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.

Bibliography

American College of Medical Genetics Newborn Screening Expert Group. (2006). Newborn screening: Toward a uniform screening panel and system—Executive summary. *Pediatrics, 117*(5 Pt 2), S296–S307.

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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.