Michigan Department of Health and Human Services

Newborn Screening News

Spring 2018

The Michigan Department of Health and Human Services (MDHHS) Newborn Screening Follow-up Program works together with the State Newborn Screening Laboratory and coordinating centers to find and treat infants who need early medical care.

NBS Quarterly Reports and Stellar Performance

During the fourth quarter of 2017, thirteen hospitals met all six of the NBS performance goals. We would like to congratulate the following hospitals on their impressive efforts!

- **Beaumont Hospital—Trenton**
- **Beaumont Hospital—Troy**
- **Crittenton Hospital**
- **Henry Ford Allegiance Health**
- **Lakeland of Niles**
- **McLaren Flint**
- **McLaren Port Huron—Special Care Nursery**
- **Mid-Michigan Medical Center Gratiot**
- **Mid-Michigan Medical Center Midland**
- **Munson Healthcare Cadillac Hospital**
- **Tawas St. Joseph Hospital**
- **Spectrum Health United Hospital**
- **Providence Park Hospital**

Performance Goals for NBS Quarterly Reports

1. <2% of screens are collected >36 hours after birth
2. >90% of screens arrive in the state laboratory by the appropriate day
3. <1% of screens are unsatisfactory
4. >95% of electronic birth certificates have the NBS card number recorded
5. >90% of specimens have a returned BioTrust for Health consent form that is completed appropriately
6. >90% of newborns with a dried blood spot have pulse oximetry screening results reported

We hope you will be able to use information in the quarterly reports to improve your part of the NBS system. If you have any questions, please call the NBS Follow-up Program at 517-335-4181.

Old look

New Look, Same Great Information

The Newborn Screening Program recently redesigned our Newborn Screening brochure. Much of the content has stayed the same, but the look was updated. As hospitals start to run out of the old brochures, they will be replaced with these new brochures. Please let us know if you have any questions once you receive these new brochures.

NBS Follow-up Program Contact Information
Phone: 517-335-4181
Email: newbornscreening@michigan.gov

MDHHS Michigan Department of Health & Human Services
RICK SNYDER, GOVERNOR | NICK LYON, DIRECTOR

ACHDNC Vote on Spinal Muscular Atrophy

On February 8, 2018, the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) voted to recommend the addition of spinal muscular atrophy (SMA) to the Recommended Uniform Screening Panel (RUSP). SMA is an autosomal recessive motor neuron disease, affecting approximately 1 in 11,000 individuals. It is the leading genetic cause of death in infancy. SMA is caused by a defect in the survival motor neuron 1 (SMN1) gene that leads to the loss of motor neurons in the spinal cord and brainstem. As a result, affected individuals develop progressive muscle weakness and wasting, especially involving the voluntary muscles used for head control, sitting, walking, breathing, and swallowing.
There are four main types of SMA, varying in age of onset and clinical severity. The most severe and common form, SMA Type 1, is usually fatal by two years of age if not treated. SMA can be detected in the newborn period by DNA based testing. Approximately 95% of all cases are associated with a specific homozygous deletion in the SMN1 gene. Additional analysis of a nearby gene, SMN2, may be useful in predicting onset and disease severity.

The ACHDNC recommendation follows the introduction of Spinraza, the first FDA-approved treatment for SMA. Infants and children treated with Spinraza have been shown to demonstrate improved achievement and maintenance of motor milestones, decreased need for respiratory support, and increased survival. Early identification of affected infants through newborn screening will allow treatment to begin prior to the onset of symptoms, when Spinraza can be most effective. While the ACHDNC awaits approval of its recommendation from the Director of Health and Human Services, the Michigan NBS Program will begin its review process to consider the addition of SMA to Michigan's newborn screening panel.

Coming Soon: Guanidinoacetate Methyl Transferase (GAMT) Deficiency

Coming soon to Michigan's Newborn Screening Program is guanadinoacetate methyl transferase (GAMT) deficiency. GAMT deficiency is an inborn error of metabolism that affects creatine synthesis. When the guanidinoacetate methyltransferase enzyme is damaged, creatine cannot be synthesized and guanidinoacetate (GAA) accumulates. This creates a shortage of creatine in the body and excessive amounts of GAA which is a neurotoxin. Creatine is an essential metabolite for the brain, heart and muscle. When the body does not get enough creatine and has extra GAA, it can cause developmental delay, speech problems, seizures and behavior issues such as autism and hyperactivity. Lack of early treatment can lead to lifelong cognitive impairments which can be severe. Treatment for GAMT deficiency consists of correcting the creatine deficiency and reducing GAA levels in the body and brain through dietary and medical interventions. Treatment is most effective if started early in life before symptoms arise. Michigan will be the second state in the United States to screen for GAMT deficiency, and screening is anticipated to begin late 2018.

Interfering Substances

An interfering substance is anything that affects a testing method, potentially causing unreliable, misleading, or clinically inaccurate results. This may cause a delay of valid test results that could have a negative impact on the health of the baby.

Some known substances that interfere with testing methods used in newborn screening include:

- Anticoagulants such as EDTA, citrate and heparin
- RBC transfusions (pre and postnatal transfusions)
- Topical anesthetic creams
- Hexachlorophene
- Povidone-iodine
- Parenteral nutrition (TPN)
- Carnitine supplementation
- Dopamine
- Steroids
- Pivalic acid antibiotic therapy

The use of capillary tubes is discouraged for NBS collection for various reasons. For example, if heparinized capillary tubes are used, heparin will interfere with the Mucopolysaccharidosis Type I (MPS I) assay causing an inconclusive test result. Using non-heparinized capillary tubes increases the chance of having an unsatisfactory-clotted sample.
In some, but not all instances, it is possible to detect when an interference is occurring. When an interferent is detected, at minimum, it requires another specimen to be collected. At worst, if an interferent is not detected, an abnormal result can be missed, resulting in serious harm or death.

Interfering substances can result in:

- Delayed valid test results that could have a negative impact on the health of the baby
- Infant distress caused by the need for a repeat specimen collection
- Unnecessary burden on parents who have to bring their baby back for a repeat screen
- Additional work for hospital and NBS staff
- Increased cost to the hospital and NBS program

References

NBS01-A6 Blood Collection on Filter Paper for Newborn Screening Programs; Approved Standard-Sixth Edition July 2013

Coming Soon: Thank You Cards

To provide a tangible reminder to hospital staff about the importance of their efforts to collect the newborn blood spot screen in a timely manner with accurate information, the NBS Follow-up Program will be sending out thank you cards. These thank you cards will be sent monthly to birth hospitals that collected a blood spot screen for an infant who confirmed with a disorder on the NBS panel. Please display these cards for your staff to see because their work helped connect a baby to specialty care providers for treatment in order to achieve the best possible health outcome.

Last month, the Michigan Department of Health and Human Services shared Mila's story on its Facebook page. Mila's experience highlights the importance of the mandatory pulse oximetry screening for critical congenital heart disease. Find out how pulse oximetry screening for critical congenital heart disease helped save Mila's life:
https://healthblog.uofmhealth.org/childrens-health/newborn-screening-catches-babys-critical-heart-condition

In case you missed it:

Upcoming Holidays:

Lower Peninsula Hospitals:
STAT will pick up your specimens following the Sunday pickup schedule on the following holidays:
Memorial Day, Monday, May 28
Independence Day, Wednesday, July 4
Labor Day, Monday, September 3

Upper Peninsula Hospitals:
There will be no UPS pickups on the following holidays:
Memorial Day, Monday, May 28
Independence Day, Wednesday, July 4
Labor Day, Monday, September 3

Submitter Code: Please remember that staff members need to record the correct hospital submitter code on the newborn screening card. The last digit indicates the type of nursery: 0 means regular nursery, 1 means NICU, and 2 means SCN. The submitter code is used to make separate quarterly reports for each unit, so it's important that we can correctly identify which unit submitted each specimen.

Phone Number: Please remember only one phone number per provider should be listed on the newborn screening card. Many requestors are using cell phone numbers or multiple different phone numbers for lines in provider offices and the laboratory is receiving new entries every day. Please have hospital staff clean up these lists and include one phone number per provider. Only include on the newborn screening card the doctor who is in charge of the baby's care when the baby leaves the hospital.

Birth Weight: Please remember to record birth weight in **grams** on the first sample newborn screening card. Enter the current weight in grams on the repeat sample card.

TECHNICAL ASSISTANCE

Lois Turbett, NBS nurse consultant, is available to work with staff in any hospital that requests help with specimen collection. She can be reached toll-free at (866) 673-9939 or by email at turbettl@michigan.gov to answer your questions. Kristen Thompson, NBS Coordinator is also available to work with hospitals on CCHD pulse oximetry screening and reporting and can be reached at thompsonk23@michigan.gov. Together we can achieve our goal that all children diagnosed through newborn screening receive prompt and careful treatment in order to live the healthiest lives possible.

Please remember to share the quarterly newsletter with staff!

If you have questions please contact the NBS Follow-up Program at 517-335-4181 or newbornscreening@michigan.gov or visit our website at www.michigan.gov/newbornscreening