Important Information for Parents about the

Newborn Screening Test








Newborn Screening Branch
Genetic Disease Screening Program
http://cdph.ca.gov/nbs

California Department of Public Health

Publication Date: January 2013

For more information about Newborn Screening and the most current list of diseases that can be detected through the Program, visit our website at **http://cdph.ca.gov/nbs**

OSP 12 125074

The California Newborn Screening Test

Newborn screening can save your baby's life or prevent serious brain damage. Newborn screening can identify babies with certain diseases so that treatment can be started right away. Early identification and treatment can prevent mental retardation and/or life-threatening illness.

What Types Of Diseases Are Screened For In California?

To protect the health of all its newborns, State law requires that all babies born in California have the Newborn Screening (NBS) Test before leaving the hospital. This test screens for specific diseases in the following groups:

Metabolic
chemical reactions in the body to create energy and build tissue

Endocrine
hormones that affect body functions

Hemoglobin
red blood cells that carry oxygen

Other Genetic Diseases
Cystic Fibrosis (CF)
Severe Combined Immunodeficiency (SCID)

In California about 1 out of every 600 babies tested will have one of these diseases.

Make Sure Your Baby Is Tested

Babies with any one of these diseases can look very healthy at birth and still have a serious disease. By the time symptoms appear, it may be too late to prevent serious damage to the baby. That is why your baby will be tested before leaving the hospital.

Babies not born in the hospital must also have this test. It should be done by the time your baby is 6 days old. The ideal time to do the test is when the baby is between 24 and 48 hours of age. Call your midwife, the baby's doctor or your local health department to have your baby tested.

How Can These Diseases Be Treated?

Effective treatment is available for most of the diseases for which we screen. Treatment may include special diets or medicine. Babies who receive early and ongoing treatment can grow up to enjoy long, productive lives. For some of the diseases found, there is no effective treatment.

What Is Screening?

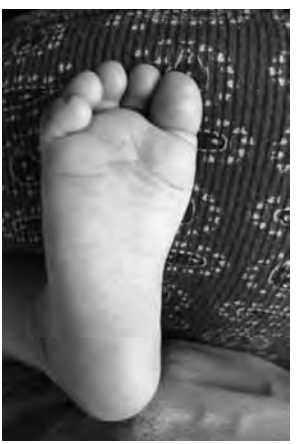
Screening is the testing of a group of people to identify those who are at risk for having a specific disease even though they may seem healthy. Newborn screening identifies most, but not all, of the babies who have one or more of the many diseases screened for by the California Program. Not every baby with a positive screening test will have one of these diseases. Further testing and evaluation by the baby's health care provider or a specialist are needed to make the diagnosis.

How Is The Test Done?

A few drops of blood taken from the baby's heel are put on special filter paper. The blood is then sent to a state-approved lab for testing.

When Should The Test Be Done?

The newborn screening test should be done when the baby is at least 12 hours of age but before 6 days of age. The ideal time to do the test is when the baby is between 24 and 48 hours of age. Blood collected before 12 hours of age is **not** always reliable for some metabolic diseases. Another blood sample must be taken later to repeat the test. If you leave the hospital or birthing center with your baby before he/she is 12 hours old, you will have to return within the next few days for a second test.



Is The Test Safe?

Yes, this is a simple and safe test. Since 1980, over 16 million California newborns have had blood safely collected by heelstick.

Is The Test Accurate?

Yes. The blood is sent to a state-approved lab for testing. The state checks the work of the testing labs closely to make sure the results are reliable. It is rare that a baby with one of the more common diseases is not found through a positive newborn screening test. For a few rare diseases, the test may find only a small number of the babies affected.

If the test is positive, you will be contacted within a few days after you leave the hospital. CF and SCID may take longer. If the test is negative, it takes about two weeks for doctors to get a copy of the results.

Can I Say No To The Test?

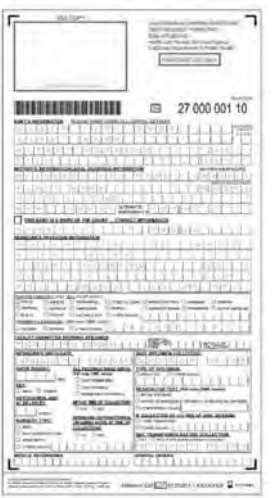
You can only say no if screening is in conflict with your religious beliefs or practices. You must then sign a special form. It states that not having the test done can result in serious illness or permanent damage to your child. It also states that you accept responsibility should this occur.

How Can I Get The Results?

You can get your baby's test results from your doctor or clinic. If your doctor does not have the results, he/she can contact the Newborn Screening Program to request a copy.

When your baby has the test done, the hospital staff will give you the pink and blue copies of the test request form. This has our Notice of Information and Privacy Practices as required by HIPAA and the information the hospital is sending about you, your baby, and your baby's doctor.

1) Review the information on the pink copy of your baby's newborn screening test request form. **Tell the nursery staff before leaving the hospital if any of the information about you, your baby, or your baby's doctor is not correct.** This will help us to contact you and your doctor right away if your baby needs more tests.



2) **Take the pink copy of this form when you go to your baby's first check up.** This will help the doctor locate your baby's newborn screening test results.

This way, you can help make sure that you and your baby's doctor gets the results from the newborn screening test.

If you move after the test is done, make sure the hospital and your baby's doctor or clinic have your new address and phone number in case they need to contact you about your baby's results.

What Do I Do If The Baby's Results Are Positive?

If the results are positive, more tests will be needed. You should receive a phone call and/or letter about what to do next. After further testing, many babies who have a positive first test are found **not** to have a disease. However, you must have your baby re-tested because babies who do have one of these diseases benefit from early treatment.

Every year in California there will be about 875 babies identified with one of these diseases each year.

Early Treatment Can Prevent Serious Problems

If these diseases are not found and treated soon after birth, the baby can have serious health problems or even die. Early treatment can prevent many of these problems.

For a complete list of the diseases screened for, visit our website at www.cdph.ca.gov/programs/nbs

Metabolic Diseases

Metabolic diseases affect the body's ability to use certain parts of food for growth, energy, and repair. The parts include **amino acids** from proteins, **fatty acids** from fats and **organic acids** from proteins, fats, and sugars. To break down or convert these substances, certain proteins called enzymes must be present. When there are not enough of the needed enzymes, some substances build up in large amounts and may be harmful to the body. Metabolic diseases have varying degrees of severity. If identified early, many of these conditions can be treated before they cause serious health problems. Treatment may include close monitoring of the baby's health, medicine, dietary supplements, and/or special diets.

These are some of the metabolic diseases screened for by the NBS Program:

- **Phenylketonuria (PKU)** – Babies with PKU have problems when they eat foods high in protein such as milk (including breast milk and formula), meat, eggs, and cheese. Without treatment, babies with PKU develop mental retardation and/or have other health problems. A special diet can prevent these problems.
- **Galactosemia** – Babies with this disease cannot use some of the sugars in milk, formula and breast milk, and other foods. Without treatment, babies with galactosemia can become very sick and die. A special infant formula and diet can help prevent these problems.
- **Maple Syrup Urine Disease (MSUD)** – Babies with MSUD have problems using fats and protein. Without treatment, MSUD can cause mental retardation or death. Treatment with a special diet can prevent these problems.
- **Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)** – Babies with MCADD are unable to convert fat into energy. Without treatment, babies can have seizures, extreme sleepiness, coma, and even die. Treatment may include frequent feedings and a special diet.
- **Biotinidase Deficiency** – Babies with this disease cannot use or recycle biotin (vitamin B) from their diet. Without treatment, babies with this disorder can have seizures, mental retardation, vision problems, hearing loss, and/or other health problems. Treatment includes daily biotin supplements.

Endocrine Diseases

Babies with endocrine diseases make too much or too little of certain hormones. Hormones are produced by glands in the body and affect body functions.

- **Congenital Adrenal Hyperplasia (CAH)** – The adrenal glands of babies with this disease do not make enough of the key stress-fighting hormone cortisol. In about two-thirds of the cases, babies also do not produce enough of the salt-retaining hormone aldosterone. As a result, affected babies can develop dehydration, shock, and even death. Treatment with one or more oral medicines can help prevent these problems. Girls with this condition may have the additional problem of having masculine-looking external genitals, which can be corrected with surgery.
- **Primary Congenital Hypothyroidism** – Babies born with this disease do not make enough thyroid hormone. Without enough hormone, babies grow very slowly and have mental retardation. These problems can be prevented by giving the baby special thyroid medicine every day.

Hemoglobin Diseases

Hemoglobin is found in red blood cells. It gives blood its red color and carries oxygen to all parts of the body. Hemoglobin diseases often lead to anemia because they affect the type and amount of hemoglobin in the red blood cells. Treatment may include medication, folic acid and close monitoring of the child's health.

These are some of the hemoglobin diseases that are part of the newborn screening test:

- **Sickle Cell Anemia and other Sickle Cell Diseases** – These diseases affect the type of hemoglobin in the baby's red blood cells. Babies with sickle cell disease can get very sick and even die from common infections. Many of the infections can be prevented with daily antibiotics. Ongoing health care and close monitoring help children with hemoglobin diseases stay as healthy as possible.
- **Hemoglobin H Disease** – This disease affects the amount of hemoglobin in the baby's blood. There is less hemoglobin, which results in smaller red blood cells. This also causes the cells to break down faster than normal. Babies with this disease can have mild to severe anemia, as well as other health problems. Treatment can include blood transfusions, taking folic acid, and avoiding certain medications and household products.

Cystic Fibrosis (CF)

This disease can affect many body organs, including the lungs and digestive system. In the first few months of life, a baby with CF can have poor absorption of milk or formula, slow growth, failure to thrive, recurrent lung infections, salty sweat, frequent runny stools, dehydration and life-threatening salt imbalance. Early treatment along with ongoing health care by a team at a special care center can alleviate many of these problems.

Severe Combined Immunodeficiency (SCID)

This immune disease is sometimes called "bubble boy" disease. Babies with SCID are born with little or no immune system.

The immune system helps fight infections. Early treatment can help prevent life-threatening infections in babies with this disease.

While newborn screening can identify some immune diseases, it does not test for HIV or AIDS.

Is Information About My Baby's Test Confidential?

Yes. There are serious penalties for any unauthorized release of private information collected during screening. For a copy of our Notice of Information and Privacy Practices please visit our website at www.cdph.ca.gov/programs/nbs.

How Much Does The Test Cost?

The cost is subject to change. Please check with your doctor, the hospital, or the NBS website for the current cost of the test. Medi-Cal, health plans and most private insurance will pay for the test. The cost is included in the hospital bill. You will not receive a bill from the NBS Program. If you have problems with your insurance, contact 1-800-927-HELP (1-800-927-4357) or if you have a prepaid health plan, contact 1-888-HMO-2219 (1-888-466-2219).

California law prevents insurance companies from refusing to issue or canceling a policy, or charging a higher rate or premium based on a person's genetic characteristics, including being diagnosed with one of the diseases found by newborn screening. If you have any of these problems, call one of the numbers listed above. It is also illegal to refuse employment based on the results of a genetic test.

Does My Baby Need Any Other Blood Tests?

If there is something that you are worried about, or know of a disease that may run in the family, talk to your doctor about what other tests can be done. The Newborn Screening Program screens for the most common treatable diseases. The Program evaluates adding other diseases as new tests and treatments become available. However, newborn screening does not test for every disease that might be found.

Due to biological variability of newborns and differences in detection rates for the various disorders in the newborn period, the Newborn Screening Program will not identify all newborns with these conditions. A negative screening result does not rule out the possibility of a disorder. Parents should remain watchful for any signs or symptoms of these disorders in their child and consult a physician. In addition to screening, babies also need regular well-baby check-ups.

Questions & Answers About the Storage of Newborn Screening Bloodspots

Why is my baby's blood spot collection card stored by the Genetic Disease Screening Program (GDSP)?

The main reason GDSP stores the used blood spots is to develop new tests to add to the newborn screening testing panel and to provide quality control for testing on an on-going basis. When the Newborn Screening Program began in the early 1980s, we tested for 3 disorders. The stored specimens were

used anonymously to develop the new tests, so that we now screen for about 80 disorders. Newborn Screening blood spot cards are **not** "DNA cards". Your child's DNA is not analyzed for our initial screening tests and his/her "DNA profile" is not stored. There is no personal information on the dried blood spot card, only a unique non-identifying number.

What if I do not want my baby's blood spot collection card used by the Genetic Disease Screening Program? What are my options?

If you decide not to allow the GDSP to use your child's unidentified dried blood spot, you may request that the specimen not be used for research and/or be destroyed by our laboratory. Please realize that if you make this choice, the spot will no longer be available should you or your doctor need it for any further health concerns with your child. Please submit this request in writing to: Chief of the Genetic Disease Screening Program, 850 Marina Bay Parkway, F175, Richmond, CA 94804.

Information About Cord Blood Banking

As a pregnant woman gets closer to her delivery date, the option of saving the baby's cord blood can be considered. Newborn umbilical cord blood contains stem cells which may be used to treat people with certain blood-related disorders. These include some types of cancer, immune system disorders, and genetic diseases.

Newborn cord blood can be collected from the umbilical cord shortly after birth. This does not interfere with the birthing process. It does not harm the health of either the baby or the mother. The collection of cord blood is safe, quick, and painless. If not collected, cord blood is discarded as medical waste.

Parents may choose to have their newborn's umbilical cord blood donated to a **public** cord blood bank. This donated cord blood can be made available to anyone who may need a blood stem cell transplant. It may also be made available to researchers who are trying to discover the causes of birth defects and other health-related problems. There is no cost for publicly donating cord blood.

Parents may instead choose to store their newborn's umbilical cord blood at a **private** cord blood bank. This cord blood could possibly be used if a compatible family member required a blood stem cell transplant. There are fees for collecting and storing cord blood at a private cord blood bank.

Both private and public cord blood banks are available in California. Parents interested in donating their baby's cord blood should talk with their prenatal care provider by the 34th week of pregnancy, or earlier.

For more information on both public and private cord blood banking, visit or call:

National Cord Blood Program:
http://www.nationalcordbloodprogram.org
866-767-6227

National Marrow Donor Program:
http://www.marrow.org
800-627-7692

Cord blood cannot be used for newborn screening.

Notes/Questions For My Baby's Doctor:

Americans with Disabilities Act (ADA)
Notice of Information and Access Statement
Policy of Nondiscrimination on the Basis of Disability
And Equal Employment Opportunity Statement

The California Department of Public Health (CDPH) complies with all state and federal laws, which prohibit discrimination in employment and provide admission and access to its programs or activities.

The Deputy Director, Office of Civil Rights (OCR), CDPH has been designated to coordinate and carry out the department's compliance with nondiscrimination requirements. Title II of the ADA addresses non-discrimination and access issues regarding disabilities. To obtain information concerning the CDPH EEO Policies or the provision of the ADA and the rights provided, you may contact the CDPH OCR by phone at 916-440-7370, TTY 916-440-7399 or write to:

OCR, CA Department of Public Health
P.O. Box 997413, MS 0009
Sacramento, CA 95899-7413

Upon request, this document will be made available in Braille, high contrast, large print, audiocassette or electronic format. To obtain a copy in one of these alternate formats, call or write:

Chief, Newborn Screening Branch
850 Marina Bay Pkwy., F175, MS 8200
Richmond, CA 94804
Phone: 510-412-1502
Relay Operator: 711/1-800-735-2929

The Genetic Disease Screening Program wants to provide quality services to the families of California and welcomes your comments and suggestions.	California Department of Public Health Newborn Screening Program 850 Marina Bay Parkway, MS 8200 Richmond, CA 94804 510-412-1502