

# Newborn Screening Update

Michigan Newborn Screening Program

July 2006

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## Introducing .....

Robert Grier, Ph.D. arrived in Michigan in October, 2004. While he is originally from Kansas, he has trained and worked across the United States. His Ph.D. training at the Medical College of Virginia in Richmond was pivotal in directing his career. The Department of Human Genetics encouraged all graduate students to be involved in the clinic, with patient interaction and counseling. The bench research resulted in the discovery of a new genetic defect in children, Biotinidase Deficiency, which is treated with the vitamin Biotin. The overall experience catapulted him into a career directing genetic testing in a variety of settings. He has directed testing of newborns for state Newborn Screening programs, testing of pregnant women to assess Down's Syndrome and neural tube defect risks, and biochemical tests to diagnose genetic defects of metabolism. He notes that the personal and professional rewards of this work have been great.

Bob is responsible for the day-to-day management of the Biochemical Genetics Lab that serves the children of Michigan who access Children's Hospital of Michigan, Detroit Medical Center and the Michigan NBS Program. He interprets test results with clinical correlation and consultation with the genetic physicians at CHM and other facilities. He is a consult to the NBS Lab in Lansing concerning Tandem Mass Spectrometry (MS/MS).

Bob notes that moving to the Detroit area last October has been a pleasant experience. The snow was pretty and daunting for their first winter. While he and his wife miss their granddaughters in Texas, they plan to visit them often. Outside the lab, he enjoys small projects around the house and time outdoors, fishing and hunting. Their church is the center of their religious and social life. He looks forward to getting to know more of you and to be of service to the patients of DMC, metro Detroit, and Michigan. Dr. Grier's contact information is 313-993-8887 or rgrier@dmc.org.

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## Why do we have so many late specimens?

This is a frequently asked question that needs to be addressed by each person or facility that obtains the Newborn Screen. The expectation is for the NBS specimen to be obtained between 24 and 36 hours of age and for the completed newborn screening card to reach the state NBS laboratory within 6 days of birth. Some things that may contribute to a delay include;

1. Obtaining the NBS when the infant is greater than 36 hours of age;
2. Holding or batching the NBS cards prior to mailing to the state NBS lab;
3. Waiting until the hearing screen is completed to send the NBS card to the state NBS lab; and,
4. Problems with the hospital mail process or the local post office, i.e., pick-up times, limited pick-up, etc..

Contact Midge McCaustland at 517-335-8588 (mccaustlandm@michigan.gov) or Tammy Ashley at 517-335-8959 (ashleyt1@michigan.gov) for suggestions on how to resolve this problem.

## **A Few Reminders ...**

### **Hepatitis Reminder...**

Be sure to complete the maternal hepatitis B surface antigen (HBsAg) test date and result on every newborn screening (NBS) card. The test date and result must be for the current pregnancy. If you have any questions, please contact Pat Fineis at 517-335-9443 or in southeast Michigan contact Kari Tapley at 313-456-4431.

### **Pamphlet available...**



Did you know that the MDCH Genetics and Newborn Screening Program offers a free pamphlet describing statewide resources and services designed for infants and toddlers who have special health care needs??

The pamphlet is available to families, hospitals, and professionals. It is an ideal family resource to have available in newborn nurseries, special care nurseries and NICUs. The resource pamphlet can be easily incorporated into educational sessions with families, or to reinforce referrals to, or enrollment in Children's Special Health Care Services (CSHCS) or Early On®.

Resources for Infants and Toddlers with Special Health Care Needs (DCH-0951) can be ordered free of charge by calling toll free 1-866-852-1247 or you can also download the pamphlet at:

[www.michigan.gov/documents/ResourcesforFamiliesBD\\_121537\\_7.pdf](http://www.michigan.gov/documents/ResourcesforFamiliesBD_121537_7.pdf)

Thank you for the important work you do in linking children and families to services!

### **Expanded newborn screening...**

The newborn screening laboratory screens all infants for more than forty (40) disorders. Effective May 1, 2006, all disorders are included on the notification of screening results.

### **Labels and/or stickers...**

Newborn Screening cards have been received in the NBS state lab with labels or stickers covering the barcode. This creates a problem with the identification of the specimen. **NEVER** place a sticker or label over the barcode on the NBS card or on any other area of the NBS card. There is no need to "seal" the flap of the NBS card as it seems some persons attempt to do.

### **Proposed NICU changes...**

The NICU draft guidelines for babies weighing less than 1800 grams is available for review. Contact Tammy at 517-335-8959 or [ashleyt1@michigan.gov](mailto:ashleyt1@michigan.gov) if you have not had an opportunity to review this document.

We will be contacting your NICU/SCN to schedule an appointment with the NICU/SCN Nurse Manager and Neonatologist to review the proposed changes and answer questions. It would be helpful if you would e-mail their contact information to Tammy or Midge at [mccaustlandm@michigan.gov](mailto:mccaustlandm@michigan.gov).

### **Hospital Newborn Screening Coordinator...**

Hospitals will be requested to identify a Newborn Screening Coordinator. This may be the person(s) at their hospital who currently has the responsibility for ensuring that the newborn screens are completed and handled appropriately. The expectation is for the Hospital Newborn Screening Coordinators to complete the NBS online tutorial or attend one of the NBS presentations. Check out Tammy's Tidbits on page 4 for more information. You can also complete the enclosed Hospital Coordinator form and return it to Tammy as directed.

### **Nursing Contact Hours...**

Effective February 1, 2006, nurse participants who complete the entire Michigan Newborn Screening Online Course, submit the posttest, evaluation and required demographic information will receive a certificate for 1.2 contact hours. This continuing nursing education activity was approved by the Michigan Nurses Association, an accredited approver of nursing continuing education by the American Nurse's Credentialing Center's Commission on Accreditation.

Nursing contact hours (1.2) can be obtained by completing the NBS Online Tutorial or by attending a NBS presentation at your facility. The information provided is also a valuable education tool for anyone interested in learning more about Newborn Screening. Contact Midge McCaustland, 517-335-8588 or [mccaustlandm@michigan.gov](mailto:mccaustlandm@michigan.gov), for information on nursing contact hours.

## A Father's Story ...

Our daughter Kaitlin was born a happy healthy baby last June 7, 2005. She was small (6lbs 1oz), but the picture of health. Her APGAR score was great and her Newborn Screens all came back passing. We were out of the hospital in the standard time and off to enjoy our new family.

Kaitlin rarely spit up let alone vomit violently until one day when she was just shy of 3 months old. This in itself was not alarming, but it kept happening. We took Kaitlin to her pediatrician where we were re-directed to the hospital. Kaitlin was diagnosed with reflux after a couple of days stay. Not all that uncommon in young babies, so armed with our new medicines, which seemed to control the problem, we went back to our normal lives.

Kaitlin went back to her pediatrician for a follow-up to the reflux and her 4-month check-up. Things looked great so one reflux medication was removed and standard immunizations were given. Kaitlin was extremely irritable over the next several days, which was unusual. She also began to vomit violently again. At this point we began making weekly visits to the pediatrician to try to assess a new battle plan. There were even a couple of trips to the ER over the next 4 or 5 weeks. We were told everything from its the reflux, to its colic, to put her back on Reglan and she will be fine.

On Thanksgiving Day 2005 Kaitlin would not calm down at all. Her appetite had been diminishing gradually over the past few weeks and now she was eating approx. 8-12 ounces in a day if we were lucky and that amount was getting smaller quickly. Kaitlin cried herself to a hoarse condition and I don't think she slept within that 24-hour period. We were waiting at the pediatrician's door on Friday morning. We said we had to be admitted to the hospital, my wife and I were at our wits end and Kaitlin seemed extremely weak (unable to hold up her head, hoarse, still irritable).

It was not clear what was ailing Kaitlin so a battery of tests was scheduled. Finally an MRI of the brain revealed that Kaitlin didn't appear to have enough myelin. Yet another panel of blood work was drawn to test for a series of Leukodystrophies. It was through this testing that we discovered that Kaitlin has Krabbe Disease, a form of Leukodystrophy. We were nearly two weeks into our stay at the hospital before the diagnosis was made and confirmed.

Since so much time had lapsed for Kaitlin and she was already showing symptoms of the disease (hypertonia, unable to hold up her head, severe vomiting, etc.) the advancements that have been made to treat this disease were really no longer an option. Great strides have been made with cord blood stem cell transplants in the first month of life of affected children. Unfortunately the treatment survival rate for children already showing symptoms is approx. 45%, with no gain in skills if the child survives the treatment. This means that in order to receive effective treatment currently a child must have already lost an older sibling to this disease. We find this cost much too high to give a child a chance at life.

Very little was known about this disease locally so we turned to the internet for additional information to help us through this difficult diagnosis and found Hunter's Hope. Hunter's Hope is an organization created to help identify and assist in finding treatments of this disease. Founded by Hall of Fame quarterback Jim Kelly and his wife Jill nine years ago after their son Hunter was diagnosed with Krabbe, Hunter's Hope continues to strive towards expanded newborn screening in all 50 states. The ultimate goal of this expanded newborn screening is to have Krabbe placed on the list of screens in all 50 states. Even if this simple test helped to save one life it would be well worth it, but it will save many, many more without the ultimate cost of losing a child to discover its presence lurking within a family.

Thanks for reading our story,

Chris, father of Kaitlin

**NOTE:** To the best of our knowledge, Krabbe Disease is not currently included in the NBS panel of disorders in any state. More information can be found at the National Institute of Health website: <http://www.ninds.nih.gov/disorders/krabbe/krabbe.htm>

## Tammy's Tidbits ...

### CHANGE IS GOOD?

I do know for sure change is stressful for us all. In the state's Newborn Screening Program we are going through a lot of growth and change this year, all for the better to make the best program ever.

Here is how we have grown so far this year:

1. Added a NBS Coordinator (Tammy)
2. Added a new computer server and updated our system to track the screens
3. Added expanded screens all to serve the states program better now greater than 40 disorders.

Next growth phase we are adding things to further improve access and speed of request for information to providers and hospitals:

1. Identify hospital coordinators in each hospital to be able to have individual contacts in each hospitals. This will make both of our jobs easier if follow-up on a newborn is needed and you will have direct contacts at the state
2. NBS results will be transferred to MICR after Jan 2007 provided hospitals include NBS kit number on birth certificate
3. E-mail newsletter and quality reports to hospital coordinators to improve distribution
4. Continue new partnerships we have made with state and local agencies to help get all newborns access to follow-up care in other states programs when needs arise

I realize change is hard and sometimes painful. Remember when we all were toddlers and began to run, we had bumps on our heads and skinned up knees. However we never stopped trying and eventually we got better at walking first then running. We at newborn screening are just beginning to run, just like the newborns we screen will someday run, it just takes time to grow.

Tammy

## Information Sharing ...

Hospitals and other providers are invited to share information on their practices, policies, procedures, and experiences in an effort to improve the NBS process. This information is not to be considered a "requirement" of the NBS program. It is merely a "sharing of ideas" that you may choose to evaluate for use in your facility. If you would like to contribute an idea to share, please e-mail the information with a contact name, phone number, and permission to include the information in the NBS Update to [mccaustlandm@michigan.gov](mailto:mccaustlandm@michigan.gov).

Many hospitals have been visited by the Nurse Consultant/Educator over the last several months. Some staff members have had very interesting ideas or suggestions - You know who You are! - that they were going to send in to be shared with others in the NBS Update. While time constraints are understandable, wouldn't you like to take a few minutes to share your ideas. It is a great way to recognize all of the excellent work that is being done with regard to Newborn Screening, infants, and families. Think about it and share your idea with others.



If you would like to receive the NBS Update, have previously requested to be placed on the mailing list, have additions, corrections or deletions, please *complete the information below* and *return this page* to the address listed below.

If preferred, you may also send an e-mail: [mccaustlandm@michigan.gov](mailto:mccaustlandm@michigan.gov)

Please indicate if your preference is to receive this newsletter by e-mail or regular mail.

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