



# Michigan Department of Community Health

# Newborn Screening News

## Spring 2015

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

## Thank you, Hospital Staff!!

Throughout 2015, Michigan will be celebrating its 50th Anniversary of Newborn Screening. This yearlong celebration will acknowledge all of the hard work done by hospital staff, the Michigan Newborn Screening Laboratory, Follow-Up and Medical Management centers as well as by the pioneers of Michigan screening, all of which has led to improving and saving the lives of newborns across the state.

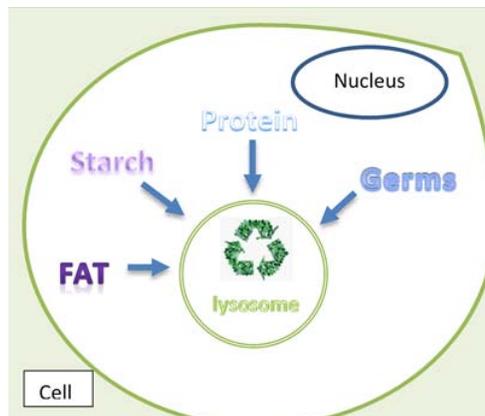
If you would like to host an event during the year or want materials to post around your hospital, please contact Kristy Tomasko at (517) 241-0332 or by email at [tomaskok@michigan.gov](mailto:tomaskok@michigan.gov).

Events will be occurring across the state. Up to date details can be found at: [www.michigan.gov/newbornscreening](http://www.michigan.gov/newbornscreening).

### National NBS News: Mucopolysaccharidosis Type 1 (MPS1) Recommended for the National NBS Panel

The Discretionary Advisory Committee on Heritable Disorders in Newborns and Children (DACHDNC) is the national group that considers new conditions nominated for inclusion on the Recommended Uniform Screening Panel (RUSP). In February 2015, the DACHDNC heard final scientific and clinical evidence and testimony from parent advocates regarding MPS1. The DACHDNC voted to recommend MPS1 to be included on the RUSP. The proposal is made to the U.S. Secretary of the Department of Health and Human Services, who makes the final decision to add, decline or refer to Interagency Coordinating Committee for additional discussion regarding implementation.

MPS1 is a rare inborn progressive condition, affecting about 1/100,000 individuals. Low activity of the enzyme  $\alpha$ -L-iduronidase (IDUA) causes MPS1. When IDUA is not working, certain macromolecules, called mucopolysaccharides, build to toxic levels. IDUA is active in a special part of the cell, an organelle called a lysosome. MPS1 is one of several dozen disorders caused by problems with lysosomal enzymes. Together, these conditions are called lysosomal storage disorders. MPS1 varies in severity and symptoms can include: heart disease, obstructive airway disease, hearing loss, loss of developmental milestones, skeletal dysplasia, joint stiffness, decreased vision, and coarsening of facial features. In severe cases, babies develop symptoms in the first few months of life and die, typically due to cardiac and respiratory disease, in early childhood. There is no cure, but stem cell transplant and ongoing enzyme replacement therapy are treatments available for MPS1.



#### About Lysosomes

Lysosomes are the cells' recycling and disposal centers. They are full of enzymes, like  $\alpha$ -L-iduronidase (IDUA) that specializes in breaking down large molecules. To do their jobs, these enzymes need the special environment found inside the lysosome.

#### NBS Follow-up Program Contact Information

Phone: 517-335-4181

Email: [newbornscreening@michigan.gov](mailto:newbornscreening@michigan.gov)

# A Note of Appreciation from Mike Finkel

It is so easy to take for granted those that helped you become who you are today. While I credit my amazing parents for all of their support and understanding throughout the years, there is also a special group of individuals who helped shape my life's goals.

I was born in 1989 and diagnosed with Classic phenylketonuria (PKU). Without newborn screening and everyone involved in those first crucial hours, my life would have turned out very differently. I would have missed out on so many wonderful opportunities. Let me share my story to illustrate how your work supporting newborn screening empowers those of us with a positive screen.

Through the care of the specialists at Children's Hospital of Michigan Metabolic Clinic, I avoided developmental delays once common in PKU patients. Having established success in the classroom, I was afforded the opportunity to attend Boston University for my undergraduate education. My time in Boston became such a valuable experience for me, both personally and professionally.

It was there, for instance, that I met my extraordinary wife, Kelsey (pictured with me on the right). We were married in June of 2013 and now reside in Mount Pleasant with our rascal-of-a-dog, Alana.

Dealing with PKU had always made me question how our bodies work at the most basic level. This intrigue led me to conduct biomedical research during college, eventually earning me my first scientific publication. I went on to graduate in 2011 with a degree in Biochemistry and Molecular Biology, and shortly thereafter was employed by University of Michigan College of Pharmacy as a research scientist.

During this time I began exploring the field of Public Health - after all, newborn screening has been very influential on my life! In 2013 I began as an intern at Michigan Department of Community of Health (MDCH) working with the Newborn Screening Program. One project I am involved in is the Diet for Life initiative to eliminate barriers to treatment for individuals living with inborn errors of metabolism.

I am now pursuing my Master of Public Health (MPH) degree from Michigan State University and expect to finish next December. One day I hope to work in preventive medicine, implementing strategies to reduce our nation's chronic disease burden.

So, on behalf of everyone in the state living with newborn screening disorders, thank you for giving us the chance to live full lives and realize our dreams!



Read more from families who are thankful for all the work hospital staff do to make newborn screening possible.  
[http://www.michigan.gov/mdch/0,4612,7-132-2942\\_4911\\_4916-308866-.00.html](http://www.michigan.gov/mdch/0,4612,7-132-2942_4911_4916-308866-.00.html)

**Makayla**



**Kreger family**



**Samantha**



## Spotlight on Unsatisfactory Specimens - Contaminated Specimens

The State NBS Laboratory received over 2,000 unsatisfactory specimens in 2014. Sometimes specimens become contaminated during the collection process.

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 *only* one side of the card
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
- Increased cost to the hospital



It appears this blood spot became contaminated after the blood had dried and something wet dropped on the card.



This blood spot may have become contaminated during the drying process when the card was placed on a wet surface.



Specimens can appear 'normal' before testing but unusual test results indicate the specimen had been contaminated during collection. This seems to happen more frequently in the NICU, perhaps since line draws are sometimes needed to collect specimens.

## Update on CCHD Screening Survey

In November, NBS coordinators and other key hospital staff were sent a link to an online survey. The survey had questions to help us learn more about hospital costs associated with CCHD screening and how staff feel about the current CCHD educational materials. We received a total of 49 responses.

Some preliminary findings from the survey are:

- 96% of respondents indicated that registered nurses perform the pulse oximetry screening for their units.
- 33% of respondents said performing the pulse oximetry screening and documenting the results takes <5 minutes on average; 52% said it takes 5-10 minutes; and, 15% said it takes >10 minutes.
- 59% of survey respondents reported providing the pulse oximetry screening results to the parents to share with their child's PCP; 11% said they sometimes provide the results; and, 30% said they do not provide the results to the parents.

Thank you to everyone who responded to the survey! Your answers will help us evaluate hospital costs related to the CCHD Screening Program and identify if any gaps exist in CCHD educational materials.



## NBS Quarterly Reports and Stellar Performance

During the fourth quarter of 2014, five hospitals met all six of the NBS performance goals. We would like to congratulate the following hospitals on their impressive efforts!

- Bronson Battle Creek Hospital
- Hillsdale Community Health Center
- St. Mary Hospital—Livonia
- Holland Hospital
- Huron Medical Center

## 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 1-517-335-4181.

## Celebrate 50 years of Newborn Screening!



### Update on NBS Customer Satisfaction Survey

In January, NBS coordinators were sent a link to an online survey and asked to share the link with their lab and unit managers. The survey had questions to help us learn more about satisfaction with NBS laboratory and follow-up services. We received a total of 48 responses from 33 different hospitals.

Thank you to everyone who responded to the survey! Your answers will help us evaluate how well the NBS Laboratory and Follow-up Programs are serving your needs.

### New look for Newborn Screening!



**MICHIGAN**  
**Newborn**  
**Screening**

With half a century of newborn screening expansion, the process has grown beyond a laboratory bloodspot screen and has expanded to point of care tests at hospital bedsides. With so many pieces to newborn screening, we wanted to make sure each of these individual screens is recognized as part of the Michigan Newborn Screening process.

The new logo helps unify the three pieces of Michigan's Newborn Screening Program. The foot is representative of the blood spot screen where 5 to 6 spots of blood are taken from the newborn and sent to the Michigan Newborn Screening Laboratory for testing. The ear represents the point of care hearing screen done before a newborn is discharged from the hospital. The newest addition to the newborn screening panel is for critical congenital heart disease using pulse oximetry, another point of care screen which is represented by the heart.



# Important Reminders!

### 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 [turbett@michigan.gov](mailto:turbett@michigan.gov) to answer your questions. Keri Urquhart, CCHD nurse educator, is also available to work with hospitals on CCHD pulse oximetry screening and reporting, and can be reached at [urquhartk1@michigan.gov](mailto:urquhartk1@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](mailto:newbornscreening@michigan.gov) or visit our website at [www.michigan.gov/newbornscreening](http://www.michigan.gov/newbornscreening)