



HEEL STICK NEWS

RAISING AWARENESS OF IOWA & NORTH DAKOTA NEONATAL METABOLIC SCREENING PROGRAMS

January 2011

Issue 8



Happy New Year!!

Welcome to another edition of the "Heel Stick News". A big thank you to the contributors to this newsletter.

In this edition we have added information about hearing screening from Iowa and North Dakota, as well as some other informative articles. Please let me know if you or others would like to be a contributor to this newsletter. I would like to have articles on hand for future newsletters. We also would like to request your assistance in the distribution of this newsletter. Please notify me at bschweit@nd.gov.

Thanks and best wishes for a great new year. *Barb Schweitzer, RN*

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Story from a PKU Mom



Tyler, PKU Patient

When we first found out that Tyler had PKU, it was overwhelming. The only thing I knew about PKU is that they do the heel stick for newborns in the hospital. I never thought twice about how important that test really is and how different our lives would be had they not done that test. Today, we are so thankful and blessed that Tyler is continuing to grow healthy and smart and is a very active, normal 3- year-old boy.

My husband, Damon, and I were worried and in denial the two days between the first phone call and the official diagnosis of PKU with Dr. Kenien at Sanford Health (then Meritcare). Our other sons, Alex who was 10 and Adam who was 9, did not have PKU, nor did anyone in our families. How could this happen to us? I remember wondering if Tyler would be OK. How would we ever be able to pay for the terribly expensive formula and special food he would need? How would I ever learn his diet or have the time to make his special food? There were many tears shed just wanting our newborn son to have a normal childhood, to be able to enjoy a pizza party with friends or share popcorn with our family at a game or movie.

After many reassuring words from our nutritionist, Cathy Breedon, our fears were eased. She was right in saying that Tyler will be OK and that I will learn his diet and it will become second nature and part of our routine. She shared the good news that our great state of North Dakota provides the formula through age 21 and can provide food also. She assured me that I would eventually be able to do Tyler's weekly blood draw myself and that my organization skills and drive to "succeed" at this disease would help me through so much.

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Story from a PKU Mom

(continued from page 1)

Thankfully, a parent can ease into learning the PKU diet. As he grew, so did the list of foods he could have, and it also gave me time to finally perfect bread, pancakes, cookies, muffins, etc. for him after much frustrating trial and error! There are foods that I certainly never thought twice about cooking or buying for our two older sons.

I am thankful for the variety of foods available for a low protein diet. When something finally is a success and Tyler likes it, what a victory it is! Tyler is learning what is “Tyler food” and knows he has special foods because he has PKU. Much planning and organization is required to manage his diet successfully. I continually try to match his food to the day care meals and what we are having at home. Outings require preplanning of where we will eat and what I should bring along, but it can be done.

I no longer need to look at my chart of phe values for the majority of foods Tyler eats right now and have learned to how to juggle if he needs more phe or less by the end of the day. I don't wait quite as nervously or anxiously for the results from his biweekly blood draw. Those first nervous times of sticking Tyler's heel and later his finger proved not to be so bad because I felt more in control of his situation. Tyler is such a brave little boy and is excited for the finger poke (only because of the Band-Aids™ he gets afterwards). Thankfully, my husband's out-of-state Blue Cross Blue Shield insurance now covers metabolic foods (they didn't at first). Of course, it took many frustrating phone calls the first few months on my part to make sure his claims went through.

We have such great support from our local clinic, First Care Health Center and from Tyler's day care, grandparents, family and friends, as well as the top-notch team we meet with annually at Sanford Health's Metabolic Clinic in Fargo and the support from the North Dakota Department of Health. Tyler received and “graduated” from Infant Development services, which was such a great reassurance that he is developing normally. Without this team effort, it would be much harder to deal with PKU.

I don't kid myself in thinking that Tyler's diet all will go smoothly handling as he continues to grow. I know there will be challenges when he goes to school and starts making food choices on his own. The teenage years will be especially challenging, but I hope that in making PKU a positive experience for Tyler, he will learn to deal with it and realize the importance of staying on his diet. We are fortunate now that Tyler loves and even asks for his “milkers,” but I am well aware how that situation could change overnight and we could start a formula battle with him. We are extremely hopeful that with the many advances there will be more treatments or a cure for PKU in Tyler's lifetime.

The other day, as Tyler was eating homemade Cool Whip™ ice cream, he said, “Mom, this is the best ice cream in the whole wide world!” My heart soared at his smile and sweet words! The times that are frustrating seem to be forgotten when you hear something like that! As a mom, we do anything for our children, especially when they have a challenge in life. I will make sure Tyler has every chance possible to live as normal as he can and continue to grow smart and healthy and strong!



Newborn Screening Quiz

The quiz is an opportunity to test your knowledge about newborn screening. The answers to the quiz are found on page 7.

1. For which disorders are newborn infants in Iowa and North Dakota screened?

Mark all that apply.

- | | |
|---|---------------------------------|
| a. Congenital Hypothyroidism | g. Galactosemia |
| b. Congenital Adrenal Hyperplasia | h. Missing Nose Disease |
| c. Hearing Loss | i. Expanded Screening Disorders |
| d. Phenylketonuria | j. Cystic Fibrosis |
| e. Hemoglobinopathies | k. Biotinidase Deficiency |
| f. Medium Chain Acyl-CoA Dehydrogenase Deficiency | |

2. Which of the following are potential sources of error when collecting newborn screening specimens? Mark all that apply.

- Applying blood to both sides of the filter paper.
- Squeezing or milking the foot of the infant.
- Using more than one drop to fill a circle.
- Touching the filter paper to a baby's heel.
- Drawing the circle in round motions when using a capillary tube.
- Warming the foot to enhance blood flow.
- Positioning the foot so that it is in a downward position from the heart.

3. Why is it important to wipe away the first drop of blood following a heel stick procedure?

- The first drop of blood is too concentrated so results will be high.
- Tissue fluids may dilute the first drop so the results may be low.
- It removes platelets that are likely to cause the blood to clot before the collection is complete.
- It is not necessary to waste blood. It is more important to collect as little as possible from the infant.

4. Match the critical symptom with the associated disorder.

- | | |
|---|---|
| 1. Salt wasting crisis, potential death | a. Phenylketonuria |
| 2. IQ lower by 5 pts each month if untreated | b. Galactosemia |
| 3. Recurrent infections | c. Congenital Adrenal Hyperplasia |
| 4. Severe mental retardation within 3 wks of birth | d. Congenital Hypothyroidism |
| 5. Potential death within first week of life | e. Hemoglobinopathies |
| 6. Fasting may bring metabolic crisis or coma | f. Biotinidase Deficiency |
| 7. Neurological damage including hearing loss and optic nerve atrophy | g. Medium Chain Acyl-CoA Dehydrogenase Deficiency |



Physician-Directed ND EHDl Meetings

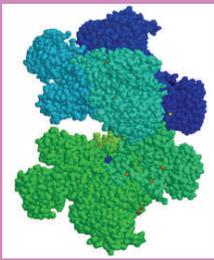


In the summer and fall of 2009, North Dakota's AAP Chapter Champion for Early Hearing Detection and Intervention (EHDl) traveled to 12 of 15 birth hospitals to conduct physician-directed EHDl meetings. Together with state ND EHDl staff, Dr. Bernard Hoggarth met with local hospital personnel and community early intervention providers to discuss each community's response to screening infants by 1 month of age, diagnosing hearing loss by 3 months of age, and intervention by 6 months of age. Each meeting was conducted face-to-face, which allowed for community with level networking to occur among medical and educational professionals. These meetings also allowed for open communication to create unique solutions in overcoming barriers and improving individual hearing screening programs.

The following direct benefits were noted:

- Follow-up appointments are now scheduled prior to discharge in two hospitals.
- OZ eSP software training was provided to three new audiologists.
- One hospital instated automatic demographical imports.
- One hospital made changes in its numbers of screeners due to high referral rates.
- One hospital made changes to its nursery parent letter to include mention of Part C in-home screenings.
- Two hospitals are looking to change their electronic records to make hearing screen results more visible.
- Two hospitals requested additional ND EHDl brochures.
- One hospital increased its communication among audiologists and early intervention providers.
- One hospital offered access to a lobbyist to promote EHDl efforts in North Dakota.
- The state EHDl program was able to clarify policies for data sharing with private audiologists.
- Two audiologists agreed to begin entering data into the statewide database.
- Communication between ND EHDl and the ND AAA (American Academy of Audiologists) was increased through a written report and an invitation to present at the state AAA meeting.
- ND EHDl was able to increase its collaboration with Tribal tracking programs.
- One pediatrician offered to incorporate office screenings as part of his practice and report results to ND EHDl.





G6PD Deficiency

Written by Mike Ramirez

Recently we have received inquiries about whether we screen for Glucose-6-Phosphate Dehydrogenase (G6PD) deficiency.

The Iowa Newborn Screening Laboratory does not screen for G6PD deficiency, and only a very few state newborn screening programs offer a test for this disorder.

G6PD is reportedly the most common human enzyme defect, estimated to affect 400 million people worldwide. Patients with G6PD deficiency are almost exclusively male, due to the X-linked pattern of inheritance. It occurs most frequently in parts of Asia, Africa, and the Mediterranean. Approximately 10 percent of African American males in the U.S. are affected.

The G6PD enzyme protects red blood cells from oxidative damage and in the absence of enzyme activity-hemolysis may occur. Most patients with G6PD deficiency are asymptomatic. For symptomatic patients, the major problem is the development of two conditions, neonatal jaundice and hemolytic anemia.

Neonatal jaundice generally occurs during the second or third day of life, with the severity ranging from subclinical to severe jaundice. When severe jaundice goes untreated for too long, it can cause damage to the brain due to high levels of bilirubin, a condition called kernicterus. Babies are successfully treated with hydration, light therapy and exchange transfusion.

Acute hemolytic anemia can occur at any age but most likely would become evident early in life. Expression of the disease is most often triggered by an infection or exposure to certain drugs, fava beans or moth balls. This condition can be managed successfully by promptly treating infections and avoidance of those chemical triggers. Informing patients and parents becomes important. Patients with severe anemia may require red blood cell transfusions.

Screening for G6PD deficiency has been implemented into screening programs in several countries in the Middle East, Eastern Europe and Southeast Asia. The magnitude of the disorder in the U.S. is unknown at this time. This fact, along with the lack of a good understanding of the natural history of the disorder in the U.S., has led scientists to recommend that G6PD not be included in the core panel of disorders for newborn screening. Large epidemiological studies in high-risk subgroups are needed to understand the true magnitude of the problem.

References:

1. Newborn Screening: Toward a Uniform Screening Panel and System. ACMG. Genetics in Medicine. 2006. Vol. 8 No. 5, Supplement.
2. Is There a Need for Neonatal Screening of Glucose-6-Phosphate Dehydrogenase Deficiency in Canada? Review by Aaron Leong. McGill Journal of Medicine. 2007, Vol. 10 No. 2.



North Dakota—Metabolic Clinic Survey Questions 2009-2010

This survey was done to gain feedback about the quarterly metabolic clinics.

1. Did you find that this clinic helps you to manage your child's/your health condition?

YES = 9

NO = 0

2. Did you feel that your child/you saw the appropriate providers? If no, whom else should they/you have seen?

Comments:

- Need someone from special ed. to get specific advice re: school, IEP planning, transition, etc.
- We especially enjoy meeting with Cathy B. and Dr. Kenien. We don't really feel that social work and psych are helpful at this point, but maybe when she's older.
- Yes, the clinic basically covers it all.
- I don't feel we need the education specialist so much now, but it was helpful before. I don't feel we need the social worker either, but I know others do.
- Definitely.

3. Through the clinic team, did you receive:

- a. **Information**

(e.g., resource materials given or questions answered)

Y or N (All nine participants answered YES.)

- b. **Written Care Coordination Plan**

(e.g., treatment or service plan)

Y or N (All nine participants answered YES.)

- c. **Assistance in accessing Resources**

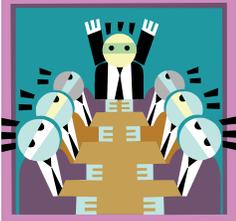
(e.g., linkage to services)

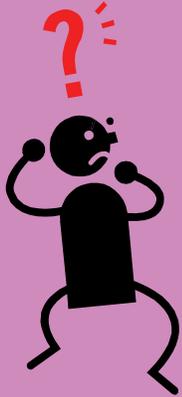
Y or N (All nine participants answered YES.)

4. Do you have any questions or concerns regarding the clinic?

Comments:

- I would like additional support/training on blood sampling.
- I feel back on track and better than I've done in a long time. I am proud of myself. I've started to tell my coworkers about my condition instead of ignoring it. That way, when I start to get a little goofy, they tell me to go drink my milk!
- It's a great thing. We absolutely love, love, love Dr. Kenien!
- Cathy and Dr. Kenien definitely make the trip worthwhile.





Quiz Answers (Quiz found on pg 3)

1. Iowa and North Dakota newborns are screened for (a) Congenital Hypothyroidism, (b) Congenital Adrenal Hyperplasia, (c) Hearing Loss, (d) Phenylketonuria, (e) Hemoglobinopathies, (f) Medium Chain Acyl-CoA Dehydrogenase Deficiency, (g) Galactosemia, (i) Expanded Screening Disorders, and (k) Biotinidase Deficiency.
2. Items (a) - (e) are sources of error in the collection process. Warming the foot to enhance blood flow and positioning the foot in a downward position from the heart will help to collect a good specimen.
3. (b) The first drop of blood is often diluted with tissue fluid. This could result in an affected child mistakenly being diagnosed as a false negative.
4. Although several of the disorders can cause mental retardation, it is the most significant in children with untreated Phenylketonuria. 1-c; 2-d; 3-e; 4-a; 5-b; 6-g; and 7-f.

Iowa's Early Hearing Detection & Intervention (EHDI) Program



EHDI Hospital Site Visits Going Well

The Iowa Early Hearing Detection and Intervention (EHDI) program staff has conducted 24 site visits with Iowa birthing hospitals. Each hospital will receive periodic visits regardless of performance. The goals of the visits are to:

- Identify strengths and best practices in hospital newborn hearing screening programs so they can be recommended to other hospitals.
- Clearly understand hospital practices for newborn hearing screening.
- Identify areas for improvement in hospital newborn hearing screening programs.
- Identify hospital technical assistance needs.
- Identify areas for improvement in the Iowa EHDI program.

The hospital site visit team includes the state EHDI coordinator or EHDI follow-up coordinator, audiology technical assistant and a parent of a child who is deaf or hard of hearing. If a parent is not able to attend, the team has a DVD taped message from a parent of a child identified with hearing loss following the newborn hearing screen at the hospital. Having a personal message from a parent is very effective. The parent reinforces that the screening, communication used at the hospital to convey the results and assistance in helping the family schedule follow-up appointments are very important and do not go unnoticed. It also reinforces the need for timely reporting.

Following is a list of strengths and areas for improvement that have been identified by the EHDI team during the hospital site visits:

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EHDI Hospital Site Visits Going Well (continued from page 7)

Strengths include:

- Sufficient or adequate data entry staff.
- Nurses discuss newborn hearing screen results with the mother prior to discharge.
- Some hospitals have established a follow-up phone call or home visit following hospital discharge to discuss how the child is doing and the importance of follow-up appointments, including hearing screening.
- A number of hospitals are bringing children born at their hospital who require a repeat hearing screen back to the hospital for the hearing rescreen.
- Many hospitals are helping families schedule the hearing rescreens with the AEA or a private audiologist prior to hospital discharge.
- Discharge summaries include the results of hearing screens, and/or hospitals are sending a separate letter to primary care physicians (PCPs).
- Many of our largest hospitals have the lowest refer and miss rates
- A couple of neonatal intensive care units (NICU's) have obtained Automated Auditory Brainstem Response (AABR) equipment as recommended; others are looking into obtaining the equipment in the future.
- Positive feedback has been provided to EHDI staff on the quarterly newsletter, rubric and hospital site visits
- Some hospitals have a great relationship with the PCPs and communicate non pass hearing screen results by phone.
- More hospitals are importing demographics into eSP, which decreases errors and decreases the number of children the hospital misses putting in eSP.
- Data entry errors are decreasing.
- The number of hospitals reporting the correct primary care provider for the child has improved over the last year.
- Some hospitals have incorporated hearing screening in their yearly competencies.



Areas for improvement include:

- Some hospitals have no formal procedure in place to ensure the PCP/medical home receives a copy of the infant's newborn hearing screening results in writing. Now required by law, but some hospitals assume that because they have electronic health records, the physicians access the results from their system, as needed. Electronic medical records are not acceptable without some sort of active communication to the PCP per IDPH asst. attorney general.
- Some hospitals are still having issues with reporting the PCP who will accept responsibility for the child's care upon discharge.
- Some hospitals have no formal process in place to ensure children who referred or were missed receive information to schedule the appointment in a timely manner or assistance in scheduling prior to hospital discharge.
- Best practice for rescreening with AABR is not always implemented because their hospital does not perform outpatient rescreens and the AEAs do not have that technology.
- No yearly training or competency for newborn hearing screening and/or tracking of high refer rates by screeners.
- No adequate quality assurance checks in place to ensure all babies born at their hospital are entered into eSP (e.g. transfers, deceased).



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EHDI Hospital Site Visits Going Well (continued from page 8)

Because of the hospital site visits, the EHDI program staff have identified hospitals that are utilizing best practices; some of which have been highlighted in *Iowa EHDI News*. In addition to highlighting hospitals utilizing best practices in the quarterly newsletter, the EHDI staff has also shared practices during other hospital site visits. Each hospital contact and quality assurance coordinator or CEO receives a written summary of the hospital site visit, including strengths and recommendations for improvement. This helps reinforce what hospital personnel are doing well with their newborn hearing screening program and where they need additional support by management.

Site visits have been well received by all hospitals visited thus far and many hospitals have already implemented recommended changes. For example, a number of hospitals are now conducting outpatient hearing screens for children born at their facilities or helping schedule a hearing rescreen with the AEA or a private audiologist prior to hospital discharge. Other hospitals are working with their AEA to allow scheduling when the AEA is closed. In addition, a few hospitals are revising procedures to ensure all physicians receive written communication in a timely manner, rather than relying on physicians to look up the hearing screen results in the electronic medical record. Other hospitals contact the physician or the physician's nurse by phone to let them know the child needs a follow up hearing screen and convey the appointment if it has already been scheduled.

Finally, each hospital was given the opportunity to respond to a survey about the hospital site visit. Of those facilities that responded to the survey to date, 100 percent said that the site visit was helpful. Additional areas hospitals found helpful included the self-rating rubric ("the self rating rubric was a very useful tool to look at our practice and see where we might need to make improvements"), review of the hospitals refer and miss rates data, the Iowa EHDI Best Practices Manual and the report that was sent following the visit that highlighted hospital screening program strengths and areas for improvement.

By: Tammy O'Hollearn, Iowa EHDI Coordinator



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Heartland Collaborative Update

The Heartland Collaborative provided funding for Kim Turner, RN, nurse clinician specialist—supervisor, Iowa Newborn Metabolic Screening Program to attend the Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children in Washington, D. C.

The following are some highlights of discussions/reports at the meeting:

September 2010 meeting

- **Quality measures** for newborn screening were reviewed and the committee addressed what its role should be in “meaningful use.” Documentation must include the process of care – referral, usability and feasibility are key components. The NBS Saves Lives Act contains the reference for quality measures.
- **SCID, LSD & Hemoglobinopathies** are to have new LOINC codes. There is a white paper under development for Hemoglobinopathies. There is a push for standardized reports and increased use of quantitative reporting. There are currently three vendors who can provide HL7 – Perkin Elmer, Natus and Oz systems.
- **Dried Blood Spot**
The committee reviewed public comments and proposed changes to the recommendations outlined in a prior meeting on the retention and use of residual dried blood spot specimens after newborn screening. The primary education target identified is prenatal care providers. A letter is to be drafted and sent to the committee for approval soon.
- **Healthcare Reform**
Christine Brown presented information regarding the impact of healthcare reform on heritable disorders. Key points:
 - Automatic access for people with heritable disorders is not guaranteed by the new law.
 - New payer mandates eventually ensure insurance access for patients and offer potential to reduce patient out-of-pocket costs over time.
 - Loophole identified for essential health benefits
 - Patient Bill of Rights – implication: medical foods do not fall under the patient bill of rights since they are not “essential healthcare benefits”.
 - Creation of a temporary high-risk pool – federal government currently administers temporary high-risk pools for 22 states. Most states operate additional high-risk pool programs.
 - Federal high-risk pool provides more comprehensive coverage than state programs.



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Heartland Collaborative Update

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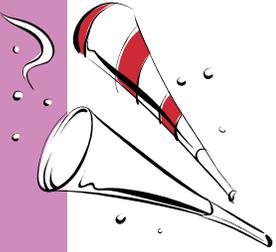
- **Single identifier**
Brad Terrell spoke regarding improving data quality and quality assurance in NBS by including the bloodspot screening collection device serial number on birth certificates. There is opposition from privacy groups. By using one number it is possible to link other healthcare information. Discussion ensued regarding a state identifier rather than national identifier. The draft paper was presented and will be voted on at the next meeting in January 2011.
- **Medical Foods**
Access to medical foods survey was presented by Susan Berry, M.D. The charge for the sub-committee is to identify barriers to short-term-follow-up & long-term-follow-up of NBS results specific to the challenges in integration of healthcare systems, financing of services and information technology; develop recommendations for overcoming barriers and recommend mechanisms for establishing accountability for NBS follow up guidelines. Reviewed FDA definition of medical food – is definition the problem? Medical foods survey sent to parents of children birth to 18 with metabolic disorders. Eighty percent of the patients of parents surveyed use at least two of the surveyed products. Products surveyed were feeding supplies, dietary supplements, modified low protein foods and medical foods. Nearly all children of the group surveyed had health care coverage – even if it did not pay for products. Most children needed more than one category of food/supplies. Coverage was variable but there were at least some out-of-pocket expenses. Families often didn't know there were caps on insurance and the dollar amount of cap. Families had a difficult time telling the survey what out of pocket expense was (data incomplete). WIC is an important source of support as well as Medicaid. Modified low protein foods are particularly poorly supported. All regions surveyed observed significant challenges to families in paying for these essential products. The Secretary's Advisory Committee for Heritable Disorders in Newborns and Children (SACHDNC) has communicated three times with the Secretary of HHS regarding medical foods. Medical Food Equity Act – House of Representative bill HR4926: 27 cosponsors; Senate bill S2766: 2 cosponsors.
- **Next steps:** anticipate Secretary of HHS response; monitor progress of Medical Foods Equity Act and Benefits Package for Affordable Health Care Act; work with FDA; extend the survey to other regions; focus on elements of highest impact; possible publication.
- **Medical Home**
Carl Cooley presented information regarding making co management explicit: integrated care and the medical home. Collaborative care needs to be made explicit – it is not only a place but a process of care that is comprehensive, coordinated and recognized by characteristics and adherence to widely accepted standards. A good guide is the medical home model. Evidence shows hospital rates of admission and emergency room visits are decreased. Communication improves outcomes and offers transparency and clarity.

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Heartland Collaborative Update (continued from page 11)

- **Subcommittee Reports**

- **The education and training subcommittee** recommends perusing the NBS Clearinghouse website. Emergency preparedness is under development. NBS Saves Lives Act requires education for primary care providers and the subcommittee is looking for ways to improve education perhaps achieving this through a national NBS awareness campaign directed at pregnant women, partners, etc. Studies indicate there is a decreased level of awareness and understanding. In the last month a group of women were polled and most did not know if the NBS had been completed for their child. The Minnesota NBS program has an education director specifically assigned to providing education for the state.
- **Health information and technology** has been looking at the work the National Quality Forum does. NQF reviews and endorses quality measures developed by organizations. Discussed working with NQF to conduct evidence review of quality measures. Currently, guidelines allow a six-month time frame to complete a referral to the specialist following an abnormal NBS result. The committee agrees this time frame is too long but took no further action on this point at this time. Hemoglobin H disease is under further review and revision to the draft recommendation.
- **Candidate Nomination – Critical Congenital Heart Disease**
Report on the candidate nomination critical congenital heart disease was presented by Alex Kemper with the external evidence review workgroup. Public comment was accepted by the SACHDNC. Heartrending stories were presented by parents and representatives of the cardiology medical community. Significant morbidity and mortality has been reported. Once this disorder is identified, it requires surgical intervention or catheterization within the first year of life. The first tier screen is pulse oximetry. Second tier or diagnostic testing is completed by echocardiogram. Discussion centered around how is the data going to be tracked, is there enough data to support adding to the NBS panel and who will be responsible for follow-up. The committee did endorse adding the disorder to the panel after an entire morning of discussion.



A happy New Year! Grant that I
May bring no tear to any eye
When this New Year in time shall end
Let it be said I've played the friend,
Have lived and loved and labored here,
And made of it a happy year.

Edgar Guest



(continued from page 11)



Children's Special Health Services



The Division of Children's Special Health Services (CSHS) is located within the Special Populations Section in the North Dakota Department of Health (NDDoH). CSHS's primary source of funding is the Maternal and Child Health Block Grant, which requires accompanying state and/or local match. Additional funds earmarked for children with Russell Silver Syndrome and early hearing detection and intervention enhancements were also appropriated for the 2009-2011 biennium.

The purpose of CSHS is to provide services for children with special healthcare needs and their families and promote family-centered, community-based, coordinated services and systems of health care.

Programs within the division include:

- **Specialty Care Diagnostic and Treatment Program** – CSHS pays for medical services for eligible children, including healthcare visits and tests to diagnose chronic health conditions early and specialty care needed for treatment
- **Multidisciplinary Clinics** – CSHS funds and administers multidisciplinary clinics that support coordinated management of nine different types of chronic health conditions.
- **Metabolic Food** – CSHS provides medical food and low-protein modified food products to individuals with PKU and MSUD.
- **Russell-Silver Syndrome Program** – CSHS pays for growth hormone treatment and medical food for individuals with Russell-Silver syndrome.
- **Care Coordination** – CSHS supports community-based programs that help families access services and resources.
- **Information Resource Center** – CSHS provides healthcare resource information.
- **State Systems Development Initiative** – CSHS enhances data infrastructure for the MCH population.
- **Children with Special Health Care Needs Service System** – CSHS supports initiatives that lead to a community-based system of services for all families, children, and youth with special healthcare needs.

Within CSHS, newborn screening long-term follow-up has primarily been addressed by the following:

Providing medical food to individuals with PKU and MSUD – Each year, CSHS provides medical food and low-protein modified food products to approximately 20 to 25 individuals with PKU and MSUD through the Metabolic Food Program. Food (formula) is provided at no cost to males under age 22 and females younger than 45. Low-protein modified food products also are provided to individuals in the above specified age ranges who are receiving medical assistance (Medicaid). Medical food is also available at cost to males 22 and older and females 45 and older, regardless of income.



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Children's Special Health Services

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Supporting quarterly Metabolic Disorders Clinics – The Multidisciplinary Clinic Program is a resource to help families manage their child's chronic health condition. Most clinics supported by Children's Special Health Services (CSHS) provide multidisciplinary team evaluations where families have an opportunity to see many different providers in one place at one time. Reports generated from the clinic visit include recommendations to help direct care for the coming year. There is no charge to families for clinic services; however, insurance or other sources of health care coverage may be used when available. All children who might benefit are eligible to attend. CSHS provides financial support through a service contract with MeritCare Children's Hospital & Clinics for four multidisciplinary metabolic disorders clinics each year. The primary target population served is children birth to 21 who have a metabolic disorder such as PKU, MSUD, MCAD or galactosemia. The multidisciplinary team at the clinic includes a registered nurse, a pediatric endocrinologist, a medical social worker, a pediatric metabolic nutrition specialist, an education specialist and a child psychologist.

Providing specialty care diagnostic and treatment program – North Dakota children birth to 21 who meet medical and financial eligibility criteria are able to access diagnostic and treatment services through CSHS. Through this program, CSHS pays for medical services for eligible children including healthcare visits and tests to diagnose chronic health conditions early and specialty care needed for treatment. All conditions currently identified through North Dakota's blood spot screening program meet medical eligibility criteria. Examples include cystic fibrosis; various Hemoglobinopathies and amino acid; fatty acid oxidation; metabolic and organic acid disorders. Children receiving treatment services through CSHS also receive community-based care coordination to help them access other needed services and resources.

Newborn Screening Blood Collection: Partnering for the Health of Babies

Written by Marcia Valbracht, MHA

All procedures in the newborn screening (NBS) process reflect a commitment to produce accurate, timely and clinically useful results. No procedure is more important than the first — specimen collection.

The following information is based on the updated standard written by the Clinical and Laboratory Standards Institute (formerly NCCLS) LA4-A5, Vol.27 No. 20, 2007, *Blood Collection on Filter Paper for Newborn Screening Programs; Approved Standard — Fifth Edition*. Copies of this standard with an instructional DVD are available from the NBS laboratory for use in training staff who collect NBS specimens.

Analyses of unacceptable specimens may yield unreliable, misleading, or clinically inaccurate results. A poor quality specimen MUST be recollected, which causes trauma to the infant and anxiety to parents and burdens the screening and collecting laboratories. The delays in testing due to recollection also could have serious medical consequences for an afflicted child, such as irreversible mental retardation, coma or death.



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Newborn Screening Blood Collection: Partnering for the Health of Babies

Written by Marcia Valbracht, MHA

The size of the blood spot is very important. See the picture below; the bottom spot has too much blood, the middle three appear layered, however the top spot is the correct size. The lab needs each spot to be large enough to punch out ;three-four discs (see top two spots).



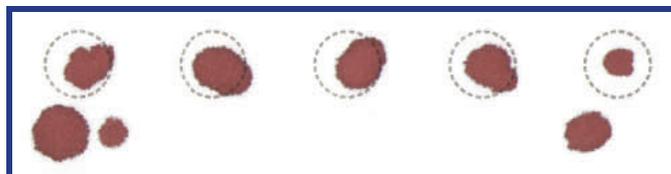
When there is insufficient quality (spots too small or insufficient number of circles filled), there will be a lack of blood for completing tests. Insufficient specimen quantity may be caused by one or more of the following reasons.

- Removing the filter paper before the blood has completely filled the circle or before the blood has soaked through to the other side.
- Applying drops that are too small.
- Spreading the blood over the surface of the circle, contributing to uneven absorption.
- Improperly applying blood to the filter paper with a device.
- See how the blood did not soak through in this example?



This can result in nonuniform analyte concentrations.

The picture below shows spots that are too small. The blood concentration is insufficient for proper analytic testing:



There is no such thing as a 'self-made' man.

We are made up of thousands of others.

George Matthew Adams

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Another reason for rejection is due to layering the blood drops as seen below. Multiple drops were applied to several circles which can cause non-uniform analyte concentrations.



Please apply only ONE drop of blood per circle. The single drop size should be approximately 75 to 100 μL in volume, which will fill the one-half inch diameter circle. Apply the blood drop in one smooth motion.

Don't spread the blood over the surface of the circle, as this contributes to uneven absorption.

Other criteria used for rejecting NBS specimens include:

- Over-saturating with blood.
- Dilution, which may be caused if alcohol is not dried on baby's heel prior to lancing.
- Contamination from invisible substances on the bench top that will dissolve into the blood. Make sure that the filter paper part of the collection form does NOT come into contact with the bench top. Always use the protective flap when placing the form on a counter both BEFORE and AFTER collection.
- Layered or clotted specimens that cause false negative and positive results for the following tests: congenital adrenal hyperplasia (CAH), hypothyroidism by thyroid stimulating hormone (TSH), cystic fibrosis (CF) screen by immunoreactive trypsinogen (IRT) and the expanded set of disorders tested using the tandem mass spectrometry (MS/MS) method.
- Tubes that contain EDTA as an anticoagulant causing false negatives for TSH and IRT and false positives for CAH. Heparin has been shown to interfere with PCR analysis in cystic fibrosis testing, so it is wisest to not use any anticoagulant when collecting NBS specimens.
- Serum rings (see picture below) or squeezing or milking the heel that causes hemolysis. Use gentle pressure.



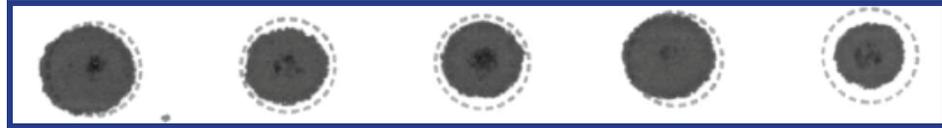
- Clotting (see picture above) can result from the improper use of capillary tubes (waiting too long to apply the blood and it clots in the tube, etc.). Apply the blood to the filter paper from each tube as it is collected. Do not draw or swirl with the capillary tube onto the filter paper.

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- Clotting (see picture below) that can result from the improper use of capillary tubes (waiting too long to apply the blood and it clots in the tube, etc.). Apply the blood to the filter paper from each tube as it is collected. Do not draw or swirl with the capillary tube onto the filter paper.



- Blood applied to both sides of the filter paper.
- Putting specimen in the envelope before drying.
- Folding the flap over the specimen before it is completely dry.
- Sending the specimen with the courier before it is completely dry. Air dry three hours before enclosing in the envelope.
- Scratching the filter paper by touching the heel to the paper or pressing a capillary tube to the paper, causing “dents” or “scratches.”
- Receipt of specimen more than 14 days after collection. There is no need to “batch” the specimens, as the courier picks up specimens seven days a week in Iowa and at least five-six days a week in North Dakota and South Dakota.

Avoid venous collections as the vessel source may affect results; veins may be needed for IV fluids, and it is more invasive than a heel stick.

Avoid using syringes to collect due to possible clotting, settling and lysing of cells. If it is necessary to use catheters, be sure to clear the line with 2 to 2.5 ccs blood prior to collection.

Umbilical cord blood may contain maternal contamination; since you must collect in the first five minutes after birth, you would still need to recollect from heel later (<24 hour age “rule”).

Unacceptable Collection Sites include:

- Arch of the foot.
- Newborn’s fingers.
- Earlobes.
- Previously punctured sites.
- Intravenous lines contaminated with interfering substances.

Acceptable Specimens look like this:





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One drop is applied to each circle $\frac{1}{2}$ inch in diameter and applied to one side of the filter paper only.

Before collecting the specimen it is important to:

- Check the expiration date on form
- Fill out the form properly and completely
- Confirm the infant's identity



Take the following precautions:

- Wash hands vigorously
- Wear powder free gloves and change between infants
- Follow safety precautions when handling and disposing of sharps

The preferred collection site is the heel. Prepare the site by warming the newborn's heel by using a soft cloth moistened with warm water (less than 42 °C) for 3-5 minutes. The infant's leg should be lower than the heart which will increase venous pressure. Wipe the heel with 70% isopropyl alcohol and air dry.

The puncture should be made WITHIN the shaded area only on the plantar surface of the heel. See the picture below:



Puncture the heel using a sterile lancet or heel incision device that is 1.0 mm deep by 2.5 mm long. Do not use scalpel blades or needles. Wipe away the first drop of blood. Allow a large drop to form and touch the paper to the blood drop ONCE and let it soak through. This is called direct application.

Apply blood to only one side of the filter paper. Continue until all circles are filled. Do not press the filter paper against the puncture site.

Examine the form by looking at both sides of the filter paper to assure the blood has soaked through. If blood is not soaking through try the process again on another circle; DO NOT reapply blood to the same circle. After collection of the entire specimen, take time to look at it and determine whether or not it is acceptable. If not, recollect it at that time.

Take care of the puncture site by elevating the foot above the body and pressing sterile gauze or a cotton swab against the puncture site until bleeding stops. Do not apply bandages that may damage the baby's delicate skin.

All NBS blood specimens should be air dried before being placed in any container, including the envelopes. Do not touch the blood spots or let them touch each other. Dry away from direct heat, in indirect light, and horizontally. Using a device such as the one pictured below may aid in proper drying of the specimen.

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When your work speaks for itself, don't interrupt.

Henry J Kaiser





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The only information we have on a baby is what you present to us on the collection form.



Managing this information is a big challenge for our laboratory since it all must be scrutinized and entered into a database. We do our best to decipher illegible handwriting and correct errors, but inaccurate information will return to you on the test report that you may have to correct later. Several hospitals have switched to electronic entry. This helps decrease data entry errors on our end. However, please note that data entry errors may occur on the hospital side as well.

When filling out the collection form, all requested information must be provided as missing information may prevent or delay test results. To properly calculate the age of baby at time of collection, we need the baby's birth date and time and the collection date and time. Collecting the specimen early affects testing results.

Information that is crucial to the interpretation of the newborn screen test results includes:

- Name
- Facility (submitter)
- Date and time of birth
- Date and time of specimen collection
- Transfusion status
- Weight of infant at time of collection
- Physician and submitter information

The date and time of birth and collection are used to calculate the infant's age at time of collection. A baby needs to be 24 hours of age or greater for accurate and reliable results for these disorders: Expanded Panel for Amino Acids [including Phenylketonuria (PKU)] & Acylcarnitines [including Medium-chain Acyl co-A Dehydrogenase Deficiency (MCADD)], Hypothyroidism, and Congenital Adrenal Hyperplasia (CAH). If a specimen is collected prior to 24 hours of age, or collection information is missing, results cannot be reported for these disorders.

These newborn screening tests are directly affected by transfusion: Hemoglobinopathy disorders, Galactosemia, Immunoreactive Trypsinogen (IRT), and Biotinidase. We cannot report these results if the transfusion status is not provided. If you know a baby is going to be transfused, collect the specimen before the transfusion, even if the baby is less than 24 hours of age. These tests can be reported on babies less than 24 hours of age. Then, you can recollect the baby after transfusion (if possible wait 2-3 days) for the Expanded Panel (PKU), Hypothyroidism, IRT, and CAH tests.

The test interpretation for CAH is weight dependent, so please remember to fill out the weight of the baby at the time of collection to receive a valid result.

Currently whenever we receive forms with inaccurate or missing information we fax the birthing facility to ask for the information. This is a time consuming step for us and for the birthing facility contact person, who must look up the information and fax it back to us.

Getting the information right the first time means you receive screening results in a timely manner and all of us can focus our efforts on improving our processes for the better health of our babies.

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