10 Things Parents Want To Know About Newborn Screening

From Their Baby's Health Professional

1. The Texas Newborn Screening Program checks all newborn babies for a number of rare disorders. The screening tests are very important for your baby's health.

2. Babies with these disorders may look healthy at birth. Many disorders can't be seen.

3. Serious problems, such as an intellectual/developmental disability, illness, or death, may be prevented if we find the disorders right away.

4. Newborns are first tested 1 to 2 days after birth before they leave the hospital and again at 7 to 14 days of age in their doctor's office or clinic.

5. To do the test, a health professional will take a few drops of blood from your baby's heel.

6. Your baby's health professional or the hospital will get a copy of the test results. Call your baby's health professional if you would like to talk about the results.

7. Some babies may need more tests. You will be notified if your baby needs more tests. *It is very important for your baby to get these tests quickly.*

8. The blood spot cards are stored for up to 2 years, and may be used to ensure laboratory tests, equipment and supplies are working right, to develop new tests, and for Department of State Health Services studies of diseases that affect public health.

9. If you give your OK, the blood spot cards will be stored for up to 25 years, and may be used for public health research outside of the Department of State Health Services. You, the parent/guardian, decide what the lab does with your baby's blood spots after testing by completing and sending in a decision form. The decision form will be given to you when the blood spots are collected. Your baby's information stays private and secure no matter your decision.

10. For more information, talk to your baby's health care provider. If you have more questions about newborn screening, call the Texas Department of State Health Services - Newborn Screening Program at 1-800-252-8023 ext. 3957. If you have more questions about bloodspot card records, call 1-888-963-7111 ext. 7333.

Texas Department of State Health Services
Newborn Screening Program
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WHAT PARENTS WANT TO KNOW ABOUT NEWBORN SCREENING

From Their Baby's Health Care Professional

The Health Professional's Guide for Brief Discussion with Parents

Quick Reference To Newborn Screening Disorders

Biotinidase Deficiency (BIOT) BIOT is an enzyme deficiency that occurs in about 1 in 60,000 newborns and can result in seizures, hearing loss, and death in severe cases. Treatment is simple and involves daily doses of biotin.

Congenital Adrenal Hyperplasia (CAH) CAH is caused by decreased or absent production of certain adrenal hormones. The most common type is detected by newborn screening in about 1 in 15,000 newborns. Early detection can prevent death in boys and girls and sex mis-assignment in girls. Treatment involves lifelong hormone replacement therapy.

Congenital Hypothyroidism (CH) Inadequate or absent production of thyroid hormone results in CH and is present in about 1 in 3,500 newborns. Thyroid hormone replacement therapy begun by 1 month of age can prevent intellectual/developmental disabilities.

Cystic Fibrosis (CF) CF is a disease that affects the flow of salt and water in the body. CF is one of the most common genetic disorders, with about 1,000 new cases diagnosed in the U.S. every year. If both parents are CF carriers, there is a 1 in 4 chance their child will have the disorder. Most people with cystic fibrosis live into their 30s, however, with new treatments and research, the life expectancy may be greater. Treatments vary depending on the stage of the disease and which organs are affected.

Galactosemia (GALT) Failure to metabolize the milk sugar galactose results in GALT and occurs in about 1 in 50,000 newborns. The classical form detected by newborn screening can lead to cataracts, liver cirrhosis, intellectual/developmental disabilities, and/or death. Treatment eliminates galactose from the diet, usually by substituting soy for milk products.

Homocystinuria (HCY) HCY is caused by an enzyme deficiency that blocks the metabolism of an amino acid that can lead to intellectual/developmental disabilities, osteoporosis, and other problems if left undetected and untreated. The incidence is approximately 1 in 350,000 U.S. newborns. Treatment may involve dietary restrictions and supplemental medicines.

Maple Syrup Urine Disease (MSUD) MSUD is a defect in the way that the body metabolizes certain amino acids and is present in about 1 in 200,000 U.S. newborns. Early detection and treatment with dietary restrictions can prevent death and severe intellectual/developmental disabilities. There is an increased risk in Mennonites.

Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency The most common disorder in the way the body metabolizes fatty acids is called MCAD deficiency. Undetected, it can cause sudden death. Treatment is simple and includes ensuring regular food intake. The incidence from newborn screening is not yet known, but is thought to be approximately 1 in 15,000 newborns.

Other Fatty Acid Oxidation (FAO) Disorders Besides MCAD deficiency, other FAO disorders may be detected through newborn screening. They are usually described in categories based on the length of the fatty acid involved. Undetected and untreated they can cause seizures, coma, and even death. The incidences of various FAO disorders are not known since it is only recently that early detection through newborn screening has occurred.

Organic Acid (OA) Disorders Organic acidemias are a group of metabolic disorders that lead to buildup of organic acids in the blood and urine and may be detected in newborn screening through analysis of acylcarnitine profiles. Restricting protein in the diet and supplementation with vitamins and/or carnitine can diminish symptoms. Because newborn screening for these disorders is relatively new, the degree of occurrence in newborns is not yet known.

Phenylketonuria (PKU) An enzyme defect that prevents metabolism of phenylalanine, an amino acid essential to brain development, is known as PKU. It occurs in approximately 1 in every 19,000 U.S. newborns. Undetected and untreated with a special diet, PKU leads to irreversible intellectual/developmental disabilities. Persons of European descent are at increased risk.

Severe Combined Immunodeficiency Disorder (SCID) SCID is one of the most serious and life-threatening immune system problems and is often fatal in the first year of life. SCID is a group of disorders with genetic causes that can occur in about 1 in 40,000 to 1 in 100,000 newborns. Although rare, SCID can be successfully treated if identified early in life. Infants who test positive for SCID should be referred to a pediatric immunologist for further testing.

Sickle Cell Disease (SCD) Sickle cell anemia (Hb-SS-Disease) is the most common SCD and causes clogged blood vessels resulting in severe pain and other severe health problems. Other common SCDs include Hb-SC-Disease and various thalassemias. Newborn screening detects about 1 in 2,500 newborns with SCD annually. Persons of African or Mediterranean descent are at an increased risk.

Tyrosinemia (TYR1) People with tyrosinemia have problems breaking down an amino acid called tyrosine, which is one of the building blocks of protein. If not treated, the condition causes severe liver disease and other serious health problems. Treatment consists of medication and a diet low in tyrosine. The estimated incidence is 1 case in every 100,000 live births.

Urea Cycle Disorders (UCD) A UCD is a genetic disorder caused by a deficiency of one of the enzymes responsible for removing ammonia from the bloodstream. Some UCDs may be detected as a part of newborn screening. They are characterized by seizures, poor muscle tone, respiratory distress, and coma, and result in death if left undetected and untreated. Because newborn screening for these disorders is relatively new, the degree of occurrence in newborns is not yet known.