

Newborn Tests That Save Lives

Every state screens newborns for life-threatening disorders, but some do more than others. Find out if your state makes the grade.

By Kristen Finello

Tests to Prepare For

On October 26, 1997, Jana Monaco, of Woodbridge, Virginia, gave birth to her third child -- a healthy 8-pound-12-ounce son. When Stephen was 3 1/2 years old, he came down with what seemed to be a normal stomach bug. It wasn't until Jana found Stephen with his teeth clenched tightly and his body as floppy as a rag doll's that she knew something far more serious was ailing him. Within two days, Stephen was in a coma, on a ventilator, and not expected to live. The cause was isovaleric acidemia, a rare metabolic condition, characterized by an inability to process certain proteins, which was present when Stephen was born. If detected early, treatment, including supplements and a special diet, can help keep kids healthy.



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At the time, the state of Virginia did not routinely screen newborns for that disorder, so Stephen's life-threatening condition went undetected. This information shocked his parents. "I said, 'You mean to tell me that if Stephen had gotten on the right diet from the start he could have been okay?'" explains Monaco. "Knowing that what happened to Stephen could have been easily prevented really compounded the tragedy."

Early Detection Makes All the Difference

The contrast between kids who receive early diagnosis and treatment for this condition and those who don't is evident in the Monaco family: Stephen survived with severe brain damage. Now 8 years old, he is in a wheelchair, unable to speak, and eats through a tube. His younger sister, Caroline, was tested prenatally and diagnosed with the same disorder, but she's a normal 3-year-old today because of early intervention.

The Monacos' story is a perfect example of the benefits of screening newborns and the consequences of not testing. "Newborn screening of babies is one of the most important things we can do," says R. Rodney Howell, MD, professor of pediatrics at the Miller School of Medicine at the University of Miami. "By identifying babies that look healthy but have one of these disorders, and by starting treatment, we can dramatically improve and even save their lives." Here's a look at the current situation in newborn screening and what you can do to ensure that your precious baby receives the tests she needs.

Screening Basics

During the first 48 hours of life, infants born in the United States are routinely screened for a number of health conditions. A hearing test is usually administered, and blood obtained through a "heel stick" is sent to a laboratory and tested for a battery of genetic, metabolic, hormonal, and functional disorders. Although these are rare conditions that affect only about 1 in 600 babies, they do occur. Usually the babies appear healthy at birth, just as Stephen Monaco did. At that point, it's only through blood testing that doctors can detect a disorder.

When Disorders Go Undetected

Not finding these conditions early on can have serious consequences. If hearing loss isn't detected at birth, it may not be apparent until a child is two or three years old. By that point, a child's speech development and language skills might be adversely affected. The stakes can be even higher for other disorders; some can result in physical problems, mental retardation, and even death if left untreated. With early detection and treatment, babies can usually lead a normal life.

For example, if newborns with hypothyroidism are identified, they can take a thyroid hormone supplement to replace what their body isn't making. This can mean the difference between serious developmental delays and normal mental growth.

"It can be devastating to learn that your new baby has an illness, but it's better to find out right away because early intervention can dramatically change your baby's life," says Siobhan Dolan, MD, associate medical director for the March of Dimes.

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State-by-State Laws

Which disorders a baby is screened for depends on where he is born. Each state has its own laws, but they all mandate screening for phenylketonuria (PKU, an inability to process a part of a protein called phenylalanine), classical galactosemia (an inability to break down the sugar in milk), congenital hypothyroidism, and three red-blood-cell disorders (sickle cell disease, sickle-C disease, and S-Beta thalassemia).

Beyond that, there's no uniformity in the number of screenings given. Some states mandate tests for fewer than 10 disorders, while other states such as Colorado, Connecticut, Iowa, Mississippi, New York, South Dakota, Tennessee, and Virginia require screening for 29 or more. In some cases, states routinely screen for conditions even when they aren't required by law to do so. For example, 49 states test all newborns for hearing loss even though it is only mandated in 28 states. In other cases, screenings may be offered only to specific populations who are considered to be at higher risk for a certain disorder.

In addition, states have different policies on when and whether to rescreen newborns. Some, such as Colorado, Delaware, and Utah, even require that all babies be rescreened after they're released from the hospital. Most states require it if the original test was done prior to 24 hours after birth. Although the screening tests are very accurate, it's possible for some conditions, such as galactosemia, to go undetected if the screening is done too soon after birth. "If a newborn eats only minimal

amounts during the first 48 hours, the tests might not detect the galactosemia because the chemical reactions need time to start in order to determine whether they're working properly," explains Dr. Dolan.

States That Screen the Most

As of early November 2005, these states mandate that all newborns be tested for 20 or more conditions on the American College of Medical Genetics's list of disorders that should be screened (*For the full list of 29 disorders, see the next page*):

States A-M:

Alaska
California
Colorado
Connecticut
Delaware
Hawaii
Idaho
Illinois
Indiana
Iowa
Kentucky
Maryland
Minnesota
Mississippi
Missouri

States N-Z:

Nevada
New Jersey
New York
North Carolina
North Dakota
Ohio
Oregon
South Carolina
South Dakota
Tennessee
Vermont
Virginia
Wisconsin

Screening Recommendations

The recent report issued by the American College of Medical Genetics recommends that all states screen for 29 disorders. They are:

Hearing
Congenital hypothyroidism
Congenital adrenal hyperplasia
Sickle cell disease
S-Beta thalassemia

Sickle-C disease

Biotinidase

Transferase deficient galactosemia (Classical)

Cystic fibrosis

5 fatty acid disorders (Untreated, they can cause brain and organ damage, coma, and death.)

9 organic acid disorders (Untreated, these conditions can cause coma and death.)

6 amino acid disorders (Untreated, these conditions can cause retardation and death.)

A major technological advance in newborn screening is tandem mass spectrometry, which gives states the ability to screen babies simultaneously for 20 metabolic disorders by breaking down blood samples into compounds with patterns that computers can analyze. "Tandem mass spectrometry is very fast and extremely accurate," says Dr. Howell. Just because this technology exists, however, doesn't mean that every baby is benefiting from it.

Arranging for More Screening

If you live in a state that doesn't offer all 29 screening tests recommended by the American College of Medical Genetics, you can arrange for additional screening tests for your newborn. Supplemental screening -- for anywhere from 20 to more than 50 disorders, depending on the lab -- is available through the organizations listed below for fees ranging from about \$25 to \$90. If you decide to have supplemental screening, discuss it with your pediatrician. If the hospital where you'll deliver doesn't have procedures for supplemental screening in place, you can order a testing kit yourself. Take it to the hospital when you deliver, and technicians can complete the tests at the same time they draw blood for the state-mandated screenings. The hospital will send the results to the pediatrician you request. For more information, please contact:

- **Mayo Medical Laboratories** 800-533-1710; mayoreferenceservices.org/mml/mml-sns-intro.asp
- **Pediatrix** 866-463-6436; pediatrixscreening.com
- **University of Colorado Expanded Newborn Screening Program** 303-724-3826; uchsc.edu/newbornscreening
- **Baylor Medical Center Institute of Metabolic Disease** 800-4BAYLOR; baylorhealth.com/newbornscreening

Crucial Tests, Changing Laws

Pushing for More Tests

As things stand now, where your baby is born will determine which screening tests she receives. This may be changing, though, thanks to an increasing awareness about the topic. Parents like Jana Monaco, whose lives have been touched by a particular disorder, are an important force in the effort to change state laws so that they will mandate screening for more disorders. "Parents are saying, 'If I lived in that state, my child would have gotten this test because it was required. But since I live here where it's not required, my baby might have died or been mentally retarded,'" says Brad Therrell, PhD, director of the National Newborn Screening and Genetics Resource Center, which is affiliated with the University of Texas Health Science Center at San Antonio.

Another driver in the push to get certain screening tests mandated across the country is the American College of Medical Genetics (ACMG), a professional organization that provides education and resources about genetic services. At the request of the U.S. Health Resources and Services Administration, the ACMG recently evaluated every condition that has ever been screened for, and they looked at key considerations such as: Is it a serious condition? Is there a good test we can use to screen for it? After the test, is there anything we can do about it?

Ultimately, the ACMG came up with a list of 29 conditions they feel every baby in the country should be

screened for (*see page 4 for the list of disorders*). And a number of states have expanded their newborn screening programs to include additional tests (*see "States that Screen the Most" on page 3*), says Therrell, who chaired the panel that oversaw the report. Find out what disorders your state screens for by visiting genes-r-us.uthscsa.edu, the Web site for the National Newborn Screening and Genetics Resource Center. Click on "Current Newborn Screening Conditions by State," which is regularly updated.

What the Results Mean

In most cases, the results of a baby's newborn screening tests are reported to the hospital or the baby's pediatrician. Parents may not hear anything unless the baby tests positive for a disorder; regardless, you can ask your doctor to inquire with the hospital or the state health department. If you are notified that your baby's screening was positive for a particular condition, don't panic. "It's important to keep in mind that these are screenings, not diagnostic tests," says Dr. Dolan. "A screening test tells you whether you're at increased risk -- that's it. It doesn't tell you if you have a disease."

Babies who test positive will need additional diagnostic testing to determine if they do, in fact, have a particular condition. "To ensure that pediatricians know what to do when a patient screens positive, we have developed materials for doctors, which parents can also read at acmg.net," says Michael S. Watson, PhD, executive director of the ACMG. After all, the more parents understand about screening tests and their results, the easier it will be to ensure that every baby is getting the testing and follow-up care he needs.

The Next Step

In the future, screening for additional conditions beyond those that are currently part of newborn screenings may be part of an infant's well-baby visits to the pediatrician. The Mayo Clinic, for example, is introducing screenings for Wilson's disease (an inherited disorder that causes copper to accumulate in the body and can cause liver disease and neurological or psychiatric problems) and familial hyper-cholesterolemia (an inherited disorder that results in highly elevated levels of LDL cholesterol starting at birth), says Michael S. Watson, PhD, executive director of the American College of Medical Genetics. Some conditions cannot be screened for effectively in the first 48 hours after birth, but these disorders could be accurately screened during well-baby visits, says Watson.

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