

NEWBORN SCREENING

Nebraska Department of Health and Human Services

May 2016

Essential Information for Practitioners

Newborn (bloodspot) screening is mandatory for every baby born in Nebraska, but every step of the process doesn't just magically happen. While the state system is in place to support you in your responsibilities, it is important for every healthcare provider who takes responsibility for the care of newborns to know what state law requires of them. Frequently today, hospitalists or rounding pediatricians who are seeing the baby during the obstetric stay, won't be caring for the baby post-discharge. So whose job is it anyway? How do we make sure no baby affected with a screened disorder goes unidentified? The law requires the attending physician to ensure the screen is done, and to follow-up and confirm results that need further action.

I ordered the screen but I'm not seeing the baby post-discharge. What's my responsibility?

Your first job is to educate the parents about the newborn screen using the materials provided by the Department of Health and Human Services (DHHS). The "Parent's Guide to Your Baby's Newborn Screening" explains how it is done, why it is done, for which conditions their baby will be screened and reasons they might need to bring baby back for a repeat or confirmatory test. Be sure your patients' parent(s) read this, and have an opportunity to ask questions. This is a good time for you to be sure they have identified a post-discharge physician or healthcare provider who will be seeing their baby. If you do not verify they have someone identified in the medical record, any follow-up becomes your responsibility as the attending practitioner.

"We appreciate it so much when medical office staff understand that when they get faxes from the Newborn Screening Program, they need to act quickly. If they haven't seen the patient, they verify with mom, the name of the healthcare provider she is taking baby to for care. (Sometimes they just haven't been in for their first visit yet.) When they notify the state program right away of who the baby's doctor will be, we can get the right information to the right provider and take it from there."

Julie Luedtke, Program Manager



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Helping Parent's Understand

The "Parent's Guide" is available at no charge from DHHS, in the following languages: English and also Spanish, Vietnamese, Chinese, Korean, Anuak, Dinka, Nuer, Karen, Nepali and Somali to help educate parents for whom English is not their primary language. We try to stay current with the most common languages represented by new immigrants and refugees relocating to Nebraska.

In addition the state program provided a DVD in English and Spanish to each hospital to use for patient education. For parents with limited literacy this may be useful. The video is current with the exception of SCID screening.

When to educate?

Ideally provide this to expecting patients during the third trimester. If you are a family physician providing obstetric care, this will help you and the parents later. Some OB/Gyn practices are including the Parent's Guide and/or a one-page NBS intro (also available at no charge) to their patients.

At a minimum during pre-admission registration or during the obstetric in-patient stay, the information must be provided. Hospitals usually support the ordering physician with this by providing the "Parent's Guide" with the packet of information. Hospitalists may wish to work with nursing personnel to be sure the education is completed.

Whose job is it anyway? It is 2:00 am and you are calling me with a positive Congenital Adrenal Hyperplasia (CAH) on a baby I only saw in the hospital. What am I supposed to do next?

Unless you know who the post-discharge physician is, you will need to contact the parents, evaluate the condition of the baby, consult with the pediatric endocrinologist on call, arrange for confirmatory testing and possibly readmit the baby for evaluation. Critical positive results for some conditions represent a neonatal emergency. CAH is just one of them. The NBS follow-up personnel will provide you with the recommended actions from the pediatric subspecialists, and the American College of Medical Genetics ACT Sheets for guidance.

Download your link to the ACMG ACT sheets now, and you'll always have ready access to recommendations for what to do when faced with a critical positive screen result for any of the rare conditions screened on Nebraska's newborn screening panel. As of 2016, twenty nine conditions are screened. Go to: <https://www.acmg.net/ACMG/Publications>

Is this really necessary? The screen was collected too early, but only 10 minutes shy of the 24 hour mark. Do I really have to get a repeat?

Under Nebraska State Law you are responsible for ensuring the screen is completed. Specimens collected at less than 24 hours very frequently have false positive results for Cystic Fibrosis (CF), Congenital Adrenal Hyperplasia (CAH) and Congenital Primary Hypothyroidism (CPH), and CPH results are not considered reliable from specimens collected at less than 24 hours of age. **The screening lab no longer tests specimens collected at less than 24 hours for those conditions, (CF, CAH and CPH) so it is imperative to get the repeat so you have a complete screen.**

Marginalizing Newborn Screening - a Deadly Risk

It can be a deadly mistake to even slightly dismiss the importance of newborn screening. It is true these conditions are rare, and true that we do have false positives ...that is the nature of screening.

Newborn Screening professionals are well aware that we cannot save every baby affected by the diseases on the screening panel. Some times even with the earliest feasible notification, things still go wrong and babies die. This has even happened here in Nebraska. Despite having screening results reported by 3 1/2 days of age, a baby with MCAD was too sick by then to recover.

Because of the urgent need to identify these babies, we continually try to optimize screening.

Every year in Nebraska alone, about 1 in every 560 babies (50 60 *every year*) are found to have a disease through the screening process. Because of early diagnosis, these babies can be entered into treatment optimizing their growth and development.

We all share the responsibility to try to make sure screening happens correctly, in a timely manner, and with the highest quality we can pro



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Are Your Patient's Medical Records Complete?

Most clinics verify at the one or two week well-baby check-up that they have all of the newborn screening results (blood spot, hearing and critical congenital heart disease). If you don't have a copy of the bloodspot screen results or an electronic record of them, there are a number of options available to you. From your affiliated hospitals you may be able to access them through the EMR or request them from medical records.

Alternatively, you can access the results for your patients directly from the PerkinElmer Genetics Inc. (Nebraska's contracted newborn screening laboratory) secure web portal. To get this access contact Susan Felinczak at PerkinElmer Genetics Inc. 412 220-2300 ext 142. You will be asked to complete and sign a form to access your patient's results, and to be set up for a secure account and establish a password. The web portal is simple and easy. You can print and scan or download your patients' bloodspot screen results into your own EMR.

You should not rely on the laboratory or newborn screening program to notify you of the need for follow-up. The program and laboratory may not have the correct information about who is caring for the baby. This can occur if the information is not collected, reported or recorded incorrectly or the mother changes her baby's provider after discharge. Ensuring you have results for every baby by the first well-baby check is one risk management tool all practices should consider adopting.

Where are the Best Resources for Practitioners

The Nebraska Newborn Screening Program has information on its website for practitioners and links to reputable associations and organizations with expertise in those conditions. You'll also find publications, reports, the state law and regulations governing newborn screening on our page. As mentioned earlier, the ACMG has online ACT sheets for just when you need it, before calling and talking with parents about a positive screen result.

Subscribers receive email notices when updates are made to the web page.

Go to: <http://dhhs.ne.gov/publichealth/Pages/nsp.aspx>

A great resource you can direct parents to is "Baby's First Test". It is the website from the National Clearinghouse for Newborn Screening Information. The site include brief informative videos that can significantly help parents understand screening.

<http://www.babysfirsttest.org/>

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Contact Us

Give us a call for more information about our services

NBS Program regulations/QA/402 471 6733

Follow up metabolic, endocrine, specimens drawn early 402 471 0374

Follow up CF, SCID and hemoglobinopathies, unacceptable specimens, 402 471 6558

Parent Education resources 402 471 9731 or order online at: <http://dhhs.ne.gov/publichealth/Pages/OrderLiterature.aspx>

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