What does the newborn blood screening test for?

- Argininosuccinic Acidemia (ASA)
- Biotinidase Deficiency
- Congenital Adrenal Hyperplasia (CAH)
- Congenital Hypothyroidism
- Citrullinemia (Types I and II)
- Cystic Fibrosis (CF)
- Fatty Acid Oxidation Disorders (12 different types)
- Galactosemia
- Hemoglobinopathies (5 different types)
- Homocystinuria
- Hyperphenylalaninemia
- Maple Syrup Urine Disease (MSUD)
- Organic Acidemias (12 different types)
- Phenylketonuria (PKU)
- Severe Combined Immune Deficiency (SCID)
- Pompe Disease

Targeted Variant Testing (TVAR)

You may already have a child with a genetic condition that is not included in standard newborn screening. The Wisconsin Newborn Screening Lab can test your baby for that genetic condition. The test is called Targeted Variant Testing (TVAR). TVAR is a tool that tests for the specific gene change that caused your other children to be affected. The following tests are available for targeted testing:

- Aicardi Goutières Syndrome
- Ataxia-telangiectasia-like disorder
- Cartilage-Hair Hypoplasia
- Cortical dysplasia-focal epilepsy Syndrome (CNTNAP2)
- Galloway-Mowat Syndrome
- GM3 Synthase Deficiency (infantile epilepsy syndrome)
- Hemophilia B – Factor IX deficiency
- Jalili Syndrome
- Mast Syndrome
- Mucolipidosis II (I-cell disease or pseudo-Hurler polydystrophy)
- Oculocutaneous Albinism (OCA)
- Primary Ciliary Dyskinesia (PCD)
- SNIP1
- BRAT1
- Nemaline Myopathy
- Nephrotic Syndrome (congenital nephrosis)
- Senior-Loken Syndrome
- HCM (hypertrophic cardiomyopathy)
- Sitosterolemia
- Troyer Syndrome (Spastic paraplegia)
- Amish Brittle Hair Syndrome

You can prepare by calling the Center for Special Children at 608-625-4039 to get more information.

Carrier screening

You may learn from the newborn screening test that your family has a history of genetic conditions found in newborn screening. In these cases, the newborn screening lab can do carrier testing on parents. The parent carrier test finds out if the parents have a copy of the genes that cause a genetic condition. This tells us if the genetic condition is passed down through the parent's genes. The Wisconsin Newborn Screening Lab tests for these conditions:

- Spinal Muscle Atrophy (SMA)
- Maple Syrup Urine Disease (MSUD)
- Propionic Acidemia (PA)
- Phenylketonuria (PKU)
- RAG1 gene change caused SCID (severe combined immune deficiency)
- Congenital Adrenal Hyperplasia (CAH)

Call the Center for Special Children (608-625-4039) to ask about testing.

Wisconsin State Laboratory of Hygiene
UNIVERSITY OF WISCONSIN–MADISON

WISCONSIN DEPARTMENT of HEALTH SERVICES

The Simple Tests That May Save Your Baby's Life

Wisconsin Newborn Screening Program

P-00446 (04/2025)

Why screen?

Many of the conditions found in newborn screening need treatment in the first days of life.

Early diagnosis and treatment of these conditions can allow your baby to live a long, healthy life. The longer one waits to screen their baby, the more likely it is that their baby will face serious issues. These conditions can cause disability and death without treatment.

Many families have a child with a genetic condition. Even if it is not your brother or sister, it may be a cousin or a grandparent. Genetic conditions are often hidden for generations. You will not know which of your children might be affected by a life-threatening genetic condition without testing them, even if you have many healthy children already.

The tests in the Wisconsin Newborn Screening Program all have treatment to allow your child to live a more normal life.

Some conditions happen more in Amish and Mennonite communities.

It is possible that someone in your family or community has a genetic condition. Do any of these sound familiar?

- Severe Combined Immunodeficiency (SCID)
- Phenylketonuria (PKU)
- Spinal Muscular Atrophy (SMA)
- Maple Syrup Urine Disease (MSUD)

What to expect?

Newborn screening tests for 48 conditions found in blood, hearing loss, and heart defects. Doctors and midwives are required by Wisconsin law to offer these screenings to improve the baby's health.

The blood spot is collected by pricking your baby's heel and dropping blood on five circles of filter paper. It hurts for only a short time, and you can comfort your baby while the sample is collected. To get the most accurate results it should be done between 24–48 hours of life.

The hearing screening happens while your baby is asleep. It is harmless. A small probe the size of a cotton swab is inserted into your baby's ear to see how your baby responds to sounds. It is best done in the first two weeks of life.

The screening for heart defects is done by wrapping a sensor around the right hand and either foot of your baby. It is harmless and measures the oxygen level in your baby's blood to determine if they may have a heart defect. This test is best done between 24–48 hours of life.

How do I get newborn screening for my baby?

Ask your midwife or doctor to complete newborn screening for your baby.

If your midwife or doctor does not offer these screenings, some other options are below.

Contact the Center for Special Children to find someone who does all three newborn screens.

Mail:
Center for Special Children
206 N. Mill St. La Farge, WI 54639
Call: 608-625-4039

Contact the Newborn Screening Program for more information about newborn screening.

Mail:
Wisconsin Newborn Screening Program
Division of Public Health, PO Box 2659
Madison, WI 53703
Call: 608-266-8904

What if something is found?

Sometimes your baby will need additional blood testing.

At times, it is another heel prick to repeat the lab work. Other times, your baby will need a blood draw to get a confirmed diagnosis or rule out a problem. Your child may need treatment or hospitalization if a diagnosis is confirmed. If your family has a history of having children with certain conditions, we may arrange the test done sooner than 24 hours after birth to make sure that your new baby is healthy.

Your baby will be offered a second hearing screening if they do not pass the first hearing test.

Sometimes your baby will have fluid in their ear canal that prevents a complete screening. Your baby needs a diagnostic test if they do not pass two hearing screenings. This diagnostic is safe and painless. It tells you how your baby's brain is responding to sound. It tells you if your baby may have hearing loss. Your health care professional will work with you on ways to communicate and promote good hearing as your baby grows.

If the heart screening shows low levels of oxygen in your baby's blood, they will be sent to the nearest hospital.

The hospital will arrange prompt follow up for your baby to have an evaluation by a special heart doctor. Sometimes babies need medical attention right away if they have certain heart defects. Most babies live long, happy lives with medical treatment.