

## **What is Newborn Metabolic Screening?**

The Newborn Metabolic Screen is a special test used to test your baby for certain serious medical conditions. The goal of the screen is to identify babies who have these disorders before they ever get sick, and to help them get treatment as soon as possible.

## **Is the Newborn Metabolic Screen the same as the “PKU test”?**

Yes, the Newborn Screen is often referred to as the PKU test, but this is an outdated term. PKU (Phenylketonuria) was the very first condition screened for by newborn screening, back in the 1960's. Since this time many more conditions have been added to the test, which is why it should be referred to as the Newborn Metabolic Screen.

## **How is the Newborn Metabolic Screen performed?**

The Newborn Metabolic Screen is performed by pricking your baby's heel and putting a few drops of blood onto special filter paper. The filter paper is allowed to dry and is then sent to the State Health Department. The blood is analyzed by the lab to identify babies who are at higher risk to have a medical condition. If the screen indicates your baby might have a medical problem, a member of the newborn screening follow-up unit will call your baby's doctor with the results. If we cannot identify the baby's doctor, we may call you directly to get this information.

## **Will the test hurt my baby?**

A nurse or medical assistant will use a small sterile lancet to prick your baby's heel. The only pain the baby may feel is that of a quick prick, and it will go away quickly. Remember that the benefits of newborn screening, such as preventing learning delays and serious medical problems, greatly outweigh the discomfort the baby may feel due to the small heel prick.

## **Are there any side effects of the testing?**

There is a very slight risk of infection of the puncture site or heel. The heel prick should be performed with a sterile lancet on a heel that has been cleansed, which means the risk for infection would be extremely small.

## **When is the Newborn Metabolic Screening collected?**

The first test should be collected after the baby has had 24 hours of feeding. We collect the screen early to try to identify babies that have problems as soon as possible. In Maryland, we also recommend that babies get a second screen at the pediatrician's office, to help identify disorders that may not have been identified on the first screen. This is usually collected between 7 days and 1 month of age.

## **What happens if my baby has a positive test?**

If your baby has a positive test result, we attempt to contact the baby's pediatrician or primary care doctor. We find out the doctor's information and your baby's name from the form connected to the blood spot that was filled out when the baby was in the nursery. It is important to make sure the nurse in the nursery fills out the paper completely and has your most accurate contact information so that we can reach you easily if there is a problem. If we do not know who the baby's doctor is, we will call you directly to find out this information. If you do not have a working number listed, we will send a certified letter to your address.

## **Does a positive test mean my baby has the condition?**

No, Newborn Metabolic Screening is only a “screening” test, it is not a “diagnostic” test. This means that the test was made to find babies who may have a medical problem. If a baby has a positive screening test, he/she should get the recommended follow-up testing right away. If the follow-up testing continues to indicate the baby has a problem, your doctor will refer your baby to a specialist for additional testing. Depending on the results of the initial Newborn Screen, the follow-up unit may refer the baby immediately to a specialist or the emergency room.

## **My baby's follow-up testing was normal, is there anything else I need to do?**

No, as long as all of the recommended follow-up tests were normal, no additional testing is needed. This means that the initial test was a “false positive”.

### **Do I get to see my baby's results?**

We do not send the results directly to parents. However, you should talk to your baby's doctor and make sure he/she has a copy of the results. You can ask the baby's doctor to show you a copy of the results.

### **What if my baby looks healthy?**

Most babies who have these conditions look healthy and act normally in the newborn period. The point of the test is to try to find babies that have these problems before they get sick. Just because your baby looks healthy does not mean he/she is healthy and it is important to listen to your doctor's advice.

### **No one in my family has these conditions, does my baby still need to be tested?**

Yes! Most babies identified by Newborn Metabolic Screening will not have a family history of the disorder. Many of them may even have healthy siblings. Having a normal family history does not necessarily mean that the baby will be healthy.

### **My baby was in the NICU (neonatal intensive care unit) for a while, do they still get the newborn metabolic screen?**

Yes! All NICU babies still have newborn metabolic screening. The timing is usually slightly different. Babies in the NICU often get tested as soon as possible, they then usually get an additional test between 2-3 days of age, another around 10 days and, if they are still in the hospital, at one month of age.

### **Should all babies born in Maryland get the Newborn Metabolic Screen?**

Yes! All babies born in the state of Maryland should get a Newborn Metabolic Screening. Most babies get their first screen at the hospital and their second at the pediatrician's office. If you choose to deliver your baby at home it is important to talk to your pediatrician or family doctor about getting your baby a screen as soon as possible.

### **Does the Newborn Metabolic Screen test for all things that could make my baby sick?**

No, the Newborn Metabolic Screen tests only for some disorders that could make your baby sick. The conditions on the newborn screen are decided by a group of specialists. They choose conditions that can be detected through the dried bloodspots and have an effective treatment. Unfortunately, there are some problems that can make children sick that we do not have treatment for, or are not able to detect using current newborn screening technology.

### **What conditions are on the Newborn Metabolic Screen?**

There are over 50 conditions on the newborn screen. [Click here to learn more.](#)

### **How do they decide which conditions should be added to the newborn screen?**

Each state makes its own decision about the conditions that are included in newborn screening. However, states base their decision on recommendations provided by a national group of specialists. The states consider things like: if treatment is available, if they can detect the condition in dried bloodspots, the cost of testing for the condition, and how good the method for testing is at picking up all children that have the condition (sensitivity) but not picking up those that do not (specificity).

### **Why does my baby have to have a repeat screen?**

In Maryland all babies should get a repeat screen. This should be done after your baby is 7 days of age or older.. This allows the lab to pick up some conditions that may not have shown up on the first screen. Other reasons babies may have a repeat screening are if something was abnormal on the first screen and the lab wants to re-check a value or if the first sample was not able to be run because of a problem with the way it was collected (unsatisfactory specimen).

### **My baby was not born in Maryland, did they get a Newborn Metabolic Screen?**

Your baby probably had a Newborn Metabolic Screen. You will need to check with the hospital where your baby was born. If your baby was born outside of the United States, he/she may or may not have had a newborn screen.

**Who has access to my baby's test results?**

The doctor caring for your baby, the hospital where your baby was born, and the people who work with the newborn screening laboratory at the state health department are the only people who have access to your baby's test results. If your baby needs to see a specialist, the results of the Newborn Screen will be provided to the specialist.

**I do not know if I want my baby to have a Newborn Metabolic Screen, what can I do about this?**

Speak to your pediatrician and ask questions. Make sure you have all the information before making this important decision. Remember, most babies look healthy, even if they have a condition on the newborn screen that will cause them significant medical harm. If you decide you really do not want your baby to have newborn screening, someone from the state health department will also contact you to discuss newborn metabolic screening. You do have the right to decline screening. Remember, your baby is counting on you for this important decision.