Frequently Asked Questions

Is the Newborn 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Yes, the Newborn Screen is often referred to as the PKU test, but this is an outdated term.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 and 70's. Since this time, many more conditions have been added to the test, which is why it should be referred to as the Newborn Screen. Calling the screen the PKU test might be confusing for families or other providers, and children may be incorrectly initially labeled as having PKU when, in fact, they have a different condition on the Newborn Screen.

Why do we get two screens in Maryland?
Historically, the 2nd screen was initiated because many babies were discharged before 24 hours of age and, therefore, did not have a satisfactory initial screen. The 2nd screen is important in these cases so that a baby can have at least one satisfactory screen. Over the years, the two screen system has been supported by the fact that about 10% of congenital hypothyroidism is missed on the initial screen but is picked up by the subsequent screen. The 2nd screen may also pick up milder variants that are not evident at the time of the initial screen. The 2nd screen picks up several children each year with conditions that are not detectable on the initial screen.

Beginning in 2006, the state of Maryland began testing for cystic fibrosis (CF) by implementing the IRT/IRT system. Using this system, two elevated IRT levels are needed to determine if a baby is at increased risk for cystic fibrosis. This means that the two screen system is a vital part of CF testing

What is the ideal timing for newborn screening for a term baby?
The first Newborn Screen (NBS) should be collected when the baby has had 24 hours of feeding. Ideally, this is the day the baby is discharged from the hospital. The 2nd screen should be collected when a baby is greater than 7 days old, ideally between 10 days and two weeks. A screen collected at less than 24 hours is not a valid for several conditions on the NBS.

What is the ideal timing for newborn screening for babies in the NICU?
All babies admitted to the NICU should have a Newborn Screen collected as soon as possible after admission. The Newborn Screen should be collected prior to transfusion and initiation of antibiotics. Additional screens should be collected between 2-3 days of age, at the 10 day interval, and then again at 1 month or discharge (whichever comes first).

How old is too old for the Newborn Screen?
The cutoff values that the laboratory uses are only valid for babies that are 8 weeks or less. If you send a screen on a baby older than 8 weeks, the laboratory does not have appropriate cut-offs for interpretation. However, if there is an elevation, the follow-up staff will contact you to make recommendations.

What conditions are on the Newborn Screen?
There are over 50 conditions on the Newborn Screen. Go here to learn more XXX

If a baby has an abnormal result how soon do I have to get them in?
Timing is crucial in newborn screening so that babies are identified and treated as soon as possible BEFORE they get sick. If you are contacted by someone at the health department about an abnormal screen, please do not delay and get the baby in as soon as possible. The follow-up team at the health department can give you more details when they call you about an abnormal result.

How do I contact a specialist to get advice?
There are many specialists available to help if you have questions about a Newborn Screen or if you need to refer a baby. For a list of providers that can help you, click here XXX. The follow-up group is also always available to assist you with these referrals.

I have a result that shows a borderline elevation that I never received a phone call about, what does this mean?
Sometimes a level is elevated on the NBS, but it is not high enough to be considered an "urgent abnormal" result. In these situations, the text of the report itself will request that you collect a repeat Newborn Screen, and you will not receive a call from the follow-up team. IT IS IMPORTANT that you READ EACH REPORT to assure that you follow-up appropriately with borderline results. A borderline result is still not normal and should still be followed up promptly with a repeat screen. If you have questions the follow-up team members are always available to help you.

The baby is noted to have an elevated IRT, but I never received a phone call about this, what does this mean?
Results are not called out to providers when babies have an abnormal first screen for cystic fibrosis (represented by an elevated IRT aka Immunoreactive Trypsinogen). Instead the newborn screening result is sent out calling the level 'borderline' and asking for a repeat to be collected. A baby is not considered to have a positive screen for cystic fibrosis until the baby has two abnormal IRT levels. Ideally the first is collected at the birth hospital and the 2nd should be collected after 10 days. IT IS IMPORTANT that you READ EACH REPORT to assure that the baby is followed appropriately.

If a baby has an unsatisfactory Newborn Screen at the birth hospital, how soon do I need to get them in for a repeat screen?
If a baby has an unsatisfactory initial Newborn Screen, you should recall the patient ASAP to get a repeat screen. Remember, the purpose of newborn screening is to identify babies before they ever get sick. Just because a baby looks well at a few days of age does not mean they will have a normal Newborn Screen. Many babies who have disorders on the Newborn Screen look healthy for a few days before they get sick.

If I get a repeat due to an unsatisfactory initial screen, does the child need a 3rd screen (a total of two satisfactory screens)?

Yes, we still recommend that all babies born in Maryland have two satisfactory Newborn Screens so that we have two chances to screen the baby. If you are notified of an initial unsatisfactory screen, and the baby is due for the repeat screen shortly after this, we still advise that you bring the baby in ASAP. You can then wait one to two weeks to collect the next screen.

How do I order more newborn screening kits (forms + Filter paper)?
To order more kits please fill out the following form (hyperlink-Screening Kit Request Form) and fax it back to the newborn screening office at 410-333-7112. You can also call 410-767-6099 to request a copy of the form.

How can I get the results of the screen for a specific patient?
If you are the baby's primary provider but you are not the provider that submitted the Newborn Screen (i.e. it was submitted by the birth hospital or another provider), please fax your request on letterhead to 410-333-7112. Please include as much information as possible including:

- The baby's name
- The mother's name
- The date of birth
- The birth hospital.

If you are the baby's primary provider and you sent the Newborn Screen from your office or you wrote the order for the screen, the result will be sent to you by mail. However, if you would like it faxed to you sooner, or for some reason you did not receive a copy of the results, please call 410-767-6099.

If you have a MyLims account and you were the ordering provider, you can access the results through the following website- http://starlims.dhmh.state.md.us/starlims10.mynbs/. If you would like to create a MyLims account, please call the main Laboratory number to set this up at 410-767-6099.

If you have questions regarding an abnormal result- please call the follow-up unit and speak with Johnna Watson, RN, BSN- 410-767-6736; Carrie Blout, MS, CGC 410-767-6708; or Julie Kaplan, MD- 410-767-3060.

How do I get training on proper blood spot collection?
If you would like a nurse to come and educate your center about proper blood spot collection, please contact Johnna Watson, RN, BSN at 410-767-6736. You may also look at the newborn screening training education materials located on this website (bottom right box located on the newborn screening home page).

What is the blood spot storage policy?
Please see (XXX) for the official blood spot storage policy.

Are there DNA tests performed using the Newborn Screen bloodspots?

No, currently the State of Maryland is not conducting DNA testing on the newborn screening blood spots to test for the disorders. We use other methods to test for the conditions on the Newborn Screen. As technology begins to change, DNA testing may be implemented in the future.