Newborn Screening

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What is newborn screening?

Newborn screening is the practice of testing all babies in their first days of life for certain disorders and conditions that can hinder their normal development. This testing is required in every state and is typically performed before the baby leaves the hospital. The conditions included in newborn screening can cause serious health problems starting in infancy or childhood. Early detection and treatment can help prevent intellectual and physical disabilities and life-threatening illnesses.

To learn more about newborn screening:

MedlinePlus from the National Library of Medicine can point you to resources related to newborn screening (https://medlineplus.gov/newbornscreening.html).

The following websites and organizations also provide information about newborn screening:

- National Newborn Screening & Global Resource Center (http://genes-r-us.uthscsa.edu/)
- Baby's First Test (https://www.babysfirsttest.org/)
- Save Babies Through Screening Foundation (http://www.savebabies.org/)
How is newborn screening done?

Newborn screening usually begins with a blood test 24 to 48 hours after a baby is born, while he or she is still in the hospital. In some states, a second blood test is performed at a check-up appointment with the baby's pediatrician when the baby is 1 to 2 weeks old. Newborn screening is part of standard care; parents do not need to request to have the test done.

The test is performed by pricking the baby's heel to collect a few drops of blood. There are very few risks associated with this procedure, and it involves minimal discomfort to the baby. The blood is placed on a special type of paper and sent to a laboratory for analysis. Within 2 to 3 weeks, the test results are sent to the baby's doctor's office or clinic.

If a baby is born outside a hospital (for example, at home or in a birthing center), a doula or midwife may collect the blood sample needed for the newborn screening test. Otherwise, the required testing can be performed at the baby's doctor's office or at a hospital.

In addition to the blood test, most states also screen newborns for hearing loss and critical congenital heart disease. These tests are also done shortly after birth. The hearing test uses earphones and sensors to determine whether the baby's inner ear or brain respond to sound. The test for critical congenital heart disease, called pulse oximetry, uses a sensor on the skin to measure how much oxygen is in the blood. Low oxygen levels suggest that an infant may have heart problems. The hearing and pulse oximetry tests are painless and can be done while the baby is sleeping.

To learn more about how newborn screening tests are performed:

Additional information about newborn screening procedures (https://www.nichd.nih.gov/health/topics/newborn/conditioninfo/how-done) is available from the Eunice Kennedy Shriver National Institute of Child Health and Human Development at NIH.

These resources offer specific information about the tests used to screen for hearing loss:

- It's Important to Have Your Baby’s Hearing Screened (https://www.nidcd.nih.gov/health/your-babys-hearing-screening) from the National Institute on Deafness and Other Communication Disorders at NIH
- Purpose of Newborn Hearing Screening (https://www.healthychildren.org/English/ages-stages/baby/pages/Purpose-of-Newborn-Hearing-Screening.aspx) from Healthychildren.org, a service of the American Academy of Pediatrics
What disorders are included in newborn screening?

The disorders included in newborn screening vary from state to state. Most states test for all of the conditions specified by the Health Resources and Services Administration (HRSA) in their Recommended Uniform Screening Panel. These conditions include phenylketonuria (PKU), cystic fibrosis, sickle cell disease, critical congenital heart disease, hearing loss, and others. Some states test for additional disorders that are not part of the HRSA panel.

Most of the conditions included in newborn screening can cause serious health problems if treatment is not started shortly after birth. Prompt identification and management of these conditions may be able to prevent life-threatening complications.

Parents can ask their baby's healthcare provider about expanded (supplemental) screening if they live in a state that screens for a smaller number of disorders. Supplemental screening is typically done by commercial laboratories. It is separate from the testing done by the state, although it often uses a blood sample drawn at the same time.

To find out more about the disorders included in newborn screening:

These resources list the disorders included in each state's newborn screening panel:

- Baby's First Test: Conditions Screened by State (https://www.babysfirsttest.org/newborn-screening/states)
- National Newborn Screening & Global Resource Center: Newborn Screening by State (http://genes-r-us.uthscsa.edu/resources/consumer/statemap.htm)

Additional information about supplemental newborn screening is available:

- Baby's First Test: What is additional screening? (https://www.babysfirsttest.org/newborn-screening/conditions#2)
- National Newborn Screening & Global Resource Center: Commercial and Non-Profit Organizations Offering Expanded Newborn Screening Tests (http://genes-r-us.uthscsa.edu/resources/newborn/commercial.htm)
Who pays for newborn screening?

Newborn screening is performed on every infant regardless of the parents' health insurance status or ability to pay. The fees for newborn screening vary by state, from less than $15 to about $150. Some states do not charge a fee for this testing. When there is a fee, it is often covered by private health insurance plans. This testing is also covered under the Children's Health Insurance Program (CHIP) and Medicaid for those who are eligible.

If a parent chooses to have supplemental screening done through a private laboratory, that testing is not covered under the fees charged by each state for newborn screening. The costs of supplemental testing are charged by the laboratory that performs the tests. Parents should check with their health insurer to find out whether supplemental newborn screening is a covered service.

If a newborn screening test comes back positive (abnormal), further testing needs to be done to determine whether the baby has a particular condition. This additional testing involves separate costs that may be covered by health insurance plans.

To find out more about the costs of newborn screening:

The National Newborn Screening & Global Resource Center lists the cost of newborn screening in each state (http://genes-r-us.uthscsa.edu/resources/consumer/statemap.htm).


Baby's First Test has state-specific information about who pays for newborn screening. Choose your state (https://www.babysfirsttest.org/newborn-screening/states) to find out more.
What happens if a newborn screening test comes back negative?

Within 2 to 3 weeks after newborn screening tests are performed, results are sent to the baby's doctor's office or clinic. A negative result means that all of the tests are in the normal range, and they do not indicate any increased risk. Other words for a negative test result are "passing," "in-range," or "normal."

In most cases, families are not notified of negative results. Parents can contact their baby's healthcare provider if they wish to confirm that the results were negative. Usually no follow-up testing is necessary.

Rarely, the results of a newborn screening test can be a false negative. "False negative" means that a disease was missed by the screen; the test results came back negative, but the child actually has the disease. Possible reasons for a false negative result include laboratory errors, such as mixing up samples, and doing the test too early. Because false negatives are possible, further testing should be done if a baby has a family history of a particular disease or shows signs and symptoms, regardless of the newborn screening result.

Learn more about negative newborn screening test results:

Baby’s First Test provides more information about test results and what they mean (https://www.babysfirsttest.org/newborn-screening/responding-to-results).

The Minnesota Department of Health provides details about interpreting the results of several types of newborn screening tests (https://www.health.state.mn.us/people/newbornscreening/families/screeningresults.html).
What happens if a newborn screening test comes back positive?

Within 2 to 3 weeks after newborn screening tests are performed, results are sent to the baby's doctor's office or clinic. A positive result means that at least one of the tests came back outside the normal range. Other words for a positive result are "failing," "out-of-range," or "abnormal."

The healthcare provider will notify parents of a positive test result. A positive result does not mean that a baby definitely has a disease, but it indicates that further testing (called diagnostic testing, because it is used to diagnose a disease) should be performed as soon as possible. If the baby does have the disease, quick follow-up testing can allow treatment or management, such as a special diet, to begin very soon after birth.

Often when there is a positive screening test result, follow-up diagnostic testing shows that the baby does not have the disease. In these cases, the results of the newborn screening test are described as "false positive," meaning that the test suggested an increased risk of the disease when the baby does not actually have the disease. False positive test results occur because screening tests are designed to identify as many babies affected with treatable diseases as possible. Because it is critical not to miss affected babies, some babies who are unaffected also have a positive screening result.

Occasionally, the results of a newborn screening test are reported as "borderline." These results are not quite normal, but they are not clearly abnormal, either. In these cases, the baby’s healthcare provider may repeat the test.

Learn more about positive newborn screening test results:

Baby's First Test provides more information about test results and what they mean (https://www.babysfirsttest.org/newborn-screening/responding-to-results).

The Minnesota Department of Health provides details about interpreting the results of several types of newborn screening tests (https://www.health.state.mn.us/people/newbornscreening/families/screeningresults.html) and a fact sheet about borderline test results (https://www.health.state.mn.us/people/newbornscreening/families/familyfsnewborderline.pdf).

Save Babies Through Screening offers more information about positive test results and follow-up testing (http://www.savebabies.org/ips_faqs.html).
A description of false positive results is available from Baby's First Test (https://www.babysfirsttest.org/newborn-screening/false-positives).
What is newborn genomic sequencing?

Newborn genomic sequencing is an approach currently under study to collect and analyze large amounts of DNA sequence data in the newborn period. Genomic sequencing, a technology used to determine the order of DNA building blocks (nucleotides) in an individual's genetic code, is already available to test for genetic disorders in children and adults. Researchers have proposed using this technology to screen all newborns for health conditions they may have or be at risk of developing in childhood.

Newborn genomic sequencing would not replace standard newborn screening on page 3, which tests for a recommended 34 health conditions (although the exact number varies by state). Like current newborn screening, newborn genomic sequencing would allow doctors to identify health conditions very early in life. This technique would expand significantly the number and scope of health conditions that could be diagnosed soon after birth, potentially allowing doctors to start treatment and other follow-up as soon as possible.

As interest in newborn genomic sequencing grows, researchers and ethicists have identified possible ethical, social, and legal issues that need to be considered before the technology is widely adopted. These include the following considerations:

- Some genetic changes will have implications for the health of not only the infant, but of his or her parents and other family members.
- The interpretation of genomic data is constantly evolving, and right now it is unclear whether some changes in the genome are relevant to a person's health or not.
- While some genetic changes have immediate significance for an infant's health, other changes only influence the risk of developing health problems later in life. Infants are unable to provide informed consent, which is generally required when testing for adult-onset diseases.
- Newborn genomic screening raises issues of privacy and potential genetic discrimination if genomic data becomes part of a baby's medical record.

Newborn genomic sequencing may also have other risks and limitations that have not yet been recognized. All of these issues are under study as newborn genomic sequencing becomes increasingly feasible on a large scale. The NIH has sponsored several research studies to explore potential benefits, limitations, and ethical concerns in their Newborn Sequencing in Genomic Medicine and
Public Health (NSIGHT) program. The NSIGHT studies are funded through August 2018.

Scientific journal articles for further reading


Learn more about newborn genomic sequencing and the NSIGHT program:


NSIGHT clinical trials:

- Genomic Sequencing for Childhood Risk and Newborn Illness (BabySeq) (Study overview (https://www.genomes2people.org/research/babyseq/)) (ClinicalTrials.gov (https://clinicaltrials.gov/ct2/show/NCT02422511))
- Clinical and Social Implications of 2-Day Genome Results in Acutely III Newborns (ClinicalTrials.gov (https://clinicaltrials.gov/ct2/show/NCT02225522))
- Perinatal Precision Medicine (NSIGHT2) (ClinicalTrials.gov (https://clinicaltrials.gov/ct2/show/NCT03211039))