**Answers to Frequently Asked Questions**

**Currently more than 4 million babies are born each year in the United States. Even though every baby born in the United States receives a newborn screen, how many diseases screened for is determined by the state in which that baby is born. Thus, where a baby is born may determine whether or not an affected baby lives a healthy, normal life. Without proper screening, affected babies will likely suffer mental retardation, physical disability, or even death. Most affected babies can lead normal, healthy lives when diagnosed early and started on treatment shortly after birth.**

**What Is Newborn Screening (NBS)?**
Newborn Screening is a medical procedure which screens babies for serious disorders. The procedure should be performed when your baby is 24-48 hours old. NBS is sometimes incorrectly referred to by medical professionals as the “PKU test.”

**Why does my baby need Newborn Screening?**
The screening is done to find out if your baby has a serious disorder or condition for which early detection and treatment can prevent death, mental retardation, or physical disability.

**How will my baby be screened?**
The screening is performed by pricking your baby’s heel and putting a few drops of blood on special filter paper. The paper is allowed to dry and then sent to a newborn screening laboratory where many different screening tests are performed.

**But we have no family history of these disorders...**
Parents who have no family history of problems and/or who have already had healthy children may still have babies with these disorders or conditions. In fact, most babies with these disorders come from families with no previous history of the condition.

**But my baby looks healthy...**
Most babies with these disorders look and act normal and seem perfectly healthy at birth. The newborn screening helps your doctor catch a problem with your baby before it makes him or her sick. Most babies that are diagnosed and treated early do well. The earlier the detection, the greater the chance that the baby will grow up healthy with normal development.

**Why do some babies need a second Newborn Screening?**
If the result of your baby’s first screening is abnormal or unusual, a repeat screening test is usually required. A request for a rescreening does not necessarily mean your baby has a disorder, but it is possible. If you are asked for a rescreening, it is important that you take your baby for the rescreening as quickly as possible. In addition, some states routinely require a second screening to ensure that you have the most accurate information for the health of your baby.

**How will I know the results of my baby’s newborn screening?**
Generally, parents are notified of the screening results only if there is a problem. However, it is a good idea to call your doctor and request the results of the screening test. This is important to ensure that your baby’s screening results have not been lost or misplaced. If your baby’s screening shows an abnormal result, you will be notified immediately and given directions about what to do next. Follow the directions of your doctor very carefully. If your baby’s screening test is abnormal, additional screenings and diagnostic tests are usually necessary to verify if your baby has a disorder. It is important that you advise your doctor if you move or change phone numbers soon after your baby is born in case your baby has a positive test result for a disorder. If the results of your baby’s screening test are normal, they will be mailed to your baby’s physician approximately two weeks after the screening test was performed.

**Will my baby get the same Newborn Screening no matter where I live?**
No. Although all states require newborn screenings be performed on all babies born in their state, each state may screen for different disorders. Newborn screening is capable of testing for more than 60 disorders, yet many states still screen for less than 10, some for less than 40 or even as few as 30.

**How can I find out what my state’s routine newborn screening covers?**
Visit [www.savebabies.org](http://www.savebabies.org) or call us toll free at 1-888-454-3383.

**What is Supplemental Newborn Screening?**
Supplemental Newborn Screening is an additional screening for disorders which are not routinely screened for in your state. Supplemental Newborn Screening initiated by your baby’s hospital shortly after birth does exactly what it says. It “supplements” the screening already done in compliance with state law. In states screening for more than 50 disorders, supplemental screening is not always necessary. You can discuss this with your doctor. However, in states which screen for less than 50 disorders, supplemental newborn screening is crucial in order to prevent damage from undiagnosed disorders that are not required for screening in your state. Please visit [www.savebabies.org](http://www.savebabies.org)/parent_packets.html to determine whether Supplemental Newborn Screening is recommended for babies born in your state.

**How can I get my baby screened for all available disorders?**
Contact the laboratory below to obtain a parent packet, which contains a supplemental screening kit and information for you and your physician. The screening kit should be obtained prior to the baby’s delivery and taken to the hospital with you. Let your doctor know you would like to have an extra blood sample drawn at the time of your baby’s routine newborn screening. Then, when your baby’s routine heelstick is performed, a few extra drops of blood will be applied to the supplemental screening test card. As with your baby’s routine screening, if the supplemental screening is abnormal, your baby’s physician will be contacted immediately. If the supplemental screening is normal, results will be sent through the mail to you or your physician.

PerkinElmer Genetics – 866-463-6436
[http://www.perkinelmergenetics.com](http://www.perkinelmergenetics.com)

**What if my baby’s doctor doesn’t know about Supplemental Newborn Screening?**
Because supplemental screening has not been available everywhere until recently, your doctor may not know about it. Be prepared that you may have to supply your doctor with information on supplemental screening.

**Newborn Screening and SIDS**
It is estimated that at least 5% of all cases of Sudden Infant Death Syndrome (SIDS) are actually cases of undiagnosed metabolic disorders that are detectable through newborn screening. Most routine newborn screenings do not screen babies for the disorders that have been linked to SIDS. Because these babies appear normal and healthy, it is not suspected that they have an underlying disorder that could cause sudden unexplained death. Even after death, many times these babies are undiagnosed or said to be cases of SIDS. Proper universal comprehensive screening would allow diagnosis and treatment of these disorders, thus allowing most of these babies to avoid early death and disability, and to live healthy lives.
LeA's Story
LeA was born in 1996. As a newborn, she seemed perfectly healthy. LeA ate and slept like all other newborn babies. She began to notice the world around her. Her smiles and happy cooing delighted her parents, family, and friends. Within a few months LeA's physical condition changed dramatically. Her body became rigid. She cried constantly, and she would not eat, frequently choking. 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 deficient enzymes that normally eliminate unwanted substances in the cells of the body. Because her diagnosis took months, LeA was past the period when she could receive treatment. Her physical condition worsened to where she needed feeding tubes, specialty formulas, more than 30 doses of medicine a day, oxygen, suctioning, and in-house nursing. After a quarter of a million dollars worth of medical bills, LeA died at the age of two. Early diagnosis and intervention could have made a difference in LeA's life.

Damian's Story
When Damian was born, he was the youngest of six healthy, happy children in his family. While his parents knew the nurses had taken a heelstick for his newborn screening, they thought nothing of it – previous screenings had shown no issues for Damian’s older brothers and sisters. However, when Damian was five days old, his pediatrician contacted his parents to urgently share the news that Damian had screened positive for 3-MCC deficiency, an inherited disorder in which the body is unable to process certain proteins properly. Infants with 3-MCC deficiency appear normal at birth but usually develop signs and symptoms in infancy or early childhood. If untreated, 3-MCC deficiency can lead to delayed development, seizures, and coma. Many of these complications can be prevented with early detection and lifelong management with a low-protein diet and appropriate supplements. After more screening and diagnostic tests were performed, Damian's diagnosis of 3-MCC was confirmed. Although it was hard for his parents to accept that such a seemingly healthy baby could have a life-threatening disease, they see Damian's early diagnosis as a blessing. Through treatment including diet, supplements, and special precautions, Damian is able to lead a healthy life, and is an adorable, strong little boy today!