



Michigan Department of Community Health Newborn Screening News

Fall 2012

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

MICHIGAN HIGHLIGHT

NBS Courier Service Update



A goal of the NBS program has been to decrease specimen transit time (time from specimen collection to receipt in the NBS laboratory) for all NBS specimens. As of July 2012, 48 hospitals in the Lower Peninsula have specimens picked up on Sundays by A-1 International. Having specimens picked up on

Sunday allows for more specimens drawn over the weekend to arrive at the MDCH NBS Laboratory in Lansing early Monday morning. This decrease in transit time leads to earlier confirmation and treatment of NBS disorders. For specimens drawn on the weekend, the average transit time has been reduced by approximately 12 hours.

With the transition to Sunday pick-up, Quest Courier Saturday pick-up will be discontinued at the 48 hospitals that now have Sunday pick-up.

Please remember to maintain a courier sign-in log (next to your pick-up-site basket) for the courier, so that you are able to monitor the pick-up times and record the name of the courier. This NBS log is important for monitoring purposes and informs your staff when to prepare the envelopes and place them into the basket for pick-up. Please do not ask couriers to place the NBS specimens in the envelopes as they are not OSHA-trained. Also, please use a (wire) basket for the NBS envelopes to reduce the number of specimens that get misplaced. A missing specimen creates additional work for both NBS and hospital staff and could delay confirmation and treatment initiation. All specimens must be placed in the MDCH NBS Laboratory white envelopes for transport by the courier. The envelopes can be ordered by calling Val Klasko at 517-241-5583. The NBS envelopes have a printed UPC bar on them which allows for tracking by both the courier and the state NBS laboratory.

If you encounter any NBS transit issues, we encourage you to first call Quest at 1-866-MYQUEST (1-866-697-8378) for Monday through Saturday pick-up or A-1 Courier Services at 1-248-786-2042 if you have a concern with a Sunday pick-up. If necessary, please call the NBS Follow-up Program at 1-517-335-4181 to address any concerns.

NATIONAL HIGHLIGHT

Pompe Disease

Pompe Disease, a lysosomal storage disorder also referred to as Glycogen Storage Disease Type II, is being considered for inclusion in the Recommended Uniform Screening Panel (RUSP) by the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC). The SACHDNC provides recommendations to the Department of Health and Human Services (HHS) on disorders for which there is sufficient evidence to justify inclusion in state newborn screening panels. Pompe Disease will be reviewed at the SACHDNC meeting in January and a final recommendation on inclusion in the RUSP is expected in 2013.

If the SACHDNC recommends inclusion of Pompe Disease, MDCH will review the recommendation with the state's Newborn Screening Advisory Committee in 2013 for possible addition of this disorder to the newborn screening panel in 2014.

Pompe Disease is caused by a deficiency of the lysosomal enzyme acid alpha-glucosidase (GAA) which results in a build up of glycogen in the body's cells. This accumulation of glycogen in organs and tissues impairs their ability to function normally. The estimated incidence is 1/40,000 births. Infants typically develop symptoms by 2 months of age and die at an average age of 9 months. The FDA approved enzyme replacement therapies (recombinant forms of GAA, Myozyme and Lumizyme) are now available for treatment of infantile and late onset forms of Pompe Disease. Several laboratory methods applicable to newborn screening have been developed and are being evaluated in Illinois, Missouri and New Mexico. Taiwan has provided reliable newborn screening for Pompe Disease since 2005 using a fluorometric method.

NBS Follow-up Program Contact Information

Phone: 517-335-4181

Email: newbornscreening@michigan.gov

Michigan Department
of Community Health



Rick Snyder, Governor
James K. Haveman, Director

Urea Cycle Disorders

The Urea Cycle is composed of six enzymes and two carrier/membrane transport proteins. Three of the six enzymes are located within the mitochondrial matrix and three in the cytosol. Newborn screening is targeted at the detection of defects in the three cytosol enzymes (arginosuccinic acidemia: ASA, citrullinemia type I: CIT-I, arginase deficiency: ARG) and one of the carrier proteins citrin (citrullinemia type II: CIT-II).



The urea cycle is the main pathway for removal of highly toxic ammonia in the form of urea. Enzyme or carrier protein defects in the urea cycle can lead to rapid intoxication of the central nervous system due to hyperammonemia. These disorders

lead to the most severe metabolic crises of all the newborn screening disorders and often result in metabolic deterioration during the first week of life. Since 2006, nine newborns with urea cycle disorders have been diagnosed through Michigan's newborn screening program. For these newborns, the average time from birth to receipt of specimens at the newborn screening laboratory was 4 days (range from 2-7 days).

The newborn screening program's goal is to screen, diagnose and initiate treatment for urea cycle disorders within the first week of life. It is very important that specimens are obtained within 24-36 hours of life, dried and sent immediately by courier to the newborn screening laboratory.



Please Note: Upcoming State Holidays

November 22, 23, 2012—Thanksgiving
December 24, 25, 2012—Christmas Holiday
December 31, 2012—New Year's Eve
January 1, 2013 — New Year's Day
January 21, 2013— Martin Luther King, Jr. Day

Michigan BioTrust for Health

Thank You! to the 146 staff from 32 hospitals who took the time to share opinions about the BioTrust parent consent process. Based on the results, parts of the process seem to



be working well, some areas need improvement and some facts need to be communicated better. A full report with results from our parent evaluation will be on the BioTrust website (www.michigan.gov/biotrust) in 2013. Below is a sneak peak at our findings.

Spotlight on Survey Results

The majority of hospital staff returning the survey self-identified as nurses, and most (86.3%) stated they felt the BioTrust consent booklet, *After Newborn Screening*, answered all of their patients' questions. High rates of satisfaction were also reported with the BioTrust consent form found on the back of the NBS card. Several helpful suggestions for revision were provided for the consent booklet and form. **We hope to have revised BioTrust parent consent materials distributed in 2013 that incorporate suggestions from all stakeholders including parents and nurses.**

Most survey respondents (80.1%) stated they did not know where to find forms for parents to opt-out of the use of archived blood spots (collected from July 1984 to May 1, 2010) through the BioTrust. **Forms to opt-out are available on the newborn screening and BioTrust websites or by calling 866-673-9939. Additional information about archived blood spots will also soon be added to BioTrust parent educational materials.**

The responses for reasons MDCH does not receive a parental decision about the BioTrust included "parent refusal". When a parent refuses the BioTrust, hospital staff needs to document this decision by marking the "parent declined" checkbox on the BioTrust consent form.



Please remind staff of the importance of this and let us know if we can help in any way!



We would like to thank all of Michigan's hospitals and healthcare providers for your work to implement the BioTrust parental consent process. To date, we are receiving 91% of consent forms. Of those, 64% are signed, 20% marked parent decline and 16% are returned blank. Please make sure staff are documenting parental decisions and returning completed consent forms from all parents. Let's continue towards reaching our goals for returning over 95% of forms with over 90% filled out properly.

Using Pulse Oximetry to Detect Critical Congenital Heart Disease (CCHD) in Newborns.



Congenital heart defects (CHDs) are conditions present at birth that affect how a baby's heart is made and how it works. The US Secretary of Health and Human Services recently recommended that every well newborn be screened for seven Critical Congenital Heart Defects (CCHD). Screening for CCHD uses a technology called pulse oximetry to measure oxygen saturation in the blood. A newborn baby who appears healthy, but has low blood oxygen levels, may have a heart condition or other illness needing prompt treatment.

MDCH, together with participating Michigan birthing hospitals, is exploring the implementation of CCHD newborn screening this year in a pilot project. The Michigan CCHD Advisory Committee includes neonatal nurses, hospitalists, neonatologists, pediatric cardiologists, primary care providers, parents and advocates, as well as state public health. This committee has been meeting since early this year, forming workgroups to address data and reporting and development of educational materials. Coming to consensus on an algorithm for newborn screening for CCHD by pulse oximetry in Michigan was the group's first task.

The CCHD Advisory Committee and MDCH NBS staff emphasize that pulse oximetry is performed on a well newborn as a screening tool. Babies with symptoms should be evaluated accordingly. Factors that remain to be addressed include screening guidelines for babies in the NICU, home births and babies born out of state, among others.

PULSE OXIMETRY PROTOCOL

Pulse ox on right hand (RH) and one foot (F) for newborns with no cardiac or respiratory symptoms.

Result 90-94% in RH and F or >3% difference between RH and F = LOW

Result 89% and below in RH or F = LOW

Result 95% and above

PASS

If low (x1)
REPEAT within 1 hour

If low (x2)
REPEAT within 1 hour

If low (x3)

Refer for evaluation—assess for cardiac, respiratory, infectious causes



NBS Quarterly Reports and Stellar Performance

During the second quarter of 2012, four hospitals met all seven NBS performance goals. We would like to congratulate the following hospitals on their impressive efforts!

Huron Medical Center
Huron Valley-Sinai Hospital
McLaren Greater Lansing Hospital
William Beaumont Hospital-Troy

Performance Goals for NBS Quarterly Reports

- <2% of screens are collected >36 hours after birth
- >90% of screens arrive in the state laboratory ≤4 days after collection
- <1% of screens are unsatisfactory
- <2% of envelopes are batched (i.e., contain screens with collection dates >2 days)
- >95% of electronic birth certificates have the NBS card number recorded
- >95% of specimens have a returned BioTrust for Health consent form
- >90% of returned BioTrust for Health consent forms are completed appropriately

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 1-866-673-9939.



NBS Lab NEWS



All laboratories that perform testing on human samples are required by federal regulations to comply with the Clinical Laboratory Improvement Amendments of 1988 (CLIA 88). CLIA 88 offers laboratories the option of either being surveyed by the Centers for Medicare & Medicaid Services (CMS) or meeting the laboratory requirements of an accrediting agency such as the College of American Pathologists (CAP). After careful evaluation of the pros and cons of making this change, the MDCH Bureau of Laboratories has determined that meeting the accreditation requirements of CAP offers significant advantages over our current CMS certification. The primary reason for this change is that CAP places a greater emphasis on establishing and maintaining the various components of a laboratory quality management system (QMS). Implementation of a QMS based on CAP requirements will serve to strengthen the overall quality of our laboratory testing and reporting. We are therefore in the process of seeking CAP accreditation. We anticipate this process to be complete by Spring 2013. Newborn Screening Laboratory and follow up staff are working together toward this goal.



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Introducing Our New Staff

Keri Urquhart is the new Critical Congenital Heart Disease nurse educator. Keri received her nursing degree from U of D and is currently working towards her MPH at Michigan State. She worked as a NICU nurse and in local public health before starting with the NBS program in September.

Kidist Dimore is a Public Health Associate who was assigned by CDC to the NBS Program. She received her undergraduate degree in Global Studies from the University of Minnesota. She will be assisting the NBS and BioTrust programs with prenatal education, data entry and surveys.

Important Reminders!

Specimens collected at less than 24 hours of life are considered early specimens and will require a repeat specimen. This includes specimens collected at 23 hours and 59 minutes.

The *MDCH Newborn Screening Guide* includes certain clinical circumstances that require obtaining an early specimen. However, a repeat specimen will always be requested as follow-up to an early specimen.

NEWBORN SCREENING GUIDE

The updated version of the Michigan Department of Community Health Newborn Screening Guide can be found at: http://www.michigan.gov/documents/mdch/MI_NBS_Guide_368636_7.pdf The online NBS guide will always be the most up to date version. Since we often update the content, we encourage you to bookmark the site and refer to the most recent electronic version rather than a printed copy that may be obsolete or outdated.



Please hand out the NBS and BioTrust brochures to all parents.
Is your supply running low?

Contact Val Klasko at 517-241-5583 or email: MDCH-NBScards@michigan.gov

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