Newborn Screening

Newborn screening tests look for serious developmental, genetic, and metabolic disorders so action can be taken during the critical time before symptoms develop. Most of these illnesses are very rare but treatable if caught early.

In the United States, individual states regulate newborn screening, so the diseases screened for vary considerably from state to state. Most states require three to eight tests, but organizations such as the March of Dimes and the American College of Medical Genetics suggest more than two dozen additional tests.

The most thorough screening panel checks for about 40 disorders. All 50 states screen for congenital hypothyroidism, galactosemia, and phenylketonuria (PKU).

Screening tests do not diagnose illnesses. They identify which babies need additional testing to confirm or rule out illnesses. Remember, a negative screen does not mean that the disease is not present—it may mean that the baby was screened too early or that there were insufficient amounts of metabolites in the blood to reach the threshold for a positive result (sensitivity of the test). If the baby is showing signs and symptoms of concern, always do follow-up testing. If follow-up testing confirms that the infant has a disease, appropriate treatment can be started right away, before symptoms appear.

Normal values for each screening test may vary depending on how the test is performed. Each state laboratory should have established "normal ranges" for the newborn screening program and individual tests.

Each state has independent screening programs. To find out specifics on your state screening program, visit http://genes-r-us.uthscsa.edu.

Screening tests in state programs may include

- amino acid metabolism disorders
  - arginosuccinic acidemia
  - citrullinemia
  - homocystinuria
  - maple syrup urine disease
  - PKU
  - Tyrosinemia type I
- biotinidase deficiency
- congenital adrenal hyperplasia
- congenital hypothyroidism
- cystic fibrosis (see also neonatal cystic fibrosis screening)
- fatty acid metabolism disorders
  - carnitine uptake deficiency
  - long-chain L-3-hydroxyacyl-CoA dehydrogenase deficiency
  - medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
  - trifunctional protein deficiency
  - very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)
- galactosemia
- glucose-6-phosphate dehydrogenase deficiency (G6PD)
- organic acid metabolism disorders
  - 3-hydroxy-3-methylglutaric aciduria (HMG)
  - 3-methylcrotonyl-CoA carboxylase deficiency (3MCC)
  - beta ketothiolase deficiency
  - glutaric acidemia type I
  - isovaleric acidemia
  - methylmalonic acidemia
  - multiple carboxylase deficiency
  - propionic acidemia
- sickle cell disease and other hemoglobinopathy disorders and traits.

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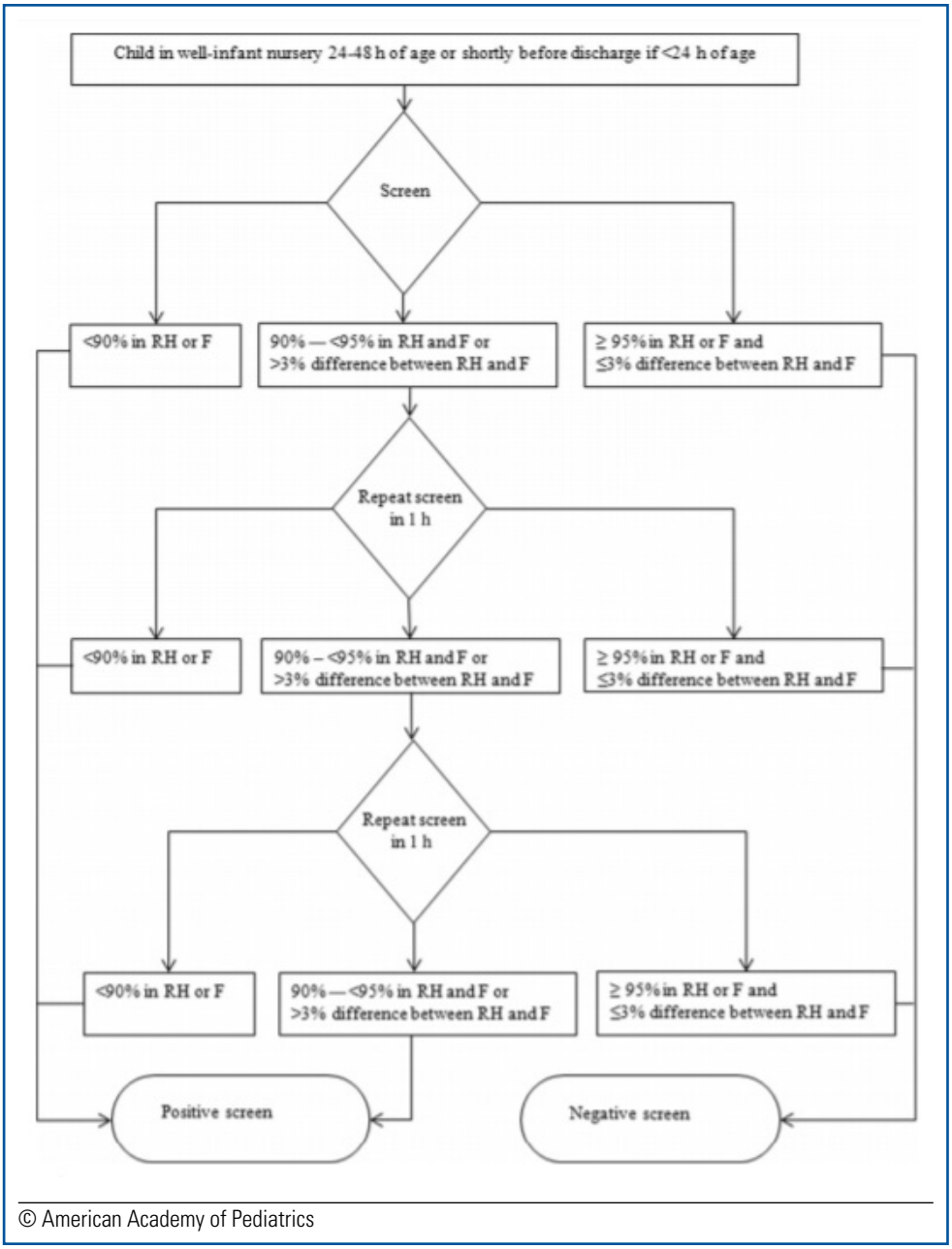
Critical Congenital Heart Defects (CCHD) Screening

Approximately 18 of every 10,000 babies are born with a critical congenital heart defect (CCHD). CCHD is life threatening and requires intervention in infancy. However, CCHD is not always detected prenatally or upon exam in the nursery. As a result, some infants with CCHD are discharged from the nursery to home, where they quickly decompensate. To improve the early detection of CCHD, the Secretary of Health and Human Services recommended that CCHD screening be added to the uniform newborn screening panel (American Academy of Pediatrics, 2017).

Screening with pulse oximetry can identify a number of types of CCHD, including coarctation of the aorta, double outlet right ventricle, Ebstein anomaly, hypoplastic left heart syndrome, interrupted aortic arch, pulmonary atresia, single ventricle, tetralogy of Fallot, total anomalous pulmonary venous return, transposition of the great arteries, tricuspid atresia, and truncus arteriosus.

Current published recommendations focus on screening infants in the well-baby nursery and in intermediate care nurseries or other units in which discharge from the hospital is common during an infant's first week of life, although not all states require this screen. Timing the screening around the time of the newborn hearing screening can help improve efficiency.

A pulse oximeter is used to measure the percentage of hemoglobin in the blood that is saturated with oxygen (Centers for Disease Control and Prevention, 2017). A failed screen occurs when there is an oxygen saturation measure that is less than 90% (in the initial screen or in repeat screens); oxygen saturation is less than 95% in the right hand and foot on three measures, each separated by 1 hour; or a greater than 3% absolute difference exists in oxygen saturation between the right hand and foot on three measures, each separated by 1 hour. Any infant who fails the screen should have an evaluation such as an echocardiogram for causes of hypoxemia.



A passed screen occurs when the oxygen saturation measure is greater than or equal to 95% in the right hand or foot with an equal to or less than 3% absolute difference between the right hand and foot. Pulse oximetry screening does not detect all CCHDs, so it is possible for a baby with a passing screening result to still have a CCHD or other congenital heart defect.

References

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Newborn Screening: Information for Parents

All newborn babies are given tests before they leave the hospital to identify possible serious or life-threatening conditions that they may have, even if they don't have symptoms. Serious diseases are rare. Some disorders can slow down an infant's normal physical and mental development in a variety of ways. Parents can pass along the gene for a certain disorder without even knowing that they carry the gene. Most disorders can be treated if found early, but not all disorders found during screening can be treated.

The following are excellent resources if you need more information about genetic screening:

- National Newborn Screening and Genetics Resource Center: www.genes-r-us.uthscsa.edu
- American College of Medical Genetics: www.acmg.net
- March of Dimes: www.marchofdimes.com/pnhec
- Baby's First Test: www.babysfirsttest.org

Most tests use a few drops of blood collected on a special sheet of paper by pricking the baby's heel. If a screening test suggests a problem, your baby's provider will follow up with more testing. If those tests confirm a problem, the provider may refer you to a specialist for treatment. Even though these conditions are considered rare and most babies are given a clean bill of health, finding disorders early and giving proper treatment can make the difference between lifelong challenges and healthy development for your child.

Although individual states may not perform all screening tests, parents can have additional tests done by qualified laboratories at large medical centers. Private laboratories also offer newborn screening. Parents can find out about extra newborn screening tests from their provider or the hospital where their baby was born, as well as through organizations such as the March of Dimes.

An abnormal result means that the baby should have additional testing to confirm or rule out the condition, but remember, normal value ranges may vary slightly among different laboratories. Talk to your baby's provider about the meaning of your specific test results.

Critical Congenital Heart Defects (CCHD) Screening

Congenital heart defects are heart conditions that a baby is born with. Critical congenital heart disease (also called CCHD) is a group of severe congenital heart defects. These defects can affect the shape of a baby's heart, the way it works, or both. Babies with CCHD need treatment within the first few hours, days, or months of life. Without treatment, CCHD can be deadly. Your baby may be tested for CCHD as part of newborn screening before he or she leaves the hospital after birth (older than 24 hours), though not all states require this screen. Babies are screened for CCHD with a small machine called a pulse oximeter (also called pulse ox) that uses sensors attached to your baby's hand and foot. This testing is painless and only takes a few minutes.

The pulse oximeter checks your baby's oxygen level in the blood. Low levels of oxygen in the blood can be a sign of a CCHD. If a low level of oxygen is detected in your baby's blood, your baby's doctor may order further testing, such as an echocardiogram (a heart ultrasound), to check for any severe congenital heart defects.