



## CHAPTER 4 : NEWBORN SCREENING

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Almost every child born in the United States undergoes state-mandated newborn screening. For each state, a small blood sample (“heel stick”) is collected from each newborn within 48 hours of birth and sent to a laboratory for testing for a panel of medical conditions. Newborn screening programs screen for an ever-increasing number of conditions, including phenylketonuria (PKU), sickle cell disease, and hypothyroidism. Every year, about 3,000 newborns test positive for one of these conditions. In the event that a newborn screens positive, 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 New England.

In the U.S., about 3,000 newborns test positive each year for a medical condition detected via newborn screening. Currently, the conditions screened in each state vary; efforts are underway to develop a national newborn screening program. This chapter provides an overview of the newborn screening programs and procedures in the six New England states.

#### 4.1 THE IMPORTANCE OF NEWBORN SCREENING TESTS

By law, all newborns are tested for several rare but serious medical conditions. Babies with these conditions may look healthy at birth. If not treated, these conditions can cause health problems such as mental retardation, slow growth, and even death. With treatment, these problems may be prevented.

#### 4.2 TESTING PROCEDURE AND FOLLOW-UP

A nurse or other medical professional will take a few drops of blood from the baby's heel. This blood sample is sent to a newborn screening laboratory. The blood should be drawn after the baby is 24 hours old, but before the baby leaves the hospital.

The baby's doctor will contact the parent(s) if the results are positive for one of the screened conditions. Follow-up testing may be required.

#### 4.3 RETESTING

Sometimes, a baby needs to be tested 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
- There was a problem with the way the blood sample was taken
- The first test showed 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.



## 4.4 TESTS PERFORMED

The tests that are done vary from state to state. In general, the conditions that are tested for fall into one of the following groups:

- Metabolic conditions, which affect how the body processes food
- Endocrine conditions, which affect the levels of important hormones
- Hemoglobin conditions, which affect the blood and cause anemia, infections, and other health problems
- A pulmonary condition, which affects growth and the lungs

For information on the diseases tested for in a particular state, contact that state's newborn screening program. Testing for more conditions may be available at other laboratories for a fee.

## 4.5 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.

## 4.6 NEWBORN SCREENING PROGRAMS

### Connecticut

State of Connecticut,  
Department of Public Health  
410 Capitol Avenue, MS #11 MAT  
P.O. Box 340308  
Hartford, CT 06134-0308  
(860) 509-8081  
[www.dph.state.ct.us/bch/nbs/nbs.htm](http://www.dph.state.ct.us/bch/nbs/nbs.htm)

### Maine

Maine Newborn Screening Program  
11 Statehouse Station  
286 Water Street  
Augusta, ME 04333  
(207) 287-5357  
[www.maine.gov/dhhs/boh/cshn/cshn](http://www.maine.gov/dhhs/boh/cshn/cshn)

### Massachusetts

New England Newborn Screening Program  
University of Massachusetts Medical School  
305 South Street  
Jamaica Plain, MA 02130-3515  
(617) 983-6300  
[www.umassmed.edu/nbs](http://www.umassmed.edu/nbs)

### New Hampshire

Maternal & Child Health Section  
29 Hazen Drive  
Concord, NH 03301  
(603) 271-4225  
[www.dhhs.state.nh.us/dhhs/mch.htm](http://www.dhhs.state.nh.us/dhhs/mch.htm)

### Rhode Island

Rhode Island Department of Health  
3 Capitol Hill, Room 302  
Providence, RI 02908-5097  
(800) 942-7434  
[www.health.ri.gov/genetics/newborn.php](http://www.health.ri.gov/genetics/newborn.php)

### Vermont

Division of Health Improvement,  
Children with Special Health Needs  
108 Cherry Street, P.O. Box 70  
Burlington, VT 05402  
(802) 951-5180

## 4.7 NEWBORN HEARING SCREENING

Hearing loss is a common condition that is present in as many as 1 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.

Each of the six New England states has a program to provide hearing screening to all newborns. Five states (all except Vermont) have a law mandating this screening. Babies are usually screened in the first few days of life, before they are discharged from the hospital. The testing, 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. Babies who do not pass the first screening are retested and may be referred to an audiologist (hearing specialist).



## 4.8 NEWBORN HEARING SCREENING PROGRAMS

### Connecticut

State of Connecticut,  
Department of Public Health  
410 Capitol Avenue, MS #11 MAT  
P.O. Box 340308  
Hartford, CT 06134-0308  
(860) 509-8081  
[www.dph.state.ct.us/bch/ehdi/b\\_unhs.htm](http://www.dph.state.ct.us/bch/ehdi/b_unhs.htm)

### Maine

Maine Newborn Hearing Program  
11 Statehouse Station  
286 Water Street  
Augusta, ME 04333  
(207) 287-5357  
[www.maine.gov/dhhs/bob/cshn/cshn](http://www.maine.gov/dhhs/bob/cshn/cshn)

### Massachusetts

Massachusetts Universal Newborn Hearing  
Screening Program  
Bureau of Family and Community Health  
250 Washington Street  
Boston, MA 02108  
(800) 882-1435  
[www.mass.gov/dph/fch/unhspl/index.htm](http://www.mass.gov/dph/fch/unhspl/index.htm)

### New Hampshire

Early Hearing Detection and Intervention  
Program (EHDI)  
Maternal & Child Health Section  
29 Hazen Drive  
Concord, NH 03301  
(603) 271-1037  
[www.dhhs.state.nh.us/dhhs/mch.htm](http://www.dhhs.state.nh.us/dhhs/mch.htm)



**Rhode Island**

Rhode Island Hearing Assessment Program  
Women and Infants Hospital  
Dudley Street  
Providence, RI 02905  
(401) 274-1122 x1844  
[www.health.state.ri.us/family/hearing/universal.php](http://www.health.state.ri.us/family/hearing/universal.php)

**Vermont**

Vermont Universal Newborn Hearing  
Screening Program  
108 Cherry Street, P.O. Box 70  
Burlington, VT 05402  
(800) 660-4427 x1330  
[www.healthvermont.gov/family/hearing/newborn.aspx](http://www.healthvermont.gov/family/hearing/newborn.aspx)

**SELECTED REFERENCES****Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children**

[www.mchb.hrsa.gov/programs/genetics/committee/](http://www.mchb.hrsa.gov/programs/genetics/committee/)

**American Academy of Pediatrics: Metabolic/Genetic Screening**

[www.medicalhomeinfo.org/screening/newborn.html](http://www.medicalhomeinfo.org/screening/newborn.html)

**Center for Disease Control's Early Hearing Detection and Intervention Program**

[www.cdc.gov/ncbddd/ehdi/](http://www.cdc.gov/ncbddd/ehdi/)

**National Newborn Screening and Genetics Resource Center**

<http://genes-r-us.uthscsa.edu/>



