Newborn Screening

For Your Baby's Health

NEW YORK STATE | Department of Health

Why is my baby tested?

To help make sure your baby will be as healthy as possible. The blood test provides important information about your baby's health that you and your doctor might not otherwise know. The Newborn Screening Program identifies infants who may have one of several rare, but treatable diseases that don't show symptoms right away. With early diagnosis and medical treatment, serious illness, and even death, can often be prevented, so it is very important for us to test your baby's sample and report the results to your baby's doctor. Ask your baby's doctor for your baby's results.

Is newborn screening new?

No. Every state has a newborn screening program. The New York State program is mandated by Public Health Law, and it began in 1965. Some diseases can affect a child very early in life – even within the first few days. Timely testing and diagnosis are important for treatments to work the best.

For how many diseases is my baby tested?

The number has increased from one in 1965 to more than 45 today. They are listed in this booklet. Although these diseases are rare, 1 in 300 babies born in New York every day has one of these diseases. Most of the diseases are serious and can even be fatal. Some may slow down a baby's development, cause intellectual disabilities, increase a baby's risk for infection, or cause other problems if undetected and untreated.

That is why:

Early treatment is very important!

But my baby seems very healthy. Are these tests still needed?

Yes. Most infants with a disease identified by the Newborn Screening Program show no signs of the disease right after birth and look healthy. With these special laboratory tests, we can identify a baby who may have one of these diseases and tell the baby's doctor of the need for more tests and special care. Most of the time, it is very important to start treatment before your baby shows symptoms or becomes sick. Many of the diseases are genetic, and they are inherited from the baby's parents.

Every baby has two sets of genes – one from their mother and the other from their father. Sometimes only one set of the genes has a problem, but because the other set doesn't, the baby is not sick. These babies are called carriers. Although these babies are not sick, this means that at least one or sometimes even both of their parents are also carriers. Newborn screening tests can identify carriers for some genetic

diseases some of the time, but the program is not designed to find **all** carriers. *It is important to get genetic counseling if your baby has a carrier result because most parents who are carriers do not know. Counselors can help you understand this information.*

But children in our family have never had any of those health problems.

Parents who have already had healthy children do not expect any problems, and they are almost always right. But there is still a chance your new baby may have one of these diseases. Each of these diseases is very rare, and the chances are excellent that your child will not have one of them, but altogether 1 in 300 babies born in New York every day does have one of them. A negative newborn screen for your new baby does not guarantee that your future children will be negative too. Some babies and parents can be carriers for diseases even if no one in your family has a disease. Many families go for genetic counseling to better understand these risks to their future children and other family members. *It is also important to remember that newborn screening does NOT find all babies who are carriers for these genetic diseases.* The tests are designed to find most babies with these genetic diseases.

How is my baby tested?

All the tests are performed on a tiny sample of blood taken by pricking the baby's heel. The blood is put on a special filter paper. The sample is usually taken when the baby is one or two days old. The sample is sent for testing to the laboratory at the State Health Department in Albany.

Will I get the test results?

Be sure to tell the nurse at the hospital the name and office information for your baby's doctor or clinic. This doctor will be told of the results and will contact you immediately if anything is wrong. To be sure, ask about the result when you bring your baby to the doctor or clinic for his or her first check-up. The hospital nurse should give you a pink form, which will tell you how to get the test results from your baby's doctor.

If all the tests are screen-negative, does that mean my baby will be healthy?

The Newborn Screening Program only looks for a few of many diseases a baby could have. In addition, some babies with these diseases may not be identified for several reasons. You should bring your baby to the doctor or clinic for all their check-ups. Always watch your baby for unexpected symptoms or behavior, and call the doctor immediately if things don't seem right.

A negative newborn screen for your new baby does not guarantee that your future children will not have a disease. Also, newborn screening does NOT find all babies who are carriers for these genetic diseases. Carriers have one gene mutation but are healthy. Babies and their parents can be carriers without any family history of a disease. Many families go to genetic counseling to better understand disease and carrier risks to their future children and other family members.

screen is also abnormal. On very rare occasions, because a disease may cause a baby to become very sick quickly, the doctor will treat the baby immediately while waiting for the results of the second series of tests. If you are asked to have your baby retested, please bring in your baby as soon as possible, so the repeat test can be done immediately, to determine if your baby needs treatment.

1 in 300 babies born each day in NYS has one of the screened diseases!

Does a "repeat test" mean my baby may have a disease?

Not necessarily. Repeat testing may be needed for a number of reasons. The most common is that the blood was put on the special filter paper incorrectly. Usually this does not mean there is anything wrong with your baby. It simply means that another blood sample must be taken as soon as possible.

When the first test results suggest a problem, the results are not considered final until the screening tests are done again. This requires a new blood sample. In general, a doctor will discuss the need for further diagnostic testing only after a baby's second

What if my baby has one of these diseases?

The tested diseases all have treatments that can lessen the effects of the disease.

Sometimes the symptoms can be completely prevented if a special diet or other medical treatment is started early. Most of these diseases are very complicated to treat, and medical care should be coordinated by a doctor who specializes in the specific disease.

If my new baby has a disease, will my future children have it?

That depends on the disease. Most of these diseases are genetic and inherited by children from their parents. A negative newborn screen does not guarantee that future children will not have the disease. Also, newborn screening does NOT

find all babies who are carriers for these genetic diseases. Carriers have one gene mutation and are healthy. Babies and their parents can be carriers without any family history of the disease. Many families seek genetic counseling to better understand how their child got the disease, and to understand disease and carrier risks to their future children and other family members. Some diseases are not inherited. For example, congenital hypothyroidism has many causes, while HIV infection is caused by a virus, not a gene mutation.

Why is my baby tested for HIV?

We test the baby for HIV antibodies. If the test is positive, that means the mother has the virus and we want to be sure the baby is not infected with the virus. HIV can be transmitted by an infected mother to her baby before it is born, during delivery or from breastfeeding. In NYS, most pregnant women are tested for HIV before the baby is born. Ideally, the mother should get medicine during pregnancy and labor to protect the baby from the HIV infection.

How much will these tests cost me?

Nothing. These tests are done at no cost to families.

How can I make it easier for the doctor to help my baby?

First, be sure you tell the nurse at the hospital where your baby was born the name of your baby's doctor so we can contact the doctor if we need to. If you change your doctor, let us know by emailing or calling us (see back of this booklet). If your doctor asks you to bring your baby in for a repeat test, do so as soon as you can. If your baby ***does*** have a disease, quick action is very important.

If you do not have a telephone, give your doctor the phone number of someone who can contact you immediately. If you move soon after your baby is born, tell your doctor or clinic your new address and phone number right away. Then your doctor will know where to reach you if your child needs more tests or treatment.

Remember, time is very important. As a parent, you can help the Newborn Screening Program make sure that your baby is as healthy as possible by making sure your baby's doctor knows how to reach you.

Be Informed: Get your baby's newborn screening results from his or her doctor!

See our website for much more information on newborn screening.

Diseases Identified by the New York State Newborn Screening Program

Group		Diseases
Endocrinology		Congenital adrenal hyperplasia (CAH)
		Congenital hypothyroidism (CH)
Hematology, Hemoglobinopathies		Hb SS disease (Sickle cell anemia)
		Hb SC disease
		Hb CC disease
		Other hemoglobinopathies
Infectious Disease		HIV exposure
Inborn Errors of Metabolism	Amino Acid Diseases	Homocystinuria (HCY)
		Hypermethioninemia (HMET)
		Maple syrup urine disease (MSUD)
		Phenylketonuria (PKU) and Hyperphenylalaninemia (HyperPhe)
		Tyrosinemia (TYR-I, TYR-II, TYR-III)
	Fatty Acid Oxidation Diseases	Carnitine-acylcarnitine translocase deficiency (CAT)
		Carnitine palmitoyltransferase I (CPT-I) and II (CPT-II) deficiencies
		Carnitine uptake defect (CUD)
		2,4-Dienoyl-CoA reductase deficiency (2,4Di)
		Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
		Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
		Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)
		Medium/short-chain hydroxyacyl-CoA dehydrogenase deficiency (M/SCHAD)
		Mitochondrial trifunctional protein deficiency (TFP)
		Multiple acyl-CoA dehydrogenase deficiency (MADD) (also known as Glutaric acidemia type II (GA-II))
		Short-chain acyl-CoA dehydrogenase deficiency (SCAD)
		Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Group		Diseases
Inborn Errors of Metabolism	Organic Acid Diseases	Glutaric acidemia type I (GA-I)
		3-Hydroxy-3-methylglutaryl-CoA lyase deficiency (HMG)
		Isobutyryl-CoA dehydrogenase deficiency (IBCD)
		Isovaleric acidemia (IVA)
		Malonic acidemia (MA)
		2-Methylbutyryl-CoA dehydrogenase deficiency (2-MBCD)
		3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)
		3-Methylglutaconic acidemia (3-MGA)
		2-Methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency (MHBD)
		Methylmalonyl-CoA mutase deficiency (MUT), Cobalamin A,B (Cbl A,B) and Cobalamin C,D (Cbl C,D) cofactor deficiencies and other Methylmalonic acidemias (MMA)
		Mitochondrial acetoacetyl-CoA thiolase deficiency (Beta-ketothiolase deficiency) (BKT)
		Multiple carboxylase deficiency (MCD)
		Propionic acidemia (PA)
	Urea Cycle Diseases	Argininemia (ARG)
		Argininosuccinic acidemia (ASA)
		Citrullinemia (CIT)
Other Genetic Diseases		Adrenoleukodystrophy (X-linked)
		Biotinidase deficiency (BIOT)
		Cystic Fibrosis (CF)
		Galactosemia (GALT)
		Guanidinoacetate methyltransferase deficiency (GAMT)
		Krabbe Disease
		Mucopolysaccharidosis type I (MPS I)
		Pompe Disease
		Severe Combined Immunodeficiency Disease (SCID)
		Spinal muscular atrophy (SMA)

For more information on the New York State Newborn Screening Program and the diseases in the panel please visit our webpage at **www.wadsworth.org/programs/newborn**

The New York State Newborn Screening Program is a service provided by the State Health Department to families with newborn babies.

Important: Questions about newborn screening? Need to let us know information about your baby's doctor? Write, call or visit our website:

Newborn Screening Program
Wadsworth Center
New York State Department of Health
P.O. Box 22002
Albany NY 12201-2002

Email: nbsinfo@health.ny.gov

www.wadsworth.org/programs/newborn

Dear Parents,

Your child's specimen(s) will be stored by the Newborn Screening Program for up to 27 years under secure conditions where access is strictly controlled. Should the need arise, the specimen(s) may be used for diagnostic purposes for your child with your consent. A portion of the specimen will also be stripped of all information that might identify your child and may be used in public health research that has been reviewed and approved by a Board charged with overseeing compliance with all applicable laws and ethical guidelines. You may arrange to have your child's specimen(s) destroyed or prevented from being used in public health research by calling (518) 473-7552 for instructions. You may visit our website for more information or to download a copy of the form we need to honor your written request. Note: Upon request, we will completely destroy specimens. We cannot do this until 8 weeks after you give birth.