NEWBORN SCREENING

MISSOURI DEPARTMENT OF
HEALTH &
SENIOR SERVICES

Health.Mo.Gov/NewbornScreening

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NEWBORN SCREENING

Newborn screening refers to screenings performed on newborns shortly after birth to protect them from the dangerous effects of disorders that otherwise may not be detected for several days, months or even years. Missouri law requires all babies born in the state to be screened for certain rare, but serious, conditions. All newborns are screened for more than 70 disorders, including hearing loss and critical congenital heart disease. A small sample of blood is collected from your baby's heel shortly after birth and is then sent to the Missouri Department of Health and Senior Services (DHSS) State Public Health Laboratory (SPHL) for testing.

Whether your baby is born in the hospital or at home, the newborn screen should be collected between 24 and 48 hours after birth. The results of your baby's blood screen will be given to your baby's health care provider and the birthing hospital or midwife. Sometimes, more than one newborn screen is needed. If an additional newborn screen is needed, you will be notified either by your baby's health care provider, the hospital, midwife or staff from DHSS. If so, it is very important that you bring your baby back for a repeat newborn screen as soon as possible.

The results of your baby's blood spot screen will be given to your baby's health care provider and the birthing hospital or midwife. If an additional newborn screen is needed, it is very important that you bring your baby back for a repeat newborn screen as soon as possible.

FREQUENTLY ASKED QUESTIONS

Why is newborn screening important?

The disorders your baby will be screened for are rare. However, these disorders can result in severe injury to the brain, organs or nervous system, and some may result in death if not treated. Newborn screening helps to identify babies needing diagnosis and treatment, such as a special diet or medication. Since symptoms are not generally noticeable at birth, the only way to find these disorders before permanent damage occurs is by newborn screening. Early treatment will help your baby grow up as healthy as possible.

What if the screening results are abnormal?

A “positive” or abnormal screening result only means that a baby might have a disorder. Sometimes positive screening results are found in babies that do not have the disorder. A diagnosis of a disorder is not made with the first lab test. Further testing will be necessary to determine if your baby actually has the disorder.

If you are asked to repeat the newborn screen or to take your baby to have additional testing, please act quickly so tests can be completed and final results obtained. If needed, treatment must be started as soon as possible to prevent the onset of developmental delays or other damaging results.

Is there a cure for the disorders?

There are no cures for most of these disorders. If these disorders are found and babies get treatment early, serious problems can be prevented or reduced. If babies with these disorders get early and continuous treatment, most can grow and develop normally and live healthy lives.

When is rescreening needed?

When newborn screening samples arrive at the SPHL, they are carefully reviewed to make sure the test results will be accurate and reliable. Parents may be asked to bring their babies back for rescreening if:

- The specimen was collected too early (less than 24 hours after birth).
- The baby was given treatment or medicine that interfered with the results.
- A problem occurred with the quality of the sample due to issues with the collection or handling of the blood spots.

A rescreening may also be requested if the results indicate a borderline or low risk result. This means the results were almost normal, but not quite, and a repeat screen is needed to determine if there is cause for concern.

What can parents do?

The goal of the Newborn Screening Program is to prevent serious health problems through early screening. You can help!

- Leave two phone numbers (your number and one for a relative, neighbor or friend) with the hospital or midwife and health care provider to assure you can be contacted regarding the screening results.
- Let the hospital or midwife know the name of your baby's health care provider.
- Make sure your baby is screened before you leave the hospital or 24 to 48 hours after birth if your baby is born at home.
- Ask your baby's health care provider or midwife about your baby's newborn screening results.
- Listen to your baby's health care provider and follow recommendations if more tests or medical appointments are needed.

Do not rely on others to make sure your baby gets tested!

DISORDERS INCLUDED IN NEWBORN SCREENING

Amino Acid Disorders

This group of disorders prevents a baby's system from breaking down certain waste products in their blood such as phenylalanine, ammonia or other amino acids. Buildup of amino acids and/or bi-products of amino acid metabolism in the blood causes severe medical complications. In each of these disorders, the lack of early identification and treatment may result in serious medical consequences, including developmental delays, failure to thrive and/or death. Early identification and treatment with a special diet or medications can help a baby to grow and develop as healthy as possible.

Amino Acid Disorders screened for include:

- 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, cystathionine beta synthase)
- 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)

Biotinidase Deficiency

A disorder found in babies who are missing the enzyme biotinidase. This can lead to seizures, developmental delay, eczema and hearing loss. Symptoms can be prevented if babies are started on treatment with a vitamin called biotin in the first few weeks of life.

Congenital Adrenal Hyperplasia

A disorder caused by an enzyme deficiency that results in the adrenal gland producing too little of one hormone and too much of another. Some babies with this disorder are at risk of sudden death. Problems can be prevented with hormone treatment started early.

Congenital Hypothyroidism

A disorder caused by not having enough thyroid hormone. The most common effects of congenital hypothyroidism are developmental delays and poor growth. If treatment with thyroid medication starts in the first few weeks of life, these children usually develop normally.

Cystic Fibrosis

A disorder that causes thick, sticky mucus to build up in the lungs, digestive system and other organs of the body. This can lead to respiratory and digestive problems, which can be very serious. Early detection and treatment are important for lung health, growth and development. Newborn screening may detect some, but not all, carriers of cystic fibrosis.

Fatty Acid Oxidation Disorders

This group of disorders causes a baby to have trouble using fat for energy. This leads to a buildup of toxic fatty acids, which may cause metabolic crisis. A metabolic crisis can lead to seizures, failure to breathe, cardiac arrest, serious brain damage or death.

However, screening can provide diagnosis before symptoms occur. Early diagnosis and treatment of these disorders allows for proactive treatment and management to prevent or control metabolic crisis effectively.

Fatty Acid Oxidation Disorders screened for include:

- 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)

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

Classic Galactosemia

A disorder in which a simple sugar called “galactose” cannot be broken down in the body. Galactose is found in breast milk, many formulas and milk products. If it remains at high levels in the body, galactose will harm the baby’s eyes, liver and brain. If left untreated, classical galactosemia results in death. When started early, a special diet can prevent these problems.

Sickle Cell Disease

Sickle cell disease is mainly found in African Americans and those of Mediterranean background. In sickle cell disease, the red blood cells change from the normal round shape to an abnormal sickle shape. These sickled cells can clog blood vessels so not enough oxygen can be carried to organs and tissues. Babies with sickle cell disease are more likely to have anemia, pain, strokes and life-threatening infections. Early treatment with antibiotics, immunizations and parent education can help. These measures can prevent serious infections in childhood and reduce health problems. It is important that affected children be under the care of a health care provider early in life for medication and other treatment.

Disorders that may be detected include:

- Sickle cell anemia
- Sickle hemoglobin-C disease
- 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 Hereditary Persistence of Fetal Hemoglobin disease
- 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 disease
- Double heterozygous beta thalassemia disease

Newborn screening may also identify carrier or trait conditions associated with sickle cell disease and other hemoglobin variants.

Lysosomal Storage Disorders

Lysosomal Storage Disorders (LSDs) are a group of genetic disorders that result in enzyme deficiencies within the lysosomes of the body's cells. Lysosomes are the recycling bins of the cells and they use very specific enzymes to break down and recycle large unwanted waste products. In the LSDs, one of these enzymes is absent or not working. This causes a buildup of waste products in the lysosomes that results in damage to muscles, nerves and/or certain organs in the body. If left untreated, the severe infantile forms of these disorders lead to critical and irreversible disabilities, requiring continuous and extraordinary care, and ultimately lead to death. Treatments may be available for these disorders if caught early, before symptoms begin to appear in the infant.

Lysosomal Storage Disorders screened for include:

- Fabry disease
- Gaucher disease
- Hunter syndrome
- Hurler syndrome
- Krabbe disease
- Pompe diease
- Acid Sphingomyelinase Deficiency (ASMD)

Organic Acid Disorders

This group of disorders prevents a baby's system from removing certain waste products of proteins and other substances from their blood. These disorders can have a variety of mild to severe effects on babies including metabolic crisis and problems with the heart, muscles and some organs. Babies with these disorders may become rapidly sick, have seizures, go into a coma and could die without treatment.

Organic Acid Disorders screened for include:

- 2-Methyl-3-hydroxybutyric acidemia (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)
- 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)

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

Severe Combined Immunodeficiency

Severe Combined Immunodeficiency (SCID) is a rare, but very serious genetic defect characterized by extremely low counts of T-cells, which are a specific type of white blood cell. White blood cells are responsible for your immune system's ability to fight off infection. Babies affected by SCID may appear normal and healthy for the first few months of life until the immunity given to them by their mother begins to disappear. Children with a normal immune system can fight off everyday germs, but children with SCID will not be able to recover from something as simple as the common cold. Without early detection and treatment, children affected by SCID will die of infections before their first or second birthday. Treatment with bone marrow transplant within the first few months of life, before infection has occurred, can cure SCID in most cases.

Spinal Muscular Atrophy

Spinal muscular atrophy (SMA) is a condition that affects the motor neurons of the spinal cord. Motor neurons are specialized nerve cells that control the muscles used for activities such as breathing, crawling and walking. Babies affected by SMA gradually lose motor neurons which causes progressive weakness and atrophy (wasting). Without treatment, babies with the most severe form of SMA typically die in early childhood. Although SMA cannot be cured, it can be treated with therapies or medication. Early intervention and treatment of SMA is important because the therapies and medication are most effective in improving children's quality of life when started within the first few months of life.

Adrenoleukodystrophy

Adrenoleukodystrophy (ALD) is an X-linked genetic disease that most severely affects males. This disease mainly affects the nervous system and the adrenal glands. ALD often causes progressive loss of the myelin sheath, the fatty covering that acts as an insulator and surrounds the nerves in the brain and spinal cord. This can cause a variety of neurological problems including cognitive, mobility and sensory issues. If left untreated, the severe form of ALD (cerebral ALD) can lead to critical and irreversible disabilities that can require continuous and extraordinary care and ultimately lead to death. ALD may also cause adrenal insufficiency, a deficiency of certain hormones due to damage to the adrenal glands. Adrenal insufficiency can cause weakness, weight loss, skin changes, vomiting and coma.

Early diagnosis through newborn screening allows for proactive care and consistent monitoring. Adrenal insufficiency can be treated with medication and bone marrow transplant can halt the progression of the cerebral form of ALD once detected.

Newborn Hearing Screening

All babies receive a safe and painless hearing screening shortly after birth to identify possible hearing loss. Hearing loss is one of the most common birth defects, occurring in one per 1,000 newborns screened in Missouri. If hearing loss is not detected and managed early, it can impede speech, language and cognitive development. If the final screening results indicate your baby may have hearing loss, it is important to work with your health care provider to make an appointment with an audiologist to perform a thorough hearing test before your baby is three months old.

Do you know the results of your baby's hearing screening?

Critical Congenital Heart Disease

Critical congenital heart disease (CCHD) is the name given to specific congenital heart defects. These defects in the heart occur before birth, cause blood to flow in an abnormal pattern, and may lead to blockage of blood flow throughout the body. If left untreated, these defects can lead to death or can cause serious developmental delay.

CCHD screening is a simple bedside test to determine the amount of oxygen in the baby's blood. The test is painless and takes just a few minutes. This screening test does not detect all CCHDs, so it is possible to still have a CCHD or other congenital heart defect with a negative screening result.

NEWBORN SCREENING SAMPLES

Missouri Newborn Screen Sample Storage and Release Policy

Once the newborn screening test is completed, the Missouri State Public Health Laboratory will store the remaining newborn screening sample for five years. The storage is secure. Missouri state law allows for the stored sample to be used for research. The research may help improve methods for detecting illnesses. The research may also find better ways to test, treat and cure major childhood diseases. Your baby is not identified to the researcher in any way. After five years, the rest of the newborn screening sample will be destroyed.

The law allows the parent or legal guardian the option of not having their baby's leftover newborn screening sample stored or studied. You may ask the state laboratory to:

Give the extra newborn screening sample back to you.

Destroy the newborn screening sample after the newborn tests are completed.

Store the extra newborn screening sample for five years but do not release it for study.

If you choose NOT to allow your baby's leftover newborn screening sample to be studied, select one of the three options from above and write to the laboratory director at: Missouri State Public Health Laboratory, Newborn Screening Laboratory, P.O. Box 570, Jefferson City, MO 65102.

Include this information:

- Baby's name.
- Mother's first and last name.
- The option you selected from above.
- State if you are the parent and/orlegal guardian.
- Baby's date of birth.
- Please where baby was born.
- Your current address.
- Your signature and current date.

If you have questions or need assistance, contact the Newborn Screening Laboratory at 573-751-3334 or visit Health.Mo.Gov/lab/newborn.

Benefits of Storing Newborn Screening Samples

There are many reasons why newborn screening samples are kept, many of which benefit your family and other Missouri families. In some cases, samples are requested by the family or the baby's health care team. The baby's sample is available to you for other health related testing within five years of storage. The baby's sample is available to help identify a missing or deceased child within five years of storage. If your child has an illness and is enrolled in a research study, parents may request that their baby's newborn screening sample be returned to them in order that they may send it to the researcher within five years of storage.

For research purposes, all identifying information is removed from the samples (baby's name, parent's name, parent's address, hospital of birth, etc.). The researcher does not know who the baby is. These samples may be used to:

- Provide quality assurance in newborn screening.
- Perform public health studies and research to help develop newborn screening tests and better understand diseases for the benefit of the general public.
- Search for new markers for chronic diseases such as childhood leukemia, sickle cell disease, autism and diabetes.

Only those research projects that undergo careful scientific and ethical review will be given approval to use newborn screening samples.

SPECIAL NOTES

The newborn screening tests are not diagnostic. They are a “screen” designed to detect newborns who need further testing to determine if they have certain disorders. The screening tests are very efficient and provide newborns with the best opportunity for having the disorders identified early. However, like most laboratory tests, the tests used for newborn screening cannot guarantee that every affected newborn will be identified, or that only infants at higher risk of being affected will be identified. Therefore, it is important to recognize that there will be some “false positives” (newborns with a positive or abnormal screen result who are later found to have normal results), and the possibility of “false negatives” (newborns with normal screening results who are later found to have one of the conditions).

Medical Homes

The Missouri Department of Health and Senior Services supports and encourages access to a medical home for all children, with and without special health care needs. A medical home is defined by the Health Resources and Services Administration as “a home base for any (person’s) medical and non-medical care. The medical home is a cultivated partnership between the patient, family and primary provider in cooperation with specialists and support from the community.”

For more information on newborn screening, contact:
Missouri Department of Health and Senior Services at
800-877-6246 or visit the
Health.Mo.Gov/Newbornscreening
For more information on medical homes, visit:
https://medicalhomeinfo.aap.org/overview/Pages/Whatisthemedicalhome.aspx.

MISSOURI DEPARTMENT OF
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For information regarding other Maternal and
Child Health Services call: 800-TEL-LINK (800-835-5465)

This project is/was funded in part by the Missouri Department of Health and Senior Services Title V Maternal Child Health Services Block Grant and is/was supported by the Health Resources Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) under grant #B04MC52935, Maternal and Child Health Services for $12,742,189, of which $0 is from nongovernmental sources. This information or content and conclusions are those of the author and should not be construed as the official position or policy of, nor should any endorsements be inferred by HRSA, HHS or the U.S. Government.

#14 (09-2026)