PARENT'S GUIDE

To Your Baby's Newborn Screening

Pregnancy and the period after can be overwhelming for families. Newborn screening is just one piece of the care your baby will receive after birth, but it's a key to ensuring your baby has the healthiest start possible.

April 2024

NEBRASKA
Good Life. Great Mission.
DEPT. OF HEALTH AND HUMAN SERVICES

WHAT IS NEWBORN SCREENING?

Newborn screening is done for every baby born in Nebraska. It is a set of tests done to find certain conditions that can cause problems for your baby. You and your baby's healthcare professional can't tell by looking at your baby if they have any of these conditions. The tests (blood tests, hearing tests, and heart tests) are needed to find them so that problems can be prevented or reduced. This brochure will explain these three kinds of testing.

Who needs to be screened? All babies born in Nebraska must have the Blood-spot and Heart Disease testing. Because the tests are so important to protecting the health and safety of babies who are affected with the diseases, newborn screening is mandatory. The hospital is also required by law to offer hearing screening for your baby.

What happens during the bloodspot screen? This part of newborn screening is also referred to as the "heel stick". Five drops of blood are taken from your baby's heel. The blood sample is sent to a newborn screening laboratory and tested for the conditions described later.

Why are the screening tests important? The law requires screening to help prevent intellectual disability, brain and organ damage, seizures, stroke, other long-term problems and even infant death. The conditions screened for can usually be controlled by early and proper treatment. *(There are other causes of these health problems, which will not be found by these tests).*

When should my baby be screened? Your doctor will have the blood collected for the tests before your baby leaves the hospital. If you and your baby go home before your baby is 24 hours old, your baby will need to have some tests repeated. Some of the tests can give reliable results before 24 hours of age but not all of them. The hospital may schedule this appointment before you leave. **If not, you should contact your doctor and have the tests repeated when your baby is 24 to 48 hours old.**

Babies born at home: Babies who are born at home must also be screened for these diseases. The parent(s) or the person registering the birth must arrange with a healthcare professional to order the tests. The blood should be collected between 24 and 48 hours of birth, so that any needed treatment can be started as soon as possible. If a baby is older, they must still get the screening. Even though it is less ideal in terms of timing, older babies can still benefit from treatment for certain conditions.

How will I find out what the results are?
Your baby's doctor should receive the results from the hospital where the blood was collected. Usually by the time of your first "well-baby" check-up. Make sure to ask your baby's doctor about the newborn screening results.

What if any of the screening results are abnormal?

A "positive," abnormal or inconclusive screening test result only means that your baby ***might*** have one of the conditions mentioned later in this booklet. Sometimes positive screening results are found in babies that do not have the condition. A diagnosis of a condition is not made with the first lab test. Further testing will be necessary to determine if your baby actually has it.

If you are asked to have your child re-tested, ***please act quickly*** so tests can be repeated and final results obtained. If needed, treatment must be started as soon as possible to prevent the onset of problems for your baby's health and development, or in some cases to prevent death in the newborn, infant or early childhood period.

Why else might I be asked to have my baby "re-tested?"

Some things can cause problems with the tests. When these things happen, we can't be very sure of some of the results. Some of the reasons parents are asked to bring their babies back for re-testing are:

- ✓ The sample was collected too early for some conditions to be tested (less than 24 hours of age).
- ✓ The sample wasn't collected before a blood transfusion or too soon after a blood transfusion; or

- ✓ A problem occurred with the quality of the sample (e.g. problem with collection and/or handling of the dried bloodspots).

What do I need to do to help my baby?

Before you leave the hospital:

Your Baby's Doctor:	**John Doe M.D.**
Phone Number:	**402-555-1212**

- ✓ Make sure the hospital knows who your baby's healthcare provider will be. That is important in case the newborn screening program needs to notify them of any additional tests or evaluation needed.
- ✓ Make sure the hospital *and* your baby's doctor have your current contact (phone) information.

Mother's name:	**Jane New Mom**
Phone Number:	**402-444-1212**

After you go home, if your baby's doctor's office calls:

- ✓ Write down whatever instructions the doctor or nurse provide you.
- ✓ Remember there are a lot of reasons why babies sometimes need to have repeat testing. It does NOT necessarily mean your baby has a disease.

- ✓ Do not delay. Follow your baby's doctor's instructions. If your baby would have one of the screened diseases, the earlier you get to diagnosis and treatment the better for your baby.

PROTECTIONS FOR YOUR BABY'S BLOOD SAMPLE

We want you to be confident your baby's bloodspot is used only for its intended purpose, which is to find information that can help your baby. Nebraska law and regulations require the following: The laboratory must keep the newborn screening specimens at least 90 days. Within 30 days after this 90 day period, the laboratory must get rid of the sample. The blood spots are disposed of in a way that they cannot be linked to identifying information (they are burned). This process helps protect your baby's health information.

Your baby's blood specimen can only be used for public health research if all of these things have been obtained:

- ✓ Your written consent
- ✓ Written assurance that your baby's confidentiality is preserved
- ✓ Institutional Review Board approval following protections of human subjects from research risks under subpart A of part 46 of 45 Code of Federal Regulations
- ✓ Approval of the State Newborn Screening Advisory Committee
- ✓ Approval of the Chief Medical Officer of the Nebraska Department of Health and Human Services

Retrieving Your Baby's Blood Spot

As you will read on the next pages, your baby's hearing will also be tested. For babies who are "confirmed" with a hearing loss, additional testing on the blood spot might be helpful. Cytomeg-

alovirus (CMV) infection present at birth can be one cause of hearing loss. Testing for congenital CMV is best accomplished on blood samples obtained shortly after birth. This is because later samples can't tell the difference between "congenital" or "acquired" CMV. Therefore, if your baby does not pass the hearing screening, it is important to get repeat hearing screening and audiologic diagnostic testing done as soon as possible. That way, if your baby does have hearing loss, you can ask your baby's doctor to request the dried bloodspot to be returned for more tests before the bloodspots are destroyed.

✧ A Special Note: ✧

The newborn screening tests alone **cannot be used to make a final diagnosis.** They are a "screen," designed to detect newborns who need further testing to determine if they have certain disorders. The screening tests are very efficient and provide newborns with the best opportunity to identify the disorders early and obtain treatment.

However, like most laboratory tests, the tests used for newborn screening cannot guarantee that every affected newborn will be identified, or that only infants at higher risk of being affected will have positive tests. Therefore, it is important to recognize that there will be some "false positives" (newborns with a positive, abnormal or inconclusive screen result who are later found to not have the suspected disorder), and the possibility of "false negatives" (newborns with normal screening results who are later found to have one of the conditions).

WHAT CONDITIONS ARE INCLUDED IN NEBRASKA'S ROUTINE BLOODSPOT SCREENING PANEL?

Amino Acid Disorders

Phenylketonuria, (FEE-nil-KEE-tone-u-ree-ah), or PKU, is an amino acid disorder. Babies with PKU can't break down phenylalanine or phe. Phe is an amino acid found in protein foods like milk, meats, eggs and cheese. The high levels of phe may cause nerve and brain cell damage and can result in intellectual disability. If detected early and the baby is started on a special diet, intellectual disability is prevented. Other Amino Acid Disorders screened for include: • Argininosuccinic Acidemia (ASA Lyase deficiency) • Citrullinemia (CIT) • Homocystinuria (HCY) • Maple Syrup Urine Disease (MSUD) • Tyrosinemia (TYR)

Biotinidase Deficiency (BIO) (BYE-o-TIN-I-days) is a disorder found in babies who are missing the enzyme biotinidase. This can lead to seizures, developmental delay, eczema, and hearing loss. Problems can be prevented with biotin (a vitamin) treatment started in the first few weeks of life.

Congenital Adrenal Hyperplasia (CAH)

(Congenital A-DREE-nal Hyper-PLAY-zia) is a disorder caused by an enzyme deficiency that results in the adrenal gland producing too little of one hormone and too much of another. Some babies with this disorder are at risk of sudden death. A hormone treatment started early can prevent problems.

Congenital Primary Hypothyroidism (CPH)

(HIGH-po-THIGH-roid-ism), or CPH is a disorder caused by not having enough thyroid hormone. The most common effects of CPH are intellectual disability and poor growth. If treatment with thyroid medication starts in the first few weeks of life, these children usually develop normally.

Cystic Fibrosis (CF)

(SIS-tic FY-bro-sis) is a genetic disorder that affects people in a variety of ways. They may have persistent coughing, wheezing or shortness of breath. They may have an excessive appetite, but have poor growth and greasy, bulky stools. With early treatment those who would have had problems gaining weight, can have improved growth and development. With early diagnosis from newborn screening, some may have fewer hospitalizations. Regular monitoring may prevent or reduce lung infections.

Fatty Acid Oxidation Defects (FAO's)

Medium Chain Acyl-CoA Dehydrogenase Deficiency (Medium Chain A-seal Co-A Dee-HIGH-dra-gen-AZE Deficiency - MCAD) is a disorder of fatty acid metabolism. When babies and children with MCAD become ill or have long periods of fasting, blood sugars become dangerously low and they are at risk of having a "metabolic crisis." A metabolic crisis can lead to seizures, failure to breathe, and cardiac arrest. These can result in serious brain damage or death. However, screening can provide diagnosis before symptoms occur. Parents can then prevent the fasting periods, and know when to seek early medical care, to prevent the crisis. A special diet supplement may be added to the baby's diet to help prevent problems. Other Fatty Acid Defects screened for include: • Carnitine Uptake Defect (CUD) • Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD) • Trifunctional Protein Deficiency (TFP) • Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)

Galactosemia (GAL)

(Guh-LAK-toe-SEE-me-ah) is a disorder in which a simple sugar called "galactose" can't be broken down in the body. Galactose is found in breast milk, many formulas and milk products. If it remains at high levels in the body, it can harm the baby's eyes, liver, and brain, and could be life threatening. When started early, a special diet can prevent these problems.

Hemoglobinopathies (HGb's)

(He-ma-glow-bin-OP-ah-thees) are a group of disorders of the red blood cells, including sickle cell anemia. Babies with this are more likely to have anemia (low number of red blood cells), episodes of pain, strokes, and life-threatening infections. Early treatment with antibiotics, immunizations and parent education can help. These measures can prevent serious infections in childhood, and reduce health problems.

Lysosomal Storage Diseases (LSD's)

Pompe (Pom-pay) Disease and Mucopolysaccharidosis (Muu-co-pah-lee-sack-a-ry-doh-sis) type I (MPS-I) are Lysosomal storage disorders. Lysosomes are the parts of cells that store, break down, and recycle unwanted molecules using specific enzymes. The enzymes in children with these diseases are missing or not working well. As a result, unwanted molecules build up. This results in irreversible damage to muscles, nerves and/or certain organs in the body. If caught early, there are treatments to help prevent damage.

Organic Acid Disorders (OA's)

These conditions can have a variety of effects on babies from mild to severe including metabolic crisis and problems with the heart, muscles, and some organs. Babies with these disorders may become rapidly sick, have seizures, go into a coma and could die without treatment. The Organic Acid Disorders screened for include: • Beta Ketothiolase Deficiency (BKT) • Glutaric Acidemia

type I (GA-I) • IsovalericAcidemia (IVA) • Methylmalonic Acidemia (mutase) & (Cbl B&C) • Multiple Carboxylase Deficiency (MCD) • Propionic Acidemia (PA) • 3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC) • 3-Hydroxy-3 Methylglutaric Aciduria (HMG)

Severe Combined Immune Deficiency (SCID)

Sometimes referred to as the "bubble-boy" disease, SCID is a serious life-threatening condition. Babies with SCID lack a functioning immune system and suffer from repeated infections. With no treatment, these babies usually die within the first two years of life. However, early intervention and bone marrow stem cell transplant can cure these babies. The screening test may also pick up other T-cell immune deficiencies for which other treatments can help.

Spinal Muscular Atrophy (SMA)

Spinal Muscular Atrophy affects the nerves in the spinal cord that send signals to the muscles to tell them how to work. When these nerves don't work, muscles can't do their job and become very weak. Babies born with SMA received missing or faulty SMN1 gene copies from both parents. SMA is a progressive disorder, and if it is not treated, it will worsen. Early treatment of babies with SMA, before symptoms occur, is most effective. As a result, screening of all newborns for SMA gives babies the best chance for a healthier life.

X-linked Adrenoleukodystrophy

X-linked Adrenoleukodystrophy (a-DREE-no-luke-o-DIS-tro-fee) is a genetic disorder mainly affecting males. Patients with this disease can't break down certain long-chain fatty acids. As the fatty acids build up, this leads to neurologic problems from the breakdown of the protective cover of nerve cells. There are three main types of X-ALD, ranging from serious childhood onset with cerebral damage, to patients whose adrenal system is mainly affected, to adult onset. Early diagnosis, monitoring, and treatment can prevent much of the damage from this disease.

NEWBORN HEARING SCREENING

All hospitals offer newborn hearing screening during the birth admission or by arrangement with another facility after the birth admission.

Hearing loss is one of the most common major birth defects. If not identified early deafness or hearing loss will affect language development, social-emotional growth, and educational performance in children. The earlier children are identified as deaf or hard of hearing and start receiving early intervention, the more likely they will stay on track with their hearing peers and reach their full potential.

The goal is for:

- ✓ All infants to receive a hearing screening by 1 month of age.
- ✓ All infants who "refer" (or did not pass) the hearing screen to receive a diagnostic evaluation by 3 months of age.
- ✓ All infants who are identified as deaf and hard of hearing begin receiving early intervention services by 6 months of age.

The hearing screening is a safe procedure that can be done in a very short time. There are two methods that might be used: auditory brainstem response (ABR) and/or otoacoustic emissions (OAE). Both are done when the baby is asleep or quiet. Responses to sounds sent through tiny earphones are counted and averaged by a computer. It is important to remember that while many newborns may not pass the hearing screening screening, only further audiologic testing and evaluation can determine which ones really have hearing loss. Therefore, it is important that you work with your baby's doctor to get further testing if the screening results indicate "refer" (or did not pass).

If your baby passes the hearing screening but has risk factors for hearing loss, you and your baby's doctor will want to monitor your child closely. Some risk factors include a family history of deafness or hard of hearing, low birth weight or certain other medical conditions. The newborn hearing screening will not pick up hearing loss that develops later in life, for example, as a result of serious infection or illness.

If your child is suspected of or found to be deaf or hard of hearing, Nebraska's Early Intervention Program called the Early Development Network (EDN) can help you coordinate services with local school systems and providers for evaluations, early intervention services and assistive technology.

The EDN provides services and supports based on the needs of children birth to age 3 and their families with the belief that parents know what is best for their families. Call **(402) 471-2471** or call toll-free **1-888-806-6287** to reach the EDN.

NEWBORN SCREENING FOR CRITICAL CONGENITAL HEART DISEASE (CCHD)

Congenital heart disease is the most common birth defect affecting about 8 in every 1,000 babies born. Some forms of congenital heart disease can be found when examined by the baby's healthcare professional. However, sometimes the baby won't show signs of a problem until they are more affected.

The screening is done to find these before symptoms develop. Not all forms of heart disease can be detected by the newborn screening test for CCHD, but the screen increases the chance of early detection for 7 of the most serious forms of congenital heart disease. The screening test for CCHD involves placing a pulse oximeter on your baby's foot and hand.

Simple and painless, the pulse oximeter measures the amount of oxygen in the blood and the heart rate. Babies with low levels of oxygen in their blood should get further evaluation. The screening test should be done at 24 hours of age or shortly thereafter. Babies sent home without the screening who have undetected CCHD can suffer serious brain and other organ damage, or even die when deprived of oxygen. Catching CCHD early means you can help prevent these problems and get treatment for your baby to correct the heart defect.

For More Information on BLOOD-SPOT SCREENING:

- ✓ Call **402-471-6558**
- ✓ E-mail dhhs.newborn.screening@nebraska.gov
- ✓ Check out our web site at: https://dhhs.ne.gov/Pages/Newborn-Screening.aspx or scan
- ✓ When in the hospital, ask your nurse about watching a short video on newborn screening.

For More Information on HEARING SCREENING:

- ✓ **Refer to the brochure: "Can Your Baby Hear?"** included with your newborn packet.
- ✓ If you did not receive this brochure or have questions about the Nebraska Early Hearing Detection and Intervention
- ✓ (NE-EHDI) Program, contact the Nebraska Department of Health and Human Services at: 402-471-6746.

For questions about Nebraska's **Early Intervention** Program

- ✓ Contact "Nebraska CHILDFIND" at 1-888-806-6287
- ✓ Visit: https://dhhs.ne.gov/Pages/Hearing-Detection-and-Intervention.aspx or scan

For More Information on CRITICAL CONGENITAL HEART DISEASE SCREENING:

- ✓ Talk to your baby's doctor or other healthcare professional
- ✓ The Centers for Disease Control and Prevention have a good website at: www.cdc.gov/ncbddd/heartdefects/screening.html or scan

To see a VIDEO on Newborn Screening,

go to the web-site: https://www.babysfirsttest.org/ or scan

and watch https://youtu.be/6skxqLUjFDg or scan

"Newborn Screening Saves Lives, One Spot at a Time"

Or check out the **"One Foot at a Time"** video at

https://vimeo.com/34898727 or scan

To order more PARENT INFORMATION booklet/brochures:

Call **402-471-9731** or fax order to "**NNSP**": fax **402-471-1863** or e-mail dhhs.newbornscreening@nebraska.gov.

Please specify the brochure you'd like to order:

- ✓ Parent's Guide booklet
- ✓ Hearing Screening brochures ("Can Your Baby Hear?" or "Your Baby Needs Another Hearing Screening")
- ✓ Also available in Spanish in print

This program is supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) as part of an award totaling $244,000 with 0% financed with non-governmental sources. The contents are those of the author(s) and do not necessarily represent the official views of, nor an endorsement, by HRSA, HHS, or the U.S. Government. For more information, please visit HRSA.gov.

NEBRASKA
Good Life. Great Mission.
DEPT. OF HEALTH AND HUMAN SERVICES

U.S. Department of Health and Human Services
HRSA
Health Resources and Services Administration

The Nebraska Department of Health and Human Services is committed to affirmative action/equal employment opportunity and does not discriminate in delivering benefits or services.