

More than 4 million babies are born each year in the United States. Most of these babies have newborn screening done to test for certain medical conditions. Without screening, affected babies may suffer mental or physical disabilities, or even death. Most affected babies can lead normal, healthy lives when diagnosed and treated early in life.

# FAQs

## FREQUENTLY ASKED QUESTIONS ABOUT NEWBORN SCREENING

### ■ What is Newborn Screening (NBS)?

Newborn Screening identifies babies who may have serious medical conditions.

### ■ Why does my baby need Newborn Screening?

For each of the conditions in the screening program, the newborn benefits most when the condition is detected early and treatment is started in the first few days of life. Most babies should get their first newborn screen done before going home from the hospital or birth center, ideally between 24 and 48 hours of age.

### ■ How will my baby be screened?

Your baby will have a blood screening test, a hearing screen, and possibly a screen for heart problems. The blood screening is done by pricking your baby's heel and collecting a few drops of blood onto a special filter paper. The filter paper is sent to a newborn screening laboratory where all of your state's screening tests are done. You will receive the blood screening results shortly after you leave the hospital. Trained staff members will

usually do the other screening tests in the hospital. The results will be shared with you at the time of the screen.

### ■ Why is NBS sometimes referred to as the “PKU test?”

Early NBS testing included only a single condition called PKU. Today's screening tests look for many additional conditions.

### ■ What if we have no family history of these conditions?

Most babies with these conditions are born to families with no history of problems or who already have healthy children. **Damian's story** (*see below*) tells about a family learning how true this is.

### ■ Should I still get screening if my baby looks healthy?

Most babies with the disorders that can be found by newborn screening seem normal and healthy at birth. The newborn screening helps detect the condition in a baby before problems start. Most babies who are diagnosed and treated early do well.

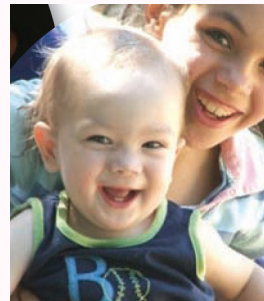
### ■ Why do some babies need a second newborn screen?

If a first screen is abnormal or unclear, a repeat screen is usually needed. A request for a repeat NBS does not always mean your baby has a condition. If you are asked to repeat the screening for your baby or to go to your doctor for other tests, it is important that you respond as quickly as possible. Some states routinely require this second screening test.

### ■ How will I know the results of my baby's newborn screen?

Generally, parents are notified within a few days if the screening detects a possible problem. You will be given directions about what to do next. Follow these directions very carefully. Additional tests will be necessary to confirm whether your baby has a condition. If the results are normal, you will be informed usually by two weeks after the blood test is collected. The results will be mailed to the birth hospital (*continued on next page*)

## Surprised and Supported



**DAMIAN'S STORY** Damian's parents knew the nurses had taken a blood sample for his newborn screening, but they thought nothing of it. Screening had shown no issues for Damian's five older brothers and sisters. When Damian was five days old, his pediatrician contacted his parents to tell them that he had screened positive for 3-MCC deficiency, an inherited condition that makes the body unable to use certain proteins.

Further tests soon confirmed the positive screening re-

sults. Infants with 3-MCC deficiency appear normal at birth but may develop feeding difficulty, muscle weakness, delayed development, and seizures, usually between ages 3 and 5 and accompanying an illness. Many of these complications can be prevented with early detection and lifelong management, including a low-protein diet and supplements.

It was hard for Damian's parents to accept that such a healthy-appearing baby could have a medical condition, but they see Damian's early diagnosis as a blessing. With treatment and special precautions, Damian is leading a healthy life and today is an adorable, strong little boy.

(continued from preceding page) or to your baby's doctor. You can ask the doctor for the results of the screening test by telephone or during the first well-baby check if you have not received them by two weeks.

Be sure to put your correct telephone number and address on the newborn screening form in the hospital. Update the doctor or health care provider if you move or change phone numbers soon after your baby is born. Be sure that your baby's full name is listed correctly on all screening paperwork. This information is needed so you can be reached if your baby's NBS test results show a possible problem.

### ■ Will my baby have the same NBS no matter where I live?

No. All states require newborn screening of babies born in their state. There is list of conditions recommended for NBS by a federal NBS advisory panel and by the Secretary of Health and Human Services. However, each state may screen for a slightly different list of conditions.

### ■ Can I get my baby screened for all disorders detectable by NBS?

First, ask your baby's doctor about screening done in your state, or check with your state's Department of Health or [www.savebabies.org](http://www.savebabies.org) to learn about the conditions in your state's screening program. If your state requires screening for only some of the conditions recommended by the Secretary of Health and Human Services, you may want to ask about additional screening options. Contact Save Babies Through Screening Foundation at 1-888-454-3383 or [email@savebabies.org](mailto:email@savebabies.org) for further assistance.

### ■ Is it important to give permission to have my baby's blood spot card stored after screening is complete?

There are many reasons for storage of blood spot cards. Having a newborn blood sample may benefit your family directly if your baby is diagnosed with a medical condition later in childhood. The sample may be used to help the lab confirm the accuracy of their newborn screening tests and to help develop new newborn screening tests.

These and other types of research may benefit your future children and other families, while helping us all better understand health and illness. **LeA's story** (at right) shows how important research can be.

Our parent-targeted newborn screening education DVD, **One Foot at a Time**, is available upon request at no charge. Please contact us for additional information.

## “If only I had known.”

**LeA's STORY** Imagine learning too late that the disease that took your child's life could have been identified by newborn screening. LeA's family learned this terrible lesson, and hopes that you will learn from their story.



LeA was born in 1996 and seemed perfectly healthy. She had the routine newborn screening that was available in her state at the time. She ate and slept well and began to notice the world around her. Her smiles and happy cooing delighted her family and friends. But within a few months LeA's physical condition changed dramatically. Her body became rigid and she cried constantly. She would not eat, frequently choking on what little she tried to eat. At 10 months, LeA was diagnosed with Krabbe disease, which is part of a group of more than 40 rare diseases known as lysosomal storage diseases (LSDs).

LSDs are caused by very low levels of enzymes that help remove stored fats from cells in the body. By the time her disease was diagnosed, LeA was not eligible for treatment using bone marrow transplant. She slowly got worse. She needed a feeding tube, special formulas, multiple medications each day, oxygen, and in-home nursing. LeA died at the age of two. Early diagnosis and intervention could have made a difference in LeA's life.

Lysosomal storage disorders were not part of the newborn screen when LeA was born, but research using stored blood spots from newborn screening is happening right now to make newborn screening for these disorders possible in the future. LeA's family strongly supports this research to make safe, effective newborn screening and follow-up care for these conditions available to all babies. They urge you to have your baby screened.

## About the Save Babies Through Screening Foundation

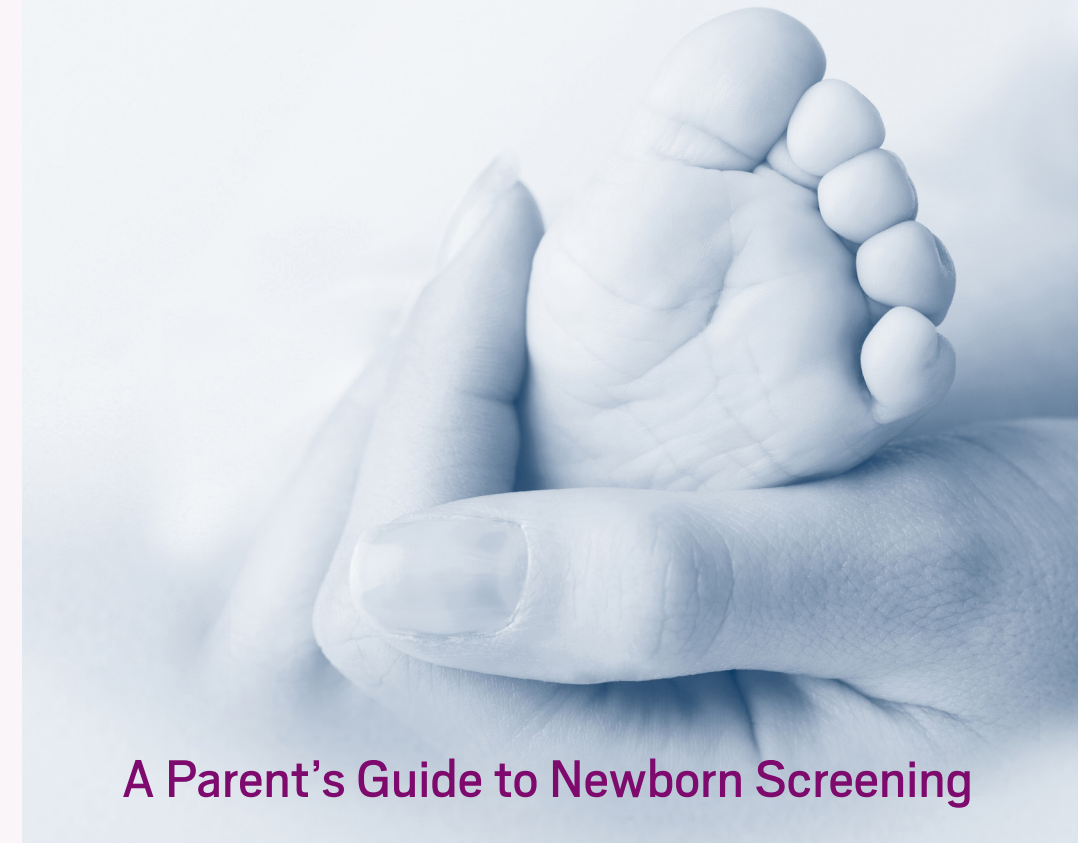


Founded in 1998, Save Babies Through Screening Foundation is the only national non-profit organization devoted exclusively to newborn screening advocacy. The Save Babies Through Screening Foundation's mission is to improve the lives of children by working to prevent death and disabilities resulting from disorders detectable through newborn screening tests. The Foundation is a leader in the national grassroots advocacy movement and media awareness. The Foundation actively participates at local, state, and federal levels to improve newborn screening. The Foundation's goal is to see that every child born is screened successfully, effectively, and comprehensively.



Save Babies Through Screening Foundation

# A Simple Test Could Save Your Baby's Life



A Parent's Guide to Newborn Screening