Newborn screening (NBS) is a group of tests that check all babies for certain rare conditions where there may not be immediate symptoms, but if the condition goes untreated can lead to serious health and developmental issues. Some of the medical conditions on NBS panels can be life-limiting if not treated. Most of the conditions covered by NBS are genetic, and the majority have treatment options that are available. With early diagnosis and treatment, most babies with these disorders can lead healthy lives.

While the field of genetics has rapidly expanded in recent years, NBS has been done since the 1960s. At this time, it became possible to screen newborns for a genetic condition called PKU (or phenylketonuria). PKU prevents that body from breaking down a substance called phenylalanine, which can lead to it building up in various parts of the body. If untreated, individuals with PKU can develop intellectual disability, seizures, behavioral issues, and mental health disorders. If identified and treated shortly after birth, most children with PKU will experience no health issues or developmental concerns.

As our understanding of various genetic disorders and their treatments has expanded, NBS has gone from just testing for PKU to being able to test for over 30 different medical and genetic conditions.

Health Conditions Covered by NBS

The disorders covered by NBS are often grouped into several categories based on their signs, symptoms, or underlying causes. Many, but not all, affect a person's metabolism or the way the body changes food into the energy it needs to breathe, digest food, and grow. These are lifelong conditions, so they will not go away as someone gets older.

There are many different types of genetic conditions that are included in newborn screening tests. Some more common types of disorders include:

- Metabolic disorders
    - Amino acid disorders
    - Organic acid disorders
    - Fatty acid oxidation disorders
    - Other metabolic disorders
- Cystic fibrosis
- Spinal Muscular Atrophy
- Severe combined immune deficiency (SCID)
- Endocrine (hormone) disorders
- Hemoglobinopathies (inherited anemias)

- Congenital (from birth) hearing loss
- Congenital (from birth) heart defects

NBS by State

What conditions are included on a NBS panel depends on the state where the baby is born. The NBS program (including coming up with the conditions that are tested for and following up with results) is overseen by the Department of Health in each state. Although individual states determine what disorders their NBS testing will include, the Secretary of the Department of Health and Human Services (HHS) recommends that all states include a list of disorders called the Recommended Universal Screening Panel, or RUSP.

The Department of HHS uses three main criteria to determine what conditions should go on the RUSP list:

- The potential net benefit of screening for the disorder,
- The availability of effective treatments, and
- The ability of states to screen for the disorder.

Currently, there are 35 health conditions that are on the RUSP. Each state's Department of Health can use this list as a guide to determine what conditions they will include on their NBS panels, and can include more or fewer conditions than those that are on the RUSP.

To see a listing of the specific disorders your baby is screened for, click on the state where you delivered or plan to deliver:

Alabama	Alaska	Arizona	Arkansas	California
Colorado	Connecticut	Delaware	District of Columbia	Florida
Georgia	Hawaii	Idaho	Illinois	Indiana
Iowa	Kansas	Kentucky	Louisiana	Maine
Maryland	Massachusetts	Michigan	Minnesota	Mississippi
Missouri	Montana	Nebraska	Nevada	New Hampshire
New Jersey	New Mexico	New York	North Carolina	North Dakota
Ohio	Oklahoma	Oregon	Pennsylvania	Puerto Rico
Rhode Island	South Carolina	South Dakota	Tennessee	Texas
Utah	Vermont	Virginia	Washington	West Virginia
Wisconsin	Wyoming			

Frequently Asked Questions

Here are some frequently asked questions about NBS:

How and When is Newborn Screening Done?

Newborn screening is typically done after birth within the first few days of life by poking a baby's heel to place a few drops of blood onto a special filter-paper blood spot card. The blood spot card is then sent to the state-designated laboratory for testing. While some states require a single sample, many require a second sample be collected within the first two weeks after delivery. This is especially important if a baby's first sample was drawn very close to their time of birth, or if they were born at very low birth weight.

In addition to the NBS blood test, the tests for congenital hearing loss and congenital heart defects are also done in the first few days of life. Both are painless and can even be done while a baby is sleeping. The newborn hearing test uses earphones and sensors to determine whether the baby's inner ear or brain responds to sound. The test for congenital heart disease, called pulse oximetry, uses a sensor on the skin to measure how much oxygen is a baby's blood.

Does your baby have to have newborn screening?

Most states mandate by law that NBS be performed on all infants who are born within their borders. Exceptions include Minnesota and Vermont, which allow parents to opt out of newborn screening for any reason. Idaho allows parents to opt out for religious reasons only. Maryland is the only state that requires a parent's consent to perform newborn screening.

How will you get your baby's NBS results?

Many state NBS programs will send your baby's pediatrician the results of their newborn screen. Others send them to the hospital or birth center where you delivered your baby. In most states, if your baby's result signal they are at low-risk for the things they were tested for, you will not hear directly about the results ("No news is good news").

If your baby's results indicate he or she could have one of the conditions included in your state's newborn screening panel, you will get a call from the NBS program or your baby's healthcare provider. You may also get a call if your baby's initial sample did not contain enough blood, or if test results are inconclusive. In either case, you will be given specific directions about what to do next, and it is important to do any follow-up immediately. The sooner any repeat or further testing is completed, the sooner treatment can begin if your baby is ultimately found to have one of the conditions. This will lead to the best possible outcome for your baby.

If you do not receive information about your baby's newborn screening results and would like to confirm that the results were negative, contact your baby's primary healthcare provider.

What does a negative ('in-range') NBS result mean?

A "negative" or "in-range" result means that your baby's blood test DID NOT show any signs of the conditions included on the NBS panel.

In terms of the newborn hearing and congenital heart defect screening, often an in-range or negative result will be referred to as "passing" the newborn hearing and cardiac screen. This means that when your baby was tested, his or her hearing and oxygen levels were normal.

What does a positive ('out-of-range') NBS result mean?

A "positive" or "out-of-range" result means that your baby's results signal they have a higher risk to have one or more of the conditions screen for, and further tested is needed to get more information. It does not mean with certainty that your baby will have the condition(s). While many babies who receive positive NBS results do not have any medical conditions, to ensure the best health for your baby it is important for you to follow any recommendations for follow-up immediately. The closer look often includes additional testing, including a repeat newborn screening sample or other specialized tests of the baby's blood, urine, or even sometimes their sweat. It may also involve an evaluation at a regional genetic or metabolic center with physicians that are experts in identifying and managing NBS disorders.

In terms of the newborn hearing and congenital heart defect screening, often an out of range or positive result will be referred to as “not passing” the newborn hearing or cardiac screen. This means that when your baby was tested, his or her hearing and oxygen levels were less than expected on the days tested. Further testing is usually recommended, and may include a more extensive hearing evaluation and imaging tests (like an echocadiogram).

If your baby really has one of these conditions, what do you do next?

If you find out our baby has one of the conditions that were tested for on NBS, know that you are not alone. Each year, over 5,000 babies are found to have one of these conditions. There are specialists and many healthcare providers available for any questions or concerns that may come up.

If your baby is found to have one of these conditions after the additional testing, the next step will be to meet with experts who specialize in that condition. This may include a medical geneticist, nutritionist, cardiologist, endocrinologist, hematologist, audiologist, or other specialist. They can help to explain the condition and treatment plan in more detail. They will work with you and your pediatrician to help your baby stay healthy.

Children who are found to have metabolic conditions can require more appointments within their first 6 months of life. This is helpful to make sure the baby is getting the right treatment, including recommendations for diet or medications. Blood and urine testing may also be necessary to measure different markers in the baby’s body. These appointments are usually less frequent as a child gets older. There are typically only a few centers in your state who specialize in treating children with these conditions. Because of this, families who have a child with one of these medical conditions may have extra time and expense requirements for travel to doctor’s appointments. Often times, particularly if a patient is coming from a great distance away, they can see multiple specialists in the same day to try to cut down on the amount of travel. There is also often a lot of information that is covered at these appointments, so some people find it helpful to bring a friend or family member who can be an extra set of ears, or help with child care so the parent can focus on the information.

It may also helpful to bring things like snacks and toys, particularly if the child is seeing multiple doctors in the same day. It is important to write down any questions or specific concerns that you have in advance so you don’t forget when you get to the appointment.

What does this mean for other family members?

Once a genetic condition has been identified in your newborn, testing for other members of the family may be recommended. This could include the parents, siblings, or more extended family members depending on the condition. Your genetics team will review the specifics with you. This is often a blood draw, but may require other samples.

Sometimes, the child's positive test identifies an underlying condition in the mother. This will not harm the baby as they get older but it may require additional supplements or diet modifications for the baby and/or the mother.