

# Newborn Screening Update

Michigan Newborn Screening Program

April 2005

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## Introducing .....

Many of you may have had the opportunity to speak with Sue Miller when you have contacted the Newborn Screening Program office. It is her pleasant voice you hear when calling the main Newborn Screening phone line. Sue responds to phone and fax requests related to Newborn Screening results. If she is unable to answer your questions regarding the Newborn Screening Program, she will refer you to the appropriate person. Her responsibilities include physician and parent notification when a repeat Newborn Screen is necessary because the initial specimen was obtained prior to 24 hours after the infant's birth and/or the initial specimen was noted to be unsatisfactory. This responsibility involves notification from the point of acknowledgement of the case through the complete follow-up. It is also Sue's responsibility to notify the physician when a Newborn Screen was obtained after the infant received a blood transfusion and the initial specimen was noted to be unsatisfactory. She notifies the physician and Sickle Cell Disease Association of America (SCDAA), Michigan Chapter, when a Newborn Screen results in a positive hemoglobin result. Additionally, Sue is responsible for the referral notification of the physician and the Endocrine Follow-up Program (EFUP) at the University of Michigan when the Newborn Screen result for Congenital Adrenal Hyperplasia (CAH) or Congenital Hypothyroidism (CH) is borderline positive or a strong positive. She provides coverage, when necessary, for the Tandem Mass Spectrometry, Galactosemia, and Biotinidase Deficiency referral notifications to the physician and Children's Hospital of Michigan Metabolic Clinic (CHMMC).

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## What are we supposed to do with the "Submitter Copy" of the NBS card?



Here are a few ways that a hospital, or other health care provider, can choose to utilize the "Submitter Copy" of the NBS card.

The "Submitter Copy" may be useful for the hospital's Quality Improvement initiative. The copy is a replica of the card that was sent to the state lab and therefore can be used to determine if the information documented on the card was correct and complete. The hospital may choose to have the person who checks the card for completeness prior to mailing initial the copy so that, if a problem is noted, that person would be easily identified.

The "Submitter Copy" can be maintained as a part of the infant's medical record.

The "Submitter Copy" can be maintained as documentation that the NBS sample had been obtained.

If your hospital has found another use for the "Submitter Copy" send an e-mail with your ideas to [mccaustandm@michigan.gov](mailto:mccaustandm@michigan.gov) so that it can be shared with other hospitals and health care providers.

### Contacts

Program Director  
William Young, PhD  
(517) 335-8938

NBS Educator  
Midge McCaustland, RNC, MSN  
(517) 335-8588

Laboratory Manager  
Harry Hawkins  
(517) 335-8095

NBS Accountant  
Valerie Klasko  
(517) 241-5583

## A Few Reminders ...

### **REPEAT SAMPLE EXPIRATION DATE DISCREPANCY**

Be sure to check the **filter paper** for the **correct expiration date** when using the pink repeat NBS cards.

### **MAILING OF NBS SPECIMEN CARDS**

It is important to mail the NBS specimen cards to the state laboratory within 24 hours after the specimen has been obtained. One way to monitor your compliance with this request is to maintain a NBS log that would include the date the NBS specimen was obtained and the date the NBS card was mailed. This log could then be used to compare the data included in the quarterly quality reports that you receive.

### **SEPARATE NICU DATA REPORTING**

Your hospital can request to receive separate quarterly quality reports for the NICU and Mother/Baby Units. This may be helpful in identifying problems related to specimen collection, incomplete information on the NBS card, and late specimens. Call 517-335-9205 to request this change. Your NICU will be issued a new hospital code to be entered on the NBS card.

### **NBS BROCHURE**

Check your supply stock to ensure that you have the most current copy of the NBS brochure, entitled "A First Step to Your Baby's Health." The brochure can be ordered, free of charge, by contacting Valerie Klasko at 517-241-5583.

### **NEWBORN SCREENING EDUCATION**

Midge McCaustland, RNC, MSN, is available to provide a variety of educational materials and presentations. She is available for one-on-one consultation and group presentations. If you would like her to speak to your staff or at your conference, call her at 517-335-8588 or send her an e-mail to [mccaustlandm@michigan.gov](mailto:mccaustlandm@michigan.gov).

### **NEWBORN SCREENING EDUCATION PROGRAM**

You may access the NBS Online Tutorial two ways: 1) go to the NBS website at [www.michigan.gov/newbornscreening](http://www.michigan.gov/newbornscreening) or 2) go directly to the site at [www.training.mihealth.org](http://www.training.mihealth.org).

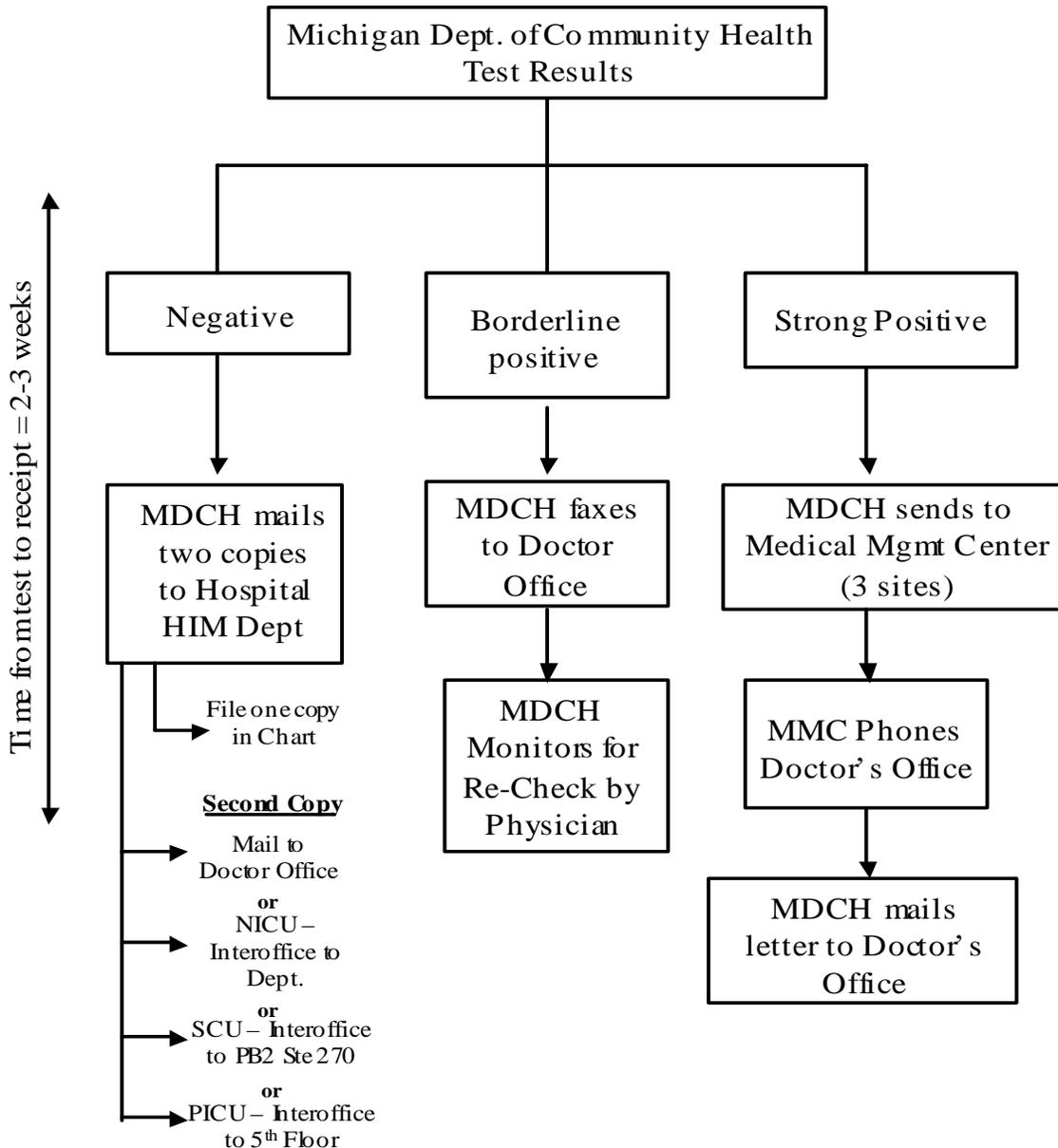
## Information Sharing ...

Beginning with this issue, we plan to provide information that hospitals or other providers wish to share 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. Page 3 of this issue contains a flow chart that identifies the process used by St. John Hospital's Health Information Management when NBS results are received.

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).

## Ideas to share ...

# St. John Hospital & Medical Center Newborn Screening Results Distribution Process and Physician Notification



A special THANK YOU to Karen Sowislo and Bonnie Brown, Health Information Management, St. John Hospital, Detroit, for sharing their flow chart.

## A Child's Story ...



Hi, my name is Anna. I am 7 years old, and in the first grade. I live in Mid-Michigan. I also have MSUD. That means I can't metabolize the branched-chain amino acids, so I have to follow a special diet, and drink a special formula, that I call my milk. I don't really always like it, but I know that if I don't drink it, I will get sick. I love to play with my Barbies, and to play house. In the summer, I love to go swimming, and to ride my bike. I really like to watch Spongebob Squarepants, and my favorite movie is Shrek 2. I would love to be pen pals with someone! If you would like to be my pen pal send your name and address in an e-mail to:

[NBS-Parent@michigan.gov](mailto:NBS-Parent@michigan.gov) and they will send it to me. I can't wait to hear from you!

Submitted by Anna's mom, Paula, who is thankful for the Newborn Screening Program.

If you know a parent, child or clinician who would like to submit a story to the NBS Update, please e-mail [mccaustlandm@michigan.gov](mailto:mccaustlandm@michigan.gov) with the information and permission to publish.

Also, the NBS Program has parent consultants who are available to speak with parents or family members. To reach a parent consultant, send an e-mail to [NBS-Parent@michigan.gov](mailto:NBS-Parent@michigan.gov) or call 1-866-852-1247 and ask to speak to a NBS parent consultant.



STATE OF MICHIGAN

DEPARTMENT OF COMMUNITY HEALTH  
LANSING

JENNIFER M. GRANHOLM  
GOVERNOR

JANET OLSZEWSKI  
DIRECTOR

## NOTICE OF PILOT PROJECT FOR EXPANDED NEWBORN SCREENING

The Michigan Department of Community Health will initiate a pilot project on April 18, 2005 to expand the current dry blood spot screening panel of 11 disorders to include 27 additional fatty acid oxidation and organic acid disorders. The purpose of the project is to evaluate the feasibility of tandem mass spectrometry in detecting these disorders and to assure that there are follow-up and medical management systems in place for early diagnosis and treatment. We hope to eventually screen all Michigan newborns for these disorders in accordance with a March 8, 2005 federal recommendation that all states should consider adding these additional disorders to their current screening panel. The additional disorders are:

### FATTY ACID OXIDATION DISORDERS

Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)  
Long-chain L-3-OH acyl-CoA dehydrogenase deficiency (LCHAD)  
Tri-functional protein deficiency (TFP)  
Carnitine uptake defect (CUD)  
Short chain acyl-CoA dehydrogenase deficiency (SCAD)  
Glutaric acidemia type II (GA2)  
Medium/short-chain L-3-OH acyl-CoA dehydrogenase deficiency (M/SCHAD)  
Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)  
Carnitine palmitoyltransferase II deficiency (CPT II)  
Carnitine: acylcarnitine translocase deficiency (CACT)  
Carnitine palmitoyltransferase I deficiency (liver) (CPT IA)  
Dienoyl-CoA reductase deficiency (DE RED)

### ORGANIC ACID DISORDERS

Isovaleric acidemia (IVA)  
Glutaric acidemia type 1 (GA1)  
3-OH 3-CH3 glutaric aciduria (HMG)  
Multiple carboxylase deficiency (MCD)  
Methylmalonic acidemia (mutase deficiency) (MUT)  
3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)  
Methylmalonic acidemia (Cbl A,B)  
Propionic acidemia (PROP)  
B-Ketothiolase deficiency (BKT)  
Methylmalonic acidemia (Cbl C,D)  
Malonic acidemia (MAL)  
Isobutyryl-CoA dehydrogenase deficiency (IBG)  
2-methyl 3-hydroxy butyric aciduria (2M3HBA)  
2-Methylbutyryl-CoA dehydrogenase deficiency (2MBG)  
3-Methylglutaconic aciduria (3MGA)

Due to the specificity of tandem mass spectrometry screening, few false positives are anticipated during the pilot project. Therefore, if your office is notified by fax of a positive screen for a fatty acid oxidation or organic acid disorder, it is important that prompt action be taken in accordance with the action steps included in the fax notification. Follow-up referrals will be to the Children's Hospital of Michigan Metabolic Clinic in Detroit (313-745-4513; toll free 1-866-442-4662).

If you have questions about the expanded screening pilot project, please telephone William Young, Ph.D., at (517) 335-8938 or e-mail at [youngw@michigan.gov](mailto:youngw@michigan.gov). 4/11/2005

3423 NORTH MARTIN LUTHER KING JR. BLVD. • P.O. BOX 30195 • LANSING, MICHIGAN 48909

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HowardA@michigan.gov

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3423 N. Martin Luther King, Jr. Blvd.  
PO Box 30195  
Lansing, MI 48909  
Phone: (517) 335-9205  
Fax: (517) 335-9419  
Email: mdch-newbornscreening@michigan.gov  
Website: www.mi.gov/newbornscreening

