



Health Services Newsletter

Newborn Screening: a Public Health Success Story

Newborn screening is the process of testing newborn babies for some serious, but treatable, conditions. The testing process identifies conditions that can affect a child’s long-term health or survival. Each year, millions of babies in the U.S. are routinely screened, using a few drops of blood from the newborn’s heel, for certain genetic, endocrine, and metabolic disorders. Newborns are also tested for hearing loss and critical congenital heart defect prior to discharge from a hospital or birthing center.

Newborn screening can include a heel stick, hearing screen, and pulse oximetry. The conditions that newborn babies are screened for varies by state. It is just one of many things that happen in the first few days after a baby is born.

Get the facts at www.BabysFirstTest.org

Newborn screening is designed to detect disorders in infants who might not show symptoms. Healthcare providers are unlikely to recognize these conditions before symptoms arise if screening does not occur. The conditions detected by newborn screening are individually rare; however, they collectively affect about 25 million people in the United States. Rare diseases are a common cause of neurological and intellectual disabilities, and many have no cure. Screening is an effective public health approach to prevent adverse outcomes and developmental consequences for affected children and their families.

An “abnormal” result (also called out-of-range or non-pass point of care) on the newborn screening panel is not a confirmed diagnosis. Prompt action may be needed to confirm the diagnosis and initiate timely intervention because some congenital conditions can be rapidly disabling and even fatal. Early diagnosis and treatment of certain conditions can make a significant difference in improving health outcomes for affected children.

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What are states responsible for?

The state level newborn screening programs are public health systems that encompass much more than a blood test. As public health programs, they require a coordinated system of follow-up, diagnosis, and treatment designed to assist hospital, pediatric healthcare providers and state surveillance teams in connecting families to needed services. State by state information is available on the www.BabysFirstTest.org website.

How do pediatricians and healthcare providers work with the states to support identification and care coordination?

It is important that pediatricians and all child health providers ensure processes are in place for documenting that newborn screening was done and that results, and if necessary next steps, are discussed with the family/primary caregivers. Newborn screening fact sheets were developed by the Council on Genetics and Birth Defects of the American Academy of Pediatrics. These fact sheets provide information to assist pediatricians and other professionals who care for children in performing their essential role within the newborn screening public health system. They are available online at www.pediatrics.org/cgi/content/full/118/3/e934).



It must be emphasized that “normal” results of newborn screenings do not rule out the presence of these disorders, in all cases. The clinical judgement of the pediatrician remains the most important tool in the diagnosis of all of these conditions. Pediatric healthcare providers play an important role in the education of family members and support through the diagnosis and lifelong care of the infant/child.

What is a point-of-care newborn screening test?

Most conditions identified through newborn screening are done so with a heel stick dried blood spot test. However, beginning with newborn screening for congenital hearing loss in the early 1990s and also newborn screening for critical congenital heart defects after 2011, newborn screening was expanded to include point-of-care tests. Most babies receive one or both of these point-of-care tests before they are discharged from the hospital or birthing center.

Point-of-care screening encompasses physiologic tests that are administered and interpreted outside of a laboratory but close to the site of direct delivery of medical care (i.e. birth hospital/nursery). Point-of-care newborn screening describes those practices in which results are obtained at the bedside with oversight from public health agencies for the detection of a state-specified list of conditions.

Early Hearing Detection and Intervention

Hearing screening is a test to tell if babies might have hearing loss. Hearing screening is easy and not painful. In fact, babies are often asleep while being screened. It takes a very short time — usually only a few minutes. All babies should be screened for hearing loss no later than 1 month of age. It is best if they are screened before leaving the hospital after birth. If a baby does not pass a hearing screening, it's very important to get a full hearing test as soon as possible, but no later than 3 months of age. Learn more about Early Hearing Detection and Intervention from the National Center for Hearing Assessment and Management (NCHAM) (www.infanthearing.org).



Critical Congenital Heart Defects

Babies identified with one of the Critical Congenital Heart Defects (CCHDs) are at significant risk for death or disability if their heart defect is not diagnosed and treated soon after birth. Some of these heart conditions can potentially be detected using pulse oximetry screening, which is a test to determine the amount of oxygen in the blood. Most hospitals routinely screen all newborns using pulse oximetry screening. Learn more about newborn screening for CCHDs from the American Academy of Pediatrics (www.aap.org/pehdc/cchd).

Newborn Screening Continues to Expand

While newborn screening has been around for over 50 years, the conditions on the recommended universal screening panel (RUSP) continue to expand. Newborn screening was introduced in the 1960s when states first started screening for PKU (phenylketonuria), with the prick of a heel. Since then, the list of conditions for which a blood test has the ability to screen has expanded to over 60 conditions.

Every state has its own newborn screening program. To encourage uniformity, the American College of Medical Genetics and the Health Resources and Services Administration manage the consensus based process establishing the conditions that are on the Recommended Uniform Screening Panel (RUSP). All states screen for 29 core conditions. As technologies advance in treatment and detection, more conditions have been considered for the RUSP, which now includes 34 core conditions and 26 secondary conditions. Secondary conditions are detected during the screening for core conditions but do not reach the evidence threshold for inclusion as a primary target for screening.

Newborn Screening Resources for Parents

The website [BabysFirstTest.org](http://www.babysfirsttest.org) has resources that can help prepare expecting parents for the newborn screening process and answer common questions, such as: What should I do Before Birth? What are the Screening Procedures? How should I respond to the Results? What are the Screening Outcomes? What happens to the Blood Sample?— See more at: <http://www.babysfirsttest.org/#sthash.eM12n2Ek.dpuf>

Newborn Screening Fact Sheet for Providers and Families

The federal Maternal Child Health Bureau, Health Resources and Services Administration and health literacy researchers at Louisiana State University developed the following materials to help health care professionals provide parents with easy to understand newborn screening information.

- Provider Material
[Newborn Screening Disorders: What Parents Want To Know About Newborn Screening Disorders](#)
- Parent Material
[Newborn Screening Tests: These Tests Could Save Your Baby's Life](#)

Sickle Cell Disease

Sickle cell disease (SCD) is a group of inherited red blood cell disorders and is one of the most common genetic disorders in the United States affecting approximately 70,000 to 100,000 children and adults, predominantly of African and Hispanic descent. Approximately 1,000 babies are born with SCD each year in the United States.

Healthy red blood cells are round and move through small blood vessels to carry oxygen to all parts of the body. In someone who has sickle cell disease, the

red blood cells change shape to look like a C-shaped farm tool called a “sickle” and some become hard and sticky. The sickled cells break up in the blood stream, which causes a constant low level of red blood cells (also called anemia). Further, when they travel through small blood vessels, they get stuck and clog the blood flow. This may cause pain and other serious problems such as stroke, acute chest syndrome, and organ damage leading to an increased risk of infection.

Universal newborn screening allows SCD patients to be diagnosed early and to implement advances in routine care, including twice daily prophylactic penicillin and immunization against serious bacterial infections, which has significantly decreased the death rate in young children with SCD. The early administration of a daily medication called hydroxyurea can reduce long-term complications from SCD. Screening with transcranial Doppler (an ultrasound looking at blood flow to the brain) starting at two years of age has been shown to identify children at higher risk of stroke who can be offered preventative treatment with chronic transfusion therapy.

Screening for sickle cell disease in the newborn may pick up other disorders of red blood cells, such as thalassemia, which requires the review of a specialist in childhood blood disorders (pediatric hematologist).

However, the more important step in the management of abnormal neonatal hemoglobin screen is recognition of the need for referral to an expert for diagnosis.



Further, newborn screening for hemoglobinopathies, including sickle cell disease, not only identifies children with disease but also many more infants with trait conditions such as sickle cell trait. Early identification of a newborn with trait provides the opportunity for the general pediatric provider to inform and educate the family about the presence of trait, and the genetic risk to the next generation of a child with two parents with trait. This is, of course, a risk for the couple that has had a child with disease, and potentially with trait. Thus, newborn screening is the key to giving families all of the information about their child, so that they have the data to make informed choices about future pregnancies.

The Green Family Story

When AJ was born in 1992, there was a real lack of information and education about newborn screening and sickle cell disease. They just told us he was having a PKU shot and then the doctor actually called on the telephone and gave us his results.

Read about the importance of screening for sickle cell and AJ Green's experience with the disease.
<http://www.cdc.gov/features/SickleCellDisease/>

Sickle Cell Disease: Information for Parents

<https://www.healthychildren.org/English/health-issues/conditions/chronic/Pages/Sickle-Cell-Disease-in-Children.aspx>

American Sickle Cell Anemia Foundation

<http://www.ascaa.org/index.php>

Sickle Cell Disease: Information for Providers

AAP Clinical Report

<http://pediatrics.aappublications.org/content/109/3/526>

Management of Children With Sickle Cell Disease:

A Comprehensive Review of the Literature

<http://pediatrics.aappublications.org/content/128/6/e1552>

Sickle Cell Disease: 10 Things You Need to Know

<https://eclkc.ohs.acf.hhs.gov/hslc/tta-system/health/health-literacy-family-engagement/family-education/SickleCellDisea.htm;YXdpbGxpYW1zMjI3>



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School readiness begins with health!