Almost every child born in the United States undergoes state-mandated newborn screening. In each state, a small blood sample (“heel stick”) is collected within 48 hours of birth. The sample is sent to a laboratory and tested for a panel of medical conditions. State newborn screening panels include testing for an ever-increasing number of conditions. Every year, over 100,000 newborns have an abnormal screen for one of these conditions. In the event that a newborn is affected by one of the diseases screened for, early medical intervention can reduce the severity of the condition and possibly even prevent symptoms from occurring. This chapter provides an overview of newborn screening programs in the New York – Mid-Atlantic region. In the U.S., newborn screening programs are state-mandated, and each state’s list of screened conditions varies. Efforts are underway to develop a consistent panel to be used throughout the U.S. New technologies have enabled substantial expansion of newborn screening programs.
4.1 Overview of Newborn Screening

By state law, all newborns are screened for various serious medical conditions. Babies with any of these conditions may look healthy at birth; but, if left untreated, these conditions can cause health problems such as mental retardation, slow growth, and even death. These outcomes may be prevented with treatment and long-term follow-up.

Newborn screening programs began in the U.S. in the 1960s with the work of Dr. Robert Guthrie, who developed a screening test for phenylketonuria (PKU). PKU is an inherited metabolic disease caused by a mutation of the gene for an enzyme responsible for metabolism of the amino acid phenylalanine. Children who are identified early can avoid foods with phenylalanine, thereby avoiding buildup of the amino acid, which would otherwise lead to brain damage and mental retardation. When Dr. Guthrie introduced a system for collecting and transporting blood samples on filter paper, cost-effective, wide-scale genetic screening became possible.

4.1.1 Screening Procedure and Follow-up. A nurse or other medical professional takes a few drops of blood from the baby’s heel. The blood should be drawn after the baby is 24 hours old, but before the baby leaves the hospital. This blood sample is sent to a newborn screening laboratory. The baby’s doctor contacts the parent(s) if the results are not in normal range for any of the screened conditions. If this scenario occurs, follow-up testing may be required.

4.1.2 Retesting. Sometimes a baby must be screened again. This does not necessarily mean that a medical condition is present. Retesting may need to be done if:

• The blood sample was taken before the baby was 24 hours old
• A problem occurred with the way the blood sample was taken
• The first test showed risk of a possible medical condition

The baby’s doctor or the state’s newborn screening program will contact the parent(s) if retesting is necessary. It is important to get this testing done right away.

4.1.3 Clinical Evaluation and Diagnostic Testing. Occasionally, the results of the newborn screen strongly suggest that the infant has one of the conditions. The newborn screening program notifies one of four specialty-care centers, depending on which test was abnormal. The specialties are metabolic, cystic fibrosis, endocrine, and hematology. The parents will be notified by the newborn screening program, the primary physician, the hospital of birth, or the specialty-care center, depending on the newborn screening program’s protocol. If this happens, it is extremely important that the parents bring their child to the specialist as soon as possible, sometimes that very day, for further evaluation and laboratory testing.

4.1.4 Treatment. The treatment for each condition is different and may include a special diet, hormones, and/or medications. It is very important to start the treatment of affected infants as soon as possible.
4.1.5 Tests Performed. Completed tests vary from state to state. Typically, each state has an advisory committee that reviews and selects which conditions are screened for based on current scientific and clinical data. Social and ethical issues are also included in the decision-making process. Increasingly, tandem mass spectrometry is being used for newborn screening. This technology is capable of screening for over 50 metabolic conditions from dried blood-spot specimens. In 1999, the American College of Medical Genetics released a report commissioned by the U.S. Health Resources and Services Administration recommending a uniform screening panel of 29 genetic conditions. Efforts are under way to examine the feasibility of instituting a uniform newborn screening policy so that every infant is screened for the same conditions, regardless of the state in which he or she is born. In general, the conditions on newborn screening panels fall into one of the following groups: metabolic conditions, endocrine conditions, hemoglobin conditions, and pulmonary conditions.

For information on the diseases tested for in a particular state, contact that state’s newborn screening program or the National Newborn Screening and Genetics Resource Center (genes-r-us.uthscsa.edu). Screening for more conditions may be available at other laboratories for a fee.

4.2 Newborn Screening Programs

**Delaware**
Delaware Health and Social Services, Division of Public Health
Delaware Public Health Laboratory
30 Sunnyside Road
P.O. Box 1047
Smyrna, DE 19977
Ph: 302.223.1520

**District of Columbia**
District of Columbia Department of Health
Newborn Screening Program
825 North Capital Street, NE
Washington, DC 20002
Ph: 202.650.5000
[www.dchealth.dc.gov/doh/site/default.asp](http://www.dchealth.dc.gov/doh/site/default.asp)

**Maryland**
Maryland Department of Health and Mental Hygiene
Division of Newborn and Childhood Screening
201 West Preston Street, Room 1A6
Baltimore, MD 21201
Ph: 410.767.6099
[www.fha.state.md.us/genetics/newprog.cfm](http://www.fha.state.md.us/genetics/newprog.cfm)

**New Jersey**
New Jersey Department of Health and Senior Services
Public Health and Environmental Laboratories
Newborn Genetic and Biochemical Screening Program
Health and Agriculture Building
Market & Warren Streets, P.O. Box 371
Trenton, NJ 08625
Ph: 609.292.4811
[www.state.nj.us/health/fhs/nbs/index.shtml](http://www.state.nj.us/health/fhs/nbs/index.shtml)

**New York**
New York State Department of Health
Wadsworth Center
Newborn Screening Program
Empire State Plaza, P.O. Box 509
Albany, NY 12201
Ph: 518.473.7552
[www.wadsworth.org/newborn](http://www.wadsworth.org/newborn)

**Pennsylvania**
Pennsylvania Department of Health
Bureau of Family Health
Division of Newborn Screening
Health and Welfare Building
7th and Forster Streets
7th Floor, East Wing
Harrisburg, PA 17120
Ph: 717.783.8143
[www.dsf.health.state.pa.us/health/cwp/view.asp?a=179&q=232592](http://www.dsf.health.state.pa.us/health/cwp/view.asp?a=179&q=232592)
4.3 NEWBORN HEARING SCREENING

Hearing loss is a common condition present in as many as one in every 300 babies. When hearing loss goes undetected, even for just a year or two, serious delays in speech and language can result. When hearing loss is discovered in infancy, treatment can be started early enough to prevent or lessen these delays.

4.3.1 Screening Procedure. Babies are usually screened in the first few days of life, before they are discharged from the hospital. The screen, which is quick and painless, is done by one of two methods: otoacoustic emissions (OAE) or automatic brainstem response (ABR). Both of these methods involve placing tiny earplugs in the ear canals or earphones on the ears and using a computer to measure the baby’s reactions to sound. The OAE test measures how the baby’s inner ear responds to sound, and the ABR test measures how the brain responds to sound. Typically, testing is done when the baby is asleep and unaware of the testing. Passing the hearing screening indicates that the baby’s hearing is within the normal range at the time of the test. However, some babies with a family history of hearing loss, repeated ear infections, or serious illness may develop hearing loss later. The child's hearing and speech should be monitored as he or she grows.

4.3.2 Retesting. Babies who do not pass the first screening are retested and may be referred to an audiologist (hearing specialist). The second screening should occur while the baby is still in the hospital or within two weeks after leaving the hospital. If the baby does not pass the initial hearing screening, it does not mean that the baby has permanent hearing loss since most babies who do not pass the first hearing screening pass the second screening. Often, babies can have fluid, blockage, or debris in the ear that clears naturally. If further testing shows that a baby has hearing loss, an audiologist along with an ear, nose, and throat specialist can best determine the next steps.

4.3.3 Treatment. Treatment will depend on the type and degree of hearing loss. If hearing loss is permanent, treatment options include hearing aids, cochlear implants, or early intervention services.
### 4.4 Newborn Hearing Screening Programs

**Delaware**  
Delaware Health and Social Services,  
Division of Public Health  
Delaware Newborn Hearing  
Screening Program  
655 Bay Road, Suite 216  
Dover, DE 19903  
Ph: 302.741.2975  
[www.dhss.delaware.gov/dhss/dph/chca/dphnhs1.html](http://www.dhss.delaware.gov/dhss/dph/chca/dphnhs1.html)

**District of Columbia**  
District of Columbia Department of Health  
Newborn Hearing Screening Program  
825 North Capital Street NE, 3rd Floor  
Washington, DC 20002  
Ph: 202.671.5000

**Maryland**  
Maryland Department of Health and Mental Hygiene  
Office of Genetics and Children with Special Health Care Needs  
Infant Hearing  
201 West Preston Street, Room 423A  
Baltimore, MD 21201  
Ph: 410.767.6432  
[www.fha.state.md.us/genes/inf_hrg.cfm](http://www.fha.state.md.us/genes/inf_hrg.cfm)

**New Jersey**  
New Jersey Department of Health and Senior Services  
Early Hearing Detection and Intervention Program  
50 East State Street, P.O. Box 364  
Trenton, NJ 08625  
Ph: 609.292.5676  

**New York**  
New York State Department of Health  
Division of Family Health  
Early Intervention Program  
Empire State Plaza  
Corning Tower, Room 287  
Albany, NY 12237  
Ph: 518.473.7016  
[www.health.state.ny.us/community/infants_children/early_intervention/newborn_hearing_screening](http://www.health.state.ny.us/community/infants_children/early_intervention/newborn_hearing_screening)

**Pennsylvania**  
Pennsylvania Department of Health  
Pennsylvania Newborn Hearing Screening and Intervention Program  
Health and Welfare Building  
7th and Forster Streets  
7th Floor, East Wing  
Harrisburg, PA 17108  
Ph: 717.783.8143  

**Virginia**  
Virginia Department of Health  
Virginia Early Hearing, Detection, and Intervention Program  
109 Governor Street, 8th Floor  
Richmond, VA 23219  
Ph: 804.864.7713  
[www.vahealth.org/hearing](http://www.vahealth.org/hearing)

**West Virginia**  
West Virginia Department of Health and Human Resources  
Office of Maternal, Child, and Family Health  
Right From The Start Project  
Department of Health  
350 Capitol Street, Room 427  
Charleston, WV 25301  
Ph: 304.558.5388  
SELECTED REFERENCES

Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children
www.hrsa.gov/heritabledisorderscommittee/

American Academy of Pediatrics, Newborn Screening Overview
www.medicalhomeinfo.org/screening/newborn.html

Centers for Disease Control and Prevention, Early Hearing Detection and Intervention Program
www.cdc.gov/ncbddd/ehdi

March of Dimes
www.marchofdimes.com

National Newborn Screening and Genetics Resource Center
genes-r-us.uthscsa.edu