# 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](http://www.genes-r-us.uthscsa.edu/)
* American College of Medical Genetics: [www.acmg.net](http://www.acmg.net/)
* [March of Dimes: www.marchofdimes.com/pnhec](http://www.marchofdimes.com/pnhec)
* Baby’s First T[est: www.babysfirsttest.org](http://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. With- out 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.