

NewSTEPS



The development of this presentation was supported by Cooperative Agreement #U22MC24078 from the Health Resources and Services Administration (HRSA). Its contents are solely the responsibility of the authors and do not necessarily represent the official views of HRSA.



NewSTEPS

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NewSTEPS

- **NewSTEPS** is the Newborn Screening Technical assistance and Evaluation Program
- **NewSTEPS** is partnering with newborn screening programs throughout the U.S. to:
 - 👣 Provide technical assistance and resources
 - 👣 Collate and summarize data in aggregate form
 - 👣 Develop opportunities for quality improvement, locally, regionally, and nationally



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National Data Repository for Newborn Screening



Purpose: Provide tools to state newborn screening systems to adequately evaluate, analyze, and benchmark the performance of their tests and the quality of their newborn screening programs

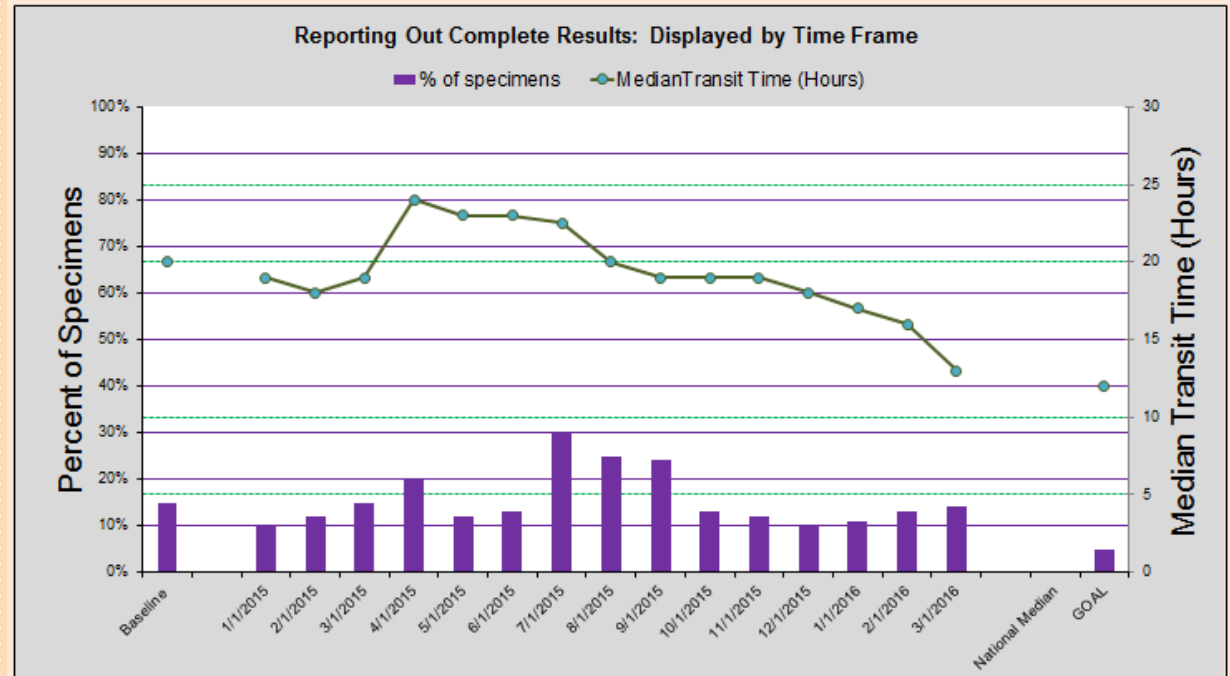


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Quality Indicator Data

- 8 Quality indicators developed vetted by the NBS community
- Track state progress over time
- Upload tools available
 - LIMS vendors developing export queries for available data
 - Colorado working with Perkin Elmer
 - Delaware working with Natus



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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

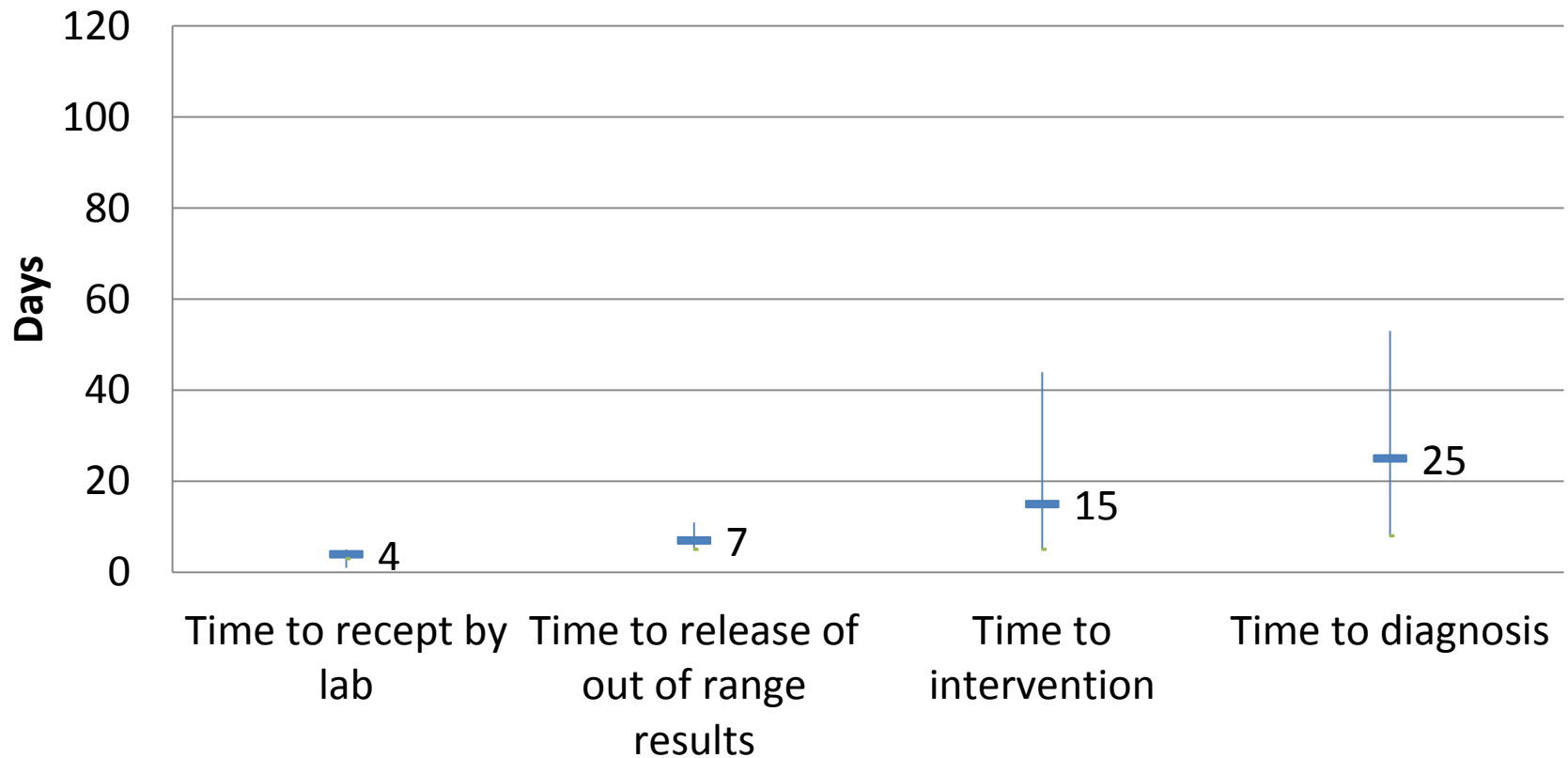


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Timeliness data

Infants identified with disorder on NBS in NewSTEPS repository



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CF Data babies with CF in NewSTEPS Repository

- Sweat test results
 - 70%: >60mmol/L
 - 5%: 30-60mmol/L
 - 2%: <30 mmol/L
 - 2%: QNS
 - 21%: NOT DONE (Or not on file with state)

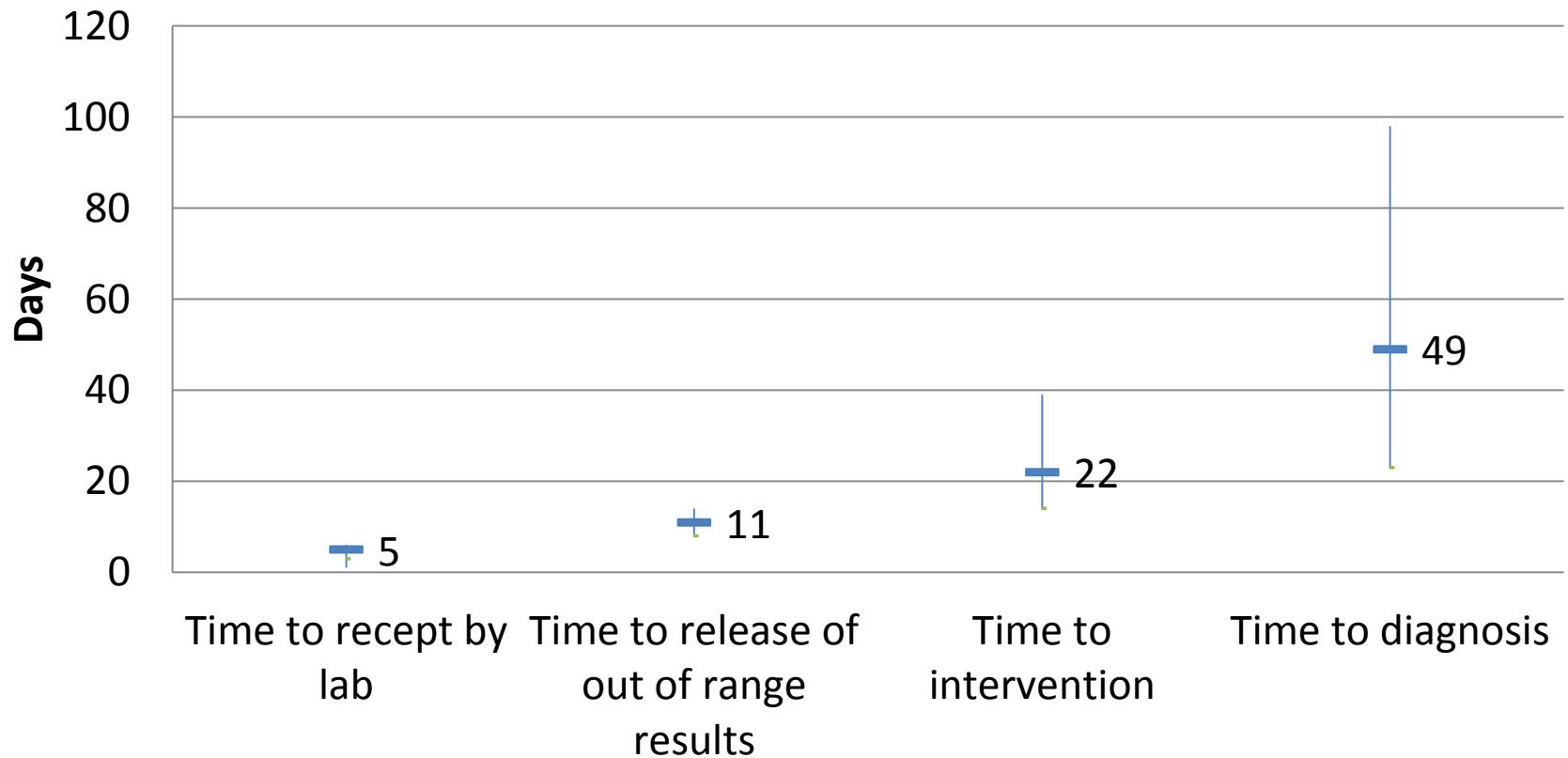


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Timeliness data

Infants identified with CF on NBS in NewSTEPs repository



Making it easier for you

- Data import templates available
- Program can extract data from their LIMS systems
- Links on NewSTEPs website will import data into appropriate fields
- Available for Case demographic/screening information data and quality indicators

[← Back To List](#)

Case Definition

State *

Illinois ▼

Disorder

Start typing a condition name

Cystic fibrosis - CF ▼

OR

Import Case Demographic and Screening information from csv file



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Case Definition – Import tool available

State unique id	0040231
Disorder	Congenital hypothyroidism - CH
Birth year	2011
Gestational age in weeks	39
Birth weight in grams	3,402
Biological Sex	Female
Race	White
Ethnicity	Not Reported

Was prenatal testing done that indicated that this infant was at risk for this disorder?	Unknown
Which newborn screen result indicated this infant was at risk for the disorder	Initial Screen
Was this individual diagnosed later in life (not identified by newborn screening)?	No
Initial Specimen Collection Information	
Hours between birth and initial specimen collection	48 hours
Days between birth and initial specimen receipt by lab	6 days
Days between birth and initial release of out-of-range results	7 days
Subsequent Specimen Collection Information	
Not Entered	
Intervention, Follow-Up, and Diagnosis	
Days between birth and intervention between appropriate medical provider	7 days
Days between birth and confirmation of diagnosis	7 days



Short Term Follow-up

- Forum for individuals working in short-term follow-up to network with colleagues
- Workgroup of individuals from all regions plan activities
 - Bi-Monthly Webinars on Short-Term Follow-up
 - Gatherings at NBSGT Symposium



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Health Information Technology

- Data import templates developed for quality indicator and case data to upload data from state LIMS systems into NewSTEPS Repository
- Plans in upcoming year:
 - Complete and ballot the HL7 Implementation Guide (November proposal to HL7)
 - Partnering with vendors to identify solutions for HL7 implementation and interoperability
 - Building tools for uploading data into NewSTEPS data repository



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CoIIN

- 8 states participating in Newborn Screening Quality Improvement CoIIN
- 18 month interactive opportunity to share ideas across states
 - Iowa participating from the Heartland
- Identifying challenges and successes, building team



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Quality Practice Resources

Case Definitions Quality Indicators **Disorder Specific Activities** Model Practices Short Term Follow Up

Disorder Specific Activities

- **Critical Congenital Heart Disease (CCHD)**
- **Severe Combined Immunodeficiency (SCID)**
- **Pompe Disease**

Educational Resources

PUBLICATIONS

Indicators of newborn screening for congenital hypothyroidism in Sri Lanka: program challenges and way forward

This article analyzed the newborn screening system in Sri Lanka in the areas of sample...

PUBLICATIONS

Newborn screening for Pompe



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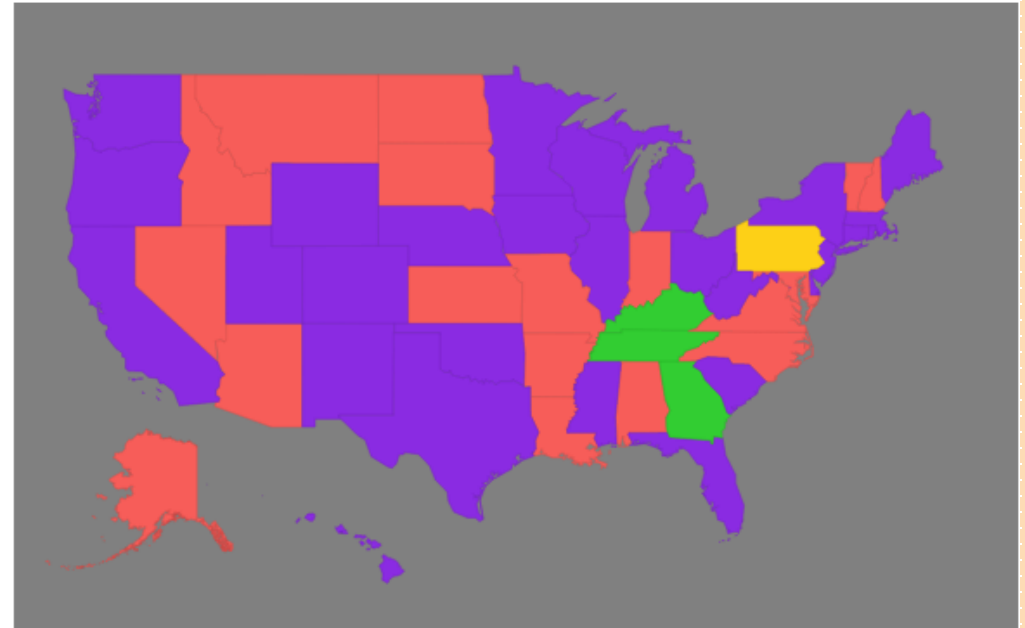
SCID

- Partnering with NBSTRN to offer technical assistance
- Funding 12 grantees to fully implement SCID (HRSA CoA)
 - In the Heartland: Kansas and North Dakota

SCID Newborn Screening Status

Updated April 1, 2015

[Click on a state for additional details on SCID NBS.](#)



Universally screening Required but not yet implemented Offered to select populations or by request Being considered but not yet approved

Territories currently not depicted on SCID map:

*Navajo Nation - Universally screening (New Mexico and Arizona)

*District of Columbia - Universally screening (January 2015).

*Puerto Rico - Being considered but not yet approved. Receiving support to further develop SCID NBS implementation through funding from APHL via a Cooperative Agreement from HRSA



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Pompe

- Added to the RUSP February 2015
- NewSTEPs developing advisory group and technical assistance activities to be debuted May 2015



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Evaluation Program

About the Program

[Request a Site Visit](#)

Evaluation Program: Request a site visit

Full name *

- Part of the on-going technical assistance provided by NewSTEPS – at no cost to state
- Not regulatory
- At the invitation of the state, a site visit is scheduled and experts chosen depending on program needs
- Reviews all aspects of the newborn screening system—lab, follow up, expert care for infants diagnosed
- Two states visited to date: both made improvements in NBS program activities as result



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NBSSLA Research Amendment

**MSGRC Annual Meeting
April 23, 2014**

**Joan A. Scott, M.S., C.G.C.
Department of Health and Human Services
Health Resources and Services Administration
Maternal and Child Health Bureau
Division of Services for Children with Special Healthcare Needs
Chief, Genetics Services Branch**



NBSSLA Research Amendment

- Federally funded research on NBS blood spots is considered research on human subjects, regardless of whether the specimens are identifiable
- Eliminates the ability of an IRB to approve alterations or waivers of informed consent
- Applies to samples collected 90 days after enacted date
- Secretary must promulgate proposed revisions to Federal Policy for the Protection of Human Subjects within six months and final regulations within two years

Research Amendment: Some Issues

- What is 'research'?
- What constitutes appropriate informed consent in this context?
- Will families confuse consenting for storage and broad research use of NBS samples with consent for clinical testing?



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NBSSLA Research Amendment Activities

- March 9 meeting sponsored by NIH for researchers
- March 24 SACHRP meeting voted on recommendations to OHRP
<http://www.hhs.gov/ohrp/sachrp/>
- May 11-12 DACHDNC meeting
- June 1-2 *A National Conversation about NBS Research and Informed Consent* sponsored by APHL



A National Conversation on Newborn Screening Research and Informed Consent

June 1-2, 2015

Renaissance Washington, DC Dupont Circle Hotel
Washington D.C.

AGENDA

- Overview of amendment and activities
- Public Health Implications on State Newborn Screening Programs
- What Constitutes Newborn Screening Research: An evaluation of essential program activities for screening of current and new conditions
- Informed Consent for Newborn Screening Research: Examples of a Broad Consent Package
- Educating the Public about Newborn Screening: Current and Proposed Communication Strategies



SACHRP Recommendations to OHRP

- Rapidly disseminate guidance for researchers
 - Make clear only applies to HHS-funded research
 - Encourage use of de-identified samples
 - Institutions continue to assess whether activities are research
 - Include NBS scenarios in the 2008 report *Engagement of Institutions in Human Subject Research*
 - Note that expedited review categories can be used
 - Emphasize that one-time permission for broad consent would simplify consent process
 - Develop an example document for broad consent
 - Emphasize the ability of IRBS to grant waivers of consent documentation



What is Research?

- *“Research means the systematic investigation, including research development, testing and evaluation, designed to develop or contribute to generalizable knowledge. Activities which meet this definition constitute research for purposes of this policy, whether or not they are conducted or supported under a program which is considered research for other purposes. For example, some demonstration and service programs may include research activities”.*



What is Research in NBS Programs?

Clinical Testing

- Lab QC & QA
- Emergency preparedness drills

Test translation and implementation

Research

- Developing a new NBS test

Course going forward

- Provide information to inform guidance on what is laboratory practice and what is research
- Provide information to inform guidance on informed consent processes



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NewSTEPS Data Repository

- MOUs - 46% of NBS programs have fully ratified MOUs (or are close to ratifying)
- Entering data
 - All States: State Profiles (disorders screened, policies, etc)
 - States with signed MOUs
 - Quality Indicators
 - Case Level Data



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Did you know...

*NewSTEPS offers a **non-regulatory customized SITE VISIT** aimed at assessing all components of a newborn screening (NBS) program including the laboratory system, birth facilities, and follow-up system?*

- To find out more, please visit: <https://newsteps.org/evaluation-program>

Evaluation Program



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