


# **NEWBORN SCREENING CASE DEFINITIONS AND QUALITY INDICATORS**

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SACHNDC Meeting  
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# NewSTEPs: Newborn Screening Technical assistance and Evaluation Program

NewSTEPs will provide quality improvement initiatives, a new innovative data repository, technical and educational resources and a comprehensive resource center to *state newborn screening programs and stakeholders.*


# Problem

- States use inconsistent definitions for calling a case a case.
  - States can't compare quality indicators because their definitions are different.
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# NBS Case Definition Project

- Rationale: Diagnoses are often not comparable from practice to practice or between newborn screening programs.
- Purpose: to develop a simple and standardized model for nominal categories of disease diagnosis.
  - Model will allow for harmonization across data systems, programs and patients.

# NBS Case Definition Project

- HRSA convened a number of condition category workgroups
  - Initiated in January 2011
  - Assist with the harmonization of newborn screening diagnoses for surveillance and epidemiologic purposes
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# NBS Case Definition Workgroups

- Metabolic
  - Pulmonology
  - Endocrinology
  - Immunology
  - Hemoglobinopathies
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# NBS Case Definition Workgroup Members

## Metabolic

- Dr. Jose Abdenur
- Dr. Nancy Leslie
- Dr. Lorenzo Botto
- Dr. Janet Thomas
- Dr. Bob Zori
- Dr. Cary Harding
- Dr. David Kronn
- Dr. Celia Kaye
- Dr. Maddy Martin
- Dr. Steve Kahler
- Dr. Sue Berry

## Endocrinology

- Dr. Kupper Wintergerst
- Dr. Stephen LaFranchi
- Dr. Marvin Mitchell
- Dr. Susan Rose
- Dr. Chanika Phornphutkul
- Dr. Phyllis Speiser
- Dr. Dan Hale

# NBS Case Definition Workgroup Members

## Pulmonology

- Dr. Richard Parad
- Dr. Hank Dorkin
- Dr. Laurie Varlotta
- Dr. Phil Farrell
- Dr. Frank Accurso
- Dr. Mike Rock
- Dr. Drucy Borowitz
- Dr. George Retsch-Bogart
- Dr. Michelle Howenstine
- Dr. Marci Sontag
- Dr. Jame Vajda
- Dr. Ron Gibson

## Immunology

- Dr. Sean McGhee
- Dr. Anne Comeau
- Dr. Vincent Bonagura
- Dr. Francisco Bonilla
- Dr. Jennifer Puck
- Dr. Rebecca Buckley
- Dr. John Routes
- Dr. Christine Seroogy
- Dr. J Church
- Dr. Luigi Notarangelo




# NBS Case Definition Workgroup Members

## Hemoglobinopathies

- Dr. Kathy Hassell
- Dr. James Eckman
- Dr. Carolyn Hoppe
- Dr. Ferdane Kutlar
- Dr. Kim Smith-Whitley
- Dr. Elliott Vichinsky

# NBS Case Definition

## July 2012

- Discussion of case definitions for the different categories of disease diagnosis with state newborn screening programs across the country.
  - Next steps: Several states have volunteered to beta test the case definition modules for the different disorder categories
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# NBS Quality Indicator Project

- Supported through a contract with Genetic Alliance and funds from the Health Resources and Services Administration
- Project started in March 2011
- Conference calls and f-t-t meeting

# NBS Quality Indicators Project

## Purpose:

- Develop and outline state newborn screening program needs including laboratory and short term follow up quality indicators (as agreed upon by the states)
- Summarize the utility of current quality indicators and outline other quality indicators that should be collected for the next generation newborn screening data repository

# NBS QI Project Meeting Participants - June 2011

- Swapna Abhyankar
- Lou Bartoshesky
- Natasha Bonhomme
- Bob Bowman
- Michele Caggana
- Sara Copeland
- Carla Cuthbert
- Roger Eaton
- Leslie Gafney
- Karim George
- Jane Getchell
- Michael Glass
- Rebecca Goodwin
- Alaina Harris
- Cheryl Hermerath
- Asha Farrah
- Elizabeth Jones
- Jelili Ojodu
- Mike Smith
- Reed Deshler
- Patrick Hopkins
- Ward B. Jacox
- Christelle Larose
- Mark McCann
- Joanne Mei
- Michele Puryear
- Deborah Rodriguez
- Debi Sarkar
- Scott Shone
- Marci Sontag
- Sharon Terry
- Judi Tuerck
- Sharon Vaz
- Sheila Weiss
- Donna Williams
- Bill Young
- Alan Zuckerman

# Activities

- Definition and Labeling of QI Categories
- Inventory QI within Category and Finalize Definitions
- Key Quality Indicator Definition
- Large Group Read Out of Key Quality Indicators by Category

# Key Quality Indicators

- Percent of unsatisfactory specimens due to improper collection
- Percent of parental refusal
- Percent of eligible infants receiving valid newborn screening test
- Percent of unsatisfactory specimens
- Rate of out-of-range results and rate of any referral to evaluation
- Rate of loss to follow-up: Unsatisfactory and out-of-range
- Frequency of condition detected at birth: first screen, second screen
- Performance of screening (false negatives, false positives)
- Timing of specimen collection, testing, reporting, disorder diagnosis.

# NBS Quality Indicator Meeting July 2012

- Focus on refining the ten national quality indicators for newborn screening programs that was proposed in June 2011 by the Newborn Screening Quality Indicators Workgroup.



# NBS QI State Experiences

- NBS Quality Indicators: New Jersey Newborn Screening Pilot Experience
  - Scott Shone, PhD (New Jersey Department of Health)
- Beta tested the ten NBS QI in the NBS program in New Jersey

# Refining the NBS QI

- Interactive session (using polling devices)
- NBS QI
  - Importance
  - Definition
  - Feasibility

# Next Steps

- Subcommittee formed to continue to refine the NBS QI
- Several states have volunteered to beta test the case definition modules for the different disorder categories
- The NBS case definitions and QI will be incorporated in the NewSTEPs database

# Questions

Jelili Ojodu

Director, Newborn Screening and Genetics

APHL

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# NewSTEPs - Structure

- NewSTEPs is funded through a five-year cooperative agreement to APHL by the Genetic Services Branch of the Health Resources and Services Administration (HRSA)
  - APHL
    - Director of NewSTEPs: Jelili Ojodu, MPH (APHL)
    - Senior Advisor: Jane Getchell, DrPH (APHL)
    - Project Manager: Sikha Singh, MPH (APHL)
    - Senior Specialist (TBD)
    - Specialist (TBD)
    - NBSGPH Liaison (Elizabeth Jones, MPH)
  - APHL staff is collaborating with Colorado School of Public Health
    - Marci Sontag, PhD (Director of Epidemiology)
    - Yvonne Kellar-Guenther, PhD (Program Evaluator)
  - NewSTEPs Steering Committee

# Established Collaborations

- State Newborn Screening Programs
  - Regional Collaboratives and the National Coordinating Center
  - Newborn Screening Clearinghouse (Baby's First Test)
  - Newborn Screening Translational Research Network (NBSTRN)
  - Center for Disease Control and Prevention (CDC)
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# Goal 1 – Information gathering, building relationships

- Identify gaps and barriers:
  - NBS education
    - PH decision makers
    - NBS program personnel (new disorders, new assays, follow-up strategies.)
  - Communication
  - Data collection and reporting
- Build and strengthen relationships among local, state, regional, and national NBS stakeholders, private partners, and NewSTEPS.
- IMPACT: Use information from the NBS community to strengthen NewSTEPS going forward. Develop a program that will support states and NBS stakeholders



# Goal 2 – Education, Networking

- Create and support a NBS network for education and communication
  - Local
  - State
  - National
- IMPACT: Networks of individuals can be utilized to improve newborn screening outcomes
  - In Person and Virtual Networking
  - Sharing Successes/Challenges

# Goal 3 – Data repository

- Design, build, and validate a data repository system
  - Evaluation of newborn screening systems
  - Quality improvement and monitoring of changes in systems
  - Disorder occurrence, practices, etc.
- Impact:
  - Use quality indicators to improve newborn screening programs at state, regional, and national level
  - Identify trends at state, regional, national levels
  - Long term: Benchmarking, networking between states

# Goal 4 – Technical Assistance

- Build on existing technical assistance and training opportunities for both individuals and programs providing NBS services.
  - Pre-analytic
  - Analytic
  - Post-analytic
  - Short term follow-up
  - Point of Care
- Voluntary Newborn Screen Quality Certificate Program

## Impact:

Site visits will be provided under HRSA funding

Improved Educational Resources

Quality Certification Program could be used as point of pride in NBS programs or identify need for more resources

# Highlights of the First Year

- July 2012
  - Quality Indicators – Modify and finalize
  - Case Definitions – Implement case definitions
  - Engage NBS community and get input
- August - October 2012
  - Define the structure and component of Data Repository, advertise RFP, and begin development
  - Hire NewSTEPS Staff
- January – May 2013
  - Work with NBS laboratory system vendors to identify possibilities for data transfers
  - Host Webinars to demonstrate repository
- May 2013
  - Debut repository at NBSGT and ISNS Symposium (Atlanta)

# Contact Us

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