Real Lives, Real Stories

Almost every family whose child dies or is disabled from a disorder detectable through newborn screening, and who then learns that their child's death or disability could have been prevented, says, "If only I had known." These families urge you to have your baby screened for all detectable disorders.

Brett's Story

At three months old Brett suffered a heart attack and spent two months on life support while doctors tried to diagnose what was wrong with him. Finally, Brett's doctors realized that he had a condition commonly called VLCADD. VLCADD is a genetic disorder that prevents the body from properly breaking down stored fats. With his condition undiagnosed, the excess fat in Brett's body swelled his organs and almost caused Brett to lose his life. Fortunately, Brett recovered and, after much rehabilitation, is doing well today. However, supplemental screening could have detected his disorder and allowed his doctors to initiate treatment that likely would have prevented the crisis that sent him to intensive care. An American Academy of Pediatrics report concluded that $400,000 in medical bills likely could have been avoided had Brett's condition been picked up early through newborn screening. Fewer than 10 states currently require screening for VLCADD in routine newborn screening tests.

Joey's Story

When Joey was eight months old, his parents took him to the doctor because he had a cold. When the doctor saw him, he realized that something more serious was wrong. Immediately, Joey was admitted to the hospital where he began to have seizures. What started out like a common cold changed Joey's life forever. Joey was diagnosed with Glutaric Acidemia Type I (GA-I) within a week of being hospitalized, but by then it was too late. Joey, who had been like any other eight month old just a few days before, had suffered massive brain damage. Today Joey is five years old. He is fed through a feeding tube, is unable to talk or walk, and is unable to control any of his movements. When GA-I is treated early, damage such as that which Joey has suffered can usually be prevented. Joey's parents are devastated by the fact that Joey likely would have lived a healthy life had he received supplemental newborn screening at birth.

Routine Screening Tests Fall Short

Fewer than 10 states currently provide comprehensive newborn screening for the more than 50 disorders presently detectable. Early diagnosis allows babies with most of these disorders to live normal, healthy lives. However, without early diagnosis and treatment, babies are at risk of mental retardation, physical disability, and even death. Treatment usually consists of dietary (eating) restrictions, special formulas, medication, and/or dietary supplements such as vitamins. It is estimated that 1 in every 1,500 babies will have a disorder detectable through comprehensive newborn screening.

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 screening tests do not screen children for the disorders that have been linked to SIDS. Because these children 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 children are undiagnosed or said to be cases of SIDS. Proper comprehensive screening would allow diagnosis and treatment of these disorders, thus allowing most of these children to avoid early death and to live healthy lives.

Delays After the Specimen is Taken

The lives of some babies are unnecessarily put at risk because their newborn screening test is mishandled. Some hospitals and doctors' offices "batch" newborn screening tests. "Batching" means they don't send the tests to the newborn screening (NBS) laboratory until they accumulate a certain number of tests, or they send them only on a specific day of the week. Many also use the regular mail to send your baby's critical test to the NBS laboratory. This causes delays in the NBS laboratory receiving the tests and may risk your baby's life. The newborn screening specimen needs approximately 4-6 hours to dry, and then a courier or overnight delivery service should immediately take it to the NBS laboratory. Request that your baby's test be sent to the NBS laboratory immediately . . . the same day as taken.

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A Simple Test Could Save Your Baby's Life

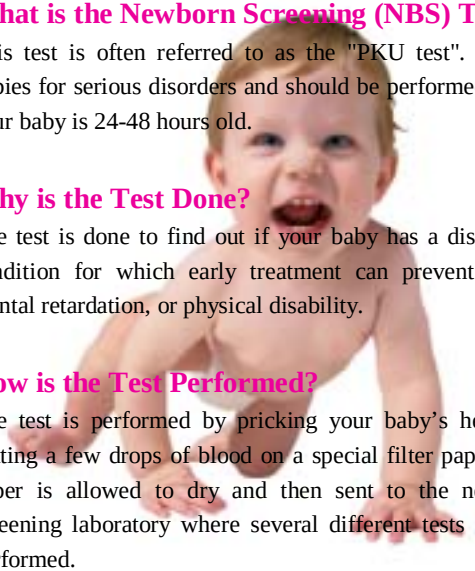
A Parent's Guide to Newborn Screening

Save Babies Through Screening Foundation
1-888-454-3383
www.savebabies.org

Answers to Frequently Asked Questions

What is the Newborn Screening (NBS) Test?

This test is often referred to as the "PKU test". It tests babies for serious disorders and should be performed when your baby is 24-48 hours old.



Why is the Test Done?

The test is done to find out if your baby has a disease or condition for which early treatment can prevent death, mental retardation, or physical disability.

How is the Test Performed?

The test is performed by pricking your baby's heel and putting a few drops of blood on a special filter paper. The paper is allowed to dry and then sent to the newborn screening laboratory where several different tests will be 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 can still have children with these disorders. In fact, most children with these disorders come from families with no previous history of the condition.

But My Baby Looks Healthy . . .

Most babies with disorders look and act normal and seem perfectly healthy. The newborn screening test 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 higher the chance of having a good prognosis.

What is a Retest?

If the result of your child's test is abnormal, a repeat test or a "retest" is usually required. A request for a retest does not necessarily mean your child has a disorder, but it is possible. If you are asked for a retest, it is important that you take your baby for the retest as soon as possible.

How Will I Know the Results of My Baby's Test?

Generally, parents are notified of the test results only if there is a problem. However, it is a good idea to call your doctor and request the results of the test. This is important to ensure that your child's test results have not been lost or misplaced.

If your child's test 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 child's test is abnormal, additional tests are usually necessary to verify if your child has the disorder. It is important that you advise your doctor if you move or change phone numbers soon after your baby is born in case there is a problem with your baby's test.

If the results of your baby's test are normal, they will be mailed to your physician approximately two weeks after the test was performed.

Will My Baby Get the Same Test No Matter Where I Live?

No. Although all states require newborn screening tests be performed on babies born in their state, each state screens for different disorders. The newborn screening test is capable of screening for more than 50 disorders, yet most states screen for less than 8. For example, if your child was born in the state of Texas in 2003, your baby was screened for 5 disorders. Yet if your baby had been born in Massachusetts, he or she would have been screened for 30 disorders.

Find out what your state's routine newborn screening test detects
on the web at www.savebabies.org or by calling toll free 1-888-454-3383.

Should I Have My Baby Screened Supplementally?

Yes. In order to prevent damage from undiagnosed disorders that are not screened for through your child's routine newborn screening test, all babies should be screened through supplemental newborn screening in addition to receiving a routine newborn screening test.

How Can I Get My Baby Screened for All Disorders?

Contact one of the laboratories below to obtain a parent packet, which contains a screening kit and information for you and your physician. The screening kit should be obtained prior to delivery and taken to the hospital with you. ****provide partial screening only***

Pediatrix Screening - 55 disorders for $89
1-866-993-2300 ext. 7

Baylor Medical Center* - 35 disorders for $25
1-800-422-9567

Mayo Medical Laboratories* - 35 disorders for $57
1-800-533-1710

University of Colorado* - 20+ disorders for $25
1-303-724-3826

Lab info www.savebabies.org/NBS/snbs.htm

Note: Prices subject to change
CPT Code for Insurance ~ 83788

Simply give the kit to your doctor, and ask to have an extra blood sample drawn at the time of your baby's routine newborn screening. Then, when your baby's routine heel-stick is performed, a few extra drops of blood will be applied to the supplemental screening test. As with your baby's routine screening, if the screening is abnormal, your baby's physician will be contacted immediately. And if the screening is normal, results will be sent through the mail.

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.