Tales from the Crib
The Texas Newborn Screening Program

March of Dimes Visiting Professorship in Nursing Conference
September 23, 2014
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Medical Director Texas Newborn Screening Program
BASICS: PURPOSE OF NEWBORN SCREENING

- Program to screen for 31 congenital and heritable disorders.
- Disorders may cause severe intellectual disability, chronic illness, or death with no clinical symptoms.
- Early detection and treatment leads to dramatic positive outcomes for most affected babies.
- Typically treatment through diet control, hormone replacement, and medical supervision.
Many disorders may cause irreparable damage in the first days of life.

Changes in diet and/or other simple interventions can prevent lifelong consequences.

If an abnormal result occurs, prompt follow-up is critical.

Accuracy in testing and correct demographic information are essential.
As of September 1, 2014 Texas screens for 31 disorders:

- 29 rare disorders: Newborn Screening blood spot specimen.
- Congenital hearing loss is a point of contact Newborn Screen.
- Critical Congenital Heart Disease is another point of contact Newborn Screen.
Newborn Screening Advisory Committee

Purpose of Advisory Committee:

To advise DSHS on strategic planning, policy, rules and services related to newborn screening tests.

• 9 member committee formed in May 2010
• 2 additional members to be added 2014

Last meeting June 20, 2014
TEXAS EARLY HEARING DETECTION AND INTERVENTION
Hearing screening by one of two tests:

- Otoacoustic Emissions (OAE).
- Automated Auditory Brainstem Response (AABR).
TEHDI: 1 – 3 – 6 MONTH PATH

BEFORE 1 MONTH SCREENING
- HEARING SCREEN at Birth Facility
  - PASS
  - 2ND HEARING SCREEN at Birth Facility before discharge
    - PASS
    - FOLLOW-UP SCREEN as an outpatient
      - PASS
      - Refer to Early Childhood Intervention (ECI)
      - Early Childhood Intervention (ECI)
      - Audiological/Hearing Diagnostic Evaluation

BEFORE 3 MONTHS CONFIRMATION DIAGNOSIS
- TO AUDIOLOGIST: DIAGNOSTIC EVALUATION Using Texas Evaluation Protocol
  - Report Results & Referrals-TEHDI
- HEARING LOSS CONFIRMED
  - Referral to Early Childhood Intervention (ECI)

BEFORE 6 MONTHS
- ENROLLED IN ECI AND RECEIVING APPROPRIATE EARLY INTERVENTION, HEARING & MEDICAL SERVICES
  - Report Services - TEHDI
- HEARING AID EVALUATION/FITTING WHEN APPROPRIATE
  - Report Services - TEHDI
Critical Congenital Heart Disease

- 20-30% of all congenital heart defects
- 2/1000 potentially lethal - “critical”
  - Requiring expert cardiac care and intervention in the immediate NB period or early infancy
- In the US, about 4800 babies are born each year with CRITICAL CHD
- One of the leading causes of death in infants < 1 year old
CCHD Screening

- US Health and Human Services (HHS) Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC)
  - In 2010, recommended that CCHD be added to the newborn uniform screening panel to identify newborns with structural heart defects associated with hypoxia that could have significant morbidity or mortality early in life with closing of the patent ductus arteriosus or other physiologic changes
  - 2011, Endorsed by Secretary of HHS Kathleen Sibelius
  - 2013 Texas HB 740 added CCHD to core panel in 83(R) session
- Implementation in Texas: September 1, 2014
Transitional Circulation
The seven defects classified as CCHD are:

1. Hypoplastic Left Heart Syndrome (HLHS)
2. Pulmonary Atresia with intact septum (PA/IVS)
3. Tetralogy of Fallot (TOF)
4. Total Anomalous Pulmonary Venous Return (TAPVR)
5. Transposition of the Great Arteries (TGA)
6. Tricuspid Atresia (TA)
7. Truncus Arteriosus communis (TAC)
How is it done?
Critical Congenital Heart Disease
Newborn Screening Algorithm

Pulse ox on right hand and foot after 24 hours

≥95% in right hand or foot and ≤3% difference between right hand and foot
PASS

90% - 94% in right hand and foot

≥3% difference between right hand and foot
Indeterminate
Repeat screen in 1 hour

<90% in right hand or foot
POSITIVE (FAIL)

Indeterminate
Repeat screen in 1 hour

Remind parents that CHD newborn screening may not find all types of problems in a baby's heart.

A Joint Educational Initiative of
The University of Texas Health Science Center at San Antonio/Department of Pediatrics, Baylor College of Medicine/Department of Pediatrics and Texas Department of State Health Services

[Image]

14
Texas CCHD Reporting Form

Critical Congenital Heart Disease Reporting Form

Chapter 37, Subchapter B of the Texas Administrative Code requires a physician, nurse, or any other individual who has information of a confirmed case of a disorder for which screening test is required, to report the confirmed cases to the department.

Instructions:
1. Complete form for all confirmed CCHD cases
2. Print form
3. Manually sign form
4. Fax signed form to 512-776-7593 Attention: CCHD Program

Facility Name: __________________ Facility Location (City): __________________

Medical Record #: __________________ Mother Texas Resident: □ Yes □ No

Facility Type: □ Hospital □ Children’s Hospital □ Birthing Center □ Home Birth

Baby’s Name: First _______ Last _______ Date of Birth: __________

Baby’s Ethnicity: □ White □ African American □ Hispanic □ Asian □ Native American □ Other

Patient Age (in hours at time of screening): ___________ Sex: □ M □ F Unknown

Mother’s Name: First _______ Last _______ Mother’s Date of Birth: __________

Diagnosis: □ First report on this baby □ Update to a previously reported case

Diagnosis Target Condition

□ Hypoplastic left heart syndrome
□ Pulmonary atresia with intact septum
□ Ebstein’s anomaly
□ Total anomalous pulmonary venous return
□ Congenital diaphragmatic hernia
□ Coarctation of the aorta
□ Vascular ring

Secretary Target Condition

□ Seen during the prenatal period
□ Seen during the postnatal period

Comments:

Diagnosis Timeframe (choose only one):

□ Prenatal diagnosis
If prenataly diagnosed, did prenatal and post-natal diagnosis match? □ Yes □ No

If no what was the prenatal diagnosis?

□ Post-natal diagnosis prior to pulse oximeter screening
□ Post-natal diagnosis with pulse oximeter screening
□ Post-natal diagnosis after a normal pulse oximeter screening

Was post-natal echocardiogram performed? □ Yes □ No

Delivery Outcome: □ Live Birth □ Non-live birth

Treatment: □ Surgery □ Medical management □ Supportive care

Baby Status: □ Baby Living □ Baby Expired

Infant was transported: □ Yes □ No

If yes indicate for what purpose(s):

□ Evaluation
□ Treatment

Infant has:
□ Isolated heart disease
□ Multiple anomalies
□ Syndrome/chromosomal anomaly diagnosed

Printed name of person completing report: __________________ Title: __________________

Signature of person completing report: __________________ Date sent: __________

Fax signed form to 512-776-7593 Attention: CCHD Screening
NEWBORN BLOOD SPOT SCREEN
Disorders fall into the following categories:

- Organic acid disorders.
- Fatty acid oxidation disorders.
- Amino acid disorders.
- Hemoglobinopathies.
- Endocrine disorders.
- Other disorders.
<table>
<thead>
<tr>
<th>Condition (acronym)</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>IVA (isovaleric acidemia)</td>
<td>MSUD (maple syrup urine disease)</td>
</tr>
<tr>
<td>GA I (glutaric acidemia)</td>
<td>HCY (homocystinuria due to CBS deficiency)</td>
</tr>
<tr>
<td>HMG (3-OH 3-CH3 glutaric aciduria)</td>
<td>CIT (citrullinemia)</td>
</tr>
<tr>
<td>MCD (multiple carboxylase deficiency)</td>
<td>ASA (argininosuccinic acidemia)</td>
</tr>
<tr>
<td>MUT (methylmalonic acidemia due to mutase deficiency)</td>
<td>MCAD (medium-chain acyl-CoA dehydrogenase deficiency)</td>
</tr>
<tr>
<td>Cbl A,B (methylmalonic acidemia)</td>
<td>Hb SS (Sickle cell anemia)</td>
</tr>
<tr>
<td>3MCC (3-methylcrotonyl-CoA carboxylase deficiency)</td>
<td>LCHAD (long-chain L-3-OH acyl-CoA dehydrogenase deficiency)</td>
</tr>
<tr>
<td>PROP (propionic acidemia)</td>
<td>Hb S/C (Hemoglobin S/C disease)</td>
</tr>
<tr>
<td>TYR I (tyrosinemia type I)</td>
<td>CH (congenital hypothyroidism)</td>
</tr>
<tr>
<td>VLCAD (very long-chain acyl-CoA dehydrogenase deficiency)</td>
<td>CAH (congenital adrenal hyperplasia due to 21-hydroxylase deficiency)</td>
</tr>
<tr>
<td>Hb S/Th (Hemoglobin S/beta-thalassemia)</td>
<td>Cystic Fibrosis</td>
</tr>
<tr>
<td>TFP (trifunctional protein deficiency)</td>
<td>BIOT (biotinidase deficiency)</td>
</tr>
<tr>
<td>CUD (carnitine uptake defect)</td>
<td>GALT (classical galactosemia)</td>
</tr>
<tr>
<td>PKU (phenylketonuria)</td>
<td>BKT (beta-ketothiolase deficiency)</td>
</tr>
<tr>
<td>SCID (Severe Combined Immunodeficiency)</td>
<td></td>
</tr>
</tbody>
</table>
• Testing for CF added in December 2009.
• 297 cases confirmed as of July 15, 2014
Severe Combined Immunodeficiency
Storage and Use of Residual Dried Blood Spots
HB 411 - 82nd Legislature

New Requirements Regarding the Storage and Use of Dried Blood Spots after Completion of Newborn Screening
House Bill 411

- Strengthens DSHS management review and approval processes of proposed uses of dried blood spots after screening is completed

- Effective June 1, 2012 - Requires parental consent for:
  - External public health research uses
  - Storage longer than 2 years

- New “Decision” Form
Specimen Collection Kit

Texas Newborn Screening Parent Information

What happens to the blood spot card after testing?
- DHS keeps the blood spot cards in a secure place for up to two years. By Texas law (Health & Safety Code Sec. 33.018(b)-(c)), the blood spots may be used during that time. Uses include:
  - DHS and external quality assurance to make sure tests, equipment, and supplies are working right
  - Developing new tests; and/or
  - DHS studies of diseases that affect public health.
- If you give your OK, your baby's blood spot cards will be stored for up to 28 years, and they may be used for public health research outside of DHS.

Complete, sign, and return the "Parent Decision Form for Storage and Use of Newborn Screening Blood Spot Cards" to make your choice.

For more information, call (888) 969-7111 ext. 733 or visit:
www.dhs.state.tx.us/lab/newbornscreening.shtm
House Bill 411 (2011) includes requirements for healthcare providers for the distribution of forms to parents regarding the storage and use of dried blood spots cards after completion of the newborn screen.

8½X11 versions of form are available at:
http://www.dshs.state.tx.us/lab/nbsBloodspots.htm
Form MUST be distributed to parents upon collection of all Newborn Screening specimens.

Parents may choose:

- Specimens stored for up to 25 years and made available for possible public health research uses outside of DSHS.
- Specimens destroyed within 2 years and not allowed for research uses outside of DSHS.
FINAL STEPS

Parents may:
- Complete the decision form and return to the healthcare provider to be shipped with any regular newborn screening specimen shipment; OR
- Mail in at a later date.

Healthcare providers must:
- Check the box on the patient demographic form to indicate that the Decision form has been distributed.
IMPORTANT:

• The Use and Storage forms DO NOT allow the parent to decline NBS screening.

• The only legal reason a parent can decline the screen is for religious tenets or practices per Texas Health & Safety Code Sec. 33.012.
**TIMING**

- 1\textsuperscript{st} blood sample is collected at 24 – 48 hours after birth or before transfusion or discharge, regardless of weight or feeding status.
- 2\textsuperscript{nd} sample is recommended to be collected at 7 – 14 days of age.
- The later a specimen is drawn outside this timeframe, the greater the chance the screen may not identify a disorder.
WHY TWO SCREENING TESTS?

1st Screen

• The tests for certain disorders pick up abnormal levels produced by the stress of birth.

• Abnormal levels for some disorders may normalize by the second screen.

• Early testing may mean the difference between life and death for a patient.
2nd Screen

• Some disorders may be missed on the 1st screen due to infant physiology.

• The second screen is necessary to capture some disorders not picked up on the first screen.

• The CF testing protocol requires two screens.
IT TAKES A TEAM

- NBS Laboratory Services.
- NBS Clinical Care Coordination.
- Medical Providers/Medical Facilities.
- Parents and/or Caregivers.
NBS LABORATORY SERVICES
• Operates 6 Days a week
• Testing processes begin on all specimens within 1 business day of receipt
• Initial (critical) results available in as little as 24 hours
• All results reported within 4-5 business days
LAB TESTING THE BLOOD SPOTS

- Approximately 380,000 infants born in Texas each year.
- Approximately 750,000 newborn screens per year.
- Approximately 2,500 specimens received each day.
RESULT REPORTING

- Mailed Result Reports
- Web Application (Neometrics)
- HL7 Messaging

http://www.dshs.state.tx.us/lab/nbsRemoteDataServices.shtm
Remote Services

Web-Based System

• Available to any healthcare provider
• Username & password required
• Features:
  - Remote demographic entry/orders
  - Online access to patient results
  - Report cards to be available online soon

Goal – for all submitters to have access to the online system

HL7 File transfer capabilities

- Direct transfer of demographics and results between computer systems
- 3 large hospital systems fully implemented (~10% of all specimens)
- Several facilities waiting to start implementation
Sign Up in 3 Easy Steps

1. Download forms from:
   http://www.dshs.state.tx.us/lab/nbsRDSforms.shtm

2. Complete
   A. Security/Confidentiality Agreement (1 per facility)
      AND
   B. Web User Agreements (1 for each user)

3. Submit the completed forms:
   Fax to: 512-458-7452 ATTN: DSHS Laboratory Services;
   L457.1 Web Services
   Or e-mail to: remotelabsupport@dshs.state.tx.us
**NORMAL SCREEN REPORT**

### Overview

This is a normal screen report from the Texas Department of State Health Services. The report includes a detailed screening of various disorders, with all results indicating normalcy.

### Key Elements

1. **Screening Result:**
   - **Overall Specimen Result:** Normal

2. **Result Table:** Results in the table are listed by the category of the disorder.

3. **List of Disorders:** Complete listing of disorders screened in each category appearing in the result table.

### Table: Normal Screen Results

<table>
<thead>
<tr>
<th>Disorder Type</th>
<th>Screening Result</th>
</tr>
</thead>
<tbody>
<tr>
<td>Amino Acid Disorders</td>
<td>Normal</td>
</tr>
<tr>
<td>Fatty Acid Disorders</td>
<td>Normal</td>
</tr>
<tr>
<td>Organic Acid Disorders</td>
<td>Normal</td>
</tr>
<tr>
<td>Galactosemia</td>
<td>Normal</td>
</tr>
<tr>
<td>Biotinidase Deficiency</td>
<td>Normal</td>
</tr>
<tr>
<td>Hypothyroidism</td>
<td>Normal</td>
</tr>
<tr>
<td>CAH</td>
<td>Normal</td>
</tr>
<tr>
<td>Hemoglobinopathies</td>
<td>Normal</td>
</tr>
<tr>
<td>Cystic Fibrosis</td>
<td>Normal</td>
</tr>
<tr>
<td>SCID</td>
<td>Normal</td>
</tr>
</tbody>
</table>

### Additional Notes

- The normal screen identifies infants at increased risk for specific disorders. The reference values for all screened disorders are normal. Analytical results are only listed for those analytes that are normally and significantly elevated above the reference range. No results are reported for analytes that are not normally elevated in newborns. When the normal screening age is defined before the age of 28 years, the test may not identify some of these conditions. If there is a clinical concern, additional testing should be considered.

- An increased level of a particular analyte may be caused by a disease or condition. The presence of an increased level of an analyte does not prove the presence of a disease or condition. A normal level of an analyte does not exclude the presence of a disease or condition.

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Very low number of T-cell receptor excision circles (TREC). Please follow recommendations received from DSHS Newborn Screening Clinical Care Coordination Team.

For more information, please refer to http://www.dshs.state.tx.us/lab/newbornscreening.htm
TEXAS NEWBORN SCREENING
CLINICAL CARE COORDINATION
• Medical Director.
• Registered Nurses.
• Public Health and Prevention Specialist (PHPS).
  – Nurses and PHPS are assigned to specific disorders.
  – Nurses and PHPS are cross-trained for full coverage Monday – Saturday.
• Educators.
• Ombudsman.
SHORT TERM FOLLOW-UP

Overview

- A case is opened for each screen positive result.
- Cases are monitored until an infant is cleared or diagnosis is determined.
In Fiscal Year 2013, the DSHS laboratory screened approximately 745,000 specimens for metabolic, endocrine, and hematological disorders.

Of those screens approximately 16,000 were abnormal screens that required follow-up by Clinical Care Coordination.

There were approximately 750 diagnosed cases in Fiscal Year 2013.
FINDING THE MEDICAL PROVIDER

• Find the Medical Provider responsible for the medical care of the baby.
  • Determine if the baby is in the hospital.
• If a Medical Provider can be located:
  • Provide results.
  • Provide guidance for recommended actions.
If a Medical Provider cannot be located:

- Contact parents to obtain Primary Care Provider (PCP) information.
- If a PCP is not identified:
  - Provide results to family.
  - Direct family to an Emergency Department (ED) if necessary.
  - Clinical Care Nurse will coordinate with ED staff if family directed to ED.
WHEN ALL ELSE FAILS

If baby cannot be located:

• Utilize DSHS Regional Social Workers to assist with:
  - Locating the baby.
  - Connecting baby with health-care providers and services.

• Involve other agencies, including law enforcement and/or CPS if necessary.
RESOURCES DISTRIBUTED

Screen Positive NBS

- Information mailed to parent.
- NBS letter
- General NBS Brochures
SHORT TERM FOLLOW-UP

POSITIVE SCREEN WITH VERY ELEVATED LEVELS: MEDICAL EMERGENCY

• Reported immediately to nurses in NBS Clinical Care Coordination.

• Nurse will notify PCP by phone and fax the same day the laboratory results reports are received from the DSHS lab.

• If no PCP is on record for the newborn or cannot be located, the nurse will notify the parents directly.
RESOURCES DISTRIBUTED FOR A NEWBORN REQUIRING URGENT FOLLOW-UP

Faxed to Medical Provider

- NBS letter with:
  - NBS disorder-specific lab results.
  - Contact information for the NBS Nurse responsible for the NBS case.
  - Disorder-specific ACT/FACT Sheet.
- List of regional subspecialist consultants.
• Adapted from the American College of Medical Genetics (ACMG).
• Designed for the medical provider.
• Contain the following:
  - Differential Diagnosis.
  - Condition Description.
  - For medical emergencies, follow the instructions in the black outlined box.
• Available on the NBS Clinical Care Coordination website.
FACT SHEETS FOR PARENTS

- Each disorder has a FACT sheet that is modeled from the ACMG Fact Sheet.
- Designed for the PCP to share with the family.
- Information for the parents about symptoms, treatment, and things to remember for the specific disorder.
- Available on the NBS Clinical Care Coordination website.
- Available in English and Spanish.
MEDICAL PROVIDERS AND FACILITIES
PRIMARY CARE PROVIDER AND FACILITY RESPONSIBILITIES

Birth Hospital:
• Assist with locating baby if needed.
• Identify PCP for infant.

PCP:
• Agree to follow-up with newborn/family.
• Agree to accept patient into practice.
• Refer to subspecialists as appropriate.
PARENTS AND CAREGIVERS
- Parent provides PCP information to Clinical Coordination Staff.
- If the newborn does not have a PCP:
  - Parent is asked to identify a PCP.
  - Take infant to ED if necessary.
- Parent must follow-up to ensure newborn:
  - Attends appointments.
  - Receives treatment and care if diagnosed.
LONG TERM FOLLOW-UP

Goal: To ensure the best possible outcome for individuals with disorders identified through newborn screening.

Components:

1. Care coordination through a medical home.
2. Evidence-based treatment.
3. Continuous quality improvement.
LONG TERM FOLLOW-UP

What is Involved?
• Continuing PCP/specialist visits.
• Continuing documentation of treatment.
• Parental involvement.
• Physician/specialist participation.

How long is a child in long term follow-up?
• Begins when an infant receives a confirmatory diagnosis.
• Continues until child is 4-21 years old, depending on the disorder.
LONG TERM FOLLOW-UP

Why Track Long Term?

• Evaluate effectiveness of the NBS Program.
• Develop evidence-based treatment.
• Improve treatment of affected individuals.
• Provide continuous quality improvement.
NBS BENEFITS PROGRAM
WHAT IS THE NBS BENEFITS PROGRAM?

• Redesigned in 2007 to account for the expansion of NBS disorders screened.

• Targets families without Medicaid or private insurance.
WHO IS ELIGIBLE FOR NBS BENEFITS?

- Those with a presumed positive screen or a confirmed diagnosis of a disorder screened for in the Texas Newborn Screening Program.

- An income at or below 350% of the federal poverty income level (FPL).

- Texas resident
WHAT ARE THE NBS BENEFITS FOR PATIENTS?

- Confirmatory testing.
- Dietary supplements.
- Metabolic foods.
- Low-protein foods.
- Medications.
- Vitamins.
- Follow-up care.
What’s Next: Secondary Conditions on RUSP Panel

24 additional Secondary Conditions

- Secondary conditions are believed to be clinically significant, but some may have an unclear natural history or lack appropriate medical therapy that affects long-term outcome.

- Detected during screening for core conditions.

- The additional conditions will be detected through the same newborn screen specimen collected from a heel stick and tested at the Texas Department of State Health Services Laboratory.

- No additional blood spots will need to be collected.

- No additional fee is anticipated at this time*

*a cost estimate of the whole newborn screening panel is expected in 2015
### Secondary Conditions

<table>
<thead>
<tr>
<th>Condition</th>
<th>Diagnosis</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cbl C,D (Methylmalonic acidemia with homocystinuria)</td>
<td>MAL (Malonic academia)</td>
</tr>
<tr>
<td>IBG (Isobutyrylglycinuria)</td>
<td>2MBG (2-Methylbutyrylglycinuria)</td>
</tr>
<tr>
<td>3MGA (3-Methylglutaconic aciduria)</td>
<td>2M3HBA (2-Methyl-3-hydroxybutyric aciduria)</td>
</tr>
<tr>
<td>SCAD (Short-chain acyl-CoA dehydrogenase deficiency)</td>
<td>M/SCHAD (Medium/short-chain L-3-hydroxyacyl-CoA translocaase deficiency)</td>
</tr>
<tr>
<td>GA2 (Glutaric acidemia type II)</td>
<td>MCAT (Medium-chain ketoacyl-CoA thiolase deficiency)</td>
</tr>
<tr>
<td>DE RED (2,4 Dienoyl-CoA reductase deficiency)</td>
<td>CPT IA (Carnitine palmitoyltransferase type I deficiency)</td>
</tr>
<tr>
<td>CPT II (Carnitine palmitoyltransferase type II deficiency)</td>
<td>CACT (Carnitine acylcarnitine translocaase deficiency)</td>
</tr>
<tr>
<td>ARG (Argininemia)</td>
<td>CIT II (Citrullinemia, type II)</td>
</tr>
<tr>
<td>MET (Hypermethioninemia)</td>
<td>H-PHE (Benign hyperphenylalaninemia)</td>
</tr>
<tr>
<td>BIOPT (BS) (Biopterin defect in cofactor biosynthesis)</td>
<td>BIOPT (REG) (Biopterin defect in cofactor regeneration)</td>
</tr>
<tr>
<td>TYR II (Tyrosinemia, type II)</td>
<td>TYR III (Tyrosinemia, type III)</td>
</tr>
<tr>
<td>Var Hb (Various other hemoglobinopathies)</td>
<td>*GALE (Galactoepimerase deficiency)</td>
</tr>
<tr>
<td>*GALK (Galactokinase deficiency)</td>
<td>T-cell related lymphocyte deficiencies</td>
</tr>
</tbody>
</table>

*At this time NBS is not testing*
What’s Next:

Pompe

- DCHDNC recommended adding May 2013
- HHS Secretary Sebelius requested the ICC review Pompe NBS with recommendation by July 31, 2014

MPS1, X-ALD

- DCHDNC sent to formal evidence review, which is the next step in moving condition forward for consideration
Case 1  Hypothyroidism

BG D was the 3.4 Kg product of a uncomplicated term gestation

- DOL 2 - 1st NBS collected
- DOL 8 - Received by DSHS Lab, no PCP listed.
- DOL 11 - Results to CCC. Hospital contacted, provided PCP name. PCP notified of abnormal screen and guidance provided, parent letter mailed
- DOL 30 family call-changing PCP, but no new PCP identified
- DOL 32 family phone not answered certified letter sent to home
- DOL 35 Regional SW to home, Appointment made with new PCP
- DOL 38 PCP office noted baby scheduled for lab draw
- DOL 39 No show, PCP had difficulty reaching family
- DOL 42 SW back to home, new PCP identified
- DOL 43-1 Year Child could not be located. At 1 year of age located, admitted to PICU with severe developmental delay and malnutrition.
Baby Girl J was the 2.5 KG product of a 36 week uncomplicated gestation

DOL2 – 1st NBS obtained

DOL3 – Received by NBS Lab

DOL4 - Elevated phenylalanine and elevated phenylalanine/tyrosine ratio. PCP notified, requested repeat NBS obtained same day. Referred to metabolic MD by PCP.

DOL 6 – Evaluated by metabolic MD, confirmatory testing obtained

DOL8- 2nd NBS obtained, phenylalanine remained elevated

DOL9 – Diagnosis of classical PKU confirmed
Case 3 - CAH

Baby Boy T 3.2Kg product of uncomplicated term pregnancy

- DOL 2 NBS obtained, DOL 6 Sample received at lab
- DOL 7 Preliminary result very elevated 17-OHP
  - Birth Hospital contacted, child already discharged - newborn PCP listed as PCP, but when contacted stated not accepting new Medicaid patients
  - Family contacted- No PCP identified
  - Child referred to ED of local hospital, guidance given to ED
  - ED obtained electrolytes but refused to obtain 17-OHP, Na 133, K4.9
  - Education provided to ED, stated no Pedi Endocrine in area. Transfer to tertiary center arranged
  - Tertiary center requested 17-OHP be obtained prior to transfer, 17-OHP obtained three hours later at local ED
- DOL 8 Final 17-OHP very elevated
- DOL10 regional social worker contacted to help find PCP
- DOL11 Family directed back to ED for additional lytes by social worker. Still no PCP
- DOL17 17-OHP level returned. Still no PCP. Child obtained appropriate care
Case 4 – CF Twins

- Baby Boy’s Smith were the 1.2kg and 1.8kg products of a 31 week monozygotic twin gestation born to a 29 G1P0 female by primary C section. Pregnancy was complicated by a maternal, unrepaired menigomylocoel with associated sequelae.

- Limited prenatal care was obtained in Mexico but concern was noted of a possible twin-twin transfusion. Maternal history notes the use of oral steroids due to “maternal short stature” 3 weeks prior to delivery.
Case 4

CF TWINS

Twins were delivered by C section. Indication at time of presentation twin A was noted to have fetal ascites polyhydramnious, hydrops, and no end diastolic blood flow.

- At time of attempted placement of an umbilical line twin A had significant bleeding and the decision to transfuse before obtaining NBS was based on clinical status.

- During the first day of life child received FFP, packed irradiated RBC, and required RBC, FFP, cryoprecipitate, and platelets over the next few days.

- Child was noted to have in utero bowel rupture.

- First NBS collected at 48 hours of life. Child received a total of 220ml pRBC (183ml/kg), 125ml FFP (104ml/kg), and 20ml cryoprecipitate prior to NBS being obtained.
Case 4  CF TWINS

Twin B

- Child had initially appeared stable but was noted to have scrotal discoloration
- During w/u for scrotal discoloration fecal contents were noted in scrotum.

- Further work up noted bowel perforation.

- NBS was obtained prior to transfusion with packed irradiated RBC and FFP
<table>
<thead>
<tr>
<th>First Screen:</th>
<th>Second Screen:</th>
<th>DNA:</th>
<th>Third Screen:</th>
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<tbody>
<tr>
<td>Collected DOL 2 (prior to transfusion), Received DOL 5, Reported DOL 9</td>
<td>Collected on DOL 11, Received DOL 13, Reported DOL 20</td>
<td>Elevated IRT</td>
<td>at our request elevated IRT DOL 28</td>
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<tr>
<td>Elevated IRT</td>
<td>Elevated IRT</td>
<td>DNA: Homozygous F508 Deletion</td>
<td></td>
</tr>
</tbody>
</table>
Case 4  CF Twins

NBS on Twin A

First Screen:
Collected DOL 2
Received DOL 5
Reported DOL 7
IRT not out of range

Second Screen:
Collected DOL 10
Received DOL 14
Reported DOL 17
IRT not out of range

Third Screen:
at our request
Elevated IRT DOL 28
IRT out of range

DNA Studies
(obtained due to twin’s NBS results)
Homozygous F508 deletion
Newborn Screening System
Questions?
CASE 4