New Waves of Newborn Screening: What's Next on the Horizon!

VIRGINIA AWHONN 2016

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Special Thanks to: C. Cuthbert, A. Gaviglio, K. Olson
Plan

- Introduction & Background
- Real-life Stories & Video
- Tips - Good Card/Bad Card
- Talk about Talking with Parents
- Case Scenarios – Audience participation
- Resources for Providers & Families
- Discussion & Questions
- The End!
Newborn Screening -

• History and Background of these tests . . .
  It’s so much more than the PKU test now

Over 50 diseases that result in developmental delay and/or death if not recognized early
So much more than a PKU test!

• Disorders of amino acids, fatty acids, and organic acids
  – Detected by a sophisticated laboratory technique (Tandem-Mass Spectrometry)
• Hemoglobinopathies (e.g. sickle cell disease)
• Other conditions
  • Congenital adrenal hyperplasia
  • Congenital hypothyroidism
  • Glactosemia
  • Biotindase deficiency
  • Cystic Fibrosis
  • Severe Combined Immune Deficiency (SCID)
    • Critical Congenital Heart Disease (CCHD)
    • Hearing Disorders

NEW!!

MPS – 1 X-ALD
It’s a Team Effort

The importance of patient centered care – a program that includes the family, the bedside RN, the pediatrician, the lab, the CDC and state and national leaders all working together.
The process

Sample Collection & Logistics

Sample Receiving

Sample Testing

On-time Results & Follow-up care coordination
Follow-up

• After the Newborn Screening
  – The importance of follow-up of screen positive newborns
New Recommendations:

• Collect blood spots within 24-48 hours after birth

• Must be received in the lab within 24 hours of collection

• Critical conditions must be reported within 5 days

• All other conditions within 7 days
Where the Bottlenecks are:

• Smaller, private hospitals
  – Delivery volumes < 1000/year
• Home births/midwives
• Rural areas – distance to the state lab
• Low income, minority, Hispanic families
Deadly Delays

The nation’s newborn screening programs depend on speed and science to save babies from rare diseases. But thousands of hospitals fail short, deadly delays are ignored and failures are hidden from public view — while babies and their families suffer.

Delays at hospitals across the country undermine newborn screening programs, putting babies at risk of disability and death

By Ellen Gabler of the Journal Sentinel staff

11/16/13
Days after newborn screening reform, a baby's life is saved.

*Journal Sentinel* Nov. 8, 2014
Parent’s Perspective

• Parent’s Perspective of Newborn Screening – What every Nurse needs to know
A Parent’s Story

• The story of the Olson Family whose lives were forever changed by Newborn Screening

• Outcome would have been different if not for Nurse Kathy!

http://vimeo.com/46779932

Save Babies Through Screening Foundation

AWHONN
Promoting the Health of Women and Families
The Vital Role of Nurses in Newborn Screening

- **Heel stick** - blood sample collection on Filter Paper
- Completing newborn demographic information on the card
- **Timely submission** of blood spots to Lab — within 24 hours
- **Talking to parents** about screening
Poor outcome
We can do better!
Blood sample collection
• What the state labs wished nurses knew about sample collection
  – Reference:
    Clinical & Laboratory Standards Institute (CLSI), NBS 01-A6:
    “Blood Collection on Filter Paper for Newborn Screening Programs: Approved Standard”
  – Collect 24 to 48 hours after birth
  – Fill circle, do NOT layer successive drops of blood
NBS01-A6
Blood Collection on Filter Paper for Newborn Screening Programs; Approved Standard—Sixth Edition

This document highlights specimen collection methods, discusses acceptable techniques for applying blood drops or aliquots to the filter paper segment of the specimen collection device, and provides instructions on proper specimen handling and transport to ensure quality specimens are consistently obtained for newborn screening analysis.

A standard for global application developed through the Clinical and Laboratory Standards Institute consensus process.
Blood sample collection

• How to collect a valid blood spot sample
• What’s wrong with this picture?

Gravity is your friend -
Use it!
Other tips for Blood Spot Collection:

- Warm the heel!
- Swaddle
- Binkie
- Firm stick
- Tilt Crib Up/Foot Down
Colorado’s investigation of unsatisfactory/invalid specimens:

– Potential reasons for unsatisfactory due to collection
  • No blood applied
  • No blotter with slip
  • Clots or uneven blood
  • Serum separation
  • Contaminated
  • Multiple application
  • Incomplete submission
  • Quantity not sufficient
How to collect a good sample

- CLSI Newborn Screening DVD | Informational Video by the Clinical and Laboratory Standards Institute

https://www.youtube.com/watch?v=EnpByF_LgxA
Good Cards-Bad Cards

What to do & what not to do with blood spot cards!

• Fun, interactive activity demonstrating good and bad techniques in preparing blood spot cards

• Is this a Good Card or a Bad Card?
Is this a Good Card or a Bad Card?

What’s wrong with this card?
Is this a Good Card or a Bad Card?

What’s wrong with this card?
Is this a Good Card or a Bad Card?
What’s wrong with this card?
Valid specimen:

Allow a sufficient quantity of blood to soak through to completely fill the preprinted circle on the filter paper. Fill all required circles with blood. Do not layer successive drops of blood or apply blood more than once in the same collection circle. Avoid touching or smearing spots.
Special circumstances

• Special circumstances that can affect a sample:
  – Treatments
  – Medications
  – Prematurity
  – TPN

• The more information that we provide on the card, the better the lab interpretation
What happens next?
Time is of the Essence

• Timeliness
  – Systems & Process issues
  – Human factors

• Standards, Guidelines & Best Practices
  – Collection
  – Transit time - courier
    • Get it to the lab within 24-48 hours
  – Lab open 7 days/week
  – Public reporting
    • online “Report Cards”
Informing Parents about Newborn Screening: Tools and Strategies

1. Newborn screening saves lives.
2. Newborn screening is simple to do.
3. Newborn screening has to get done on time.
4. Remain calm.
5. Everybody’s doing it!
What to say to parents

• Strategies for discussing Newborn Screening with Parents – Simple messages

• Sample Scripting
  – “We are screening now to determine what testing your baby may need to have done later, if any”
Fill out Specimen Cards

• The importance of demographic information in Newborn Screening Cards
• Get a good address & phone #!
Got Results?

Distribute & open the blood spot cards with your individual results

• What it feels like to be “In their Shoes”
Case Scenarios
Discussion of the “In Their Shoes” cards

• Newborn Screening Case Scenarios:
  - Good news!
    Screen -
  - Mixed news
    Screen +/- Test -
  - Bad news!
    Screen +/- Test +
  - Really bad news!!
    Screen +/-??
Newborn Screening Resources

- Information and handouts for providers & families
- AWHONN - Position Statement
- AAP – Resources
- ACOG - Resources
- ACMG “ACT” sheets
- March of Dimes - NBS
More Resources

• Genetic Alliance
  http://www.geneticalliance.org/

• Baby’s 1st Test
  http://www.babysfirsttest.org/

• Save Babies through Screening
  http://www.savebabies.org

• STAR-G
  http://www.newbornscreening.info/Parents/facts.html

• NewSTEPs
  https://newsteps.org/

• State Health Departments
Virginia is leading the way!

• Our Website:
  http://www.newbornscreeningeducation.org/

• Our Grants:
  – HRSA, NewSTEPS

• Our Nurse:
  Jennifer O. Macdonald, MPH, BSN, RN
  Virginia Newborn Screening Program
  Virginia Department of Health
  109 Governor St., 9th floor
  Richmond, VA 23219
  (P) 804-864-7729  (F) 804-864-7807
Good News

• Newborn Screening Saves Lives Act
  – Funding research – new tests and treatments
  – Funding and improving screening

• CoIIN
  – The Collaborative Improvement and Innovation Network

• March of Dimes initiatives
  – Quality Improvement & Timeliness
Top Ten Reminders!

NEWBORN SCREENING
Top Ten Reminders

1. The optimum time for collection of the Newborn Screen (NBS) is when the baby is between 24 to 48 hours of age, or before a RBC transfusion. It is no longer based on feeding time.

2. Be sure to detach the top sheet from the NBS collection form and hand to the mother/parents (this is the parent's information sheet). Make sure the mother/parents also receive the two NBS brochures entitled, "Protecting Your Newborn" and "Your Baby’s First Hearing Test."

3. Fill out the collection form completely and accurately, printing clearly and in the correct boxes. Incomplete or absent sample collection date and time will not be amendable after testing has been completed, and a repeat screen will be necessary.

4. Enter the date and time of only the most recent RBC transfusion when applicable. The date and time of any antibiotics that were given is not needed.

5. Don’t confuse meconium ileus with meconium aspirate on the health status section. Check this box only if meconium ileus is present as it is often a sign of Cystic Fibrosis.

6. Please obtain two phone numbers to contact the mother, and make sure the correct Primary Care Physician is provided (the one who will be seeing the child after they are discharged). It is essential that this information is accurate in case the family needs to be immediately contacted due to high-risk screening results.

7. The heel stick is the preferred method of sample collection. New testing methods that are being added encounter interference with both anticoagulants, EDTA, and Heparin, so please avoid the use of these for newborn screening sample collection.

8. Get the sample to the state courier pickup site or mail-out site as soon as it has thoroughly air-dried (3 to 4 hours). Prompt sample transit time can be life-saving for the baby.

9. All ill and premature newborns require a repeat screen between 7 to 14 days of age. All premature newborns less than 34 weeks gestational age or less than 2000 grams at birth are recommended to have a third screen at 28 days of age.

10. Please check out the updated NBS website at health.mogs.gov/lab/newborn. It has useful and important information and provides updates in the “What’s New” tab.

If you have questions, please contact the State Newborn Screening Laboratory Manager by phone at 573-751-3660.

Source: Missouri Health Dept
Questions & Discussion

• Discussion about challenges & opportunities
HELP!!!
The Government has my DNA!
Critical Congenital Heart Disease (CCHD) screening

The 7 Targets:

1. hypoplastic left heart syndrome (HLHS)
2. pulmonary atresia
3. transposition of the great arteries
4. truncus arteriosus
5. tricuspid atresia
6. tetralogy of fallot
7. total anomalous pulmonary venous return (TAPVR)
Critical Congenital Heart Disease (CCHD) screening

- Pass/Fail vs. Neg/Positive
- How to talk to parents about this
Life Saving!

The newborn screening YOU perform can be life saving!
The End

Thank you for all you do!

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References