Michigan Department of Health and Human Services

Newborn Screening News

Summer 2019

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 first quarter of 2019, one hospital met all six of the NBS performance goals. We would like to congratulate the following hospital on their impressive efforts!

- **Henry Ford Wyandotte Hospital-SCN**

There were fewer stellar performers this quarter because of the incorrect date/time metric. In the first quarter of 2019, 2.7% of specimens collected statewide had date and/or time errors. Many of these errors were due to the incorrect year being written on the card.

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
7. <1% of specimens have errors in birth date/time and/or specimen collection date/time on the NBS card

We encourage you to use the 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.

In this Newsletter:

Save the Date: 2019 Newborn Screening Training

The Newborn Screening Program will be hosting two newborn screening trainings for hospital staff this fall. The trainings will cover a variety of topics including: tips for collecting a satisfactory blood spot specimen, spinal muscular atrophy, Michigan BioTrust for Health, follow-up on positive screens for metabolic disorders, critical congenital heart disease, and more! Lunch will be provided and nursing contact hours are available.

- October 17, 2019—St. Joseph Mercy Oakland (Pontiac)
- October 23, 2019—State Secondary Complex (Lansing)

September is Newborn Screening Awareness Month!

Thank you for your hard work ensuring that all babies in Michigan get their newborn screen.

Because of you…

- 108,277 babies received a newborn screen in 2018.
- Each year, approximately 300 babies are identified with a disorders and given the opportunity for better health outcomes.

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

MDHHS Michigan Department of Health & Human Services

Three New Disorders for Newborn Screening

Anticipated late summer 2019, all newborns in Michigan will be screened for X-linked adrenoleukodystrophy (X-ALD), guanidinoacetate methyltransferase (GAMT) deficiency, and adenosine deaminase severe combined immunodeficiency (ADA-SCID). The Newborn Screening Laboratory will use a tandem mass spectrometry method for all three of these disorders using the dried filter paper blood spots collected as part of the current newborn screening process. An additional punch from the dried blood spots will not be needed for these disorders.

X-ALD: X-ALD is the most common peroxisomal disorder, primarily affecting males. Infants with positive newborn screens for X-ALD will be referred to the follow-up coordinating center at Michigan Medicine to arrange diagnostic testing and medical management. It is anticipated that newborn screening will detect approximately 7-9 affected infants per year in Michigan, including both males with X-ALD and female carriers. Early diagnosis allows boys to be monitored pre-symptomatically to ensure treatment is initiated at the appropriate time to be most successful.

GAMT Deficiency: GAMT deficiency is an inborn error of metabolism that affects creatine synthesis. When the GAMT 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, developmental delay, speech problems, seizures and behavior issues such as autism and hyperactivity may occur. 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.

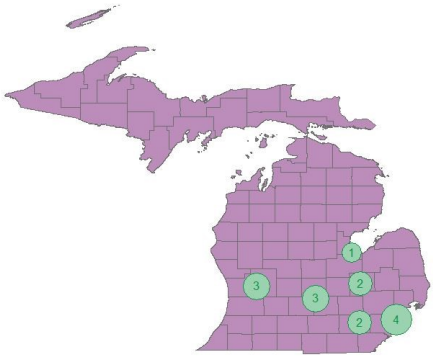
Michigan will be the third state to screen for GAMT deficiency, after Utah and New York.

ADA-SCID: All Michigan newborns have been screened for most types of severe combined immunodeficiency (SCID) since October 2011. SCID represents a group of inherited disorders that lack functional T cells and B cells responsible for cellular and humoral immunity, respectively. Newborns with SCID are unable to mount an immune response to infection by viruses, bacteria and fungi. Treatment options for SCID vary and may include preventive medications for certain types of pneumonia, intravenous immunoglobulin, enzyme replacement therapy, hematopoietic stem cell transplantation, and gene therapy.

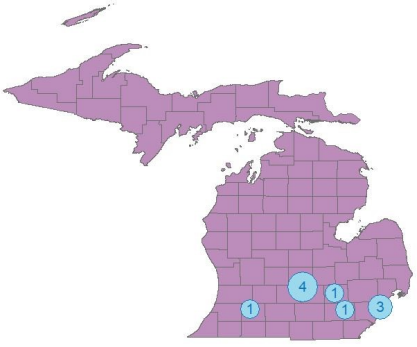
Currently the screening method for SCID and other primary immune deficiency disorders is the enumeration of T-cell receptor excision circles (TRECs) using real time polymerase chain reaction. A very low or absent TREC number could indicate SCID or other primary immune deficiency disorders. However, the TREC number is not always a good indicator for one type of SCID, caused by adenosine deaminase (ADA) deficiency.

ADA-SCID is an inherited autosomal recessive disorder of purine metabolism. Nucleosides adenosine (ADO) and 2'deoxyadenosine (dADO) are biomarkers for this disorder and will be elevated in affected newborns.

NBS Provider Educational Events, 2018-2019



NBS General Public Educational Events, 2018-2019



In 2018 and 2019, the Newborn Screening (NBS) Program exhibited at fifteen provider educational events and ten general public educational events in six and five cities, respectively.

Provider events allow us to educate healthcare professionals involved with the NBS process. If you know of any opportunities for the NBS Program to speak to healthcare professionals involved with the screen, please contact Lois Turbett, NBS Nurse Consultant, at turbettl@michigan.gov.

General public educational events allow the Program to increase parents' awareness of the newborn screen. If you know of any events that prospective and/or new parents regularly attend, please share them with Kristen Thompson, Newborn Screening Coordinator, at thompsonk23@michigan.gov.

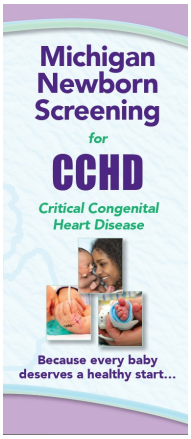
Critical Congenital Heart Disease (CCHD) Screening Quarterly Report Ten Day Metric

All newborns should have pulse oximetry screen results reported to the state less than 10 days after the screen is conducted. This is one of the metrics on the new CCHD quarterly report. To meet this metric, your hospital must submit at least 90 percent of pulse oximetry results to the state within that timeframe. It is vital that every newborn receives a pulse oximetry screen and those results are sent to the Newborn Screening Program. Timely reporting of the screening results enables the Program to identify and follow-up on infants who potentially did not receive a screen or to ensure infants who failed the screen are receiving appropriate follow-up care. The table below is an example of what this metric looks like on the quarterly report. We appreciate your staff and all of your efforts in ensuring this program is a success. Kristen Thompson, Newborn Screening Program Coordinator, is available to assist with this reporting process. If you are concerned about any particular obstacles in reporting the results of the pulse oximetry screen, please contact Kristen at thompsonk23@michigan.gov or 517-284-4992.

Reported on Time: All newborns with a right hand and foot pulse oximetry screen reported to the state less than 10 days after screen date divided by the total number of newborns with a bloodspot screen. (Target = 90%)

	Oct	Nov	Dec	Total	Goal Met
Screen Reported on Time—Your Hospital (N)					
Births—Your Hospital (N)					
Percent Reported on Time—Your Hospital (%)					
Percent Reported on Time—State (%)					

Translations for the CCHD Brochure and Parent Directive Form



The *Critical Congenital Heart Disease (CCHD) Brochure* and *Residual Newborn Screening Dried Blood Spot Directive* are available in English, Spanish, and Arabic.

The CCHD brochure has a new look! This brochure provides an overview of newborn screening for CCHD for expecting and new parents. These brochures can be ordered free of charge through our online ordering system (www.michigan.gov/nbsorders).

The *Residual Newborn Screening Dried Blood Spot Directive* form allows families to request that the Newborn Screening Program destroy their child's blood spots once testing is completed. These forms are available on the newborn screening website (www.michigan.gov/newbornscreening) in the "Resources for Hospitals and Health Professionals" tab. Please see the links below for direct access to the translated documents:
Arabic: https://www.michigan.gov/documents/mdhhs/MDHHS-5683-AR_PDF_654685_7.pdf
Spanish: https://www.michigan.gov//documents/mdhhs/MDHHS-5683-SP_PDF_654687_7.pdf

Fall Newborn Screening Conference Topic Highlight:

The state newborn screening laboratory received 118,142 first and repeat sample specimens in 2018. Of those, 1,515 (1.3 percent) were deemed unsatisfactory, leading to inconclusive results and a request for a repeat screen. *How to Collect a Satisfactory Blood Spot Specimen* is a topic that will be covered during the NBS fall conferences. Beaumont Hospital – Troy Family Birth Center submitted less than 0.2 percent unsatisfactory blood spot specimens in 2018 and Bronson Methodist Birth Place submitted less than 0.3 percent. Both hospitals will share best practices on how to collect a satisfactory blood spot specimen, how to assess specimen quality, what to do if the specimen is unsatisfactory, and how to support and train staff that struggle with blood spot collection. Staff from Beaumont Troy will speak on October 17 and staff from Bronson will speak on October 23.

Upcoming Holiday Courier Schedule

Lower Peninsula Hospitals:
Thursday, July 4, 2019 – holiday/Sunday schedule
Monday, September 2, 2019 – holiday/Sunday schedule

Upper Peninsula Hospitals:
Thursday, July 4, 2019 – no UPS pickup
Monday, September 2, 2019 – no UPS pickup

Important Reminder

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