



Newborn Screening News

Fall 2016

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

CCHD Quarterly Reports

The Michigan Newborn Screening Program is in the process of developing a Critical Congenital Heart Disease (CCHD) quarterly report, similar to the newborn screening quarterly reports that are sent to each birthing hospital every quarter. The aim of the CCHD quarterly report is to improve CCHD reporting and adherence to the CCHD screening algorithm. Currently, we receive CCHD screening data on about 82% of newborns statewide. We hope the reports will help facilities track their performance and improve in certain areas, such as timely reporting; only 38% of CCHD screens are reported within a week after birth.

The CCHD quarterly report will include several metrics:

- Percent of newborns with CCHD screens reported
- Percent of pulse oximetry screens conducted in a timely manner after birth
- Percent of CCHD screens reported to the Newborn Screening Program in a timely manner

We hope the CCHD quarterly report will help hospitals understand the strengths and weaknesses of their CCHD screening process and lead to improvements. If you have any questions about the new CCHD quarterly report, contact Isabel Miller, Pediatric Genomics Epidemiologist, at hurdeni@michigan.gov.

Newborn Screening Card Fee Increase:

On October 1, 2016, the cost for the initial newborn screening card increased to \$125.16 (from \$122.35). The cost for the repeat (pink) cards changed to \$117.11 (from \$114.48). The fee supports the Newborn Screening Laboratory, Follow-up, and Medical Management Coordinating Centers. The fee is adjusted annually to reflect changes in the Detroit Consumer Price Index and to cover the cost of adding new disorders to the newborn screening panel.

The Detroit Consumer Price Index increased 2.3% this year. To reflect this change, MDHHS applied a \$2.81 increase to the cost of each initial (blue) newborn screening card and a \$2.63 increase to the cost of each repeat (pink) card purchased. There is no fee increase this year for new disorders. Within the next year, we also expect to begin screening for Pompe Disease mucopolysaccharidosis, type I and for X-linked adrenoleukodystrophy, although a fee for the last two disorders has not yet been determined.

If you have questions regarding the fee, please contact the Newborn Screening Follow-up Program at 517-335-4181.

NBS Follow-up Program Contact Information

Phone: 517-335-4181

Email: newbornscreening@michigan.gov

New 60 Mutation Panel for Cystic Fibrosis Newborn Screening

Several notices about Michigan's response to a product recall impacting cystic fibrosis (CF) newborn screening have recently been distributed. Our newborn screening laboratory has been hard at work responding to the recall and are getting ready to use a new molecular panel for CF newborn screening this October. This is a great opportunity to learn more!

Michigan's newborn screening laboratory uses an immunoreactive trypsinogen (IRT) assay as the 1st tier screen for cystic fibrosis (CF). This 1st tier screen will still be performed. Specimens with elevated IRT (≥ 96 th daily percentile) will move to the 2nd tier of testing and will be screened for CF gene mutations. This 2nd tier molecular test is needed for about 4% of infants screened, and we're excited to announce our new molecular panel will contain 60 mutations versus the original 40 mutations.

While our lab has worked to make preparations for implementation, the Florida Department of Health Laboratory has been testing all of our specimens with elevated IRT levels using this same 60 mutation panel. The mutations found in this panel are listed in the table below. The mutation detection rate is estimated to be 54.53-95.94%, depending on the patient's ethnicity. We are pleased to offer this new mutation panel as it will allow us to provide a CF newborn screen that should allow us to maintain a continued high sensitivity for detecting CF (99.2%). *It is very important to remember that only a limited number of CF carriers will still be detected through newborn screening for cystic fibrosis using our two tier screen.*

ACMG Recommended Mutations ¹ ¹ Genet Med. 2004 Sep-Oct; 6(5):387-91.			16 Most Common Additional Mutations		Broad Ethnic Coverage	
$\Delta F508$	A455E	R1162X	1078delT	1898+5G>T	CFTR dele2,3	2055del9>A
$\Delta I507$	1717-1G>A	3659delC	394delTT	2183AA>G	W1089X	S1196X
G542X	R560T	3849+10kbC>T	Y122X	2307insA	1677delTA	935delA
G85E	R553X	W1282X	R347H	Y1092X	D1152H	2143delT
R117H	G551D	N1303K	V520F	M1101K	R1158X	K710X
621+1G>T	1898+1G>A	5/7/9T	A559T	S1255X	G178R	G330X
711+1G>T	2184delA	F508C	S549N	3876delA	3791delC	Q890X
R334W	2789+5G>A	I507V	S549R	3905insT	L206W	R1066C
R347P	3120+1G>A	I506V			E60X	3199del6
					R75X	406-1G>A
						Q493X

Blank Submitter Field:


The NBS lab has noticed an increase in the number of specimen cards coming in with the submitter field blank. MDHHS is encouraging hospitals to use pre-printed labels with the hospital address and submitter code on all NBS specimen cards. Pre-printed labels will save staff time and ensure that the submitter receives the completed lab report.




Important Reminders for the BioTrust Consent Process

We have learned through statewide and national surveys that many parents support the use of residual newborn screening blood spot specimens in medical and public health research, but they prefer to be asked. Michigan has been able to sustain a parental consent process for the research use of residual newborn screening specimens since 2010 due in large part to the successful collaboration with our birth hospitals. Today over 85% of blood spot specimens are accompanied by a completed


BioTrust consent form documenting either a parent's consent or refusal for the use of their newborn's specimen in potential future research. In order for this process to be a continued success, please review these important reminders and share them with your staff involved in this process!




Provide the BioTrust consent brochure and **ensure parents read it**. *This is an integral part of the consent process!*



After the parent(s) has read the brochure, ask them to mark "yes" or "no" on the BioTrust consent form.



Collect the parent or legal representatives signature for a "yes" or "no" decision. Only the parent or legal representative of the newborn can sign the form. *No one else can sign on behalf of the parent or legal representative!*



If the parent is under emotional duress, please do not collect the consent decision.



NBS Quarterly Reports and Stellar Performance

During the 2nd quarter of 2016, five hospitals met all six NBS performance goals. We would like to congratulate the following hospitals on their impressive efforts!

- Allegiance Health
- Spectrum Health Gerber Memorial
- Holland Hospital
- Beaumont Hospital—Trenton
- McLaren Port Huron—SCN

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.



Important Reminders!

Holiday Pickup Schedule :

Lower Peninsula Hospitals

Quest **will** pick up your NBS specimens on Friday, November 11 (Veterans' Day)

Quest **will not** pick up your specimens on Thursday, November 24 (Thanksgiving Day)

A1 **will** pick up your specimens on Sunday, December 25 (Christmas Day)

A1 **will** pick up your specimens on Sunday, January 1 (New Year's Day)



Upper Peninsula Hospitals

UPS **will** pick up your NBS specimens on Friday, November 11 (Veterans' Day)

UPS **will** pick up your NBS specimens, December 24 (Christmas Eve) if you prearrange the pickup

UPS **will** pick up your NBS specimens, December 31 (New Year's Eve) if you prearrange the pickup

Call 800-877-1497 to schedule pickup Christmas Eve and/or New Year's Eve

NBSO Ordering

The Newborn Screening (NBS) Program began using the Newborn Screening Online (NBSO) in October 2015. The NBSO is an online ordering system for NBS cards and educational materials. Users were given two payment options for ordering NBS cards: eCheck and credit card. Many facilities are using credit cards for payment. However, the NBS Program is incurring large credit card fees. While the NBS Program is currently paying for those fees, this is not a sustainable option long-term. We have sent a brief survey to registered users of the NBSO to understand the barriers that hospitals may experience when switching to eChecks and how the NBS Program can help facilitate that switch. In order to avoid passing along those fees to hospitals, we are strongly encouraging the use of eChecks. If you have any questions about how to switch to eChecks, please email nbsorders@michigan.gov or call 517-335-8887.

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. 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