



NEWBORN SCREENING in Massachusetts: Answers for you and your baby

The New England Newborn Screening Program
University of Massachusetts Medical School
305 South St.
Jamaica Plain, MA 02130
617-983-6300

A Program of the
Massachusetts Department of Public Health

Available in other languages:
<http://www.umassmed.edu/nbs/>

Dear Parent,

All Massachusetts babies are provided with Newborn Screening services. This public health program helps to prevent bad outcomes of disorders that some babies may have.

Newborn screening was begun in 1962 in Massachusetts. At that time, screening looked to find just one disorder. Since then, newborn screening looks for many more disorders, and newborn screening has become a routine standard of care worldwide.

As a leader in newborn screening, Massachusetts continues to study new screening possibilities by offering you some optional screening services. After your baby is born, you will be asked whether you want the optional services, and you will get a copy of the form that is sent to us, which is a record of your answer (please see example below).

This booklet provides general information about Newborn Screening services in Massachusetts. The colored insert has a list of disorders that Massachusetts includes in newborn screening. The colored insert also has a description of current studies to help you make decisions about optional screening.

We wish you and your baby the very best.

Sincerely,

The Staff of The New England Newborn Screening Program

EXAMPLE FORM PARENT'S COPY		Pilot Studies		
	LAB ID # 100001	<input type="checkbox"/> Declines MET	<input type="checkbox"/> Declines SCID	<input type="checkbox"/> Declines OTHER
	BABY'S NAME (Last) (First)			
Dear Parent	This sheet is your record to show that a small blood specimen was taken from your baby for routine newborn screening. This routine service ensures that your baby will be screened for treatable disorders as mandated by the Massachusetts Department of Public Health.			
	In addition, this sheet shows the instructions that were sent to the New England Newborn Screening Program after you decided whether you wanted the optional newborn screening services (pilot studies).			
	<ul style="list-style-type: none">If one or more Pilot Studies is marked with an X, at top right, then your baby will NOT be tested for any diseases included in the X'd study. If no Pilot Studies are X'd, then your baby will be tested for all diseases listed in the booklet's colored INSERT.			
New England Newborn Screening Program, University of Massachusetts Medical School 305 South St., Jamaica Plain, MA 02130 (617) 983-6300				

SUMMARY

You may find that this summary is enough for you to make your decisions about newborn screening for your baby. More information may be found inside and on the colored insert.

Newborn Screening helps to prevent certain treatable diseases.

- Babies with some disorders need treatment in early infancy to prevent severe disease.
- Newborn screening helps to find the babies who have these disorders.
- Most likely, your baby does NOT have one of these disorders.

Newborn screening works by testing *all* babies who are born in Massachusetts.

- Testing all babies is important, because most babies appear healthy at birth, even the babies who have the disorders that need treatment and that can be detected by newborn screening.
- The testing for newborn screening is done on a few small drops of blood that are collected when your baby is about two days old.
- If testing shows that your baby has signs of one of these treatable diseases, your baby's doctor will call you to arrange care for your baby.

ROUTINE NEWBORN SCREENING

- In Massachusetts, ROUTINE NEWBORN SCREENING includes disorders that are believed to be treatable.
- Under Massachusetts' law, it is a requirement that *all* babies born in Massachusetts be screened for laboratory markers of these treatable diseases unless parents object on the basis of religious beliefs.

OPTIONAL NEWBORN SCREENING (Pilot Studies)

- Massachusetts also offers optional newborn screening services.
- The OPTIONAL NEWBORN SCREENING allows Massachusetts to study new screening possibilities.
- The statewide pilot study is research that is valuable for future babies and may be of benefit to your baby.
- ***There is no extra cost and no extra blood required for your baby to participate.***
- Under Massachusetts' guidelines, after your baby is born, you will be asked whether you want to take advantage of the OPTIONAL NEWBORN SCREENING.
- If, for some reason, you decide that you do not want to participate in the OPTIONAL program, your baby will still have all the benefits of ROUTINE NEWBORN SCREENING.

MORE INFORMATION ABOUT ROUTINE and OPTIONAL NEWBORN SCREENING IS INSIDE

TABLE OF CONTENTS

Routine Screening

- *What is the purpose of the Newborn Screening Program? 4*
- *What is the chance my baby has a disorder that can be detected by routine screening? 4*
- *How are the tests done? 4*
- *Who decides which disorders are included in Newborn Screening? 4*
- *May I refuse the newborn screening tests for my baby? 5*
- *How can I get information about the results of my baby’s newborn screening tests? 5*
- *My baby’s doctor says that I need to bring my baby in because of newborn screening. Does that mean that my baby has a disorder? 5*
- *What is known about the disorders that are included in Routine Screening? 6*
- *Newborn Screening Quality Assurance and Improvements.....7*

Optional Screening Services

- *Research studies of new tests (Pilot studies)..... 8*
- *What pilot studies are being conducted now? 8*
- *Can any newborn participate in the pilot studies?..... 8*
- *May I refuse to participate in one or more pilot studies?.....9*
- *How do I enroll? Or, how do I refuse to participate?..... 9*
- *What are the general benefits and risks associated with pilot studies? 10*
- *Where can I find out more about the pilot studies? 10*

Additional Information for parents About disorders included in ROUTINE and OPTIONAL services

- **PLEASE SEE COLORED INSERT**

ROUTINE SCREENING

What is the purpose of the Newborn Screening Program?

The purpose of the Newborn Screening Program is to test all newborns in Massachusetts for early signs of a number of treatable disorders (as mandated by Massachusetts Department of Public Health Regulations 105 CMR 270.000).

What is the chance my baby has a disorder detectable by screening?

The chance that your baby will have one of these disorders is very small. In the rare cases when a disorder is found, early diagnosis and treatment can usually prevent the problems associated with these disorders.

Newborn screening tests provide an early opportunity to detect certain disorders — before symptoms appear. However, we know that even the best screening cannot always detect a disorder. If your baby does not seem well, talk to your baby's doctor as soon as possible.

How are the tests done?

At about 48 hours after birth, or just before your baby is discharged from the hospital, a small blood sample will be taken. A few drops of blood are taken from your baby's heel.

Your baby's birth hospital then sends the blood samples to the New England Newborn Screening Program. Special tests for small blood samples are done and reported by the Newborn Screening Program.

Important! Babies born outside of hospitals should also be tested, preferably at about 48 hours after birth. Parents should arrange with a doctor, hospital, or midwife to have the screening done.

Who decides which disorders are included in Newborn Screening?

The Commissioner of Public Health is responsible for deciding the list of disorders. An Advisory Board, made up of doctors, nurses, scientists, ethicists, and parents, advises the Commissioner which disorders to include. For a disorder to be included in the list, the following must be true: 1) the disorder is treatable, 2) there is a good test, and 3) early medical intervention would benefit the infant.

May I refuse the newborn screening tests for my baby?

In Massachusetts, you may refuse newborn screening for religious reasons. If you do so, you may be asked to sign a refusal form. This form relieves your doctor of liability for damages that result from a disorder that could have been detected by screening.

How can I get information about the results of my baby's newborn screening tests?

Your baby's newborn screening results will be reported to the hospital where your baby was born and to the pediatrician who is listed on your baby's sample. These reports include results of all routine testing and results of any optional screening your baby had.

In addition, if your baby's test results indicate that further attention is needed (see below), we will notify the hospital where your baby was born or your baby's doctor.

My baby's doctor says that I need to bring my baby in because of newborn screening. Does that mean that my baby has a disorder?

Not always. There are several reasons why your baby's doctor may have asked you to bring your baby in. Some reasons include:

Unsatisfactory specimen: There is not enough blood on the sample sent to us to complete all the required screening tests, or the sample does not work for other reasons. Another specimen is needed.

“Too Early” specimen: If the blood specimen was collected before your baby was 24 hours old, a second sample should be taken as soon as possible. The best time for detection is at about 48 hours after birth.

Out-of-Range Test Result: An out-of-range test result means that further evaluation is needed to know if your baby has a disorder. Sometimes this means that another specimen is needed, sometimes this means that the baby will have to be seen and tested by a specialist within a few days, and sometimes it means that the baby should be seen by a specialist as soon as possible. Your baby's doctor will let you know what is recommended.

Note: Premature or low birth weight newborns are more likely to have out-of-range test results on the first specimen even if a disorder is *not* present.

What disorders are included in ROUTINE SCREENING?

A detailed list of disorders included in screening is provided on the colored insert.

What is known about the disorders that are included in ROUTINE SCREENING?

We know that the disorders included in Routine Screening are thought to be treatable.

For some of these disorders, there is a good deal of information about the outcomes of babies who have the disorders. This may be because there are many babies with the disorder, or may be because the screening has been in place for many years, or both.

For other disorders, there is enough information to know that babies with the disorder will do better if they are found and treated early, but we do not yet have full information to know what to predict for their future. This may be because there are very few babies with the disorder, or because the screening is new, or because there is a new treatment.

In order to ensure that we can provide the best information for care and for decision-making, the newborn screening program collects information about how the patients with these disorders are doing.

The kinds of information collected depend on the disorder, and includes information about whether the patients are alive and well, and whether they see a specialist regularly. Other information collected helps the newborn screening program to provide knowledge about the disorder to babies' health care providers and families.

NEWBORN SCREENING QUALITY ASSURANCE AND IMPROVEMENTS

Newborn screening programs need to know that they are working well and need to know how to improve. This means that programs need to know whether the screening results match diagnostic results. This also means that programs need to know how babies who are diagnosed with newborn screening disorders are doing and whether they continue to get the care they need. Information on diagnoses and outcomes is collected for program-wide improvements.

Your baby's information and leftover blood may be stored for at least 10 years. Sometimes, the information or leftover blood will be used to make sure that newborn screening tests are working well. Sometimes the information or leftover blood will be used to make better tests for the newborn screening program. Other times, the information or leftover blood will be used for health studies. For any health studies, your written permission is needed before we release your baby's name to an external researcher.

In addition, if any information or leftover blood is going to be used for a study, the study has to be approved by two groups of people who make sure that your baby's rights are protected. These groups of people are called "Human Subjects Review Committees". One Human Subjects Review Committee is at the Department of Public Health, and the other is at the University of Massachusetts Medical School. The Federal Government sets the rules and regulates each Committee. For any proposed study, Human Subjects Review Committees decide whether your permission is needed. If either Committee decides that your permission is needed, the New England Newborn Screening Program will contact you before proceeding with the study.

Contacting you: We know that for many reasons, parents change health care providers and may change the name of their baby. If your baby has been diagnosed with a newborn screening disorder, or is being followed to find out if your baby has a newborn screening disorder, you may receive a letter from the New England Newborn Screening Program to ensure that your baby's information is up-to-date.

OPTIONAL SCREENING SERVICES

Research studies of new tests (Pilot studies)

The Massachusetts Department of Public Health may authorize and direct research studies of new tests in the Newborn Screening Program. Research studies of new tests, called pilot studies, are done when the Department of Public Health expects they could benefit both individuals and the public health. *No additional blood will be taken from your baby*, but these tests will screen for a number of disorders in addition to the routine testing already described.

Results of pilot studies are reported with routine screening results. As with routine newborn screening, if there was an out-of-range result, your baby's doctor would work with the right specialists to manage any special care that your baby might need.

What pilot studies are being done now?

Please see the colored insert.

Why is newborn screening for some disorders being offered as pilot studies rather than being mandated?

The Massachusetts Department of Health has determined that there is not yet enough evidence to require (mandate) routine newborn screening for the disorders included in the pilot studies. They need more information on one or more of the following questions:

1. What is the extent of benefit from newborn screening for these disorders? (Does it save lives? Does it prevent serious life-compromising outcomes? Do the treatments work as expected?)
2. How often do these disorders occur in Massachusetts?
3. How good are the laboratory tests used to screen for these disorders?

Can any newborn participate in the pilot studies?

Yes, any newborn who would be included in routine newborn screening on or after February 1, 2009 can participate.

What are the general benefits and risks associated with the pilot studies?

Possible Benefits

- The most important individual benefit for your baby is the following:
If your baby does have one of the disorders included in the study, your baby will have the earliest opportunity for detection of the disorder.
- Other benefits may include your own satisfaction that you are helping to answer important questions that may help other babies.

Possible Risks

- The most important individual risk for your baby is rare:
If your baby does have one of the disorders included in the study and the study testing does not detect your baby's disorder, there could be a delayed diagnosis. This is always a risk of any screening. Not detecting a disorder should be an unusual event, whether the testing is tried and true or still being studied. If your baby does not seem well, or does not seem right, talk with your baby's health care provider.
- Other risks include the possibility that the screening will show information that your baby has a disorder or a condition that we were not looking for, but that showed up as a by-product of the screen. Some believe this to be a benefit and for others, it is a risk. Finding by-products can happen with most any testing. Such results are reported to your baby's health care provider.
- The most common risk is that an out-of-range screening result may require additional testing and may cause you to worry, even if it turns out that your baby does not have a newborn screening disorder.

Where can I find out more about the Pilot studies?

See the colored insert and the section entitled "More about the current pilot studies."

I have some suggestions or I have some comments. How can I be sure that my comments will be considered?

You should address your written comments to any of the following committees or programs, and the Department's Newborn Screening Advisory Committee or a representative will review them:

Chairperson
Newborn Screening Advisory Committee
Massachusetts Department of Public Health
250 Washington St.
Boston, MA 02108-4619

Commissioner of Public Health
Massachusetts Department of Public Health
250 Washington St.
Boston, MA 02108-4619

Director
New England Newborn Screening Program
University of Massachusetts Medical School
305 South St.
Jamaica Plain, MA 02130

**Additional Information for parents
about disorders included in routine and optional services
effective in February 2009**

List of disorders included in ROUTINE SCREENING mandated by MA DPH:
Your baby will be screened for laboratory markers of the following 30 disorders:

- (1) Argininemia (ARG)
- (2) Argininosuccinic acidemia (ASA)
- (3) β -Ketothiolase deficiency (BKT)
- (4) Biotinidase deficiency (BIOT)
- (5) Carbamoylphosphate synthetase deficiency (CPS)
- (6) Carnitine uptake defect (CUD)
- (7) Citrullinemia (CIT)
- (8) Congenital adrenal hyperplasia (CAH)
- (9) Congenital hypothyroidism (CH)
- (10) Congenital toxoplasmosis (TOXO)
- (11) Cystic fibrosis (CF)
- (12) Galactosemia (GALT)
- (13) Glutaric acidemia type I (GAI)
- (14) Homocystinuria (HCY)
- (15) 3-hydroxy-3-methyl glutaric aciduria (HMG)
- (16) Isovaleric acidemia (IVA)
- (17) Long-chain L-3-OH acyl-CoA dehydrogenase deficiency (LCHAD)
- (18) Maple syrup disease (MSUD)
- (19) Ornithine transcarbamylase deficiency (OTC)
- (20) Phenylketonuria (PKU)
- (21) Sickle cell anemia (Hb SS)
- (22) Hb S/C disease (Hb SC)
- (23) Hb S/ β -thalassemia (Hb S/ β Th)
- (24) Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
- (25) Methylmalonic acidemia: mutase deficiency (MUT)
- (26) Methylmalonic acidemia: cobalamin A, B (Cbl A,B)
- (27) Methylmalonic acidemia: cobalamin C,D (Cbl C,D)
- (28) Propionic acidemia (PROP)
- (29) Tyrosinemia type I (TYR I)
- (30) Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Screening for these 30 disorders may show information about the following disorders and conditions (by-products of mandatory screening):

- a) Atypical cystic fibrosis (includes CBAVD)
- b) 2-Methyl 3-hydroxy butyric aciduria (2M3HBA)
- c) 2-Methylbutyryl-CoA dehydrogenase deficiency (2MBG)
- d) 3-Methylcrotonyl-CoA carboxylase Deficiency (3MCC)
- e) 3-Methylglutaconic aciduria (3MGA)
- f) Benign hyperphenylalaninemia (H-PHE)
- g) Carnitine palmitoyltransferase IA deficiency (liver) (CPT IA)
- h) Citrullinemia type II (CIT II)
- i) Defects of biopterin cofactor biosynthesis (BIOPT BS)
- j) Defects of biopterin cofactor regeneration (BIOPT Reg)
- k) Galactokinase deficiency (GALK)
- l) Galactose epimerase deficiency (GALE)
- m) Glutaric acidemia type II (GA2)
- n) Hypermethioninemia (MET)
- o) Isobutyryl-CoA dehydrogenase deficiency (IBG)
- p) Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)
- q) Multiple carboxylase deficiency (MCD)
- r) Short-chain acyl-CoA dehydrogenase deficiency (SCAD)
- s) Trifunctional protein deficiency (TFP)
- t) Tyrosinemia type II (TYR II)
- u) Tyrosinemia type III (TYR III)
- v) Variant Hb-pathies (Var Hb)
- w) Carrier status of any of the conditions listed in 1-30 or a-w.

ROUTINE SCREENING

Disorders included in routine newborn screening can be grouped according to the cause or treatment of the disorder.

- **AMINO ACIDOPATHIES:** Babies and patients with these disorders cannot use regular food because their bodies cannot break down one of the amino acids found in regular food. They are given special food. A metabolic specialist or a biochemical geneticist usually treats these babies.
- **CONGENITAL INFECTIOUS DISEASES:** Babies with these disorders are infected with a kind of bacteria, virus or parasite. The infection of the baby can occur during pregnancy or at birth. An expert in infectious disease usually treats these babies.
- **CYSTIC FIBROSIS (CF):** Babies and patients with this disorder develop thick sticky mucus in their lungs and other organs. The mucus clogs the lungs, making a home for lung infections. Mucus in the digestive system causes problems with absorbing food. A pulmonologist or a CF specialist at a CF Center usually treats these babies
- **ENDOCRINOPATHIES:** Babies and patients with these disorders cannot make one of the body's hormones. If a baby's body cannot make a hormone, the baby needs help and is usually given medicine containing the hormone. These babies are usually treated by an endocrinologist or by a pediatrician who is working with an endocrinologist.
- **ENZYME DEFICIENCIES FOR VITAMINS AND SUGARS:** Babies and patients with these disorders cannot process some sugars, vitamins or other nutrients. A metabolic specialist or a biochemical geneticist usually treats these babies.
- **FATTY ACID OXIDATION DISORDERS:** Babies and patients with these disorders cannot use the fats that they have stored in their body for emergency energy. When a person with such a disorder does not eat for a while, there is a risk that important functions of their body will stop working. A metabolic specialist or a biochemical geneticist usually treats these babies.
- **HEMOGLOBINOPATHIES:** Babies and patients with these disorders have a change in their red blood cells that causes problems such as sickle cell disease. It means the baby is more likely to have anemia, episodes of pain, strokes, and life-threatening infections. Treatment with penicillin may prevent serious infections in early childhood. These patients are usually treated by a hematologist.
- **ORGANIC ACID DISORDERS:** Babies and patients with these disorders cannot use the part of regular food called branched chain amino acids or lysine. The patient needs help and is usually given special food and other treatment. These patients are usually treated by a metabolic specialist or a biochemical geneticist.
- **UREA CYCLE DISORDERS:** Babies and patients with these disorders are unable to remove nitrogen from their bloodstreams. These patients have high levels of toxic ammonia in their blood and need immediate help. These patients are usually treated by a metabolic specialist or a biochemical geneticist.

List of disorders included in OPTIONAL SCREENING offered by MA DPH

(pilot studies)
(more information on back)

You will be asked whether you want your baby to be screened in the MET and SCID pilot studies:

“MET” pilot study

If you say yes, your baby will be screened for laboratory markers of the following 5 disorders.

- (1) Carnitine palmitoyltransferase II deficiency (CPT II)
- (2) Dienoyl-CoA reductase deficiency (DE RED)
- (3) Hyperornithinemia, Hyperammoninemia, Homocitrullinemia Syndrome (HHH)
- (4) Malonic acidemia (MAL)
- (5) Medium/short-chain L-3-OH acyl-CoA dehydrogenase deficiency (M/SCHAD)

Screening for these 5 disorders may show information about the following disorders and conditions (byproducts of optional MET screening)

- a) Carnitine: acylcarnitine translocase deficiency (CACT)
- b) Carrier status of any of the 5 MET pilot study disorders or CACT.

“SCID” pilot study

If you say yes, your baby will be screened for laboratory markers of the disorder called:

- (1) Severe Combined Immunodeficiency (SCID).

Screening for SCID may show information about the following disorders and conditions (by products of optional SCID screening).

- (a) Non-SCID primary immunodeficiencies or other conditions associated with low T cells.
- (b) Carrier status of SCID.

“OTHER” pilot study

This space is reserved as a place to hold future studies that might be offered. Right now, the only pilot studies are the MET and SCID studies.

MET PILOT STUDY

Background:

- 1) Carnitine Palmitoyl Carnitine palmitoyltransferase II deficiency (CPT II),
- 2) Dienoyl-CoA reductase deficiency (DE RED)
- 3) Medium/short-chain L-3-OH acyl-CoA dehydrogenase deficiency (M/SCHAD)

These three disorders belong to a group of metabolic conditions known as the "FATTY ACID OXIDATION DISORDERS". Patients with this condition cannot change certain fats in the food we eat into energy and depend completely on glucose. Babies and children with these disorders may become very ill when the glucose is not available (as in fasting) or when higher amounts of energy are required (as during infections). It is believed that early treatment may be able to prevent death and disability in some cases. These patients are usually treated by a metabolic specialist or a biochemical geneticist

4) Hyperornithinemia, Hyperammoninemia, Homocitrullinemia Syndrome (HHH): Patients with HHH are unable to remove nitrogen from their bloodstreams. As a result ammonia in the blood may rise to toxic levels. Patients may become very sick without immediate treatment. These patients are usually treated by a metabolic specialist or a biochemical geneticist

5) Malonic Acidemia (MAL): Patients with MAL are not able to produce fatty acids as needed or utilize the fats present in food properly. This may cause a low blood sugar, enlarged heart, poor muscle tone, vomiting, diarrhea, dehydration or seizures. These patients are usually treated by a metabolic specialist or a biochemical geneticist

Purpose

The purpose of the MET PILOT study is to understand whether early identification by newborn screening is clinically beneficial.

SCID PILOT STUDY

Background

SCID, or Severe Combined Immunodeficiency, is a disorder that severely affects the immune system. Unless treated, babies with this disorder will die at a few months of age because they cannot fight off the usual infections that all babies get. With treatment, most babies live.

Treatment for a SCID baby includes a bone-marrow transplant. This allows the baby to live because it can make T cells that untreated SCID babies cannot make.

Purpose

The purpose of the SCID pilot is to determine the best way to find SCID babies.

We believe that a molecular test will help us to know which babies are making T cells and which babies are not. We may try other tests to see if they can help us to predict which babies have SCID and which do not. The molecular test means that we will be looking for a piece of DNA that is present in most babies. If we cannot find the piece of DNA in a baby, then we may ask for another sample to be sure of the result or we may recommend that the baby be seen by an expert in immunology to have some additional testing done. If the baby has SCID, then the immunologist will work with specialists in bone marrow transplantation for the best plan for the baby.

We expect that once we start testing to find SCID babies, we may find babies with other immune diseases. If we do find such babies, they would also be seen by an immunologist.

New England Newborn Screening Program

