Caring for Your Baby

Newborn Screening

Newborn babies are screened for a variety of conditions. The conditions are all rare, but affected babies greatly benefit from early diagnosis and treatment. These tests require the collection of a blood specimen obtained by pricking the heel of the infant. The blood specimen is taken near the time of hospital discharge but before 7 days of age. (If the initial specimen was collected before 24 hours of age, a second specimen is collected before 2 weeks of age.*

Testing for newborn hemoglobinopathies (red blood cell disorders), including sickle cell anemia and beta thalassemia, is also done in a majority of states. States and provinces vary widely in the number and types of conditions for which they screen. The American College of Medical Genetics recommends twenty-nine conditions be targeted for screening. The March of Dimes offers parents information about all these conditions and tests.** The National Newborn Screening and Genetics Resource Center provides information about commercial and non-profit organizations offering newborn screening tests that parents may use to test their infants for conditions not targeted by their state or province's testing.***

Test or Procedure	What It Is	Comments
Test for PKU (phenylketonuria) Done by all U.S. states and Canadian provinces.	PKU is an inherited metabolic disease occurring in 1 infant in every 25,000 in the United States.** With this disease, the infant is unable to digest phenylalanine (an amino acid), which builds up in the blood.	If untreated, PKU causes mental retardation. It's treated by diet. A low phenylalanine formula or combining breastfeeding with a low phenylalanine formula is used during infancy. Once solid foods are introduced, a special diet followed through adolescence is very effective in preventing retardation.*
Test for hypothyroidism Done by all U.S. states and Canadian provinces.	Hypothyroidism (low production of thyroid hormone) occurs in 1 in every 5,000 newborns in the United States.** The condition can be transient or long-term.	There is a higher incidence among females, offspring of mothers with thyroid disorders, and those who have other children with thyroid disorders. Treatment with replacement hormone avoids serious long-term effects, including mental retardation, growth failure, deafness, and neurological abnormalities.*†
Test for galactosemia. Done in the majority of U.S. states.	Galactosemia (inability to digest galactose, a sugar in milk) is a hereditary disorder and occurs in 1 in 50,000 infants.**	Symptoms include vomiting, diarrhea, jaundice, and poor weight gain. Treatment includes a diet that is galactose free. Without treatment, galactosemia is fatal.*

*American Academy of Pediatrics Committee on Genetics, "Newborn Screening Fact Sheets," *Pediatrics* 2006;118;934-963.
†J. H. Dussault, "Screening for Congenital Hypothyroidism," *Clinical Obstetrics and Gynecology* 40(1) (March 1997): 117-23.
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