NEWBORN Screening

Important **Newborn Screening** Information for Parents in Washington State

Washington State Department of Health

www.doh.wa.gov/NBS DOH 304-007 May 2020

Newborn Screening Program

The Newborn Screening Program is located at the Washington State Public Health Laboratories in Shoreline, Washington. Every year, the program tests about 175,000 blood samples to help catch serious conditions early and give Washington babies a healthy start.

Congratulations on the birth of your baby!

As you begin life with your child, there are many things to consider. Newborn screening is one important way you can ensure that your baby has the healthiest start possible.

What is Newborn screening?

Newborn screening can prevent serious health problems or even save your baby's life. Newborn screening helps identify a number of rare and serious health conditions right after birth. With this information in hand, parents and doctors can begin any needed treatment before harmful effects occur.

1 out of every 480 newborns has a serious condition and can benefit from early diagnosis and treatment thanks to newborn screening.

How Is Newborn Screening Done?

A health care provider collects a few drops of blood from your baby's heel onto special filter paper, called a newborn screening card. This is a safe and routine procedure that can be done at a hospital, clinic, laboratory, birth center, or at home with a midwife. Once the blood dries, the card is sent to the Newborn Screening Program at the Washington State Public Health Laboratories for testing.

Why Is Newborn Screening Needed?

Babies who appear healthy at birth and come from healthy families can still have serious health conditions. Newborn screening identifies these disorders and helps ensure that babies get the treatment they need right away so they can grow up to be as healthy as possible. If these disorders are not caught early, they may cause potentially life-threatening health issues as the baby grows.

All Washington Babies Are Screened

Newborn screening is the fastest way to identify these rare conditions before the baby gets sick. To ensure Washington newborns are as healthy as possible, state law requires that all babies born in the state get screened before they are two days old. This allows the baby to get needed treatment as soon as possible.

Babies should be screened twice.

1

The **first** small blood sample is collected shortly after birth, when your baby is about one to two days old. **This screening is critical for detecting severe forms of the conditions.**

2

The **second** sample is usually collected at a well-child visit, when your baby is one to two weeks old. **This screening will help detect conditions that were not detectable right after their birth.**

How Will I Know My Baby's Screening Results?

Ask your baby's health care provider for the newborn screening results. The hospital, clinic, lab, or midwife that submitted your baby's screening test should be notified of the results within about a week. The Newborn Screening Program will contact your baby's health care provider sooner if there are immediate concerns.

What Will the Newborn Screening Results Tell Me?

Most newborn screening results are normal. A **positive, abnormal,** or **inconclusive** result only means that your baby might have one of the conditions. Additional tests will be needed to make a final determination. The Newborn Screening Program will work with you, your baby's health care provider, and doctors who specialize in treating these conditions to ensure your baby has the best care.

Newborn screening tests are very accurate, but no screening test is perfect. If your baby does not seem well or you have questions about the results from your baby's newborn screening, please contact your baby's health care provider.

How Are These Disorders Treated?

Each disorder is different. Some disorders are treated with special diets and others are treated with medications or other medical services, such as physical therapy or surgery. Treatment early in life will allow your baby the best opportunity for healthy growth and development.

How Much Does Screening Cost?

There is a one-time fee that covers screening done in Washington State. Typically, this fee is included with other delivery or birth fees at the hospital. However, some medical facilities may charge an additional fee for collecting the blood sample. These fees are usually covered by insurance.

Visit the Newborn Screening Program's website for the current fee and more information: **doh.wa.gov/NBS/ScreeningCost**

What Happens to the Blood Samples?

According to state law, the newborn screening cards are kept in secured storage for 21 years and then destroyed. The lab may use the blood samples to improve newborn screening testing. No identifiable information will be used without written consent from the parent or guardian. You may request that your baby's sample be destroyed at any time after newborn screening has been completed.

Visit the Newborn Screening Program's website for more information on privacy policies: **doh.wa.gov/NBS/PrivacyPolicies**

Can I Refuse to Have Newborn Screening Done on My Baby?

Due to the importance of preventing the devastating effects of these conditions, parents can only refuse newborn screening for their baby if it conflicts with their religious beliefs or practices. To refuse newborn screening on those grounds, a parent or guardian must sign the refusal section on the back of screening card and send it to the Newborn Screening Program at the Washington State Public Health Laboratories.

What Disorders Does the Newborn Screening Test Identify?

Newborn screening helps identify the following conditions. You can learn more at doh.wa.gov/NBS/disorders

Amino Acid Disorders

1 IN 10,000 BIRTHS

These disorders affect the body's ability to use certain parts of food called amino acids for growth and energy. This causes substances to build up in the body and seriously impact health, growth, and learning, and can sometimes result in death.

- Argininosuccinic acidemia (ASA)
- Citrullinemia (CIT)
- Homocystinuria (HCY)
- Maple syrup urine disease (MSUD)
- Phenylketonuria (PKU)
- Tyrosinemia type I (TYR-I)

Fatty Acid Oxidation Disorders

1 IN 11,000 BIRTHS

Babies with fatty acid oxidation disorders cannot use fats in the body for energy. This can cause severe damage to the heart, liver, and other organs. If untreated, this will result in serious health problems and sometimes death.

- Carnitine uptake deficiency (CUD)
- Long-chain L-3-hydroxy acyl-CoA dehydrogenase (LCHAD) deficiency
- Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
- Trifunctional protein (TFP) deficiency
- Very-long chain acyl-CoA dehydrogenase (VLCAD) deficiency

Organic Acid Disorders

1 IN 29,000 BIRTHS

Babies with organic acid disorders cannot digest certain parts of protein found in food. Harmful substances can build up in the body and have serious impacts on health, growth, and learning, and can result in death.

- 3-hydroxy-3-methylglutaric aciduria (HMG)
- Beta-ketothiolase deficiency (BKT)
- Glutaric acidemia type I (GA-1)
- Isovaleric acidemia (IVA)
- Methylmalonic acidemias (CbI A, B and MUT)
- Multiple carboxylase deficiency (MCD)
- Propionic Acidemia (PROP)

Lysosomal Storage Disorders

1 IN 32,000 BIRTHS

Babies with lysosomal storage disorders are unable to break down the basic components of food, such as sugars and fats. This can lead to a build-up of these materials in the body, which causes problems with the baby's organs or body systems such as their brain, bones, muscles or heart.

- Mucopolysaccharidosis type I (MPS-I)
- Glycogen storage disease type II (Pompe)

Endocrine Disorders

Congenital hypothyroidism (CH)

1 IN 950 BIRTHS

Babies with congenital hypothyroidism do not produce enough thyroid hormone for healthy growth and development. If untreated, it causes severe intellectual disability and growth problems.

Congenital adrenal hyperplasia (CAH)

1 IN 14,000 BIRTHS

Babies with congenital adrenal hyperplasia cannot make enough of the hormones needed for healthy body function. These infants can have life-threatening episodes of dehydration and coma. Female babies may have abnormal genitalia.

Other Disorders

Sickle cell and Hemoglobinopathies (HGB)

1 IN 4,700 BIRTHS

Babies with sickle cell disease or other hemoglobinopathies have abnormal red blood cells. The blood cells are unable to carry oxygen efficiently throughout the body. These disorders can cause frequent infections, severe pain, anemia, and other complications.

Cystic fibrosis (CF)

1 IN 5,500 BIRTHS

Babies with cystic fibrosis develop poor lung function and struggle with malnutrition. This leads to serious health problems and a shortened lifespan.

Biotinidase deficiency (BIO)

1 IN 86,000 BIRTHS

Babies with biotinidase deficiency cannot use an important vitamin called biotin. If untreated, this can cause rashes, hearing loss, seizures and developmental delay.

Galactosemia (GALT)

1 IN 11,000 BIRTHS

Babies with galactosemia cannot digest galactose, a sugar present in milk. When babies drink milk, including breast milk, galactose builds up in the body and can cause blindness, brain damage, or death.

Severe combined immunodeficiency (SCID)

1 IN 88,000 BIRTHS

Babies with severe combined immunodeficiency are born without a working immune system. They cannot fight germs that cause disease and even the most common infections can be life-threatening.

Spinal muscular atrophy (SMA)

1 IN 15,000 BIRTHS

Babies with SMA lose motor nerve cells in their spinal cord. When these cells cannot send messages to the muscles, they no longer work. SMA can be mild or severe. Without treatment, SMA can cause missed motor milestones, difficulty breathing or eating, or even death.

X-linked adrenoleukodystrophy (X-ALD)

1 IN 17,000 BIRTHS

Babies with X-ALD cannot break down certain fats in the body. These build up and cause nerve damage, leading to problems learning, muscle weakness, coma and even death. X-ALD primarily affects males, but females may have symptoms in adulthood and can pass X-ALD onto their children.

Newborn screening saved my life.

"When I was born, this technology detected a congenital metabolic disorder called hypothyroidism. I entered this world with no precursors for this abnormality that would have been disastrous to my growth and development had I not been diagnosed and treated within hours of birth. Without newborn screening I never would have grown to be as tall as I am, would have never gone to college, and would not have competed as an athlete. I encourage every family to take advantage of this simple, timely, life-saving test and thank the wonderful staff at the hospital and the Public Health Laboratories who carry out the newborn screening process. I am forever thankful of the work these people do."

—Aidan, detected with congenital hypothyroidism through Washington State Newborn Screening in 1998

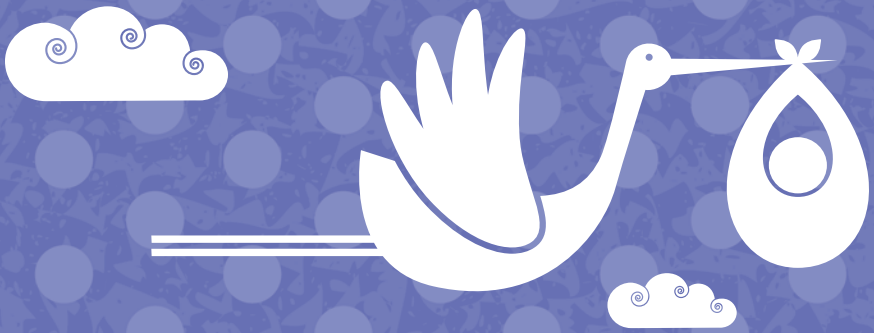
Newborn Screening Checklist

- ☐ Make sure the newborn screening blood sample was collected after birth.
- ☐ Check that your hospital or midwife has the right phone number and address to reach you.
- ☐ Choose a health care provider for your baby's pediatric care and provide their information to your hospital or midwife.
- ☐ Bring your baby in when they are one to two weeks old for a well-child visit to get their second newborn screening done.
- ☐ Ask your baby's health care provider about the newborn screening results.
- ☐ Follow any instructions for more tests, appointments, or follow up care.

Other Screenings Are Available for Your Baby

There are other health issues that you can get your baby screened for, such as critical congenital heart defects and hearing loss. For more information on heart defects screening, contact your medical provider, for hearing screening information, go to **doh.wa.gov/earlyhearingloss**.

Notes & Questions to Ask



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This work is supported by the Association of Public Health Laboratories through the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) under grant number # UG9MC30369 New Disorders Implementation Project for $4,000,000. 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.