State of Connecticut
Department of Public Health
Genetics Newborn Laboratory Screening Program
Family Fact Sheet

FOR YOUR BABY'S HEALTH

In 1964, the CT Statewide Newborn Screening Program (NBSP) was implemented with the screening for PKU and galactosemia. Subsequently, six other screens were added to the panel: Congenital Hypothyroidism (1976); Hemoglobinopathies (1990); Maple Syrup Urine Disease, Homocystinuria, and Biotinidase Deficiency (1993); and Congenital Adrenal Hyperplasia (1997). Over 1.5 million newborns have been tested since the program began. Connecticut State Law mandates that all newborns delivered in Connecticut be screened for these eight disorders. The goal of newborn screening is early identification of infants at increased risk for selected metabolic or genetic diseases, so that medical treatment can be promptly initiated to avert complications and prevent irreversible problems. Infants with abnormal screening results are referred to designated State Regional Treatment Centers for comprehensive testing, counseling, education, treatment, and follow-up services.

Why is this test done?
By law, all newborns are tested for several rare, but serious, conditions. If not treated these conditions can cause mental retardation, slow growth, or death.

Who should be tested?
Every baby should have this test.

When should it be done?
The test should be done before the baby goes home or by the 4th day of life.

How is the test done?
Your baby's heel is pricked. A few drops of blood are put on a special filter paper.

What happens with the results?
Results are sent to the hospital where the baby was born. Your baby's doctor will call you if the results are not normal. **This does not necessarily mean that your baby has a disease.** The doctor will discuss the need for more testing.
More testing and treatment may be done at a State Regional Treatment Center. If your baby does have one of these conditions, it is important that treatment starts as soon as possible.

How are these diseases treated?
Each disease is different. Treatment may include a special diet, hormones and/or medicine.

Can I say no to the test?
The law says that parents may refuse the test for religious reasons.

What disorders are screened for? For more information on the individual diseases, see Fact Sheets.

Biotinidase Deficiency
With this condition, the body cannot make enough free biotin. Taking the vitamin biotin daily helps to prevent severe skin rashes, eyesight and hearing problems, and brain damage.

Congenital Adrenal Hyperplasia
With this condition, the body cannot make enough of certain hormones. Taking the missing hormones helps to prevent severe illness or death.

Congenital Hypothyroidism
This condition is caused by a lack of thyroid hormone. Taking the missing hormone helps to prevent slow growth and mental retardation.

Galactosemia
With this condition, the body cannot use a sugar found in milk, infant formula, breast milk, and other foods. A special diet helps prevent damage to the brain, eyes and liver.

Hemoglobinopathies
These conditions cause problems with red blood cells and can lead to anemia, infections, pain, slow growth and even death. Special medical care and penicillin help to prevent these problems.

Amino Acid Disorders
Babies with one of these disorders cannot break down certain amino acids. The amino acids build up in the urine or blood. Amino acids are found in foods like meat, milk, baby formulas, and breast milk. Special diets, vitamins, and special medicines help to prevent serious problems. The problems may include vomiting, diarrhea, unusual odor or color of urine, a buildup of acid in the body, slow development, or mental retardation.
Phenylketonuria (PKU) is an example of an Amino Acid Disorder.

Fatty Acid Oxidation Disorders (FAOD)
Babies with one of these disorders have trouble using fat for energy. Fatty acids are a part of fat in the food we eat and from fat in our tissues. Oxidation is the process that breaks down fatty acids to release energy needed for body functions. Each step of the oxidation process is set in motion by a specific enzyme. Fatty acid oxidation disorders occur when one of these enzymes is missing. This can lead to drowsiness, poor tone, vomiting, low blood sugar, liver failure, and muscle problems or death. Treatment depends on the disorder a baby has but may include avoiding fasting, a special low fat diet, and a medicine called carnitine.

Organic Aciduria Disorders (OA)
Babies with one of these disorders cannot use certain amino acids and fatty acids. This can cause vomiting, poor feeding, low blood sugar, drowsiness, seizures, or death. Treatment may include a special low protein diet and/or medication.
Propionic Acidemia (PPA) is an example of an Organic Aciduria Disorder.

IMPORTANT POINTS TO REMEMBER

- ❑ Pick a doctor for your baby before he or she is born.
- ❑ **Make sure that the Newborn Hearing and Laboratory Screening tests are done before you leave the hospital.**
- ❑ In case your doctor needs to reach you after you go home, make sure you give your phone number to the hospital and doctor. If you don't have a phone, leave the number of a friend or neighbor with the doctor and hospital.
- ❑ Tell your nurse in the hospital if your baby will have a different last name after you go home.
- ❑ As soon as possible after you go home, call and make an appointment for your baby with your baby's doctor or other health care provider.
- ❑ For more information, call your baby's doctor, nurse, clinic staff, or the Newborn Screening Program at 860-509-8081.

CONNECTICUT DEPARTMENT OF PUBLIC HEALTH

Keeping Connecticut Healthy

860-509-8081