



Michigan Department of Health and Human Services

Newborn Screening News

Summer 2017

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.

National Discussion of Analyte Cutoffs

With increasing attention on false negatives in newborn screening (NBS), the federal Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) has been discussing how analyte cutoffs are set for NBS. Laboratory and follow-up staff from NBS programs across the country presented at the February and May ACHDNC meetings. If you are interested in viewing those, the presentations from the February meeting are available here: <https://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/meetings/2017/0209/index.html>.

When new disorders are added to the NBS panel, appropriate analyte cutoffs must be selected. Analyte concentrations may vary based on the newborn's age at time of specimen collection, birth weight, race/ethnicity, or feeding method. The reagents, laboratory instrumentation, and screening method also affect analyte measurement. Since all these factors may differ state to state, analyte cutoffs are unique to each state.

When on-boarding a new disorder, laboratory staff use bloodspots from known confirmed cases and controls. Population validation studies are also conducted. The analyte values from this initial work are analyzed statistically. Laboratory and follow-up staff consult with specialists to review the statistical analysis and determine cutoffs.

Once the disorder is added to the NBS panel, NBS Program staff perform continuous quality monitoring to review the assay performance and make adjustments to the cutoffs if appropriate. The best approach to optimize screening performance metrics is to have regular review with the entire NBS system, comprised of stakeholders from the NBS Lab and Follow-up Program and clinical specialists caring for identified patients. Michigan's NBS Program follows that approach. Disease-specific committees meet two-three times per year to review follow-up and treatment protocols and monitor testing and interpretation of screening results. The NBS Program staff and clinical partners are focused on ensuring that the analyte cutoffs used will optimize screening performance metrics for Michigan's newest residents.

Update on Pompe Disease and MPS I Screening in Michigan:

Pompe Disease and Mucopolysaccharidosis Type I (MPS I) are lysosomal storage disorders. People with these conditions lack certain enzyme function. The result is a build up of large molecules in the lysosomes, which damages body tissues and organs. Pompe disease affects about 2-3 in 100,000 babies. MPS I affects about 1 in 100,000 babies.

Babies with Pompe disease may be born with heart disease. They develop progressive muscle weakness affecting their ability to walk and breathe. Babies with severe Pompe disease rarely live beyond their first year or two. MPS I causes progressive cognitive impairment and progressive problems with bones and joints. The life expectancy for a child with severe MPS I is 3 to 4 years. However, with newborn screening, affected babies can begin early treatment in the first weeks of life. Pompe disease and MPS I have been added to the Michigan newborn screening panel. The MDHHS NBS lab began screening in July. While Michigan specific cutoffs are being validated, only positive screening results will be reported.

NBS Follow-up Program Contact Information

Phone: 517-335-4181

Email: newbornscreening@michigan.gov

Coming Soon: CCHD Screening Quarterly Reports

The Newborn Screening (NBS) Program will begin to send out critical congenital heart disease (CCHD) screening quarterly quality assurance reports. These reports are similar to the NBS quarterly reports, but will focus on screening for CCHD.



The first metric on the report is the percent of CCHD screens reported. Our goal is for at least 90% of infants with a dried blood spot screen to have a CCHD screen reported. Infants who have a valid reason as to why their screen was not completed (for example, prenatal diagnosis of CCHD or echocardiogram) are counted as reported. Increasing the percent of CCHD screens reported will help the NBS Program with follow-up efforts and evaluation of the CCHD algorithm.

The second metric on the report is the percent of CCHD screens reported on time. CCHD screens reported within 10 days of screening are considered on time. Our goal is for at least 90% of infants with a dried blood spot screen to have a CCHD screen reported within 10 days after screening. It is important that screens are reported in a timely manner so that the NBS Program can follow-up on failed cases and determine their outcome. Determining the outcome and final diagnosis of infants with a failed screen enables the NBS Program to evaluate the effectiveness of CCHD screening.

The third metric on the report is the percent of CCHD screens conducted within the recommended time frame after birth. All infants should be screened as close to 24 hours after birth as possible, in order to allow early intervention and decrease the false positive rate. CCHD screens completed between 20 and 28 hours after birth are considered on time. Our goal is for at least 90% of infants with a dried blood spot screen to have a CCHD screen conducted between 20 and 28 hours after birth.

The report will have a summary of the CCHD screening results at your hospital for the current quarter. Each birth hospital will receive the number of passes, rescreens, and fails as a result of all infant's first CCHD screens. Each hospital will also be provided with a list of algorithm compliance errors. These errors include missing rescreens, any rescreens conducted after a failed screen, and any rescreens conducted after a passed screen.

Thank you for all the work you do to improve CCHD screening and reporting. We hope the new CCHD quarterly reports will be a useful tool. If you have any questions about the CCHD quarterly report, please contact Kristen Thompson at ThompsonK23@michigan.gov.

Newborn Screening False Negatives in the News

In December 2016, the Milwaukee Journal Sentinel released a story by journalist Ellen Gabler called "The price of being wrong" (<http://projects.jsonline.com/news/2016/12/11/the-price-of-being-wrong.html>). The story discusses false negatives in newborn screening.

Michigan's goal is to find the delicate balance between identifying all true disease cases through screening while minimizing the number of infants (and families) affected by false positive findings. Our laboratory and follow-up staff work closely with clinicians on our advisory committees to develop and evaluate screening cutoffs. In the event of a confirmed false negative case (a very rare event), concern raised by families or clinicians, or findings from published articles relevant to NBS, Michigan program staff routinely review screening performance metrics. This review is presented at the appropriate disease-specific advisory committee for discussion and recommendations.

The NBS Program encourages all families to follow the recommended schedule of well-child visits to maintain contact with their child's primary care provider. The NBS Program educates primary care providers that newborn screening is a screen and should not replace clinical judgment, as noted on the NBS results mailer which contains the language, "If there is a clinical concern, diagnostic testing should be initiated."



We appreciate the concerns highlighted in the story and will continue to monitor our process in Michigan to encourage continuous quality improvement efforts that make the NBS experience as accurate as possible for Michigan's newborns and their families.

Thank you for all you do for NBS in Michigan!

NBS Card Replacement Process

Are You Throwing Money Away??? is the title of an article that appeared in the *Newborn Screening News Summer 2013* edition. It provided instructions on how to request a card replacement and reminded that cards sent to the NBS Laboratory for replacement may result in delayed replacement cards. However, hospital staff have continued to send cards marked 'void', 'credit', and 'parent refused' to the NBS Laboratory along with their bloodspot specimens.

To guarantee card replacement in a timely manner, cards must be submitted correctly. The *Newborn Screening Card Replacement Form* is located in the *Michigan Department of Health and Human Services Newborn Screening Guide for Hospitals*, which can be accessed through the [NBS website](#). The form is in Appendix 7 (pg 39) of the guide. Instructions for completing and mailing the request are on the form.

Points to remember:

- DO** mail just the white face sheet, not the remaining portions of the kit such as the filter paper
- DO** send 10 face sheets or less if you want a quicker turnaround time
- DO** place the face sheet in a biohazard bag if there is blood on it.
- DO NOT** send your replacement request to the NBS laboratory
- DO NOT** use courier envelopes for your replacement request
- DO NOT** use the NBS UPS account for your replacement request



NBS Quarterly Reports and Stellar Performance

During the 1st quarter of 2017, ten hospitals met all six of the NBS performance goals. We would like to congratulate the following hospitals on their impressive efforts!

- **Beaumont Hospital—Dearborn**
- **Beaumont Hospital—Trenton**
- **Holland Hospital**
- **Holland Hospital—SCN**
- **McLaren Greater Lansing**
- **Munson Healthcare Cadillac Hospital**
- **Otsego Memorial Hospital**
- **Spectrum Health Gerber Memorial**
- **Sturgis Hospital**
- **Three Rivers Area 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.

These are the upcoming State holidays:

Specimen Pick Up Schedule for Labor Day, September 4th

The State NBS Laboratory and Follow-up office will be open on Labor Day. STAT Courier will pick up specimens from Lower Peninsula hospitals following the Sunday pickup schedule. UPS **will not** pick up specimens from Upper Peninsula hospitals on Labor Day.



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 appropriate 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