
(MCD) Multiple Carboxylase Deficiency  
(MMA) Methylmalonic Acidemia (4 types)  
(PPA) Propionic Acidemia  
(MGA) Methylglutaconic Aciduria Type I, Type II, Type III, and Type IV

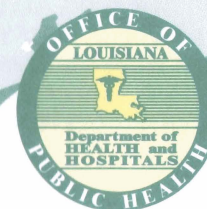


For further information, contact the Louisiana Genetic Diseases Program at (504) 219-4413 or view the Genetic Disease Program website [www.genetics.louisiana.gov](http://www.genetics.louisiana.gov)



## *Louisiana Newborn Heel Stick Screening Program*

*Make Sure Your Baby is Tested*



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**WHY DOES MY BABY NEED NEWBORN HEEL STICK SCREENING?**

Louisiana law requires that every newborn baby be screened for several diseases that if not detected and treated can slow a baby's growth, cause brain and body damage and possibly death. Early detection and treatment can help prevent these very serious problems. This test is called a **NEWBORN HEEL STICK SCREENING**.

**HOW WILL MY BABY BE TESTED?**

All babies must be screened before going home from the hospital. A few drops of blood are collected from your baby by pricking the heel. The blood drops are put onto a special paper form that is sent to the lab for analysis.

Newborn screening is most accurate if your baby's blood is tested after the first 24 hours of life. If your baby goes home from the hospital or if the test is done sooner than 24 hours after birth, another test is needed. This test should be done between 3-7 days after birth and not later than 3 weeks of age.

If your baby was born prematurely or needed special care after birth, the timing of the repeat testing may be different. If your baby is born at home, you should make an appointment to have a newborn screening test done.

**\*\*\* EVEN THOUGH YOUR BABY MAY SEEM HEALTHY, IT IS VERY IMPORTANT TO HAVE YOUR BABY RETESTED. PROBLEMS WILL DEVELOP IF YOUR BABY HAS ONE OF THESE DISORDERS AND IS NOT TREATED!**



*Please remember, newborn screening is just that – only a screening. If your baby's test is unsatisfactory or abnormal, it may need to be repeated. Your baby's doctor or nurse will talk with you about what steps should be taken.*

**WHAT DISEASES ARE SCREENED FOR BY NEWBORN HEEL STICK SCREENING IN LOUISIANA ?**

**BIOTINIDASE DEFICIENCY**  
 Biotinidase deficiency may occur in one of every 60,000 births. Babies with this problem cannot use a vitamin called biotin. Without treatment, the baby's growth and development will not be normal. A baby lacking this vitamin needs medicine containing biotin in addition to regular medical care.

**CONGENITAL ADRENAL HYPERPLASIA (CAH)**  
 An inherited condition making affected babies sick shortly after birth. The two adrenal glands of the body are affected in that they are unable to produce the correct steroid hormones, which control many systems of the body. Babies may develop severe vomiting and dehydration that may result in serious complications or death if untreated.

**CYSTIC FIBROSIS (CF)**  
 CF is an inherited disease that affects the normal airways of the body by obstructing them with mucus, making breathing difficult and making the airways easy to be infected.

Additionally thick digestive fluids also may clog ducts leading from the pancreas to the small intestine. This prevents proper food digestion leading to severe digestive problems and slow growth. All of these problems can be treated when detected, but not cured. Good treatment, of course, lengthens the life of affected children, and lets them lead close to normal lives. Untreated, CF is a fatal disease early in life.

**CONGENITAL HYPOTHYROIDISM**  
 Congenital hypothyroidism may occur in one of every 3,500 births. A baby with this problem does not make enough thyroid hormone which is needed to help the baby grow normally and stay healthy. Without the right amount of thyroid hormone, a baby's growth and development will not be normal. This problem is treated with daily medicine to replace the hormone in addition to regular medical care.

**GALACTOSEMIA**  
 Galactosemia may occur in one of every 50,000 births. A baby with this problem cannot digest the sugar galactose. If not treated, galactose will build up in the baby's body and cause serious health problems, including poor weight gain, eye, liver, and brain damage. Babies with galactosemia must not have foods containing galactose or lactose, including breast milk and some infant formulas.

This problem is treated with a special diet in addition to regular medical care.

**PHENYLKETONURIA (PKU)**  
 PKU may occur in one of every 15,000 births. Babies with PKU cannot properly use a substance found in proteins called phenylalanine. If this problem is not found or treated early, PKU can cause brain damage and learning problems. PKU is treated with special formula and diet, plus regular medical care.

**SICKLE CELL DISEASE**  
 Sickle cell disease is an inherited blood disorder that may occur in one of every 385 African American births. Babies with sickle cell disease are at risk for serious infections and other problems which can cause death if not treated with antibiotics. In addition, these babies need immunizations and regular medical care. The newborn screening for sickle cell disease may also find other changes in the blood that may or may not need treatment.

**EXPANDED NEWBORN SCREENING**

Recent advances have enabled newborn screening services to improve and expand testing to include additional treatable disorders. One of these advances is an instrument called a tandem mass spectrometer and the process this instrument controls is called tandem mass spectrometry. This instrument can detect many disorders of body chemistry from a few drops of blood that are collected on a special paper during the first few days of life. Tandem mass spectrometry is very accurate, fast and reduces the need for many different methods of testing.

These are the diseases tested for in Louisiana by tandem mass spectrometry:

**AMINO ACID DISORDERS**  
 Amino acids are the building blocks of proteins (meat, fish, eggs, milk) and have many functions in the body. Inherited disorders of amino acids can be the result of defects in either the breakdown and removal of amino acids from the body, or in the body's ability to get the amino acids into the cells. If this problem is not found or treated early, brain damage and death can occur.

These disorders are treated with special formula, other special diets, medication, plus regular medical care. The disorders are:

- (ARG) Argininemia
- (ASA) Argininosuccinic Aciduria
- (ASS) Citrullinemia or ASA Synthetase Deficiency
- (HCY) Homocystinuria
- (MSUD) Maple Syrup Urine Disease
- (PKU) Phenylketonuria)\*\*
- (Tyr) Tyrosinemia

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