

2015 Alabama Newborn Screening Conference



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The Current State of Newborn Screening

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NewSTEPS

- NewSTEPS and related activities are funded through a cooperative agreement (#U22MC24078) to APHL by the Genetic Services Branch of the Health Resources and Services Administration (HRSA)

NewSTEPS

- NewSTEPS Vision
 - Dynamic newborn screening systems have access to and utilize accurate, relevant information to achieve and maintain excellence through continuous quality improvement

NewSTEPS

- NewSTEPS Mission
 - To achieve the highest quality for newborn screening systems by providing relevant, accurate tools and resources and to facilitate collaboration between state programs and other newborn screening partners

NewSTEPS: Data to Support NBS

Birth Rates Vary Between States

- The number of live births per 1,000 population varies
- May point to different needs

What do we know about our newborn screening systems in the United States?

- 52 newborn screening programs
- 36 newborn screening labs
- Geographically diverse states
 - 663,000 Square Miles to 1,212 square miles

Consent Models for Newborn Screening

- Participation in NBS is assumed in most states
 - One state requires NBS (no option to opt-out)
 - Most states allow parents to opt-out of newborn screening (religious, other)

Storing Samples and Storing Data

Timeliness

National Timeliness Recommendations

- In order to achieve the best outcomes for babies:
 - Presumptive positive results for time-critical conditions should be communicated immediately to the child's healthcare provider but no later than 5 days of life

National Timeliness Recommendations

- Presumptive positive results for all other conditions should be communicated to the child’s healthcare provider as soon as possible but no later than 7 days of life
- All NBS tests should be completed within 7 days of life

ACHDNC Recommendations

- In order to achieve these goals and reduce delays in newborn screening:
 - Initial NBS specimens should be collected in the appropriate time frame for the baby’s condition but no later than 48 hours after birth
 - NBS specimens should be received at the Laboratory as soon as possible; ideally within 24 hours of collection

States are Changing Policies for Delivery of Samples

- “To get nearer the [recommended] time frame, the state last week deployed a new courier service that will pick up and shuttle hospitals’ newborn screening specimens on a quicker, more consistent schedule.”

Screening Practices in States

What is Covered By The Newborn Screening Fees?

- Program administration
- Laboratory tests (includes salaries of laboratory personnel, supplies, instruments and equipment maintenance)
- Information technology support (lab and general)

What is Covered By The Newborn Screening Fees?

- Short-term follow-up services (includes salaries and educational materials)
- Courier services
- Long-term follow-up services
- Bio-bank program
- Metabolic foods and formula

State Practices: Screening for Disorders

- Recommended Uniform Screening Panel
- Counting the Disorders
 - Core
 - Secondary
 - Other
- Screening for disorders on the Recommended Uniform Screening Panel

Secondary Disorders

- “Disorders that can be detected in the differential diagnosis of a core disorder”

National Progress in SCID Implementation

Alabama Receives SCID Grant

- 12 Grantees awarded up to \$150,000/year for two years from APHL for SCID Implementation
- Utilizing funding to support laboratory efforts and facilitate SCID implementation – expected by August 2016

CCHD Screening Progression

CCHD Data Reporting

- Variability throughout the country
 - No data reporting
 - All pulse oximetry values from all babies

CCHD Data Reporting

- **Alabama**
 - Submitting health care provider shall report the results of any failed pulse oximetry screening results to the Alabama Department of Public Health and shall use such forms and follow such guidelines as shall be determined by the State Health Officer

Other Disorders Screened in the United States

- **Universally Screened:**
 - Ethylmalonic encephalopathy – EME (4)
 - Hyperornithinemia with Gyrate Deficiency - Hyper ORN (5)
 - Ornithine transcarbamylase deficiency – OTC (5)
 - Prolinemia Type I/ Type II – PRO (1)

Other Disorders Screened in the United States

- Nonketotic Hyperglycinemia – NKH (5)
- Carbamoyl phosphate synthetase I deficiency – CPS (8)
- Krabbe (1)
- Fabry (2)
- Gaucher (2)
- Niemann Pick (1)

Other Disorders Screened in the United States

- Mucopolysaccharidosis I - MPS I (2)
- Glucose-6-phosphate dehydrogenase deficiency - G6PDD/G6PD (2)
- Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome – HHH (10)
- Pyroglutamic acidemia - 5-OXO (3)
- Congenital Toxoplasmosis – TOXO (5)

Other Disorders Screened in the United States

- Human Immunodeficiency Virus - HIV Exposure (1)
- X-linked Adrenoleukodystrophy (1)

Decision Making, Policies

- **Advisory committees, board of health, commissioner of health**
- **Legislators**



Composition of Advisory Committees

- Consumers or parents of patients affected by screened conditions
- Laboratory representatives of pathology and chemistry
- Pediatric, neonatology and family practitioners

Composition of Advisory Committees

- Pediatric subspecialists (e.g. Endocrine, Hematology, Metabolic etc.)
- Metabolic nutritionists
- Hospital association representative
- March of Dimes representative
- Medical ethicist
- NBS program (management, follow-up and lab) representatives

Newborn Level Data Collected within NewSTEPS

- Purpose
 - “To provide an accurate characterization of the frequency of newborn screening disorders in the U.S., along with timing of screening and diagnostic activities”
- Systematic definitions helpful at local AND national levels

Cystic Fibrosis Example

- Newborn with abnormal newborn screen:
 - IRT 105 ng/ml (normal range < 60 ng/ml)
 - NBS DNA analysis revealed F508/R117H; 7T/9T
 - Referred to CF Center for Sweat Test
 - Sweat test results: 25 mmol/L (diagnostic > 60mmol/L)

CF Diagnosis Can Vary by Clinician

- Dr. Smith: Baby likely has CF Follow monthly and repeat sweat test; tell family baby has CF
- Dr. Jones: Baby has CRMS (Cystic Fibrosis Related Metabolic Syndrome) Not CF, we should follow this baby every 6 months to see if baby develops CF symptoms

CF Diagnosis Can Vary by Clinician

- Dr. Garcia: Baby is fine, no CF, no CRMS No diagnosis, baby does not need to be seen

Surveillance Case Definitions

- This newborn would be classified as CRMS using the case definitions – at the Public Health Level
- These definitions do NOT impact clinical diagnosis
- The burden of CRMS in the U.S. is not well understood

Surveillance Case Definitions

- Classifying NBS results consistently will provide comparisons of screening programs from year to year and across the nation

Efforts to Support NBS Programs and Timeliness

Collaborative Improvement & Innovation Network: CoINN

- Eight states participating in continuous quality improvement activities to address challenges in timeliness:
 - Arizona, California, Colorado, Iowa, New Hampshire, Tennessee, Texas, Wyoming

Collaborative Improvement & Innovation Network: CoINN

- Fifteen-month project, teams of 5 individuals from states, comprised of laboratory, follow-up, hospital staff
- Sharing ideas and collaborating to find solutions



- Funding starting September 1
- Will support at least 20 state newborn screening programs to improve timeliness over three years
- Build on success of CoINN
- Competitive funding opportunity
- Applications will be released week of 9/21/15

Project Instant Gratification (PIGs)

- Giving tools back to states that help them to do their jobs
 - Did You Know E-mails
 - Run Charts
 - Personalized QI Reports



What have we learned? Where do we go from here?

- NewSTEPs is partnering with state newborn screening programs to develop solutions for strengthening the NBS System
 - Quality data
 - Technical assistance
 - Bringing people together to share ideas and expertise

NewSTEPs Team

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