Newborn Screening

All states in the U.S. have some type of newborn screening program. Tests are done to detect certain serious diseases as soon as possible after the baby is born. For many of these diseases, early treatment can avoid serious health consequences for your baby. Screening requires only a simple blood test. In most states, screening is required by law.

What is newborn screening?

There are many types of congenital diseases (present from birth), a large number of which are genetic (inherited). Genes contain information that determines our physical and mental characteristics. Genetic diseases may result when a baby inherits abnormal genes from one or both parents. Many families don't known about the abnormal genes until they have an infant diagnosed with a genetic disease. Many congenital and genetic diseases have serious complications (such as mental retardation) that can only be prevented if they are recognized and treated as soon as possible after birth.

For this reason, all states recommend certain screening tests for all newborn infants. The list of diseases tested for varies among states. The tests are done using a single blood sample obtained from a small puncture in the baby's heel.

- *Hearing loss.* Most states require a hearing test for all infants. Regardless of the cause, children with hearing loss need early intervention to promote normal development.

Tests Currently Required Statewide:

- **Congenital Hypothyroidism** - Congenital hypothyroidism results when the body does not make enough thyroid hormone. If untreated, this disorder can cause mental retardation, abnormal growth, deafness, and neurological problems. Treatment includes medication to replace the missing thyroid hormone. Early treatment can prevent the damage caused by this disease.

- **Galactosemia** - Galactosemia occurs when the body cannot use the part of milk sugar called galactose. If untreated, babies with galactosemia can have mental retardation, developmental disabilities, or may even die. Babies with galactosemia require a special diet without any galactose (must avoid milk and milk products).

- **Hemoglobinopathies, including Sickle Cell Disease** - In this group of disorders, the red blood cells are malformed. Babies with these disorders are more likely to have anemia, episodes of severe pain, and life-threatening infections. Babies with any of the hemoglobinopathies could require a variety of treatments, including antibiotics to prevent serious infections.

- **Phenylketonuria (PKU)** - Newborns with PKU cannot break down a substance called phenylalanine, which is found in the protein of many foods. If untreated, this disorder can cause mental retardation, seizures and developmental delays. Treatment consists of a diet with low levels of phenylalanine.

- **Biotinidase Deficiency** - Infants with this disorder cannot process the vitamin biotin in a normal manner. Regular medical care and treatment with a medicine containing biotin can prevent seizures, hearing loss and delays in development.

- **Congenital Adrenal Hyperplasia (CAH)** - This is a group of disorders in which the adrenal glands do not produce normal amounts of certain essential hormones. If untreated, serious loss of body salt and water, and even death, may occur. Lifetime treatment includes daily medication and close monitoring to prevent complications of the disease.

- **Cystic Fibrosis** - This disorder causes thick mucus to collect in the lungs and intestines, which can result in breathing problems, lung infections and poor digestion of food. These newborns need frequent medical monitoring, including specialist care, as well as a healthy diet. Early identification and treatment has been shown to improve growth and development in children with cystic fibrosis.

- **Maple Syrup Urine Disease** - This is a rare metabolic disorder in which a baby's body cannot break down certain proteins found in foods, including breast milk and formula. The name of this disorder comes from the distinctive maple syrup odor of the urine in an affected baby. Lifetime treatment includes a special diet of foods with low levels of certain proteins, and frequent monitoring by a doctor and a metabolic genetics specialist.

- **Fatty Acid Oxidation Disorders**
    - **Medium chain acyl-CoA dehydrogenase deficiency (MCAD)**
    - **Short chain acyl-CoA dehydrogenase deficiency (SCAD)**
    - **Long chain acyl-CoA dehydrogenase deficiency (LCAD)**
    - **Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)**

  Babies with any of these fatty acid oxidation disorders have trouble using fat for energy. A special enzyme, which converts fat to energy, is either missing or not working correctly. This can lead to a medical crisis if the baby fasts (goes without eating) for very long. This crisis can include vomiting, low blood sugar or more serious problems such as coma. Typically, a special diet and special precautions may need to be taken if the baby is sick

- **Urea Cycle Disorders**
    - **Argininosuccinic acidemia**
    - **Citrullinemia**

  These urea cycle disorders are metabolic problems where excessive amounts of ammonia accumulate in the blood. These excessive amounts of ammonia can lead to behavioral problems, mental retardation, coma and even death. Babies with these disorders need a special diet and/or medication with medical monitoring.

- **Organic Acidemias**
    - **Methylmalonic academia (MMA)**
    - **Propionic academia (PPA)**
    - **Isovaleric academia (IVA)**
    - **Glutaric academia Type I (GA –type 1)**
    - **3 - hydroxy - 3 methyl glutaryl - CoA Lyase deficiency (HMG)**
    - **3 – methyl – crontonyl – CoA carboxylase deficiency (3-MMC)**

Babies born with one of these disorders cannot remove certain waste products from their blood. This can lead to vomiting, low blood sugar, developmental delays and coma. Treatment depends on the disorder the baby has, but may include a low protein diet, vitamins, and avoiding fasting. A baby born with one of these disorders needs lifetime treatment and regular medical care.

Babies with abnormal screening results are aggressively followed by the Newborn Screening and Genetic Services Program in Special Child, Adult and Early Intervention Services, to ensure that affected children and their families are linked with a primary care provider and the regional network of specialty care centers to receive timely and appropriate services.

Early identification of all disorders screened in the newborn period is only the first step in a successful newborn screening program. Additional resources and funding to ensure immediate access to confirmatory testing, follow-up, and assurance of appropriate and comprehensive treatment services are also required. Consultants have been identified to provide comprehensive services for the various disorders and state funding has been committed to provide a statewide safety net of pediatric metabolic treatment centers, cystic fibrosis care centers, sickle cell programs, endocrine specialists and regional biochemical genetics laboratories.

Additionally, affected children must be reported to the Special Child Health Services Registry which will further link the family to community-based, culturally competent, comprehensive case management and early intervention services.

How common are these diseases?

Fortunately, many of these diseases are rare. Of the 4 million infants who are screened each year in the Untied States, only about 3000 are found to have a congenital disease, excluding hearing loss. That means that only about 1 in 1300 newborns screened has any abnormal results. Of your baby does have a congenital disease, then early detection and treatment provide the best chance to avoid future problems with health and development.

Are there any risks of testing?

Routine newborn screening has little or no risk for you infant. Most of the time, all tests can be performed, using a single blood sample: your baby will feel a little pain from a pinprick on the heel. A simple hearing test may be performed as well. *The biggest risk is not testing!* Without newborn screening, there is a small but real chance of complications occurring from unrecognized diseases.

If you have any questions or need additional information, please contact the following:

Newborn Screening and Genetic Services Program
Special Child, Health and Early Intervention Services
New Jersey Department of Health and Senior Services
P.O. Box 364
Trenton, New Jersey 08625-0364
or call (609) 292-1582

Allen Bergum - E-mail: allen.bergum@doh.state.nj.us
Public Health and Environmental Laboratories
Inborn Errors of Metabolism Laboratory
New Jersey Department of Health and Senior Services
P.O. Box 371
Trenton, New Jersey 08625-0371
or call (609) 292-4811

Where to Get Supplemental Screening

Because your child's routine newborn screening test may not be enough, supplemental newborn screening is recommended to ensure your baby receives comprehensive newborn screening.

•Laboratories

Contact one of the laboratories below to find out how to obtain a parent packet. Packets contain screening materials and information for you and your physician. The screening packet should be obtained several weeks prior to delivery so that arrangements can be made in advance for the hospital to draw an extra sample of your baby's blood when the routine newborn screening is done.

Lab information is subject to change. Contact individual labs for their most current info.

These facilities offer complete screening.*	These facilities offer partial screening only.**
Pediatrix Screening 1-866-993-2300 50+ disorders $95.35	**Mayo Medical Laboratories** 1-800-533-1710 35 disorders for $57.30 Ask for Supplemental Newborn Screen, MML Test #82594 *By physician order only* **Baylor Medical Center** 1-800-422-9567 30+ disorders for $25 *Not Available in New York*