

Understanding Your Baby's Newborn Screening Tests



Ohio
Department of Health



Why does my baby need newborn screening?

All parents hope that their babies will be healthy. Some babies who look perfectly healthy at birth may have problems that will affect how they grow and develop. Newborn screening tests babies for many medical conditions that are not obvious at birth, but need medical treatment. By finding the medical problem soon after birth, babies quickly receive the treatment they need. Newborn screening improves the health and development of babies.

Every year in Ohio, more than 500 newborns needing medical care are identified by newborn screening. About 200 infants are deaf or hard of hearing, about 80 infants have serious heart diseases, and about 300 infants have serious medical disorders that can be found by testing their blood.

What type of screening will be done on my baby?

Ohio wants to protect the health of all newborns; so state law requires that newborns receive three different types of screening before leaving the hospital:

- Hearing screening for risk of hearing loss;
- Pulse oximetry screening for finding problems with the structure of the heart; and
- Blood spot screening for risk of several different medical conditions.

What health problems does Ohio screen for?

■ Hearing screening

Hearing screening identifies risk for hearing loss. Hearing loss can be caused by many different things. Early identification of hearing loss helps your baby to develop communication, language and speech skills.

■ Pulse oximetry screening

There are many different congenital heart diseases. In Ohio, pulse oximetry screening is used to identify seven types of critical heart disease including: hypoplastic left heart syndrome, pulmonary atresia, tetralogy of Fallot, total anomalous pulmonary venous return, transposition of the great arteries, tricuspid atresia, and truncus arteriosus.

■ Blood spot screening

Ohio screens for several different health conditions from blood spots. The conditions included in the Ohio newborn screen are recommended by the March of Dimes and the US Department of Health and Human Services (a complete list can be found at the end of this pamphlet and more detailed information can be found on the website:

<https://www.odh.ohio.gov/odhprograms/phl/newbrn/nbsdisorderslist.aspx>).

How and when will my baby be screened?

■ Hearing screening

Before leaving the hospital, your baby's hearing screen will be done. It is usually done in the mother's room or nursery. It only takes 15 minutes and your baby can sleep during the screening. There are two different types of tests used to do the screening. Your baby may have one or both of the tests described below.



● Otoacoustic Emissions

(OAE): This test uses small, soft foam or rubber tips. These tips are placed inside the baby's ears and the baby hears soft sounds. Small echoes are recorded from the baby's ear to see if there is good function in the inner ear.

- **Auditory Brainstem Response (ABR):** This test uses three small patches that are placed on the baby's face, shoulder and neck; then small headphones are placed over the baby's ear or small ear tips are placed in the baby's ears and the baby hears soft sounds. This test records information from the hearing nerve.

■ Pulse oximetry screening

Pulse oximetry screening is a simple, painless test that is usually done about 24 hours after your baby is born. A machine called a pulse oximeter measures the amount of oxygen in your baby's blood through sensors placed against your baby's skin. The sensors are usually placed on a hand and a foot.

■ Blood spot screening

A few drops of blood are collected from a prick in your baby's heel. This heel prick is usually done the day after the baby is born. The drops of blood are sent to the Ohio Department of Health Laboratory where testing is done for many different medical conditions.

What if my baby was not born at a hospital?

All babies born in Ohio should receive newborn screening. If your baby was not born in a hospital, contact your midwife, doctor, or local health department for help in arranging the newborn screening tests. In many counties, the screening can be done in your home. It is important that the blood spot screening be done after your baby is at least 24 hours old, but before your baby is six days old.

How do I find out my baby's results?

■ Hearing screening

The results of your baby's hearing screen will be given to you in writing by hospital staff before you leave the hospital. Hospital staff will also help with scheduling a hearing evaluation if more testing is needed.

■ Pulse oximetry screening

Your baby's healthcare provider will give you the results of the screening. If the test results are negative, your baby has normal levels of blood oxygen. If the test results are positive, your baby has low levels of oxygen in their blood. This does not always mean your baby has a heart disease. It means that your healthcare provider will continue to monitor your baby and may repeat the test a little later or do another type of testing called an echocardiogram.

■ Blood spot screening

The results of your baby's blood spot screening will be sent to the birth hospital and to your baby's health care provider as soon as the testing is completed. The results are usually available within two weeks. For most babies, the screen results are normal and parents will get the results at their baby's health check-up. If the newborn screen is not normal, then the healthcare provider will contact the family and arrange for additional testing.

It is important to provide your hospital and healthcare provider with your correct address and phone number so they can reach you if needed.

What if my baby needs further testing?

Your baby's healthcare provider will contact you if your baby needs further testing. He or she will tell you why your baby needs to be retested and the next steps needed. If your baby needs further testing, follow the healthcare provider's advice. Some testing will need to be done right away and other tests may need to be done when your baby is a little older. In some cases, your baby will be referred to a specialist.

■ Hearing screening

If your baby does not pass the hearing screen, your baby will be referred to an audiologist for further testing within two to three weeks. If your baby has hearing loss, the Infant Hearing Program will refer your baby for home-based, early intervention services to help with the development of communication and language. The earlier a child's hearing loss is identified, the earlier that child can begin developing communication skills. This is important for development, educational achievement, literacy levels and social-emotional development.

■ Pulse oximetry screening

If your baby does not pass the pulse oximetry screening, further testing will be done to check for congenital heart disease. This testing may be done before your baby leaves the hospital or your baby will be referred to a pediatric cardiac specialist for further medical care. Critical congenital heart disease requires medical treatment within the baby's first few weeks or months of life to prevent death or disability.

■ Blood spot screening

If your baby's blood spot screening contains results that are not normal, your baby's healthcare provider will arrange for further testing right away. In some cases, your baby will be referred to a specialist. It is important to identify babies that have these conditions quickly, so they can start medical treatments that will improve their health and development as soon as possible.

What if I have questions?

- Ask your baby's healthcare provider if you have questions or concerns.
- Check the Ohio Department of Health's Newborn Screening website:
www.odh.ohio.gov/odhPrograms/phl/newbrn/nbrn1.aspx.

Does my baby have to be screened?

Newborn screening saves lives and improves the health of babies and it is required by Ohio law for all babies born in Ohio. In some circumstances, parents can refuse newborn screening.

Parents may refuse any type of newborn screening if it conflicts with their religious beliefs. Parents may also refuse hearing screening for any other reason. Parents that refuse newborn screening must sign a form stating that they are objecting to this screening for their baby.



Ohio law also gives parents the option to refuse testing for one medical condition called Krabbe disease which is included in blood spot screening. Krabbe has not been recommended for blood spot screening on a national level. Krabbe is a rare disease that results in severe damage to a person's brain and nerves over time. There is no proven cure for Krabbe disease. If it is detected shortly after birth, in some cases, a bone marrow transplant done in the first few weeks of life may slow development of the disease. Parents in Ohio have the option of refusing blood spot screening for Krabbe disease for any reason. If you do not want your baby screened for Krabbe disease, you must inform the hospital nursing staff or midwife that you refuse Krabbe screening at the time your baby's blood sample is taken. Only Krabbe screening can be refused by law; tests for all other conditions included in the Ohio blood spot screen will be performed.

Health conditions included in blood spot screening

Amino Acid Disorders are caused by a problem with the body's ability to properly use certain amino acids found in food. Dangerous chemicals build up in the body starting soon after birth and can damage the brain and other organs. Special diets or supplements may help treat these health problems and slow or prevent further damage. The Ohio program screens for the following amino acid disorders: Arginemia (ARG), Arginosuccinic Acidemia (ASA), Citrullinemia Type I (CIT) and Type II (CIT II), Homocystinuria (HCY), Hypermethioninemia (MET), Maple Syrup Urine Disease (MSUD), Phenylketonuria (PKU), Tyrosinemia Type I (TYR I), Type II (TYR II) and Type III (TYR III).

Fatty Acid Disorders can interfere with the body's ability to turn fat into energy. Babies with fatty acid disorders can have heart problems, difficulty breathing, seizures, extreme weakness and death. Special diets, eating often, and medication may help prevent symptoms. The Ohio program screens for the following fatty acid disorders: Carnitine Acylcarnitine Translocase Deficiency (CACT), Carnitine Palmitoyl Transferase Deficiency Type II (CPT II), Carnitine Uptake Defect (CUD), Glutaric Acidemia Type II (GA-2), Long-Chain Acyl-CoA Dehydrogenase Deficiency (LCHAD), Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD), Trifunctional Protein Deficiency (TFP), Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD).

Organic Acid Disorders occur when a baby's body cannot remove certain waste products from its blood. This can lead to vomiting, low blood sugar, coma or death. Treatment may include a special diet and medication. The Ohio program screens for the following organic acid disorders: 2-Methylbutyryl-CoA Dehydrogenase Deficiency (2MBG), 3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency (HMG), 3-Ketothiolase Deficiency (BKT), 3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC), Glutaric Acidemia Type I (GA-1), Isobutyryl-CoA Dehydrogenase Deficiency (IBG), Isovaleric Acidemia (IVA), Methylmalonic Acidemia (MMA), Multiple CoA Carboxylase Deficiency (MCD), Propionic Acidemia (PROP).

Biotinidase Deficiency occurs when babies cannot properly use the vitamin biotin. Problems with skin rashes, seizures, hearing loss, or developmental delay may be prevented by adding extra biotin to the diet.

Congenital Adrenal Hyperplasia (CAH) results when the body does not make certain hormones produced by the adrenal glands. Babies with this condition can develop life-threatening episodes of dehydration and coma. Baby girls can have abnormal looking genitalia. Medication may help prevent life-threatening complications of this condition.

Congenital Hypothyroidism results when the baby's body does not make enough thyroid hormone to keep the baby growing and developing. Medication may help prevent growth problems and developmental delay.

Cystic Fibrosis (CF) is an inherited disease of the lungs and digestive system that can cause recurring chest infections and malnourishment. Early detection and treatment may improve growth and decrease risk of infections.

Galactosemia occurs when a baby's body cannot break down a milk sugar (galactose). A special diet without milk sugar may prevent brain and liver damage.

Sickle Cell Disease and other Hemoglobinopathies are inherited disorders that affect red blood cells. Some of these disorders can cause severe pain and infections or other serious health problems that can lead to death. Medication may be needed to lower the chance of infection and other problems. Babies with these disorders need to be seen by a specialist.

Severe Combined Immunodeficiency (SCID) includes a group of rare but serious immune disorders. The baby's body is unable to make certain cells that protect the body from infection. Untreated infants develop life-threatening infections due to bacteria, viruses and fungi. Treatment can reduce the threat of infections.

Lysosomal Storage Disorders (LSD) are conditions that occur when the body does not make enzymes needed to break down certain large molecules into smaller, useable substances. The large molecules build up in the cells and can cause damage to the brain, nerves, heart, bones, liver and other organs. Treatment may include enzyme replacement and bone marrow transplant. Ohio screens for the following lysosomal storage disorders: Krabbe Disease, Mucopolysaccharidosis Type I, and Glycogen Storage Disease Type II (Pompe Disease).

