

# Newborn Screening

## Frequently Asked Questions

### **What is newborn screening?**

Newborn Screening is done for every baby born in Nebraska. A set of blood tests are done to find conditions that could be harmful to your child. Even after a baby is born, there are usually no signs or symptoms so parents can't tell whether or not their baby may have a condition... that's why newborn screening is so helpful. It finds things you can't necessarily see. Newborn screening has actually been around since the 1960's. Screening helps prevent mental retardation and other damaging problems in children. It can even prevent infant death from some conditions. Five drops of blood are taken from your baby's heel and placed on a special filter paper. The hospital sends this specimen to the newborn screening laboratory.

### **Why is my baby being tested?**

Nebraska law requires every baby to be tested. This is because the disorders we are looking for cannot usually be detected by looking at the baby, have very serious effects on your baby's health, but these effects can be minimized or prevented with early detection and intervention.

### **What are the specific disorders that Nebraska screens my baby for?**

Newborn Screening will screen your baby for the following disorders.

- Argininosuccinic Acidemia (ASA)
- Beta-Ketothiolase Deficiency (BKT)
- Biotinidase Deficiency
- Carnitine Uptake Defect (CUD)
- Citrullinemia (CIT)
- Congenital Adrenal Hyperplasia (CAH)
- Congenital Primary Hypothyroidism (CPH)
- Cystic Fibrosis (CF)
- Galactosemia (GAL)
- Glutaric Acidemia Type 1 (GA1)
- Hemoglobinopathies (Hgb's)
- Homocystinuria (HCY)
- Isovaleric Acidemia (IVA)
- Long-Chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
- Maple Syrup Urine Disease (MSUD)
- Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
- Methylmalonic Acidemia -Mutase (MUT)
- Methylmalonic Acidemia - Cbl A and B (Cbl A and B)
- Multiple Carboxylase Deficiency (MCD)
- Phenylketonuria (PKU)
- Propionic Acidemia (PA)
- Severe Combined Immunodeficiency (SCID)
- Sickle Cell
- Tyrosinemia (TYR)
- Trifunctional Protein Deficiency (TFP)
- Very Long-chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)
- 3-Hydroxy 3-Methyl Glutaric Aciduria (HMG)
- 3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)

### **What if I want my baby tested for something not on the required screen?**

You should discuss your wishes for additional testing with your baby's doctor/health care professional. There would be additional charges for this test. The State Newborn Screening Program would not be involved to help ensure appropriate follow-up.

### **How can I find out about the screening test results?**

Ask your baby's doctor or health care professional for the screening results. These should be available during a well-baby check-up at about 2 weeks of age.

### **What does it mean if the results were "negative?"**

This is good news. The test results indicate your baby is not at risk for any of the disorders screened. It is important to remember this is just a "screening" test. While the testing offers the best chance of identifying these disorders early, if your baby shows any signs or symptoms of illness or developmental problems, contact your baby's doctor/health care professional.



### **What does it mean if the specimen was "unsatisfactory?"**

Usually this means there was a problem with how the specimen was collected or handled before it got to the screening lab. It takes great care and skill to collect it correctly. Usually, more than 99% of specimens are collected and shipped without any problems. Just be sure to take your baby back right away for a new specimen if you are asked to do so. There won't be any new charges from the newborn screening laboratory for this repeat testing.

### **What does it mean if the specimen was collected too early?**

Sometimes babies and moms leave the hospital before the baby is 24 hours of age. If this were true for you, your baby's specimen would have been collected early. It is good to collect it before hospital discharge because it can be tested for some of the disorders that could affect the newborn in the first week of life. However, because your baby's hormone levels need time to adjust after birth, it would not be a reliable specimen to test for congenital hypothyroidism until after he/she is at least 24 hours old. Likewise, testing for congenital adrenal hyperplasia and Cystic Fibrosis is not done on the specimens collected at less than 24 hours of age. Repeating the specimen after 24 hours of age also gives better (more reliable) results for some other conditions as well. If your doctor/health care professional asks you to have your baby re-tested, please do this right away after your baby is 24 hours old.

### **What does it mean if the results are "inconclusive?"**

There are many reasons that results would be reported as "inconclusive:"

- The specimen was collected too early.
- The specimen was damaged by heat or humidity.
- The specimen was collected too soon after a blood transfusion.
- The specimen was collected while receiving intravenous feedings.
- Sometimes the results are just slightly outside of the normal or expected range. The laboratory may call these "inconclusive" and recommend a repeat test.

Be sure to take your baby back right away for a new specimen to be collected. There won't be additional charges from the newborn screening laboratory for this repeat testing.

### **What does it mean if the results are "positive"?**

There are a few things this can mean:

- Primarily, a "positive" or abnormal screening result means your baby is at higher risk for having one of the conditions screened.
- Your baby might have one of the metabolic conditions, but it can't distinguish which one.
- Your baby had a special formula or feedings around the time the specimen was collected.
- Your baby received blood a short time before the specimen was collected.
- Your baby could have a mild form of the condition.
- Your baby could be an unaffected "carrier" of a gene for the condition.
- Sometimes it is what's called a "false positive." In population-screening, the normal test result ranges are set up so we reduce the risk of missing or not identifying an affected baby. This means some babies have a screening result that looks positive on screening, but it turns out once another test is done the result is normal.



The important thing is to follow your baby's doctor or health care professional's advice right away. He/she will have you take your baby to a laboratory to collect a new specimen. Usually, this new specimen will be tested by a different method. For some conditions, the doctor/health care professional may suggest certain feeding practices with your newborn, at least until the new test results are back. Your baby's doctor or health care professional may (depending on the condition) also discuss signs or symptoms to watch for and recommend you contact him/her should they occur. Be sure to follow your doctor's or health care professional's recommendations. In some crises they may refer your baby to a pediatric specialist for the condition. Be sure to follow through with the specialist if this referral is made.

**What if my doctor says my baby needs a "repeat" test?**

A “repeat” test is testing on a new dried blood spot filter paper specimen. If this was requested by the newborn screening laboratory, there will be no additional charges from the screening laboratory. Be sure take your baby back as soon as possible for this to be done. If your baby is the next child to have one of these conditions, the sooner it’s identified, the better the outcome will be for your baby.

**What if my doctor says my baby had a positive result and needs another test?**

Follow the recommendation. He/she will have you take your baby to a laboratory to collect a new specimen. Often but not always, this new specimen will be tested by a different method. For some conditions, the doctor/health care professional may suggest certain feeding practices with your newborn, at least until the confirmatory results are back. It is important to follow your baby’s doctor or health care professional's recommendations. To help you deal with the anxiety about these results, try to keep in mind that the first screening result could be a false positive. If it turns out that your baby does have the condition, you are not alone, and there are resources available to help you and your baby. So follow your doctor’s or health care professional's advice, and continue to enjoy your newborn!



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