



The State of Connecticut Newborn Screening Program Bloodspot Screening

Make Sure Your Baby is Healthy

Checklist:

- ☐ Ask your doctor, nurse or midwife about Newborn Screening (NBS)
- ☐ Pick a doctor for your baby before your baby is born
- ☐ Make an appointment with your baby's doctor before you leave the hospital
- ☐ Ask if your baby had the NBS test done before you leave the hospital
- ☐ Ask your baby's doctor for the results of your baby's NBS test
- ☐ Give a phone number where you can be reached after you leave the hospital
 - If you do not have a phone, give a friend's or family member's phone number
- ☐ Call the doctor if your baby:
 - has trouble eating
 - vomits often
 - has skin problems
 - is very sleepy all the time
 - looks sick

More Questions?

- Talk to your baby's doctor, nurse or midwife
- Call the Connecticut NBS Program at:
(860) 920-6628
- Go to:
 - ♦ <http://savebabies.org/>
 - ♦ <http://www.babysfirsttest.org/>
 - ♦ <http://www.marchofdimes.org/baby/newborn-screening-tests-for-your-baby.aspx>
 - ♦ <http://portal.ct.gov/newbornscreening>
- Email: dph.nbstracking@ct.gov



Newborn Screening (NBS) is important!

While most babies are born healthy, some babies are born with serious but treatable medical conditions. Your baby will receive three different screening tests while in the hospital:

1. the bloodspot screen checks for conditions that can cause problems with your baby's growth and development and can sometimes cause death if not treated. You may hear someone call this the "PKU" test, but this name is outdated. PKU is just one of over 60 conditions that babies are tested for by bloodspot screening in Connecticut
2. the hearing screen
3. the pulse oximetry screen that checks for serious heart problems

This pamphlet explains Newborn Screening bloodspot screening

Answers To Your Questions about Newborn Screening

Why does my baby need NBS?

- Bloodspot screening tests for conditions that may be hidden at birth. A baby can appear healthy and still have a condition.
- Connecticut NBS tests for over 60 different conditions using a few drops of blood.
- If one of these health problems is not treated, a baby may:
 - ♦ become very sick
 - ♦ grow poorly
 - ♦ have a physical disability
 - ♦ have brain damage
 - ♦ die
- With early treatment many of these problems can be prevented.

When is the test done?

- One to three days after birth while your baby is still in the hospital.

What if my baby is born at home?

- Your midwife will collect the bloodspot specimen and send it to the State Public Health Lab. Please ask you midwife about hearing and pulse oximetry screening.

How is the test done?

- The hospital staff or a midwife will take a few drops of blood from your baby's heel and apply it to a filter paper card. The card is sent to the State Public Health Lab for testing.

Will the heel-stick used to collect the blood spots hurt my baby?

- A small needle is used to poke your baby's heel. Some babies cry when their heel is pricked, but the discomfort does not last long. The benefits of newborn screening, such as saving your baby's life and preventing health problems, outweigh the temporary discomfort that comes with the heel-stick.

Can I say "no" to this test?

- All babies born in CT automatically receive bloodspot screening
- You can say "no" to the test if it conflicts with your religious beliefs. If you say no to the test, you will be asked to sign a form stating so.

Is there a cost for bloodspot screening?

- The cost for bloodspot screening is included with the hospital birthing and nursery charges. There is no charge for bloodspot screening for a baby born at home.

How do I get the screening results?

- If your baby screens positive (has an out-of-range result) for a disorder, someone from the Newborn Screening Program will call your baby's doctor to report the result.
- A final report of screening results will be sent to your baby's doctor usually 7-14 days after birth. Ask your baby's doctor for bloodspot testing results at the first visit.

More Answers to Your Questions

What does a screen positive result mean?

- It does not mean that your baby is sick or has a disorder. Further evaluation is needed.
- There are many things that can cause a screen positive or out-of-range result
 - ♦ A screen positive or out-of-range result can happen:
 - if you took certain medicines while pregnant
 - if your baby was born early
 - if your baby's blood was collected too soon
 - if your baby had certain treatments while in the hospital
 - for many other reasons
- If your baby has a screen positive or out-of-range result your doctor may:
 - ♦ examine your baby
 - ♦ ask about conditions that run in your family
 - ♦ repeat the blood spot screening
 - ♦ order additional tests
 - ♦ have your baby see a doctor who specializes in newborn screening related disorders

What happens to any leftover blood after screening?

- When bloodspot screening is complete, a very small amount of blood is sometimes left over. The leftover blood will be stored at the CT Public Health Laboratory to allow for any necessary follow-up testing, to help make sure screening is accurate, and to develop new newborn screening tests for Connecticut. Leftover bloodspots are not used for research unrelated to newborn screening.

What does Connecticut NBS test for?

Adrenoleukodystrophy (ALD): ALD is genetic condition where the body cannot break down certain fatty acids causing them to buildup in the cells. This causes damage to the nervous system (the nerves, spinal cord and brain). ALD can also cause a problem with hormone production in the adrenal gland. There are different types of ALD ranging from mild to severe. The most severe form affects mostly boys and can cause severe disability and death. Babies with ALD will be monitored by a doctor over a period of time and treatment started if needed.

Amino Acid (AA) Disorders: Amino Acid disorders are a group of disorders that can affect an infant from birth. The body cannot use proteins in some foods like formula, breast milk and meats. If not treated, AA disorders can cause developmental delay, organ damage, breathing problems, seizures and death. A special diet and medicine can help prevent these problems. The CT NBS program tests for over a dozen AA disorders.

Biotinidase (BIO) Deficiency: The body does not have enough of the enzyme necessary to make the vitamin biotin. This can cause skin rashes, weak muscles, hair loss, trouble seeing and hearing and brain damage. A vitamin can help prevent these problems.

Congenital Adrenal Hyperplasia (CAH): With CAH the adrenal glands do not make enough of the hormones cortisol and aldosterone. Untreated, this can cause severe illness or death. CAH is treatable with medication.

Congenital Hypothyroidism (CH): With CH the body does not make enough thyroid hormone. This can cause growth problems and brain damage. Medication can prevent these problems.

Cystic Fibrosis (CF): is a disease that affects the lungs and digestive system. The body produces thick and sticky mucus that can clog the lungs and pancreas. CF can be life-threatening and people with the condition tend to have a shorter-than-normal life span. In CT, CF bloodspot screening is done through the Yale and UConn laboratories.

Fatty Acid Oxidation (FAO) Disorders: Fatty Acid Oxidation Disorders are a group of rare disorders where the body has trouble using fat for energy. This can cause sleepiness, weak muscles, vomiting, low blood sugar, liver problems and death. A special diet and medication can help prevent these problems. The CT NBS program tests for over a dozen FAO disorders.

Galactosemia (GALT): In classical GALT the body cannot use a sugar found in milk, infant formula, breast milk, and other foods. This can cause eye and liver problems, brain damage and death. A special diet can help prevent these problems.

Hemoglobin (Hb) Disease: There are many types of Hb disease where the body produces abnormal blood cells. This can cause anemia, infection, pain, poor growth and death. Medicine and special medical care can help prevent these problems. The CT NBS program tests for many types of Hb problems. Sickle cell anemia is one type of Hb disease.

Hemoglobin (Hb) Traits: Screening for Hb trait will show if you baby is a carrier of a red blood cell disease. This does not mean that your baby is sick. Your doctor will talk to you about what this means and may order additional blood tests.

Mucopolysaccharidosis I (MPS-I): With MPS-I the body doesn't make enough of an enzyme causing a build-up of certain sugars in the cells. There are different forms of MPS-I. The more severe form starts in infancy and can cause developmental delays, vision problems, damage to the bones, joints, heart and other body systems. Enzyme replacement and bone marrow transplant can help prevent some of these problems.

Pompe Disease: With Pompe the body lacks the enzyme needed to breakdown certain sugars causing a build-up in the cells. The most severe form appears in infancy and causes heart problems, breathing problems, muscle weakness and death. Babies with the severe infantile form of Pompe often appear well at birth but can deteriorate quickly. Enzyme replacement can slow the progression of the disease and improve quality of life.

Organic Acid (OA) Disorders: This is a group of disorders where the body cannot use certain proteins and fats in foods. This can cause vomiting, poor feeding, low blood sugar, sleepiness, seizures and death. A special diet and medicine can help prevent these problems. The CT NBS program tests for over a dozen OA disorders.

Severe Combined Immunodeficiency Disorder (SCID): SCID is a rare genetic disorder that causes life-threatening problems with the immune system (the body cannot fight infection). This can cause serious illness and death. Bone marrow transplant is a treatment for SCID. The CT NBS program tests for several different types of SCID.

Spinal Muscular Atrophy (SMA): SMA is a genetic condition that causes muscle weakness and muscle loss. The most severe form can affect the child's ability to crawl, walk, sit up and control head movements and can lead to breathing problems and death. Medication and gene therapy are treatments that can improve breathing, muscle function and survival.

SEE REVERSE FOR MORE INFORMATION

The Connecticut Newborn Screening Program | Phone: 860.920.6628 | Email: dph.nbstracking@ct.gov