Michigan Department of Health and Human Services

Newborn Screening News

Spring 2020

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 2019, three hospitals met all six of the NBS performance goals. We would like to congratulate the following hospitals on their impressive efforts!

- **Beaumont Hospital – Troy**
- **Bronson Methodist Hospital**
- **Henry Ford Allegiance Health**

Performance Goals for NBS Quarterly Reports

1. less than 2% of screens are collected >36 hours after birth
2. greater than 90% of screens arrive in the state laboratory by the appropriate day
3. less than 1% of screens are unsatisfactory
4. greater than 95% of electronic birth certificates have the NBS card number recorded
5. greater than 90% of specimens have a returned BioTrust for Health consent form that is completed appropriately
6. greater than 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.

In this newsletter:

Staff Highlights: Two New Newborn Screening Follow-up Technicians

We are delighted to introduce our two new follow-up technicians. The follow-up technicians answer the main Newborn Screening phone line Monday-Saturday and many holidays. If the follow-up technicians cannot answer your question, they will refer you to the appropriate person. They respond to phone and fax requests related to NBS results. They notify physicians and parents when a repeat newborn screen is necessary. They also notify physicians and clinical coordinating centers when a newborn needs to have confirmatory testing.

Becky began as a Newborn Screening Follow-up Technician in November 2019. Becky has a bachelor's degree from Michigan State University and worked at a rehabilitation center prior to joining the NBS Program.

Staci began as a Newborn Screening Follow-up Technician in February 2020. Staci worked in the laboratory central processing area of McLaren Greater Lansing Hospital for many years and has experience collecting newborn screens.

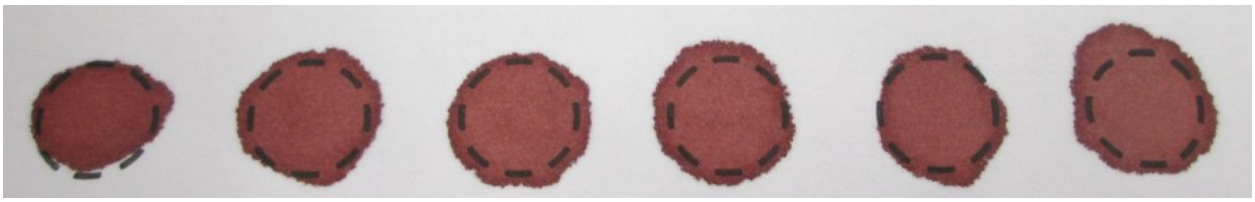
Welcome Becky and Staci!

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

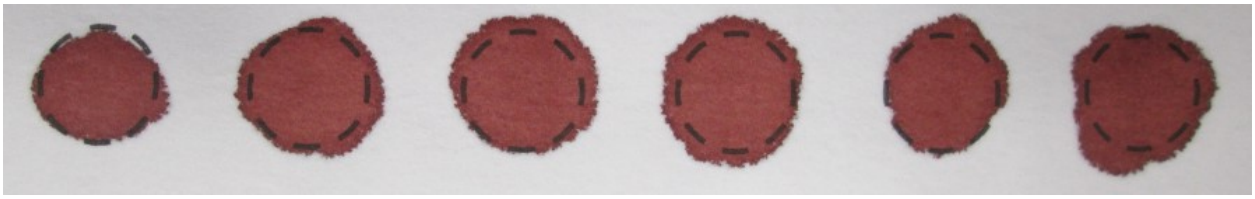
MDHHS Michigan Department of Health & Human Services

Satisfactory Blood Spot Specimens

The NBS Laboratory received 2,643 unsatisfactory first and repeat sample specimens in 2019. The unsatisfactory rate of 2.3 percent was almost double that of 2018, which was 1.3 percent.



This is the front of a satisfactory blood spot specimen, collected by staff from Michigan Medicine Brandon NICU.



This is the back of the same satisfactory specimen. Note the even saturation of one drop of blood that had been applied to each circle. Always assess the quality of the front and back of each blood spot specimen.

A satisfactory blood spot will yield four, sometimes five punches. There must be a minimum of fifteen available punches for the specimen to be satisfactory.

Uniform blood saturation is essential for accurate test interpretation. Too little or too much blood can lead to false positive or false negative determinations.

Points to remember when collecting the NBS specimen:

1. Wipe away the first drop of blood.
2. Apply *only* one large drop of blood to each preprinted circle.
3. Apply blood to one side of the card *only*.
4. Make sure the blood has soaked through to the other side.

Unsatisfactory specimens can result in:

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

Newborn Screening System Quality Improvement Project

The NBS Program was recently awarded funding for a one-year quality improvement project. The project will focus on educating expectant parents to increase parental understanding of:

- The importance of NBS
- Their role throughout the NBS process
- Their choices after NBS is complete

In order to equip parents to more actively participate in the NBS process, NBS Program staff will develop a new educational handout for expectant parents. This handout will provide a checklist of options and responsibilities parents have throughout the NBS process. The NBS Program will be partnering will three birth hospitals around the state that will distribute this handout during pre-registration tours. To determine the effectiveness of the new document, the NBS Program will survey mothers who delivered at one of the participating hospitals before and after introduction of the new handout to determine what percent completed various action items described in the handout.

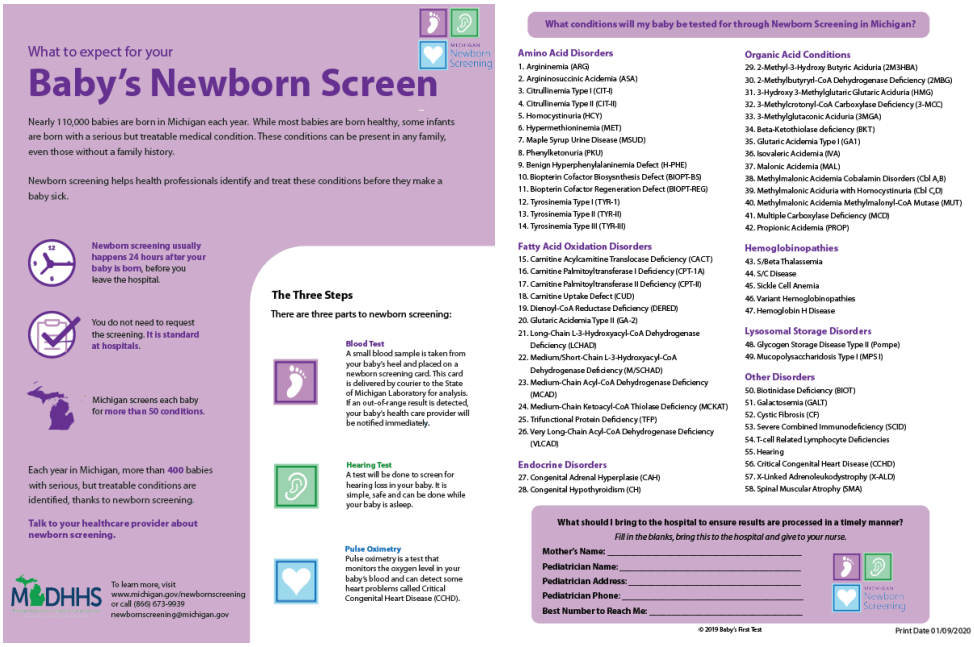
Look for updates on the project findings in future newsletters!

Critical Congenital Heart Disease Screening Algorithm Non-Compliance Follow-up

The Newborn Screening Program has implemented an additional follow-up procedure in situations where an infant was not screened in accordance with the critical congenital heart disease (CCHD) screening algorithm. Your hospital will now receive email notification if an infant did not receive a necessary rescreen or received a rescreen after failing the first screen. If you receive one of these emails, please respond and provide additional information if possible. If the baby was discharged before a CCHD could be ruled out, please promptly notify the infant's PCP, so the appropriate next steps can be taken.

A New Resource for Parents!

The Newborn Screening Program has created a new resource for parents to learn more about newborn screening. The "What to Expect" document, which is meant to complement the Newborn Screening Roadmap, is now available to order online free of charge (www.Michigan.gov/nbsorders). This document goes into more detail about each step of the newborn screen and has a complete list of disorders on Michigan's newborn screening panel. In addition, there is a spot for parents to write down the pediatrician information for their newborn. Having accurate pediatrician contact information ensures NBS Program staff can follow-up up in a timely manner if a repeat screen or additional testing is needed. The "What to Expect" document is great for expecting parents and could be handed out at prenatal classes or hospital tours.



What to expect for your

Baby's Newborn Screen

Nearly 110,000 babies are born in Michigan each year. While most babies are born healthy, some infants are born with a serious but treatable medical condition. These conditions can be present in any family, even those without a family history.

Newborn screening helps health professionals identify and treat these conditions before they make a baby sick.

Newborn screening usually happens 24 hours after your baby is born, before you leave the hospital.

You do not need to request the screening. It is standard at hospitals.

Michigan screens each baby for more than 50 conditions.

Each year in Michigan, more than 400 babies with serious, but treatable conditions are identified, thanks to newborn screening.

Talk to your healthcare provider about newborn screening.

MDHHS To learn more, visit www.michigan.gov/newbornscreening or call (866) 673-9939 newbornscreening@michigan.gov

The Three Steps

There are three parts to newborn screening:

Blood Test
A small blood sample is taken from your baby's heel and placed on a newborn screening card. This card is delivered by courier to the State of Michigan Laboratory for analysis. If an out-of-range result is detected, your baby's health care provider will be notified immediately.

Hearing Test
A test will be done to screen for hearing loss in your baby. It is simple, safe and can be done while your baby is asleep.

Pulse Oximetry
Pulse oximetry is a test that monitors the oxygen level in your baby's blood and can detect some heart problems called Critical Congenital Heart Disease (CCHD).

What conditions will my baby be tested for through Newborn Screening in Michigan?

Amino Acid Disorders
1. Argininemia (ARG)
2. Argininosuccinic Acidemia (ASA)
3. Citrullinemia Type I (CIT-I)
4. Citrullinemia Type II (CIT-II)
5. Homocystinuria (HCY)
6. Hypermethioninemia (MET)
7. Maple Syrup Urine Disease (MSUD)
8. Phenylketonuria (PKU)
9. Benign Hyperphenylalaninemia Defect (H-PHE)
10. Biopterin Cofactor Biosynthesis Defect (BIOPT-BS)
11. Biopterin Cofactor Regeneration Defect (BIOPT-REG)
12. Tyrosinemia Type I (TYR-1)
13. Tyrosinemia Type II (TYR-II)
14. Tyrosinemia Type III (TYR-III)

Fatty Acid Oxidation Disorders
15. Carnitine Acylcarnitine Translocase Deficiency (CACT)
16. Carnitine Palmitoyltransferase I Deficiency (CPT-1A)
17. Carnitine Palmitoyltransferase II Deficiency (CPT-II)
18. Carnitine Uptake Defect (CUD)
19. Dienoyl-CoA Reductase Deficiency (DERED)
20. Glutaric Acidemia Type II (GA-2)
21. Long-Chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
22. Medium/Short-Chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency (M/SCHAD)
23. Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
24. Medium-Chain Ketoacyl-CoA Thiolase Deficiency (MCKAT)
25. Trifunctional Protein Deficiency (TFP)
26. Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)

Endocrine Disorders
27. Congenital Adrenal Hyperplasia (CAH)
28. Congenital Hypothyroidism (CH)

Organic Acid Conditions
29. 2-Methyl-3-Hydroxy Butyric Aciduria (2M3HBA)
30. 2-Methylbutyryl-CoA Dehydrogenase Deficiency (2MBG)
31. 3-Hydroxy 3-Methylglutaric Glutaric Aciduria (HMG)
32. 3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)
33. 3-Methylglutaconic Aciduria (3MGA)
34. Beta-Ketothiolase deficiency (BKT)
35. Glutaric Acidemia Type I (GA1)
36. Isovaleric Acidemia (IVA)
37. Malonic Acidemia (MAL)
38. Methylmalonic Acidemia Cobalamin Disorders (Cbl A,B)
39. Methylmalonic Aciduria with Homocystinuria (Cbl C,D)
40. Methylmalonic Acidemia Methylmalonyl-CoA Mutase (MUT)
41. Multiple Carboxylase Deficiency (MCD)
42. Propionic Acidemia (PROP)

Hemoglobinopathies
43. S/Beta Thalassemia
44. S/C Disease
45. Sickle Cell Anemia
46. Variant Hemoglobinopathies
47. Hemoglobin H Disease

Lysosomal Storage Disorders
48. Glycogen Storage Disease Type II (Pompe)
49. Mucopolysaccharidosis Type I (MPS I)

Other Disorders
50. Biotinidase Deficiency (BIOT)
51. Galactosemia (GALT)
52. Cystic Fibrosis (CF)
53. Severe Combined Immunodeficiency (SCID)
54. T-cell Related Lymphocyte Deficiencies
55. Hearing
56. Critical Congenital Heart Disease (CCHD)
57. X-Linked Adrenoleukodystrophy (X-ALD)
58. Spinal Muscular Atrophy (SMA)

What should I bring to the hospital to ensure results are processed in a timely manner?
Fill in the blanks, bring this to the hospital and give to your nurse.

Mother's Name: _____
Pediatrician Name: _____
Pediatrician Address: _____
Pediatrician Phone: _____
Best Number to Reach Me: _____

Print Date 01/09/2020

BioTrust Training

The Michigan BioTrust for Health has recently created an updated online training module for the BioTrust consent process! This training is great for both new and existing staff members! It provides updated information about the BioTrust program, the consent process, and BioTrust resources that are available for health professionals and families. Though this course does not award any continuing education units (CEU), certificates of completion can be requested by emailing biotrust@michigan.gov.

Find the course at Michigan.gov/BioTrust. Click on "Resources for hospitals and health professionals", then scroll down to "*Trainings*", and click on "*Michigan BioTrust for Health Parental Consent Process*".

MDHHS Adds Spinal Muscular Atrophy to Newborn Screening

Beginning March 9, Michigan babies with spinal muscular atrophy (SMA) can be diagnosed early thanks to the addition of SMA to the state's newborn screening panel. SMA is an autosomal recessive neurodegenerative disorder affecting approximately 1 in 11,000 births. It is caused by a defect in the survival motor neuron 1 (SMN1) gene which leads to the loss of motor neurons in the spinal cord. Individuals with SMA develop progressive muscle weakness and wasting typically involving muscles that control mobility, breathing, and swallowing.

The clinical presentation of SMA can vary in terms of severity and onset. Until recently, the most severe and common form was usually fatal by two years of age. However, new FDA-approved therapies are now changing the course of the disease and giving affected individuals the best chance for improved clinical outcomes. Spinraza, the first available therapy, has been approved to treat both pediatric and adult patients with SMA. Spinraza enhances production of the SMN protein by targeting SMN2, a gene similar to SMN1. Another approved treatment, Zolgensma, is a one-time gene therapy that works by replacing the nonworking SMN1 gene with a new functional copy.

Newborn screening for SMA will be performed using a DNA-based test that identifies a specific deletion in the SMN1 gene that accounts for approximately 95% of all cases. The Newborn Screening SMA follow-up coordinating center at Michigan Medicine will be responsible for ensuring that babies with positive newborn screens receive confirmatory testing and appropriate clinical management at one of the designated MDA referral centers across the state. Early diagnosis will allow treatment to be initiated pre-symptomatically or at the first signs of disease, when it can be most effective.

To learn more about SMA, visit www.babysfirsttest.org.

Upcoming Holiday Courier Schedule:

Lower Peninsula Hospitals:
Monday, May 25, 2020 – holiday/Sunday schedule
Friday, July 3, 2020 – holiday/Sunday schedule

Upper Peninsula Hospitals:
Monday, May 25, 2020 – no UPS pickup
Saturday, July 4, 2020 – no UPS pickup

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 Michigan.gov/NewbornScreening