Newborn Screening in Texas
Overview

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Goals of Texas NBS Program

Two screening tests for each baby born in Texas

- 24 – 48 hours of age
- 1 – 2 weeks of age

Infants testing positive receive prompt and appropriate confirmatory testing.

Diagnosed infants are maintained on appropriate medical therapy.
Texas Newborn Screening Program History

• 1963 – Phenylketonuria (PKU) pilot
• 1965 – Mandated PKU screening
• 1978 – Added Galactosemia & Homocystinuria screening
• 1980 – Added Congenital Hypothyroidism screening, Recommended second screen
• 1983 – Discontinued Homocystinuria screening, added Hemoglobinopathy screening, Required second screen
• 1989 – Added Congenital Adrenal Hyperplasia screening
• 1995 – Added second-tier DNA testing for hemoglobinopathies
• 2000 – Added hearing screening
Texas Newborn Screening Program History (cont)

- 2005 - NNSGRC review – external review of NBS program
- 2005 - House Bill 790 mandated expansion to ACMG recommended core panel of 29 disorders as funding allowed
  - No funding for Cystic Fibrosis provided
  - Required cost effectiveness study
- May 2006 - Cost effectiveness study complete, testing to be performed by DSHS Laboratory
- December 2006 - 1st abnormal MS/MS results reported
  - 19 new disorders
- January 2007 - Added Biotinidase deficiency screening
Texas Newborn Screening Program History (cont)

• 2009 - HB 1672 – added provisions for
  • Disclosure to parents that specimens can be retained & used for research and QA/QC
  • Parents to request specimen destruction
  • Confidentiality of specimens and data
  • Sickle cell trait screening

• December 2009 - Added cystic fibrosis screening
• Spring 2010 - NBS Advisory Committee formed

• 2011 - HB 411 - amended HSC 33.018 related to the confidentiality and changed from an opt-out process for all residual specimen uses to an opt-in process for external research purposes.

• December 2012 – Added SCID
• 2014 – Added Critical congenital heart disease
• 2015 – Added MSMS secondary targets
• 2016 – NewSTEPs review
Currently screen for 53 disorders from DBS

- Congenital Hypothyroidism
- Congenital Adrenal Hyperplasia
- 4 Hemoglobinopathies
- Galactosemia
- Biotinidase Deficiency
- 14 Amino Acid Disorders
- 13 Fatty Acid Oxidation Disorders
- 15 Organic Acid Disorders
- Cystic Fibrosis
- SCID and T-cell related lymphocyte deficiencies

Plus 2 POC screening
- Hearing loss
- CCHD
The Logistics of Newborn Screening in Texas

• The healthcare provider requests specimen collection forms.
• DSHS Laboratory assigns form serial numbers to the healthcare provider and ships the forms.
• Healthcare provider collects the specimen and sends it to DSHS.
• Specimen is assigned a laboratory ID number in the laboratory data system.
• Demographic sheet is separated from the blood spots and sent to Demo Entry Team where the information is entered into the database.
• Specimen is sent to NBS Laboratory for testing.
Types of Kits

Medicaid / CHIP / Charity

- Newborn or mother is eligible for Medicaid
- Newborn is eligible for CHIP
- Baby doesn’t have insurance or other payment source
- Kits are free to provider
- Newborn is covered by private health insurance
- The facility orders and pays for kits and seeks reimbursement for services
- $55.24 per kit.
- Payment is due within 90 days of invoice date
NBS Fee Components

- Direct Lab Costs
  - Testing staff time
  - Reagents and consumables
  - Equipment maintenance
  - Laboratory Information Management System maintenance

- Lab Overhead
  - Specimen check-in, demo entry, reporting, QA, safety
  - Testing equipment
  - Courier
  - Laboratory building costs
NBS Fee Components

- Agency Overhead
  - Administrative
  - Purchasing
  - Legal
  - Contracts
  - IT
  - HR

- Contingency Cost

Clinical Care Coordination
Process to Change Fee

- WLU costing is performed
  - Staff time and consumables/reagents are captured
  - Lab and agency overhead and contingency cost added

- New fee is proposed in changes to Rule
  - Rules Coordination Office
  - Executive Council review
  - 30-day public comment period in Texas Register
  - Response to public comments
  - Published in Texas Register for rule adoption with effective date or 20 days after date of publication rule becomes effective.
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NBS Kit Components

• Parent Information
• Parent Decision Form
• Demographic Form (white and yellow copies)
• Filter Card
• Instructions and Fold Over Flap
Parent Decision Form

• HB411 became law in June 2011 and made major changes to NBS specimen retention and residual use.

• Opt-in for long term storage and possible research uses - effective June 1, 2012 – Use of Parent Decision Form
  • ~49% of NBS have a parent decision form returned
  • ~74% of those returned and valid give permission for longer storage and external public health research uses (36% of all newborns)
% of Births Screened

- Parents can only refuse to have their child screened if the screening conflicts with a parent’s religious tenets or practices.
- In 2014, 409,111 births were registered in TX and 8,241 (2.0%) were not linked to TX newborn screen database.
  - 645 were TX residents out of state births.
  - 901 deaths occurred within 24 hours after birth.
  - ~6,695 (1.6%) newborns not screened.
Courier Services

• First tier – Lone Star Delivery and Processing
  • Hospitals, Pediatric Clinics
  • 532 NBS submitters
  • 69% of NBS specimens (85% of 1st screens)
  • Pick-up Sun – Fri, deliver Mon - Sat

• Second tier – FedEx
  • 192 NBS submitters
  • 13.8% of specimens (14.5% of 1st screens)
  • Pick-up and deliver Mon - Sat
Pre-analytical Measures

Day 1:
- Birth

Day 2:
- Collection

Day 3:
- NBS Lab

Day 4:
- AbN Critical Results

Day 5:
- All AbN Results

Day 6:
- Complete testing

June – September 2017

96.7% 26.8%
Newborn Screening Workload 2016

- Received 782,187 specimens (~400,000 newborns)
- Specimens Assayed and Reported: 775,084
  - Average 2,573 specimens per day
  - 7,103 unsatisfactory specimens (~0.92%)
- >19,000 (2.4%) specimens reported with presumptive positive results
- ~925 cases diagnosed
- Testing & follow-up performed 6 days a week
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Texas Newborn Screening Laboratory

8 plates are distributed to 5 areas to test for 53 disorders.

**Hemoglobinopathy Screening**
One test is used to identify:
- Sickle Cell Anemia
- Sickle Hemoglobin C Disease
- Sickle/Beta Thalassemia Disease
- Various other hemoglobinopathies

**Endocrine & Cystic Fibrosis Screening**
Three tests are used to identify:
- Congenital Hypothyroidism
- Congenital Adrenal Hyperplasia
- Cystic Fibrosis

**Galactosemia & Biotinidase Screening**
Two tests are used to identify:
- Galactosemia
- Biotinidase Deficiency

**SCID Screening**
One molecular test is used to identify:
- Severe Combined Immunodeficiency
- T-cell related lymphocyte deficiencies

**Tandem Mass Spectrometry Screening**
One test is used to identify:
- 14 amino acid disorders (e.g. PKU)
- 13 fatty acid disorders (e.g. MCAD)
- 15 organic acid disorders (e.g. glutaric acidemia type 1)
Timeline of a specimen in the laboratory...

Day 1
- Specimen arrives.
- Specimen accessioned.
- Demo entry begins.
- Specimen punched.
- Testing begins for all disorders except Hemoglobinopathy (HB), Biotinidase Def (BIOT) and SCID.

Day 2
- Testing begins for Hb, BIOT and SCID.
- Testing completed for IRT and all time critical disorders.
- Demo entry is completed.
- Results for time critical disorders are released.

Day 3
- Results for most time sensitive disorders are released.
- Clinical care coordination contacts provider for out-of-range results for time critical and time sensitive disorders.
- DNA testing for VLCAD, Galactosemia (M-F) and Cystic Fibrosis (M-S), if specimen is out-of-range.

Day 4
- Remaining time sensitive results released.
- Result report is printed, sent to mailroom, and available online.
- Galactosemia & Cystic Fibrosis DNA results are sent to Clinical Care Coordination.

Hemoglobinopathy and MCAD DNA testing performed in weekly batches.
Result Reporting

- Preliminary panic values are immediately forwarded by fax for some disorders
- Final abnormal results immediately generate a case
- Clinical Care Coordination staff begins follow-up protocols with hospitals, physicians, and parents
- All results reported back to submitting provider via mail, fax, web portal and/or HL7 message
ORDERING AND REPORTING OPTIONS

Test Ordering

- DSHS Entered: 83.3%
- Web: 13.1%
- HL7: 3.6%

~130,500 per year (16.7% of NBS) Remote Ordering

Reporting

- Mailed: 68.1%
- Faxed: 16.6%
- Web: 10.4%
- HL7: 4.9%

~117,000 per year (15% of NBS) Electronic Reporting Only
Analytical Measures

June – September 2017
TX Health and Safety Code
Section 33.011

(a-1) Except as provided by this subsection and to the extent funding is available for the screening, the department shall require newborn screening tests to screen for disorders listed as core and secondary conditions in the Recommended Uniform Screening Panel of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children or another report determined by the department to provide more stringent newborn screening guidelines to protect the health and welfare of this state's newborns........The department may exclude from the newborn screening tests required under this subsection screenings for galactose epimerase and galactokinase.
New Conditions on RUSP but not TX NBS Panel

- **Pompe**
  Approved for addition to the Recommended Uniform Screening Panel (RUSP) in March 2015

- **Mucopolysaccharidosis Type I (MPS1)**
  Approved for addition to the Recommended Uniform Screening Panel (RUSP) in February 2016

- **X-linked Adrenoleukodystrophy (X-ALD)**
  - Approved for addition to the Recommended Uniform Screening Panel (RUSP) in February 2016
  - Appropriation of 1.2 million for implementation
Current Grants

- **NewSTEPs 360 grant**
  Achieve timely reporting of NBS results in 95% of newborns as recommended by ACHDNC

- **APHL New Disorders grant**
  Initiate preparation and implementation activities for X-ALD newborn screening

- **CDC SCID NextGen grant**
  Development and validation of laboratory procedures using Next Generation sequencing technologies to assess genes causing SCID
Clinical Care Coordination
CCC Organization chart
Newborn Screening Workload 2016

- Received ~782,187 specimens (~400,000 newborns)
- Specimens Assayed and Reported: ~775,084
  - Average 2,558 specimens per day
  - ~7,103 unsatisfactory specimens (~0.92%)
- ~22,820 (~2.3%) specimens reported with presumptive positive results that required follow-up by CCC
- ~925 cases diagnosed
- ~6,600 Sickle Cell Trait notifications
- Testing & follow-up performed 6 days a week
# 2015-2016 Confirmed Conditions

<table>
<thead>
<tr>
<th>Disorder</th>
<th>2015</th>
<th>2016</th>
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<tbody>
<tr>
<td>Biotinidase Deficiency</td>
<td>39</td>
<td>29</td>
</tr>
<tr>
<td>Congenital Adrenal Hyperplasia</td>
<td>87</td>
<td>92</td>
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<tr>
<td>Cystic Fibrosis</td>
<td>66</td>
<td>71</td>
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<td>Galactosemia</td>
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<td>7</td>
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<tr>
<td>Sickling Hemoglobinopathies</td>
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<tr>
<td>Various Other Hemoglobinopathies</td>
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<td>47</td>
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<tr>
<td>Hypothyroidism</td>
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<td>Metabolic Disorder</td>
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<td>Severe Combined Immune Deficiency</td>
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<td>8</td>
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<tr>
<td>T-Cell Related Lymphocyte Deficiencies</td>
<td>119</td>
<td>82</td>
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<tr>
<td><strong>Totals</strong></td>
<td><strong>934</strong></td>
<td><strong>925</strong></td>
</tr>
</tbody>
</table>
SHORT TERM Follow-up

Overview

• A case is opened for each out-of-range result

• Cases are monitored until an infant is cleared or diagnosis is determined

• Resources are provided for guidance on recommended actions.
URGENT
Follow-up

POSITIVE SCREEN WITH VERY ELEVATED LEVELS: MEDICAL EMERGENCY

• Reported immediately to nurses in NBS CCC
• On the same day, CCC Nurse will notify PCP by phone and fax the laboratory results reports received from the DSHS Lab.
• If no PCP is on record for the newborn or cannot be located, nurse will notify the parents.
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.
Finding The Family

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 the family
  • Direct family to an Emergency Department (ED) if necessary.
  • CCC 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 For Out-of-range Newborn Screen Results

**Urgent Results – Fax to Medical Provider**

- NBS letter with:
  - NBS disorder-specific lab results
  - Contact information for the CCC Nurse responsible for the NBS case
  - Disorder-specific ACT/FACT Sheet
- List of regional subspecialist consultants

**Out-of-Range NBS – Mail**

- Information to parent
- NBS letter
- General NBS brochures
Sickle Cell Trait Notification

• Required to screen by law
• Notify parents by certified letter and informational brochure
• If letter returned, resend not certified
• Contracted with sickle cell association for notification of parents in some regions of state

If certified letter returned, DSHS will utilize resources to locate family
Long Term Follow-Up

• Follow all conditions except CCHD and Sickle Trait for long term follow-up
• Long Term follow-up elements identified by specialists for condition
• Contact at varying times in first year of life
• Yearly contact after first year
• Forms returned by specialist, PCP, or parent
Long Term Follow-Up (continued)

• Girls with Hyperphenylalaninemia receive letter reminding of reproductive risks and importance of diet during pregnancy at 11 years of age
• Databases now being redesigned to reflect harmonized common data elements
• Dedicated personnel for long term follow-up
Advisory Committees

- NBS Advisory Committee was established in 2010.  
  most recent meeting 08/11/2017

- A new Sickle Cell Advisory Committee has been created by Senate Bill 200 and Senate Bill 277, 84th Legislature, Regular Session to raise awareness of sickle cell disease and sickle cell trait.  
  most recent meeting 07/21/2017
System Stakeholders

- Meet yearly with ad hoc specialty committees for technical review/assistance
  - Metabolics
  - Pulmonary
  - Immunology
  - Hematology
  - Endocrinology

- Ad Hoc meetings as needed (Metabolic when implemented secondary conditions, Immunology when SCID implemented)

- Regularly scheduled NBS system stakeholder update conference calls (MOD, TMA, TPS, THA, etc.)
NBS Support Unit

- Ombudsman
- Educators (web and external)
- NBS Program Benefits
- Contracts
- Hearing Screening
Additional DSHS Resources

• Partnered with Children with Special Health Care Needs (CSHCN) Services Program to distribute parental support resources flyer for those with confirmed conditions identified by NBS

• Regional DSHS social workers
NBS Educational Efforts

- Newborn Screening Grand Rounds
- Newborn Screening Journal Club
- Tales from the Crib
  NBS Morbidity and Mortality Conference
- Educational Outreach
  - External Grand Rounds
  - General NBS presentation
  - Webinar General NBS Grand Rounds
NBS Educational Efforts

- Texas Health Steps Modules - CME accredited Provider Education
  - Newborn Screening
  - Sickle Cell Disease and Trait
  - Critical Congenital Heart Disease
  - Newborn Hearing Screening
  - Genetic Screening, Testing, Treatment and Referral
NBS/Genetics Educational Efforts

DSHS Funds:

• Yearly State of the Art Genetics Conferences—designed for primary care providers
  Conference on Newborn Screening 2014

• Baylor Seminars with Genetics—community based genetic seminars

• Teratogen Information Program

• Clinical genetics medical student summer internships

• Hearing Screening Public Health Interns (Blue Ribbon Program)
Educational Projects

• Funded TxPOP1 project
  Tool Kit developed for CCHD screening
  Completed August, 2013

• Funded TxPOP2 project
  • Addressed NICU and rural CCHD screening
  • Developed general NBS and condition specific brochures
  Completed August, 2014
Newborn Hearing Screening Grants

CDC – 3 year $150,000/ Year 1
TEHDI MIS Enhancements

HRSA – 3 Year $250,000/ Year 1
• Texas Hands & Voices Contract
• Continued Parent Support Project with Providers and Families
• Care Coordinator at DSHS (part-time)
• TEHDI Regional Summits
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Thank you