

Propel Awardee Forum

June 12, 2024



NewSTEPS

A Program of the Association of Public Health Laboratories™



Propel Forum Planning Committee

- Heidi Elsinger | Connecticut
- Joe Huynh | Arizona
- Karli Calaway | Colorado
- Katherine Fullerton | Arizona
- Kathy Chou | New York
- Ken Coursey | Iowa
- Kim Piper | Iowa
- Nicolas Szabo | Utah





NEWSTEPS NATIONAL MEETING

ON
POST-ANALYTICAL NEWBORN SCREENING

YOU'RE INVITED
REGISTRATION OPENS SOON



February 11 - 12, 2025 Rockville, MD

FOR MORE INFORMATION

Join the NewSTEPs National Meeting Teams Site [here](#)

Contact Noah Sinangil at Noah.Sinangil@aphl.org

This meeting is supported by the Health Resources and Services Administration (HRSA) under Cooperative Agreement #U22MC24078 for \$2,725,000. This content is those of the authors and should not be construed as the official position or policy of, nor should any endorsements be inferred by HRSA, HHS or the US Government.





HRSA Newborn Screening Programs

NBS Propel

NBS Excel

Co-Propel

Debi Sarkar, MPH

Division of Services for Children with Special Health Needs

Maternal and Child Health Bureau (MCHB)

Vision: Healthy Communities, Healthy People



HRSA NBS Program Team



Alisha Keehn
Genetics Services Branch



Cdr. Leticia Manning



Kim Morrison



Cristina Novoa



Lisa Song

Branch Chief

NBS Team Lead

Public Health Analysts—Project Officers



Kathryn McLaughlin



Monica Adderley



Alaina Harris



Loraine Swanson



ARTICLES

Introducing the Blueprint for Change: A National Framework for a System of Services for Children and Youth With Special Health Care Needs

Treeby W. Brown et al

A Blueprint for Change: Guiding Principles for a System of Services for Children and Youth With Special Health Care Needs and Their Families

Sarah E. McLellan et al

Children and Youth With Special Health Care Needs: A Profile

Reem M. Ghandour et al

Progress, Persistence, and Hope: Building a System of Services for CYSHCN and Their Families Michael D. Warren et al**Health Equity for Children and Youth With Special Health Care Needs: A Vision for the Future** Amy Houtrow et al**Quality of Life and Well-Being for Children and Youth With Special Health Care Needs and their Families: A Vision for the Future** Cara L. Coleman et al**Access to Services for Children and Youth With Special Health Care Needs and Their Families: Concepts and Considerations for an Integrated Systems Redesign** Dennis Z. Kuo et al**Financing Care for CYSHCN in the Next Decade: Reducing Burden, Advancing Equity, and Transforming Systems** Jeff Schiff et al<https://publications.aap.org/pediatrics/issue/149/Supplement%207>

A SUPPLEMENT TO PEDIATRICS

Blueprint for Change: A National Framework for a System of Services for Children and Youth with Special Health Care Needs*Treeby W. Brown, MA, Sarah E. McLellan, MPH, Marie Y. Mann, MD, MPH, FAAP, and Joan A. Scott, MS, CGC, Guest Editors**Funding for the publication of this article, including editorial support (review for formatting and consistency) for this article was funded by the US Department of Health and Human Services, Health Resources and Services Administration under contract number 75R60219D00014.**The views expressed in this publication are solely the opinions of the authors and do not necessarily reflect the official policies of the US Department of Health and Human Services or the Health Resources and Services Administration, nor does mention of the department or agency names imply endorsement by the US Government.*American Academy
of Pediatrics

DEDICATED TO THE HEALTH OF ALL CHILDREN®



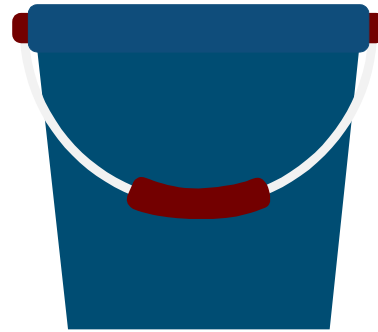
DATA - GOAL

Three Connected Buckets of Data



Input/Data sources:

Families, researchers, clinicians, public health and policy leaders



Bucket 1: NBS Lab result analysis.

Analysis of state NBS lab data/
Pre-diagnostic risk assessment.
e.g. ED3N (CDC)

Bucket 2: Notification/confirmation.

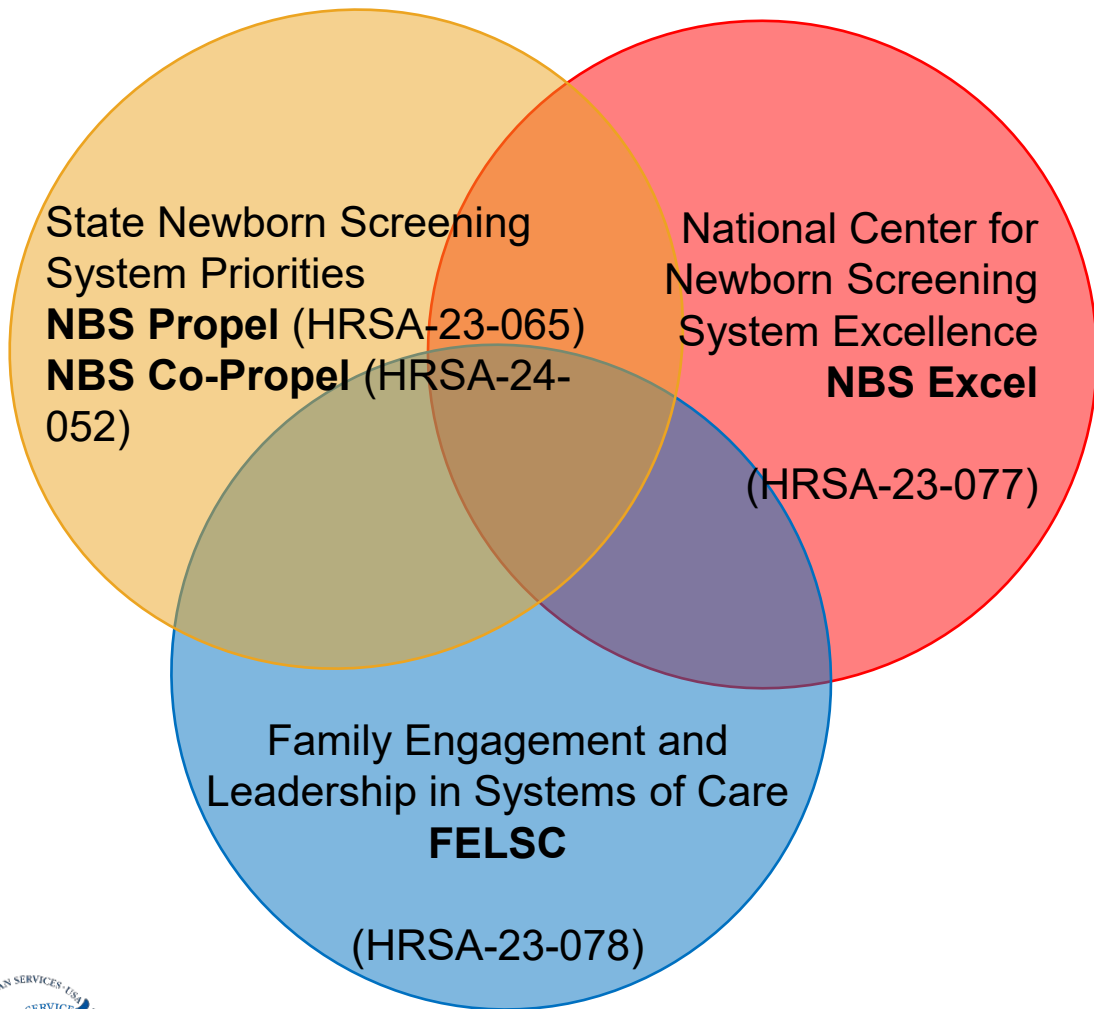
Short-term follow-up,
with families/clinicians.
e.g. NewSTEPS (HRSA),
ACT Sheets

Bucket 3: Longitudinal clinical care.

NBS-identified children
receive treatment.
e.g. Propel (HRSA), SCDC (CDC),
Registries/studies (NIH)



MCHB Newborn Screening Collaborations



NBS Propel + Co-Propel – State Priorities

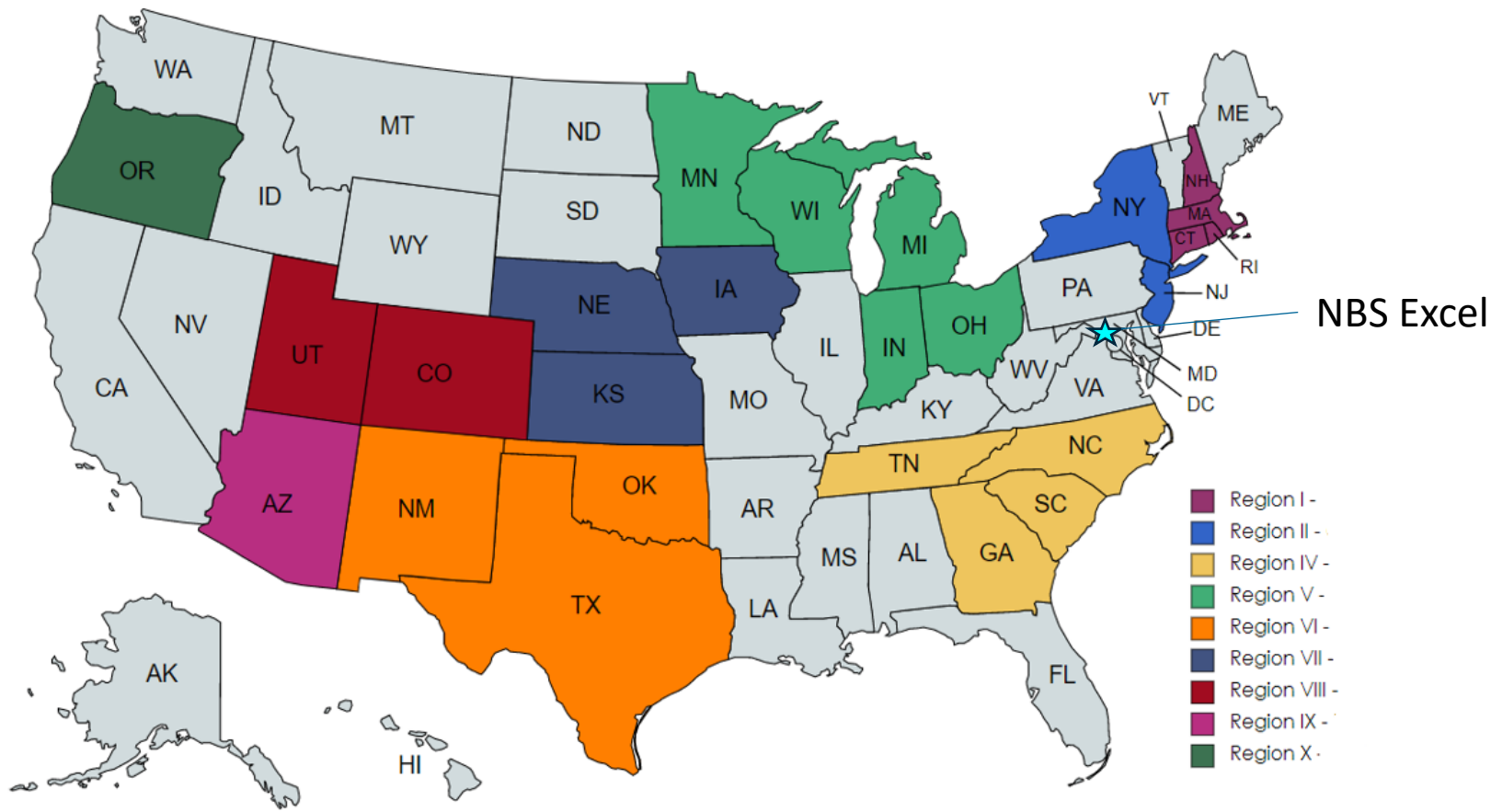
- Focus Area 1
 - screening/timeliness/implementing RUSP conditions
- Focus Area 2
 - STFU, LTFU

NBS Excel – National Center

- Data / Quality Improvement (QI)
- Technical Assistance
- Family Engagement

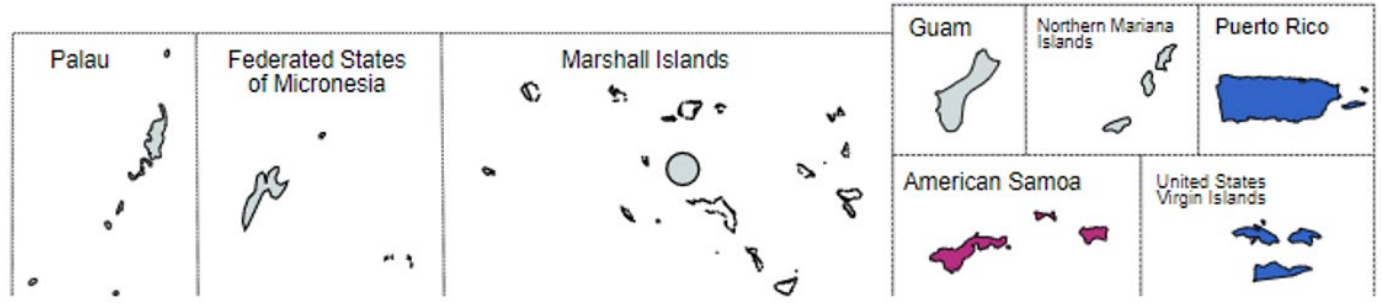
FELSC – Family Engagement and Leadership

- Project 1
 - National Leadership on Family Engagement
- Project 2
 - Technical Assistance for F2F HIC Programs



- Region I -
- Region II -
- Region III -
- Region IV -
- Region V -
- Region VI -
- Region VII -
- Region VIII -
- Region IX -
- Region X -

2023-2028 NBS Propel and Excel Grantee Map



NBS Propel--Program Overview

Number of awards

28 grants

Funding per award

Up to \$345,000.00 * per year

Total investment

Approximately \$8,625,000 per year

Period of Performance

July 1, 2023 through June 30, 2028 (5 years)

Type of award

Grant

Projected start date

July 1, 2023

**subject to the availability of appropriated funds*



State Newborn Screening System Priorities Program (NBS Propel)

NBS Propel will support NBS Programs to

1. Address state/territory-specific challenges and pursue priorities to enhance, improve and expand NBS systems.
2. Address timely collection and reporting of NBS specimens to improve early diagnosis and treatment for individuals with heritable conditions identified through NBS; and
3. Support long-term follow-up (LTFU) for individuals with Severe Combined Immunodeficiency (SCID) and other NBS conditions that link public health agencies, clinicians, and meaningfully engages and partners with families.

NBS Propel Purpose

To strengthen the NBS System to provide screening, counseling and health care services to newborns and children with, or at risk for, heritable disorders and help them achieve the best possible outcomes.

NBS Propel Goal

To Improve health outcomes by early screening, diagnosis and treatment of newborns and children with heritable disorders identified by NBS.



NBS Excel--Program Overview

Number of awards 1 (APHL)

Funding per award Up to \$2,300,000* per year

Total investment \$2,300,000 per year

Period of Performance July 1, 2023 through June 30, 2028 (5 years)

Type of award Cooperative Agreement

Projected start date July 1, 2023

**subject to the availability of appropriated funds*



National Center for Newborn Screening System Excellence Program (NBS Excel)

NBS Excel will support state NBS Programs and stakeholders on a nationwide basis by:

1. Providing leadership, technical assistance (TA), and quality improvement expertise;
2. Collecting data to identify barriers to achieving health equity and equitable access to NBS services for all infants and families;
3. Enhancing state performance in NBS; and
4. Providing subject matter expertise, TA and training, education, and other resources to youth and families in order to promote meaningful engagement of, active participation with, and leadership by families and/or individuals with heritable disorders at all levels of the newborn screening system.

NBS Excel Purpose

To strengthen the NBS System to provide screening, counseling and health care services to newborns and children with, or at risk for, heritable disorders and help them achieve the best possible outcomes.

NBS Excel Goal

To Improve health outcomes by early screening, diagnosis and treatment of newborns and children with heritable disorders identified by NBS.



NBS Co-Propel--Program Overview

Number of awards

Up to 10

Funding per award

Between \$345,000 to \$500,000* per year

Total investment

Approximately \$4,000,000 per year

Period of Performance

July 1, 2024 through June 30, 2028 (4 years)

Type of award

Cooperative Agreement

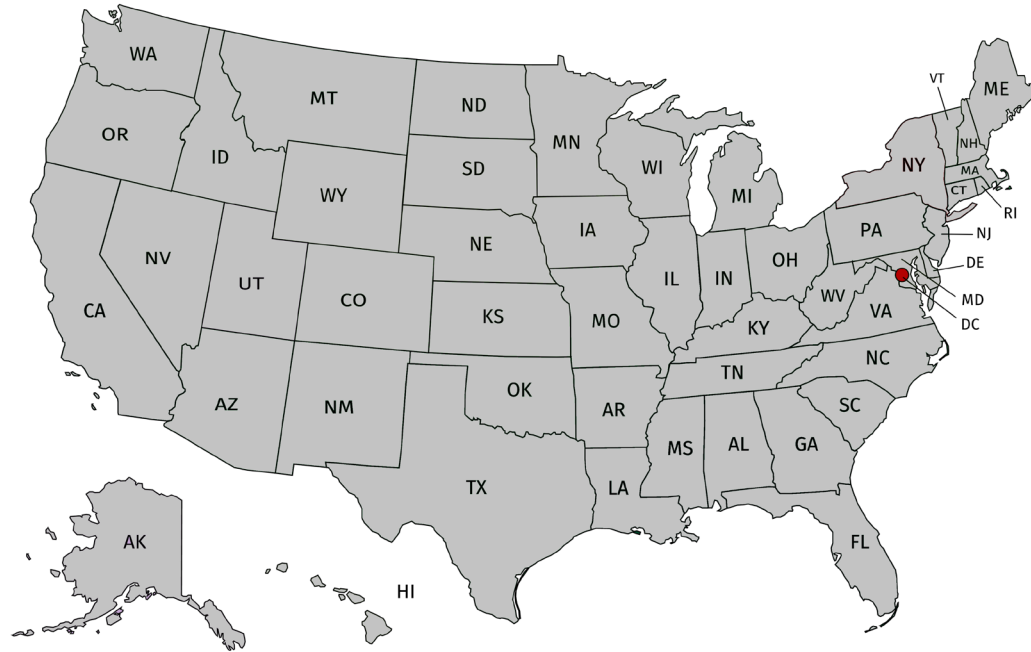
Projected start date

July 1, 2024

**subject to the availability of appropriated funds*

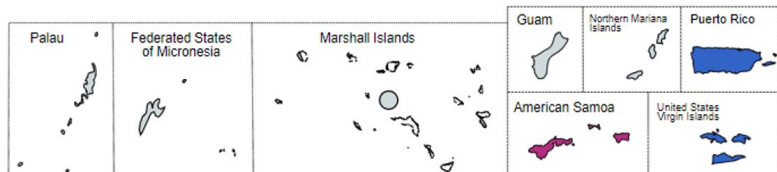


NBS Co-Propel



TBD!!!

Stay Tuned.....



Cooperative Newborn Screening System Priorities Program (NBS Co-Propel)

NBS CO-Propel will support NBS Programs to

1. Address state/territory-specific challenges and pursue priorities to enhance, improve and expand NBS systems.
2. Address timely collection and reporting of NBS specimens to improve early diagnosis and treatment for individuals with heritable conditions identified through NBS; and
3. Support long-term follow-up (LTFU) for individuals with Severe Combined Immunodeficiency (SCID) and other NBS conditions that link public health agencies, clinicians, and meaningfully engages and partners with families.

NBS Co-Propel Purpose

To strengthen the NBS System to provide screening, counseling and health care services to newborns and children with, or at risk for, heritable disorders and help them achieve the best possible outcomes.

NBS Co-Propel Goal

To Improve health outcomes by early screening, diagnosis and treatment of newborns and children with heritable disorders identified by NBS.



Propel and Co-Propel

Similarities:

- Collaborate with NBS Excel
- 2 focus areas:
 - Focus Area 1: activities related to improving collection of specimens, testing of specimens, and reporting out results, including improving timeliness of these activities; and implementing screening for newly-added RUSP conditions;
 - Focus Area 2: activities related to improving short-term through long-term follow-up and helping families understand and navigate the process from confirmation of a diagnosis to treatment, and through follow-up across the lifespan.
- Submit de-identified data annually to NBS Excel (including timeliness data).

Co-Propel additions:

- Collaborate with other Co-Propel grantees and NBS Excel to develop a data collection plan to measure the number of 3-year-old children with a heritable condition who are thriving.



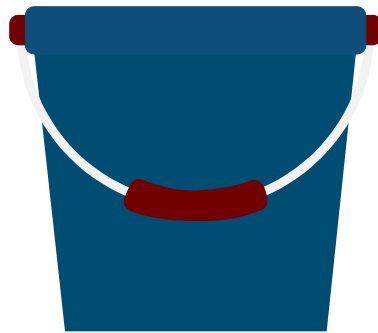
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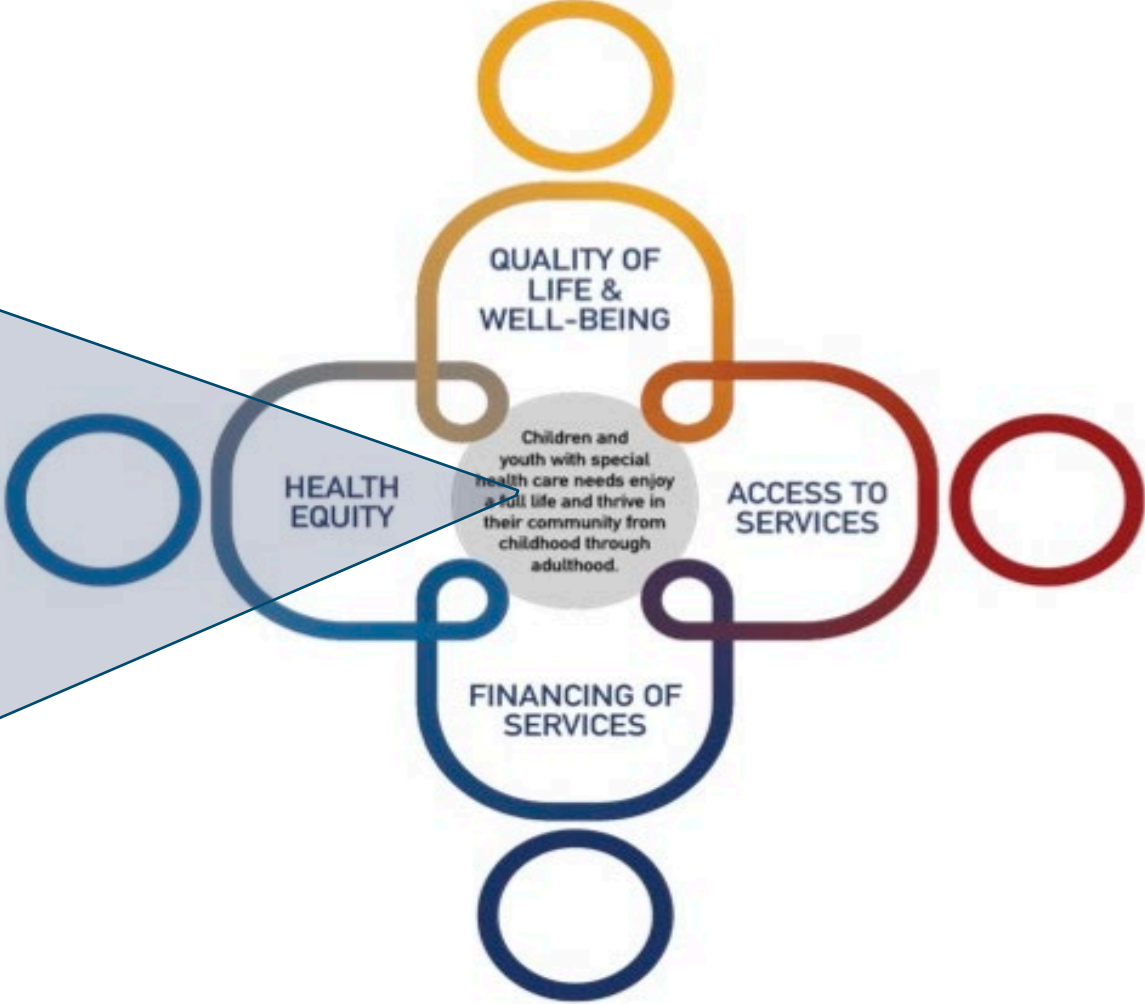
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NBS-identified children
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e.g. Propel (HRSA), SCDC (CDC),
Registries/studies (NIH)



BLUEPRINT FOR CHANGE FOR CYSHCN

Every family is able to get what their child needs so that they can play, go to school, and become a healthy adult.



Thank you!

We are grateful for your
partnership and collaboration!

Debi Sarkar, MPH
Division of Services for Children with Special Health Needs
Maternal and Child Health Bureau (MCHB)



Today's Sessions | Wednesday, June 12

10:00 am Welcome and Forum Overview

10:30 am From Awareness to Action: Engaging Families in NBS

11:30 am Explore the Long-term Follow-up Landscape

12:30 pm Break

1:30 pm (Optional) Networking Rooms

2:00 pm Quality Indicators and Improvement Strategies

3:00 pm Navigating New Disorder Implementation

4:00 pm Closing



Rowan's Story



From Awareness to Action: Engaging Families in Newborn Screening



Family Engagement in the Newborn Screening System

Marianna Raia, MS CGC
*Associate Director of
Programs*
Expecting Health

Strategies to Drive Action



1. Raise Awareness

What is Family Engagement & Why is it Important?



2. Build Knowledge

What Tools & Resources Exist to Support Family Engagement in NBS?



3. Generate Action

What can YOU do to support Family Engagement?

1. Raise Awareness

What is Family Engagement?



Family Engagement

Patients, families, their representatives, and health professionals **working in active partnership** at various levels **across** the healthcare system to improve health and healthcare.¹

This involvement **improves** individuals' and families' **access** to information and services; it is **critical to engage family leaders** at various levels across the healthcare delivery system **so their perspectives can help shape** the care and resources provided.

Family Leader

Individuals who **participate** actively in their own health care, and the care of others, raises awareness in the community, shares their story with others, advocates for a cause, **OR** is **interested in learning more** for themselves and their family members and sharing that information.

¹ Carman, K. L., Dardess, P., Maurer, M., Sofaer, S., Adams, K., Bechtel, C., & Sweeney, J. (2013). Patient and family engagement: A framework for understanding the elements and developing interventions and policies. *Health affairs, 32*(2), 223-231. <https://doi.org/10.1377/hlthaff.2012.1133>



Breakdown of Findings; 4 key Themes

EXPLORING THE ROLE OF FAMILIES IN REGIONAL GENETICS NETWORKS


*A SUMMARY OF ENGAGEMENT ACTIVITIES
AND RECOMMENDATIONS*

Final Report

National Genetics Education
and Family Support Center




- ❑ Family Engagement **Activities**
 - Tactics, methods, and organizational structure used to involve or engage families.
- ❑ Family Engagement **Facilitators**
 - Critical factors or considerations that promote family engagement.
- ❑ Family Engagement **Barriers**
 - Critical factors or considerations that hinder, inhibit or discourage family engagement.
- ❑ Family Engagement **Successes**
 - Promising family engagement techniques or initiatives that led to impacts.



Linking Families
to Genetics

Understanding Family Engagement in Genetics Services

*"Engagement is a full scope of practice from the individual, to clinical, to systems level with family and patients as partners and leaders."
— Family Leader*



National Genetics
Education and
Family Support Center

Recommendations

- Institutionalize the **definition of family engagement** within the system
- **Measure impact** and establish family engagement performance measures
- Pilot ways to categorize, assess and bolster meaningful family engagement in the healthcare delivery system
- Monitor ongoing family engagement activities by sharing best practices, promoting the sharing/solving of barriers and challenges to improve family engagement in the system authentically

<https://nationalfamilycenter.org/Understanding%20Family%20Engagement%20in%20Genetic%20Services.pdf>



Recommendations

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- ❑ Measure impact and establish family engagement performance measures
- ❑ Pilot ways to categorize, assess and bolster meaningful family engagement in the healthcare delivery system
- ❑ Monitor ongoing family engagement activities by sharing best practices, promoting the sharing/solving of barriers and challenges to improve family engagement in the system authentically

Supporting Family Leaders for Successful Engagement in Genetic Services

- I wish family involvement was *more of a priority*
- It is **hard to find groups** locally to participate in for support or advocacy
- **Keep including families**; they should share their stories with professionals. Stories should be empowering
- Not a lot of room/perceived need for families in the diagnostic odyssey to participate, especially if your children are not severely impacted by their diagnosis
- We need to be able to **help professionals** understand the difference between a family caregiver and a family leader
- The **genetics field is wide open** and just at the beginning of how we understand how genetics impacts health
- Needs to be more Genetic Counselors & Geneticists
- I think there is space for **more compensated opportunities**. So often, we contribute and work for free in this space

2. Build Knowledge

Strategies for Successful Engagement



A Framework for Engagement

Raise Awareness

- Social Media
- Awareness Campaigns
- Community Discussion Groups
- Communities of Practice

Build Knowledge

- Educational Videos
- Online Learning Modules
- Learning “Flipbook”
- Dissemination Toolkits
- Implementation Guides

Generate Action

- Steering Committees
- Workgroups
- Task Forces
- National, Regional and Local Presentations

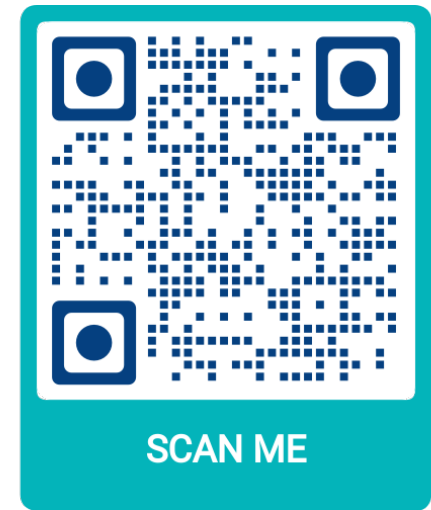
Navigate Newborn Screening
An Expecting Health Program

1. Raise Awareness

Navigate Newborn Screening: A Resource Sharing Toolkit



Navigate Newborn Screening
An Expecting Health Program



This toolkit includes multiple resources for families navigating NBS Results!
Download, use and SHARE!



Expecting Health

Sponsored ·



Knowing about newborn screening early can help you become more comfortable and know where to go if you need more information.

WHAT IS NEWBORN SCREENING?

Get the right information at the right time.

expectinghealth.org

Learn About Newborn Screening

Learn more



2

1 Share



Like



Comment



Share

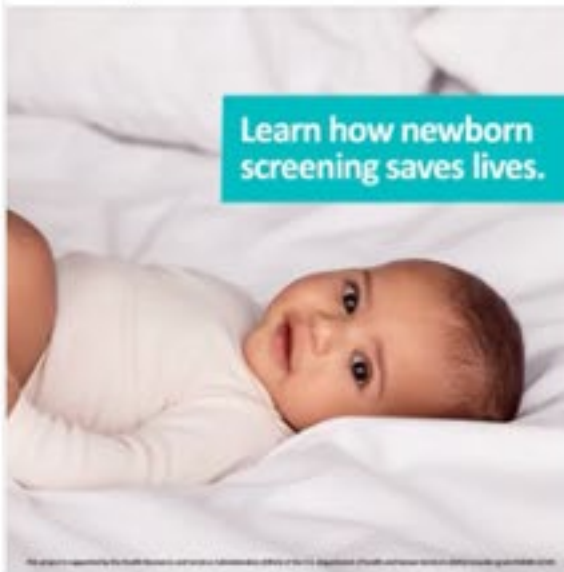


Expecting Health

Sponsored ·



So much happens in the first few days of your child's life. Get ahead by learning about newborn screening.



Learn how newborn screening saves lives.

expectinghealth.org

Learn How Newborn Screening Saves Lives

Learn more

Social Media



Expecting Health

Sponsored ·



Learning about this now can help you become more comfortable and help you know where to go if you need more information.



The right information at the right time

LEARN WHY NEWBORN SCREENING IS IMPORTANT.

expectinghealth.org

Learn Why Newborn Screening is Important

Learn more



Like

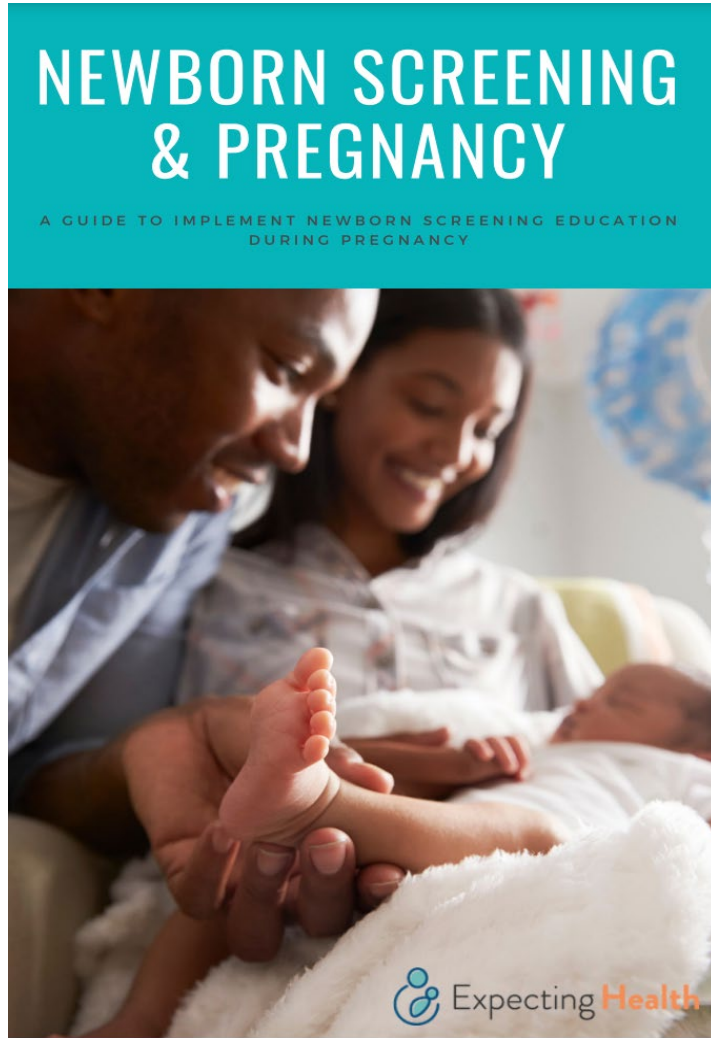


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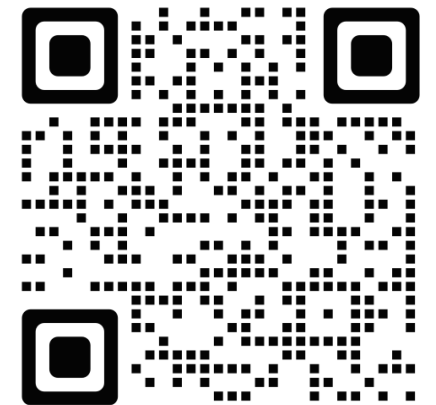


Share

2. Build Knowledge



- The toolkit is divided into 6 domains of implementation.
- Each domain contains a checklist to help guide states and their partners through implementation.
- Each section also contains tips, resources, and templates that can be used and customized to support the implementation process



Practical & Shareable Resources

Learn More Poster

This poster (16 x 20 in) is designed to hang in waiting rooms, exam rooms, bathrooms, community centers, and other family hubs so that families can learn more about newborn screening BEFORE they deliver their baby.

Hang It Up

Hang in high-traffic areas such as prenatal, pediatric and specialty care offices, community centers, WIC offices, etc.

Print & Share

Send in the mail, share at in-person events, distribute in prenatal, pediatric and specialty clinics.



Customize

Want to add local or state-specific information? Want to add your unique enrollment link? Reach out to mraia@expectinghealth.org to discuss.

Patient Information Card

The following patient information card is available in 4x6 or 5x7 and is a useful tool to helpful families gain access to prenatal education materials. These cards can be disseminated in a variety of locations.

Prenatal Clinic Visits

Distribute this card to patients as they are roomed and waiting to be seen by the clinician. Share it with them when they are having their vitals taken.

Home Prenatal Visits

Partner with doulas, WIC offices, and other home visiting groups to share this card when visiting expecting moms in their homes.

Birthing Centers

Share with staff and families that are delivering at birthing centers. Encourage families to learn about newborn screening prior to delivery.

Hospitals

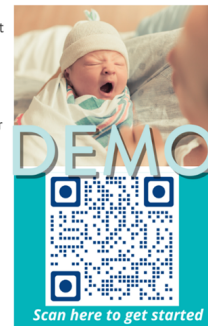
Have these cards available on delivery floors so that staff can disseminate to patients as they are administering newborn screening.

LEARN MORE ABOUT NEWBORN SCREENING!

We have partnered with *Navigate Newborn Screening* to help our patients learn more about newborn screening with a **free**, online educational book. This book will teach you more about what newborn screening is, how it works, and why it's important.

How does it work?
To access the book, scan the code in the corner with your camera. You will be taken to a short book where you can read more about newborn screening. Throughout the book, you will be asked to answer a few questions. This information will help us improve these tools and create new resources to educate families about newborn screening.

Do I have to participate?
Participation in this educational opportunity is completely voluntary. Your choice to participate (or not participate) will not change the care you or your baby receive.



This project is supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) under grant #UKSMC32105.

Customize

Want to add local or state-specific information to this patient information card? Reach out to mraia@expectinghealth.org for technical assistance.

Establishing Evaluation Measures

The purpose of this section is to support and help you establish an outcome evaluation to determine if this educational initiative (Pilot Program) is successful. This section will be based on the specific goals and objectives you set during your project planning. Examples are listed below but can be modified to meet you and your partner's needs to increasing awareness, knowledge, and perceived importance of newborn screening among pregnant women.



- To what extent does the Pilot Program increase awareness of newborn screening among medically underserved pregnant women?
- To what extent does the Pilot Program increase knowledge related to newborn screening among medically underserved pregnant women?
- To what extent does the Pilot Program increase perceived importance of newborn screening among medically underserved pregnant women?
- To what extent does the Pilot Program increase self-efficacy to find additional information about newborn screening among medically underserved pregnant women?

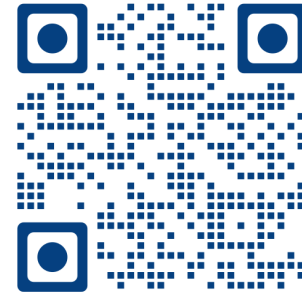
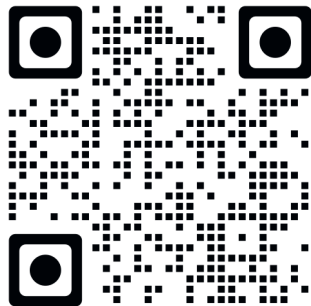
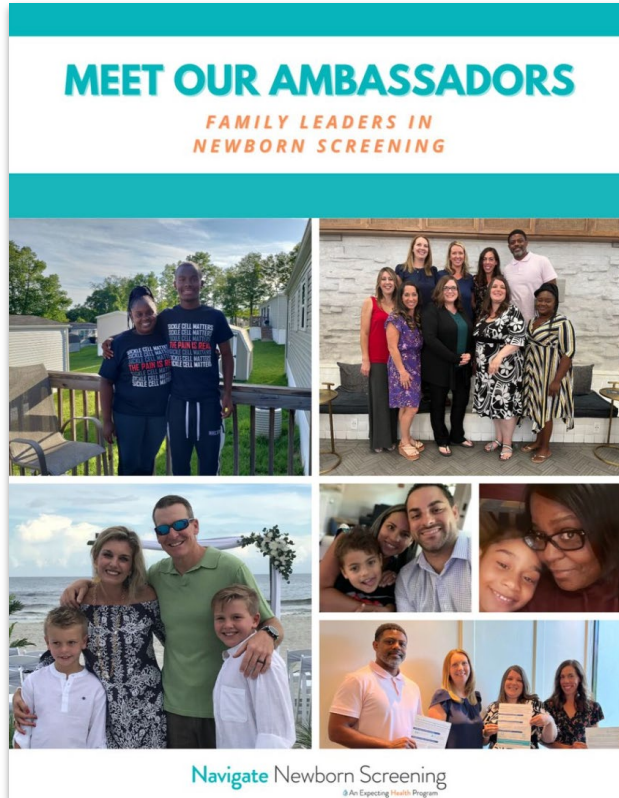
To evaluate the Pilot Program, you can elect to conduct a web-based or paper survey before the participant reviews the flip book and a second web-based or paper survey after the participant reviews the flip book. Responses from these pre- and post-tests can then be compared to assess to what extent the Pilot Program was successful.

The following pages share a copy of the questions that were used with the initial two pilot sites, and are available in both English and Spanish translations.

QUESTIONS TO CONSIDER:

- Will your patients/families need to complete this questionnaire on paper or online?
- What evaluation tool will work best for your community (Google Form, SurveyMonkey, etc.?)

3. Generate Action



- Share, embed or utilize these ambassador videos
- Read and learn from the “Meet Our Ambassadors” book
- Connect with one of our 22 trained NBS Ambassadors

Education Workgroup Charges & Activities

Objectives

- Support the development of state and family needs assessments to identify areas of interest and opportunity for education within the NBS community.
- Engaging families and individuals with lived experience in NBS.
- Evaluating ongoing needs of the NBS community and performing outreach to providers serving medically underserved and historically excluded communities.

Year 1 Activities

- Support the development of a needs assessments to identify areas of interest and opportunity for education within the NBS community
- Establish and define 3-4 key “regional” groups with emphasis on providers serving medically underserved communities (geography, tribal communities, Spanish affinity groups, etc.)
- Support and generate content ideas for regional virtual trainings for providers and families with annual focus areas in line with NewSTEPs programmatic objectives:

Workgroup Insights

Successes of family engagement

- Family participation in NBS educational video
- Formation of state-wide NBS Family Advisory Group
- Formation of state-wide Rare Disease Advisory Committee
- Dissemination of education to prenatal offices, PCPs & outpatient labs
- Updated educational content for PCPs
- Added pre-diagnosis and post-diagnosis resources to website

Benefits to programs & families

- Both families and practitioners are more informed and serve as partners
- Ensures that new programs will be utilized by those they are designed for
- Adds the patient voice to "issues" we're hoping to address (i.e. payer/advocacy issues)
- Through the sharing of stories/journeys, Lets those who are newly diagnosed know they are not alone
- Families provide a very good perspective for user friendly and culturally appropriate materials (Spanish)

Compensation & Incentives



- Stipends
- Dependent Care Support
- Parking
- Travel Costs
- Training/Leadership Development
- Employment

<https://nationalhealthcouncil.org/fair-market-value-calculator/>



Lessons Learned

- Authentic Engagement needs **Trust**, and trust takes time
- Families are **Experts**
- Sometimes you have to **Ask-** more than once
- Clear and **Consistent Communication** is an essential
- Create a **“Home”** at the table

*Authentic engagement allows us to
TEACH each other*



3. Generate Action *What Can You Do?*



A Plan to Engage:

- 1 Raise Awareness:** Gather Insights, ask questions, share opportunities
- 2 Build Knowledge:** Define terms/project, set goals, assign roles
- 3 Generate Action:** Identify the measure(s) of success



Key Questions to Consider

- *How are you building trust with families?*
- *Who are your trusted community partners?*
- *What are the biggest challenges to your current system(s)?*
- *What type of engagement are you building? (short vs long term)*
- *What are we asking families to do?*
- *Do they feel prepared or need further training?*
- *How will you know you've been successful?*

Key Strategies for Success

- Inclusive Outreach
- Co-Creation & Co-Design
- Relevant & Timely Information
- Ongoing Evaluation & Reflection
- Innovative & Scalable Methods
- Trusted Partners



Measuring “Success”



CONFIDENCE



KNOWLEDGE



INVOLVEMENT



TRUST



INVESTMENTS

ACTION: What else Can **YOU** Do?

- 1 Raise Awareness:** Download & Share Resources
- 2 Build Knowledge:** Partner with Others
- 3 Generate Action:** Identify opportunities to partner with families in your work



Stay Connected.

A follow-up survey to come



www.ExpectingHealth.org



@ExpectingHealth



@ExpectHealthOrg



@ExpectingHealth



MORE SUPPORT. MORE GUIDANCE. BETTER HEALTH



HANDS &
VOICES™



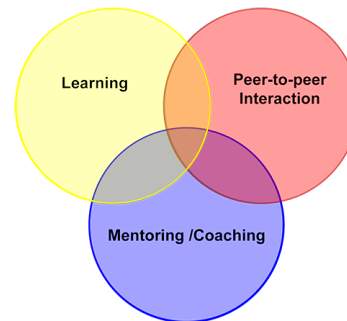
Promoting and Implementing Parent Engagement and Leadership

Lisa Kovacs – Director of Programs Hands & Voices

Hands & Voices *at a glance*



Leadership-to-Leadership L2L



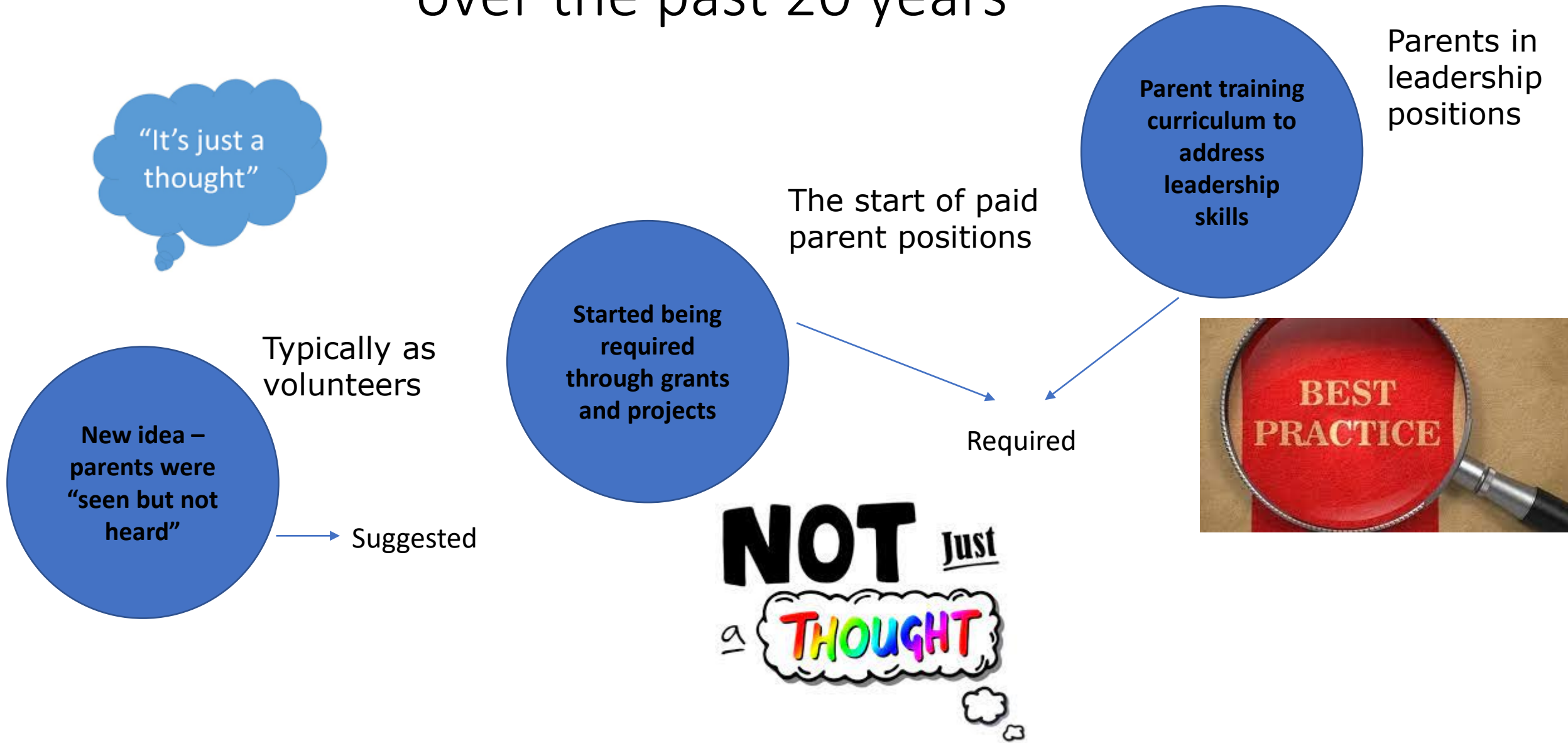


The purpose of the FL3 Center is to increase state capacity for family-to-family support, develop family leadership skills, and engage DHH adults as mentors to families across state/territory EHDI programs and other EHDI stakeholders.

New: EHDI National Network – Three national centers to advance EHDI systems nationwide so that Deaf and Hard of Hearing newborns, infants, and young children up to the age 3 receive appropriate supports and timely services to improve language acquisition outcomes.



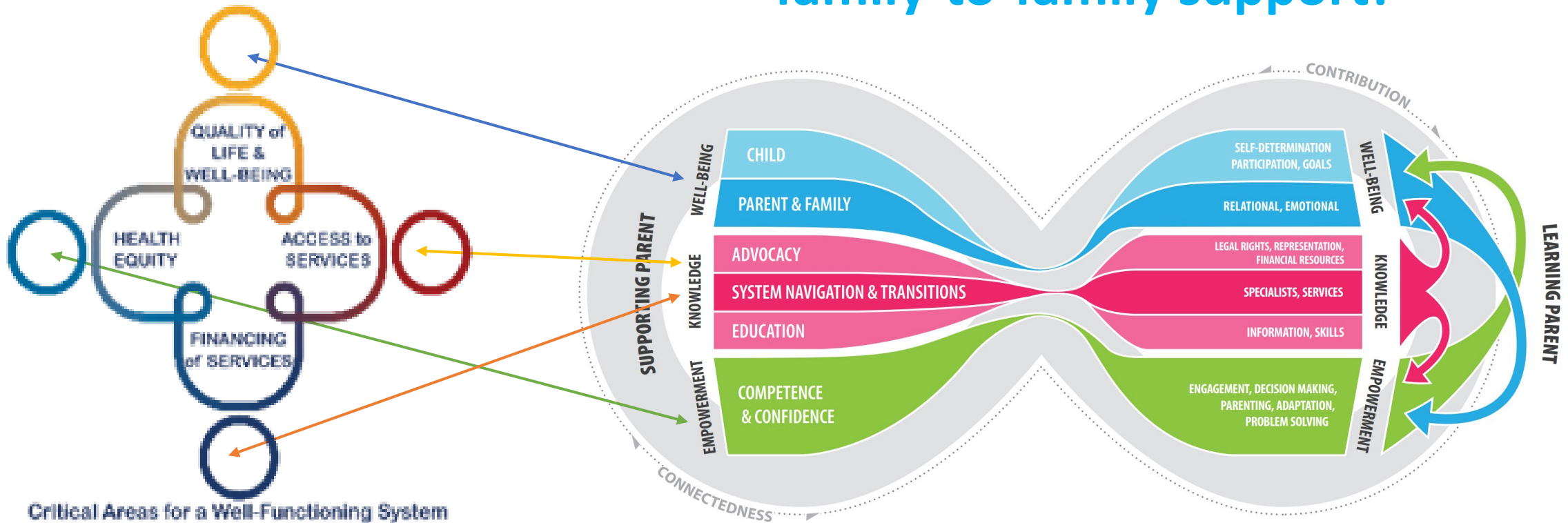
Family Engagement in the EHDI system over the past 20 years



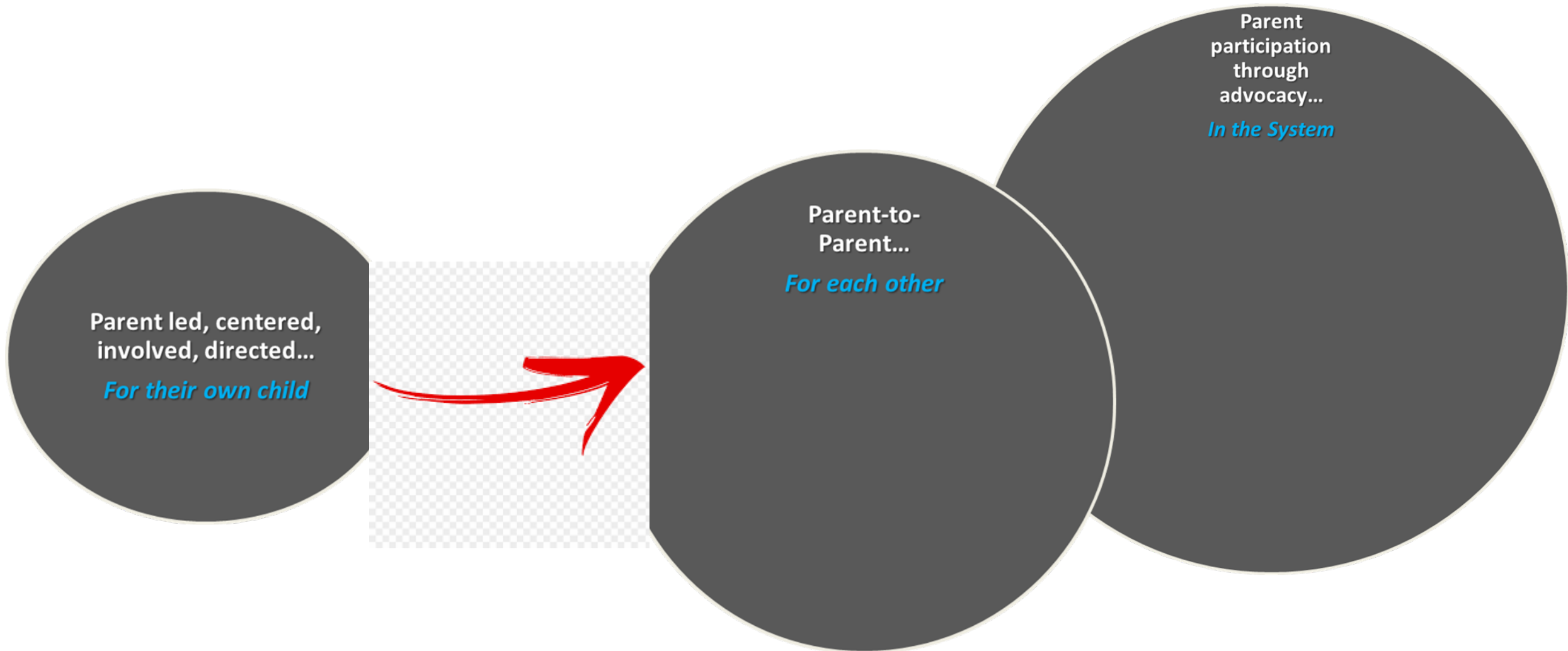
MCHB/Children and Youth
with Special Healthcare
Needs
Blueprint for Change

Family Engagement and Leadership...

It starts with families receiving
family-to-family support!



Building parent leadership for self, others, and systems



Parent
participation
through
advocacy...

In the System

- **Willingness to share insights and information about their experiences in ways that others can learn from**
- **Knowledge of diverse families - communication modes, special needs, medical/cultural viewpoints**
- **Respect for the perspectives of others**
- **Comfortable working in groups**
- **Desire to work in partnership with others**

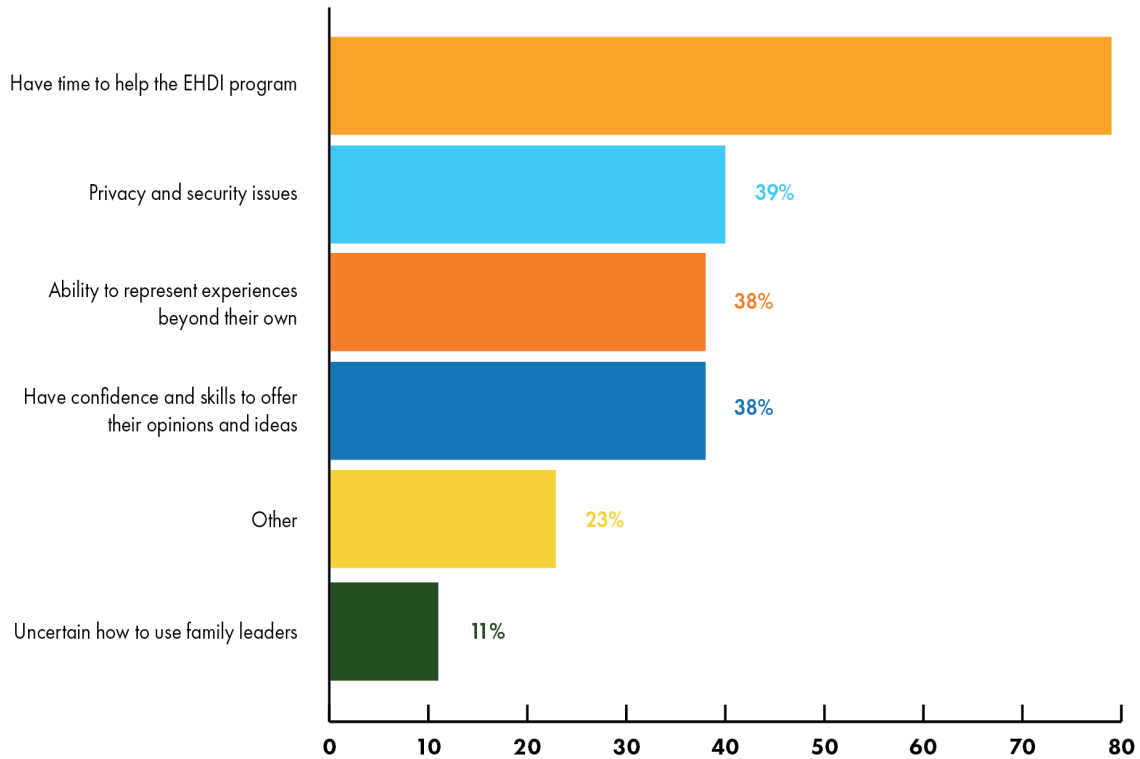


**HANDS &
VOICES™**

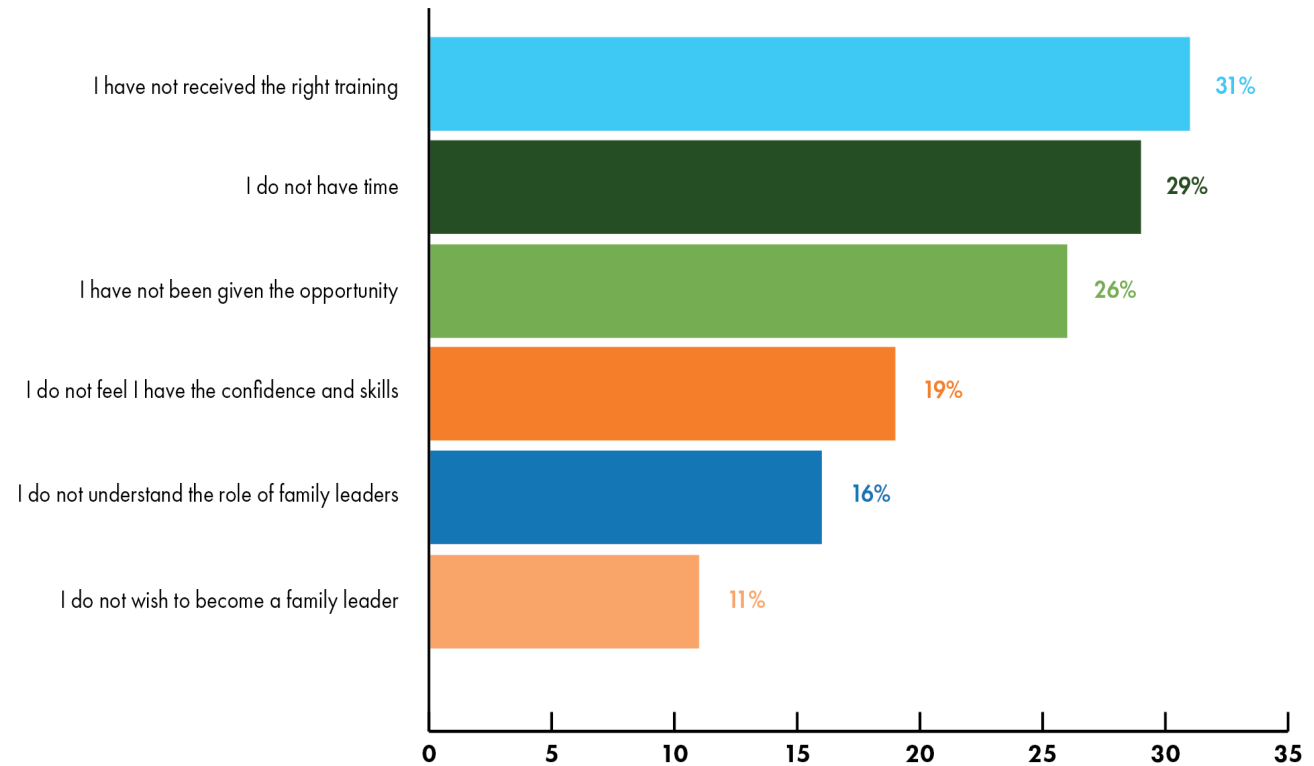
Findings from a National Needs Assessment 2018

EHDI Coordinators / FBOs Report

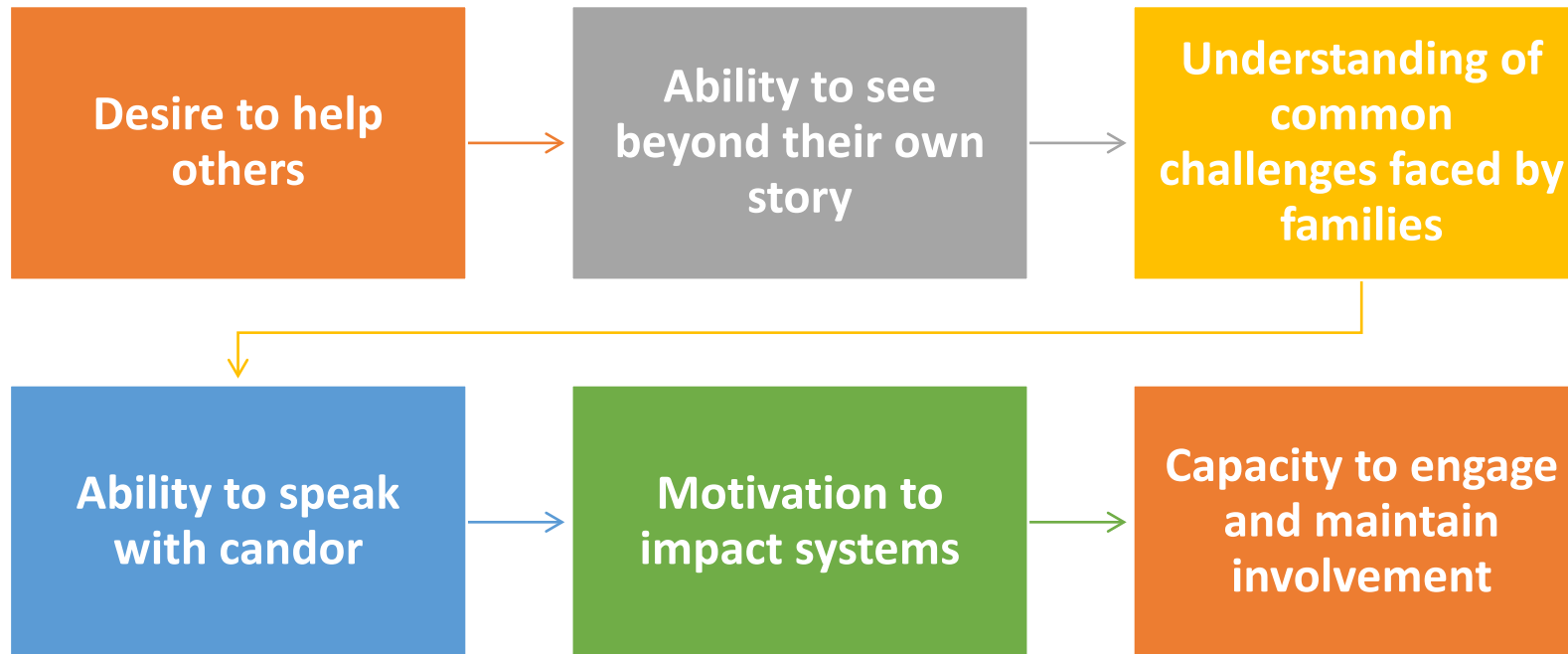
What challenges do you face in getting active family leadership in your EHDI system?



What challenges do you face in becoming a family leader?



Key qualities of a family leader



Planning/Preparation/Investment of Time Leads to Success

Initial
Assessment/gap
analysis

Recruitment

Maximizing
access

Onboarding

Meaningful
participation

On-going
assessment



TIPS FOR MEANINGFUL PARTICIPATION

TIPS FOR MEANINGFUL
PARTICIPATION BY PARENTS AND
ADULTS WHO ARE DEAF AND HARD
OF HEARING SERVING IN EARLY
HEARING DETECTION AND
INTERVENTION (EHDI) SYSTEMS*

**Specifically, Advisory Committees and Learning Communities*

H&V FL3 RESOURCES

H&V LEAD COURSE

Leadership Enhancement And Development



- This course is designed to provide participants with knowledge and resources to enhance their development as leaders in the systems serving families.
- Included in this curriculum are self-study activities covering three leadership domains: Personal Leadership, Leading Others, and Systems Leadership.
- **Register Here:**
<https://handsandvoicesfl3centercourses.mykajabi.com/offers/s4Xw7Zdo/checkout>

Building Family Leaders - Family Leader Support

Group Support

- FL3 Office Hours

<https://www.handsandvoices.org/fl3/office-hours/index.html>

- Leadership Book Club

<https://www.handsandvoices.org/about/hv-events.htm>

- Leadership Conference

<https://hvleadership2024.zohobackstage.com/HandsVoices20thAnnualLeadershipConference>

- FBO/EHDI Family Liaison Meeting at the annual EHDI Conference
- Annual Learning Community (FL3 & ICC)
- New –IDEA Lab – coming soon

Individual Support

- Onboarding new Family Leaders/EHDI Co.

- Individual TA

- H&V Online Academy

<https://handsandvoicesfl3centercourses.mykajabi.com/>

- Recorded Webinars

<https://www.handsandvoices.org/fl3/resources/trainings-webinars.html>

- Proactive TA check-ins

- Spotlight on Success

<https://www.handsandvoices.org/fl3/resources/comms-read-articles.html>

- Resource Toolbox

<https://www.handsandvoices.org/fl3/resources/toolbox.html>

*“I didn’t know
I was a
leader...Until I
found myself
leading...”*

“It is a curious thing, Harry, but perhaps those who are best suited to power are those who have never sought it. Those who, like you, have leadership thrust upon them, and take up the mantle because they must, and find to their own surprise that they wear it well.”

-J.K. Rowling Harry Potter and the Deathly Hallows

Consider something you plan to do by the end of next week that would impact family engagement, leadership, or family support in your program

Action
Changes
Things

Thank you!

fl3@handsandvoices.org
www.handsandvoices.org/fl3

Family Leadership in Language and Learning Center



“Where love, language, and learning thrive”

Explore the Long-term Follow up Landscape

LTFU Workgroup Co-Chairs



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New**STEPS**s

Exploring the Long-term Follow-up Landscape

*Jennifer Baysinger, MSN, RN
Jennifer Hauser, MPH, RN, PHN*

June 12, 2024

Objectives



1

Describe Long-term Follow-up

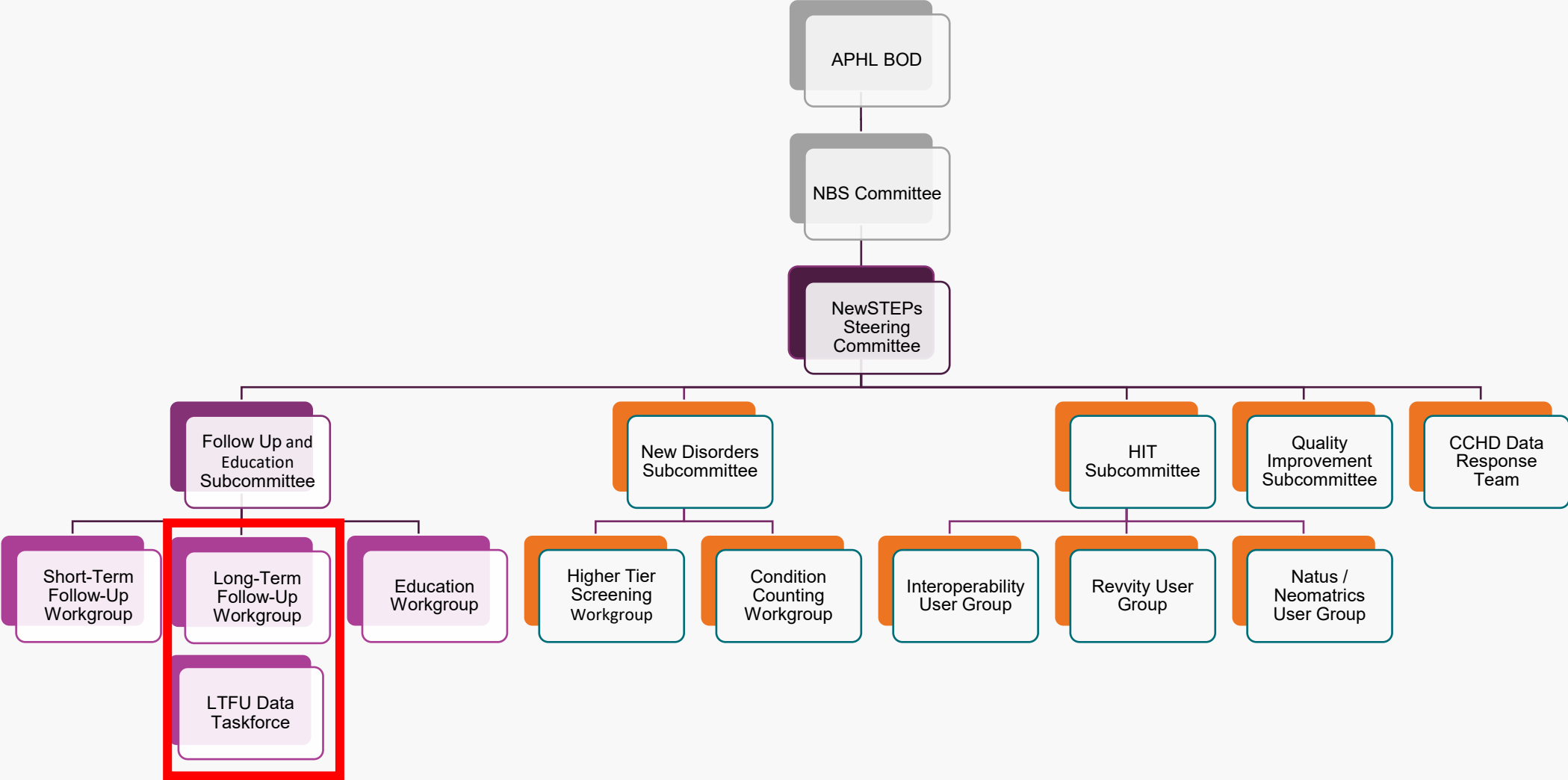
2

Provide an overview of Long-term Follow-up models

3

Describe the future landscape of Long-term Follow-up

NewSTEPS Long-Term Follow-up workgroup



LTFU Workgroup Charge & Activities

Objectives

- Develop a foundation for LTFU initiatives
- Define LTFU and its essential elements
 - Minimum data sets
 - Surveillance case definitions
- Identify existing LTFU models, LTFU elements beyond data, and feedback models for families and persons with lived experience
- Develop a LTFU fact sheet for programs to demonstrate LTFU value.
- Publish an APHL LTFU position statement.

Year 1 Activities

- Redefine LTFU culture and develop a position statement.
- Identify and distribute existing LTFU models and resources.
- Engage follow-up community members, registries, and research networks in national webinars.
- Demonstrate LTFU value to NBS programs.
- Shift NBS Culture: Public Health vs. Clinical Assessment



What is Long-term Follow-up (LTFU)?

Long-term Follow-up Definitions

Clinical and Laboratory Standards Institute (CLSI) - 2023

Long-term follow-up (LTFU) – ongoing steps taken after diagnosis to prevent morbidity and mortality in affected individuals.

- In the context of public health, LTFU most often involves periodic assessments of health outcomes as part of system evaluation. Other activities might include but are not limited to care coordination and assurance of access to treatments and/or other interventions.

CLSI. *Newborn Screening Follow-up and Education*. 3rd ed. CLSI guideline NB02. Clinical and Laboratory Standards Institute; 2023.



Long-term Follow-up Definitions, cont.

NewSTEPs Long-Term Follow-Up Taskforce (2020)

Long-term follow-up (LTFU) is an essential component of the Newborn Screening (NBS) system that begins after an infant has been diagnosed with a condition screened for by NBS and extends throughout the lifetime of the individual. Long-term follow-up serves to:

- Evaluate health and developmental outcomes in affected children and their families,
- Assess whether there are appropriate benefits to early detection and treatment for all conditions,
- Continually improve upon the NBS, public health, and medical systems to maximize benefits, and
- Reduce condition-associated morbidity and mortality through ongoing care, connection to services, and management.

LTFU relies on the collaboration and integration of both public health and clinical follow-up. LTFU may include care coordination, assuring the availability of evidence-based treatment, continuous quality improvement, and new knowledge discovery, as well as periodic assessment of the clinical and developmental outcomes in affected individuals.

[Long-Term Follow-Up Taskforce Report | NewSTEPs](#)

ACHDNC Follow-up and Treatment Workgroup (2008)

Fundamentally, long-term follow-up comprises the assurance and provision of quality chronic disease management, condition-specific treatment, and age-appropriate preventive care throughout the lifespan of individuals identified with a condition included in newborn screening. Integral components of LTFU:

- Care coordination through a Medical Home
- Evidence-based treatment
- Continuous quality improvement
- New Knowledge discovery – the “unnatural history” of the disease
- Active surveillance and evaluation of data related to care and outcomes.

Kemper, et al. Genet Med. 2008 Apr;10(4):259-61



Long-term Follow-up Goals

Public Health

- To improve life-time health and developmental outcomes for children identified with a condition screened for by NBS
- To identify how the NBS system is achieving the primary aims of preventing mortality and mitigating morbidity
- To assure equitable access, effective NBS programs, and interventions for children with conditions screened for by NBS and their families
- To use surveillance data to inform public health actions such as policy change, targeted outreach, or additional resources to address barriers

[Long-Term Follow-Up Taskforce Report | NewSTEPS \(2020\)](#)

Clinical Care

- To improve life-time health and developmental outcomes for children identified with a condition screened for by NBS
- To ensure appropriate care coordination across the clinical system
- To collect data for the purposes of evaluating and improving evidence-based treatment/care effectiveness



Public Health Surveillance Defined

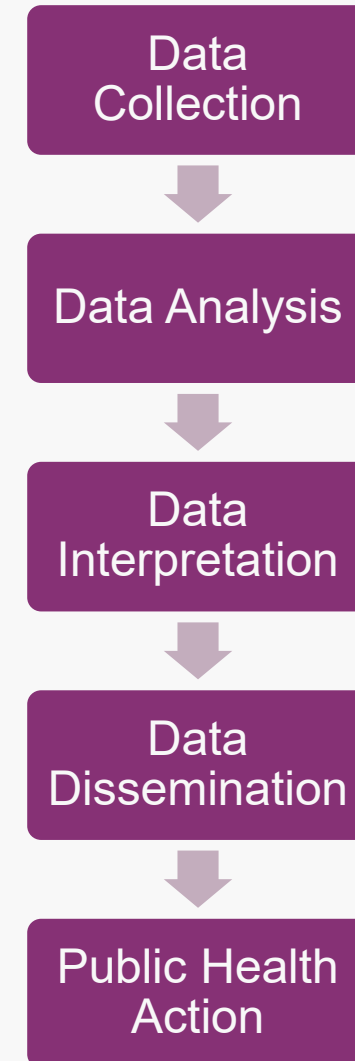
The ongoing, systematic collection, analysis, and interpretation of health-related data essential to planning, implementation, and evaluation of public health practice, closely integrated with the timely dissemination of these data.

Adapted from: Thacker SB, Birkhead GS. Surveillance. In: Gregg, MB, ed. Field epidemiology. Oxford, England: Oxford University Press; 2008.

[Introduction to Public Health Surveillance | Public Health 101 Series | CDC](#)

Goal of Public Health Surveillance

- Provide information that can be used for **action** by public health personnel, government leaders, and the public to guide public health policy and programs



Smith PF, Hadler JL, Stanbury M, et al. Blueprint version 2.0: updating public health surveillance for the 21st century. J Public Health Manag Pract 2013;19:231–9.

How can we use LTFU [surveillance] data?

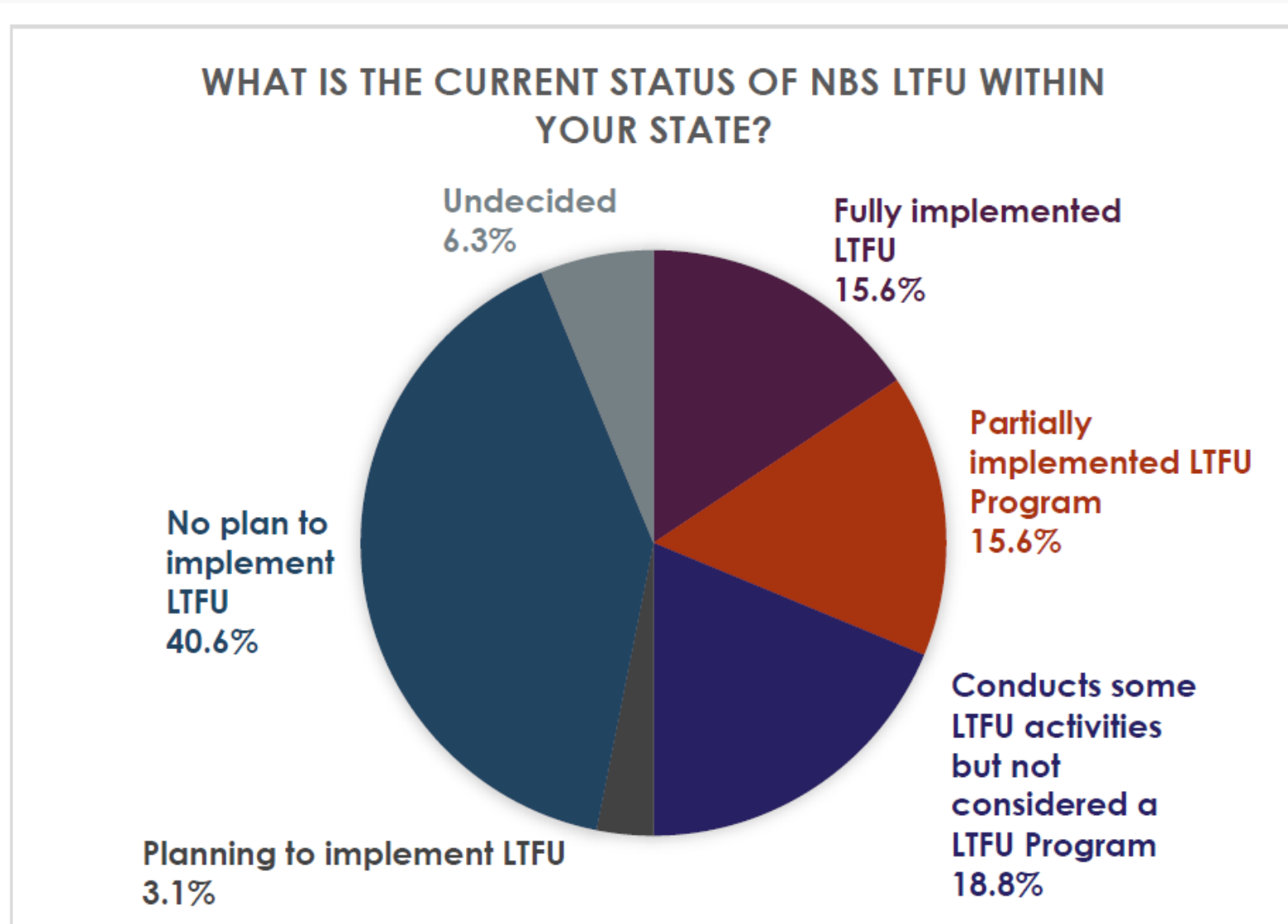
Goals of data collection

- Aid in evaluation of the benefits of early identification through newborn screening
- Better understand health care utilization
- Identify disparities in outcomes for children with newborn screening conditions
- Identify inequities in experiences, access, and resources
- Assess effects on public health & health care systems upon adding conditions to newborn screening
- Strengthen the evidence base for public health action
- Inform policy, public health practice, decision-making, and process improvements through a health equity perspective



Models for Long-term Follow-up

States Vary on Ability and Capacity to do LTFU





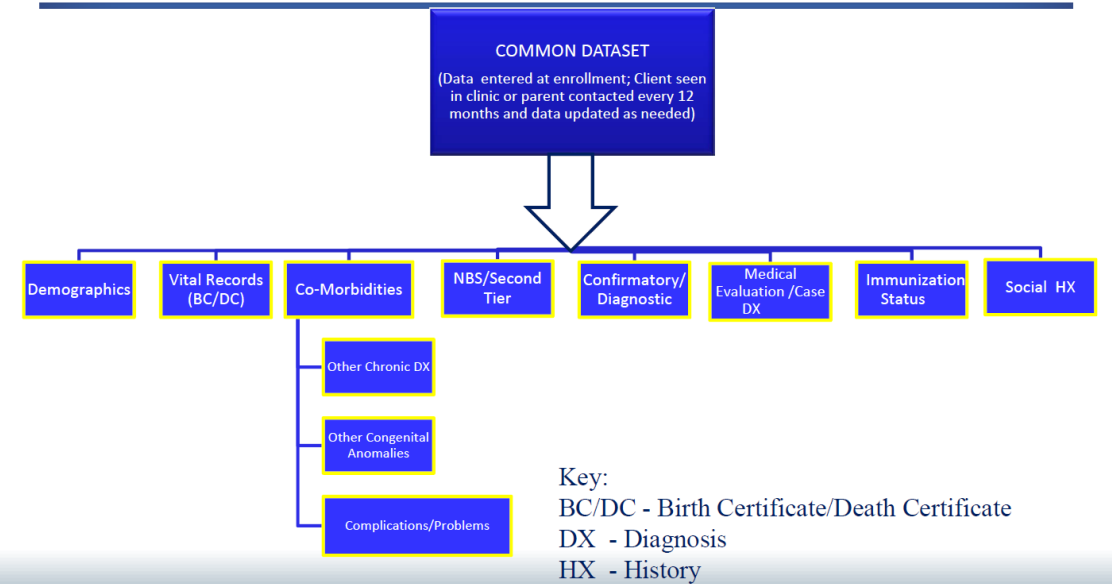
Arkansas Model

Arkansas NBS LTFU Database

- Acknowledgement to NBSTRN: Utilized common data elements developed by NBSTRN as our starting point
- Collaborated with ACH clinical experts to develop 8 disease-specific data sets
- Utilized REDCap (Research Electronic Data Capture) hosted by UAMS Translational Research Institute (NCRR/NIH 1 UL1 RR02988)
- REDCap secure, web-based application designed to capture research studies
- Database created by abstracting and compiling information from the medical record.
- Data abstraction from the medical record for each visit to ACH Specialty Clinics



Common Dataset



California Model



Table 3 Indicators of patient health status at each year of follow-up

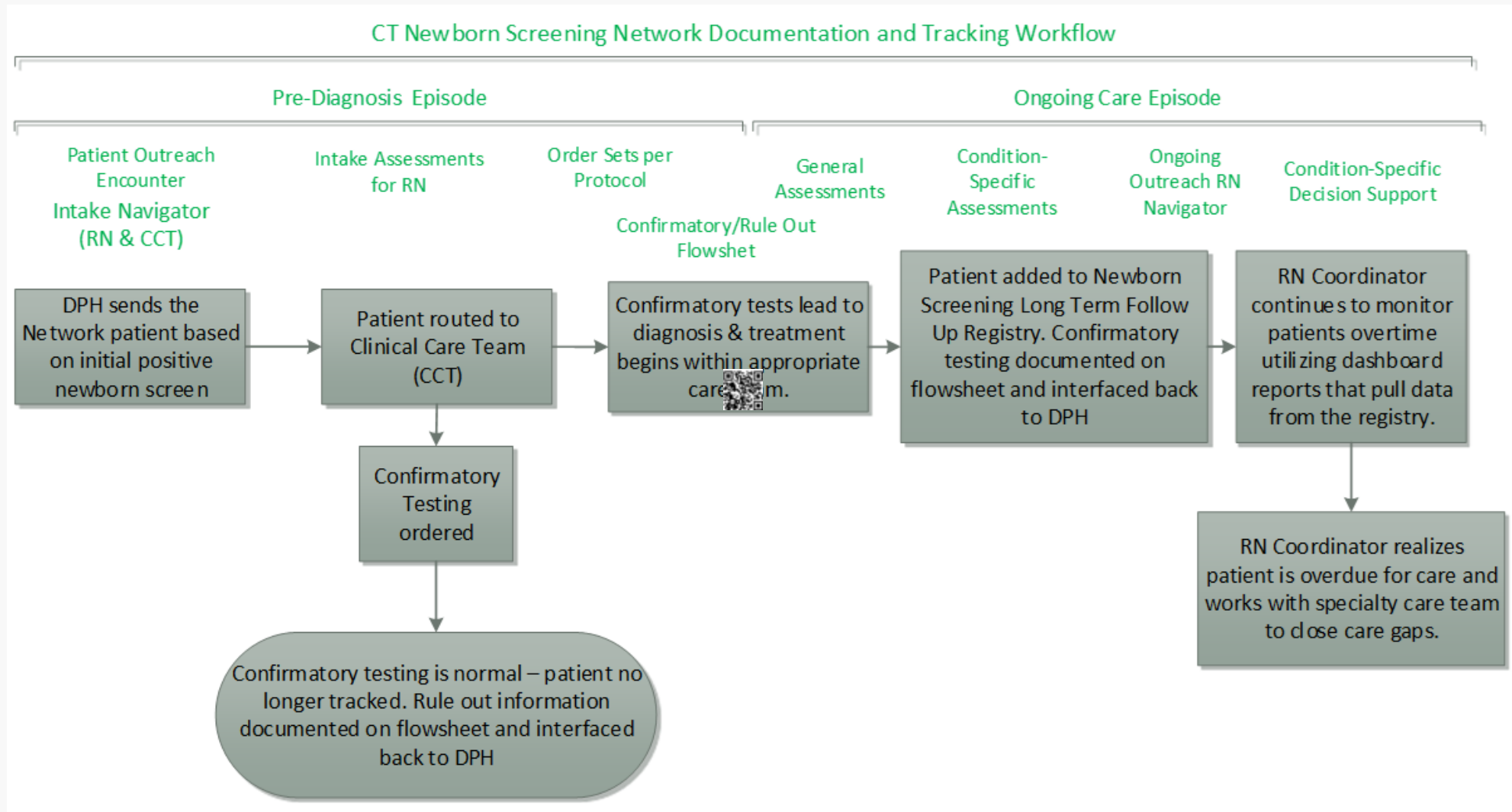
	First year, n (%)	Second year, n (%)	Third year, n (%)	Fourth year, n (%)	Fifth year, n (%)
Developmental delays (includes those with mild, moderate or severe delays)					
Speech/language	28 (10)	39 (18)	20 (22)	4 (8)	4 (10)
Physical growth	26 (9)	25 (12)	13 (14)	1 (2)	1 (2)
Mental/cognitive	26 (9)	26 (12)	14 (15)	4 (8)	2 (5)
Gross motor skills	34 (12)	27 (13)	9 (10)	None	1 (2)
Fine motor skills	27 (9)	25 (12)	9 (10)	1 (2)	None
Age appropriate development					
Speech/language	236 (83)	147 (69)	61 (68)	39 (80)	33 (82)
Physical growth	244 (86)	164 (77)	70 (78)	42 (86)	37 (92)
Mental/cognitive	239 (84)	157 (74)	68 (75)	38 (78)	36 (90)
Gross motor skills	235 (82)	161 (76)	73 (81)	43 (88)	37 (92)
Fine motor skills	233 (82)	161 (76)	69 (77)	42 (86)	38 (95)
Symptoms					
No symptoms	190 (67)	125 (59)	66 (73)	32 (65)	30 (74)
Symptoms	69 (24)	56 (26)	18 (20)	7 (14)	5 (13)
Unknown	26 (9)	31 (15)	6 (7)	10 (21)	5 (13)

Table 9 California's LTFU system can address the following questions

- What percent of children diagnosed with disorders remain in care between the ages of 1 and 5 yr old?
- What percent become lost to follow-up?
- What percent of parents refuse treatment?
- What percent died due to problems associated with the disorder?
- What percent were determined not to need ongoing treatment?
- What percent of children (combined or by specific type of disease) had age appropriate developmental status with respect to speech, physical development, mental/cognitive development, and gross motor and fine motor development?
- What percent of children were severely delayed with respect to any of the developmental measures and what year of life did the delays become apparent?
- What percent of patients experienced symptoms associated with their disorder and at what age did the symptoms become apparent?
- In any given year, what percent of children experienced the loss of skills they had previously acquired?
- What percent of children had no hospitalizations or emergency room visits in the previous year of life?
- What disorders are associated with the greatest number of hospitalizations and emergency room visits due to disorder-related complications?
- What disorders are associated with the highest utilization of metabolic center visits?
- What percent of children are receiving a multidisciplinary team of services, including nutritional counseling, health education, and social services counseling?

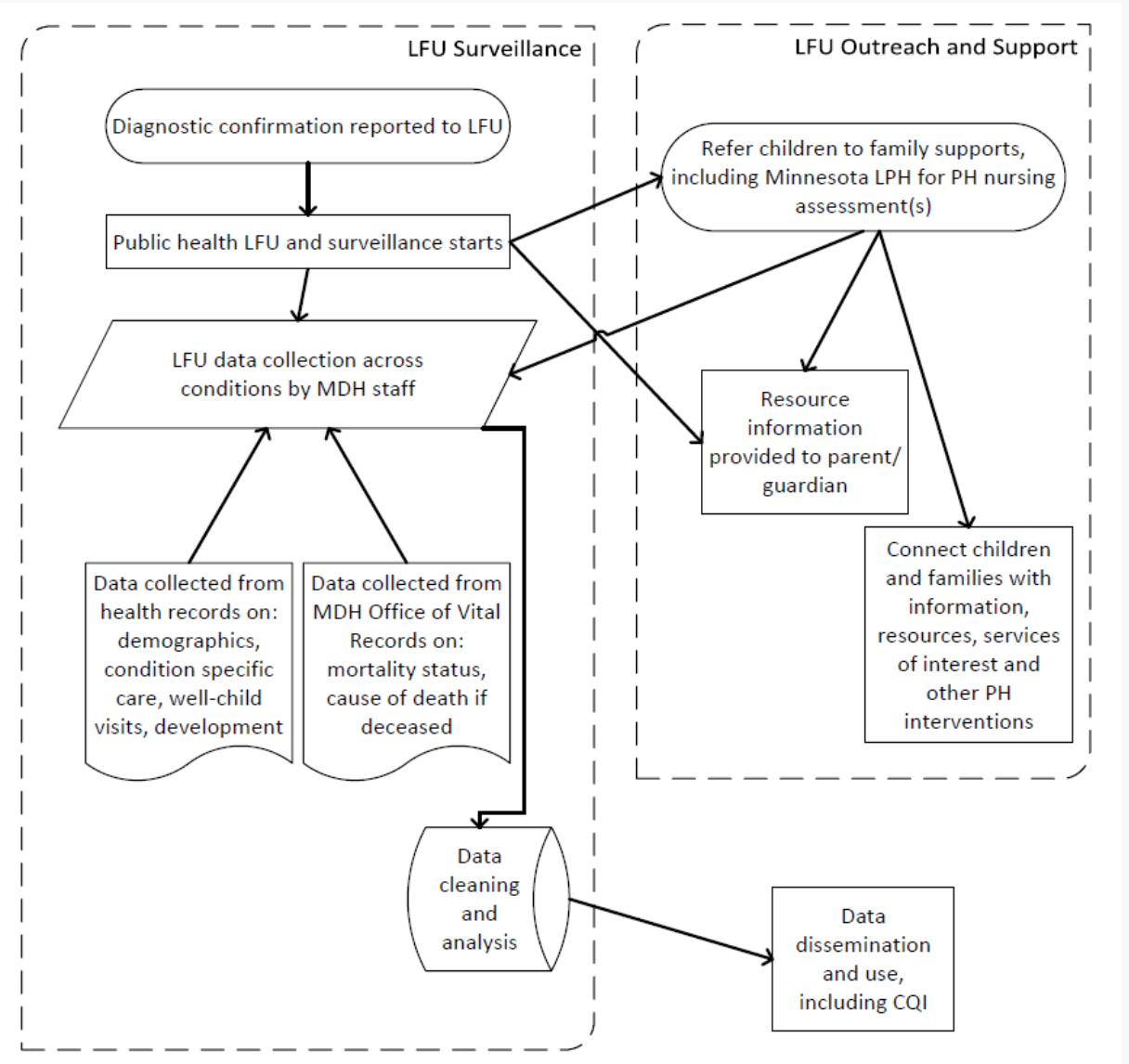
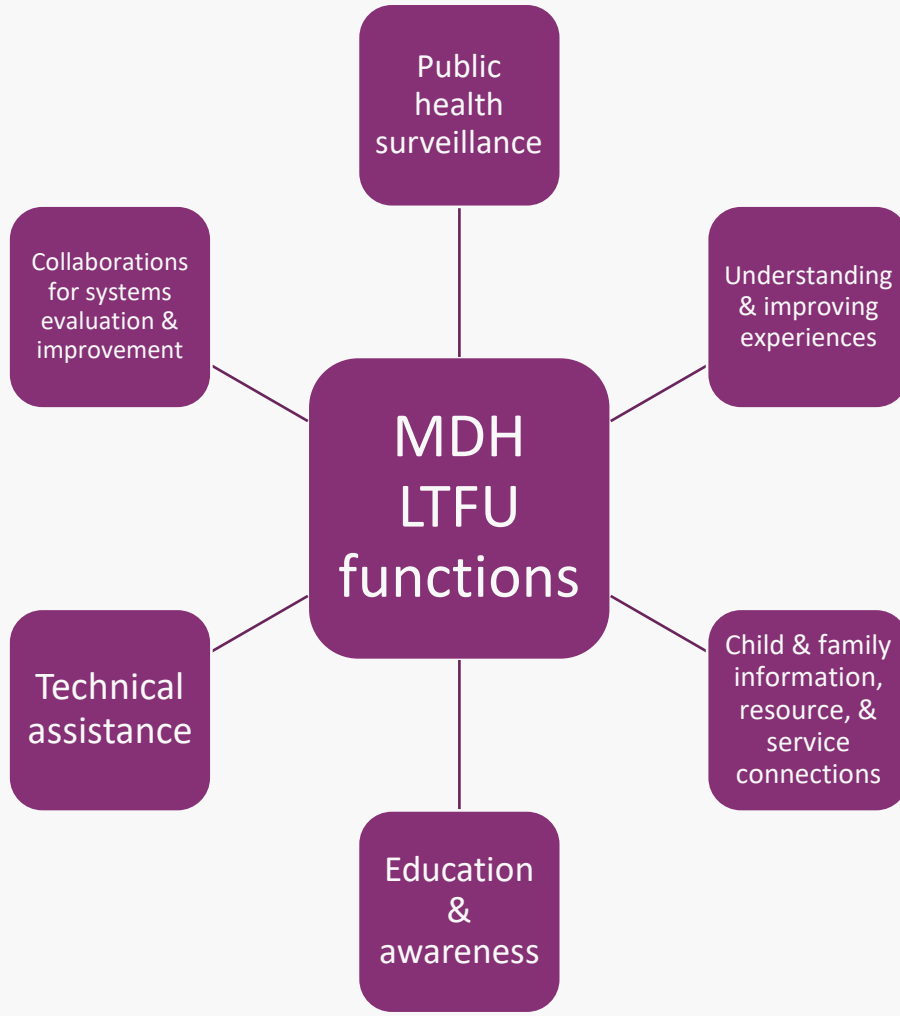


Connecticut Model





Minnesota Model



New York Model

LTFU Structure: Patient Registry

- Consent-based patient registry.
- 2022-2024: Developed data dictionaries by a group of metabolic specialists and NBSP for inherited metabolic disorders screened in NYS.
- Currently we are building an in-house LTFU data system/portal in the Health Commerce System. The [REDCap](#) resources hosted by NBSTRN has ended in Feb 2024.
- Currently SCC staff enter data using survey forms.
- Data elements: General demographic vs. Disorder-specific information

NBSP: Newborn Screening Program; **NBSTRN:** Newborn Screening Translational Research Network; **SCC:** Specialty Care Centers



General Shared Data Elements

vs.

Disorder-Specific Data Elements

- Demographics
- Socioeconomic factors
- Hospitalizations
- Development and education
- Visit measurements
- Project status

- Newborn screen results
- Diagnostic testing
- Nutrition
- Pharmacotherapy
- Care coordination
- Developmental assessment
- Lab testing



North Dakota Model

NORTH DAKOTA LONG TERM FOLLOW UP SNAPSHOT

Mission:

- To ensure identified children have access to quality, evidence-based disease management, age-appropriate preventive care, care coordination through a medical home and programmatic continuous quality improvement.

Project Outcomes:

- To build a system that ensures children identified with conditions are not lost to follow up
- To ensure that children identified with conditions have access to high quality health care & early intervention services thus leading to improved health outcomes
- To develop a data base and reports that align with national follow up efforts.

Program Objectives:

- Engage with families and specialists to ensure cohesive communication
- Refer families to valuable community supports (EI, Insurance navigation, WIC, Medicaid, etc)
- School readiness data to direct ongoing activities
- Access to experienced parents (parent referral system for additional supports)

This Photo by Unknown Author is licensed under CC BY-SA

Oklahoma Model

Baby Diagnosed

LTFU contacts parents and completes family survey twice per year

Survey data is entered into REDCap

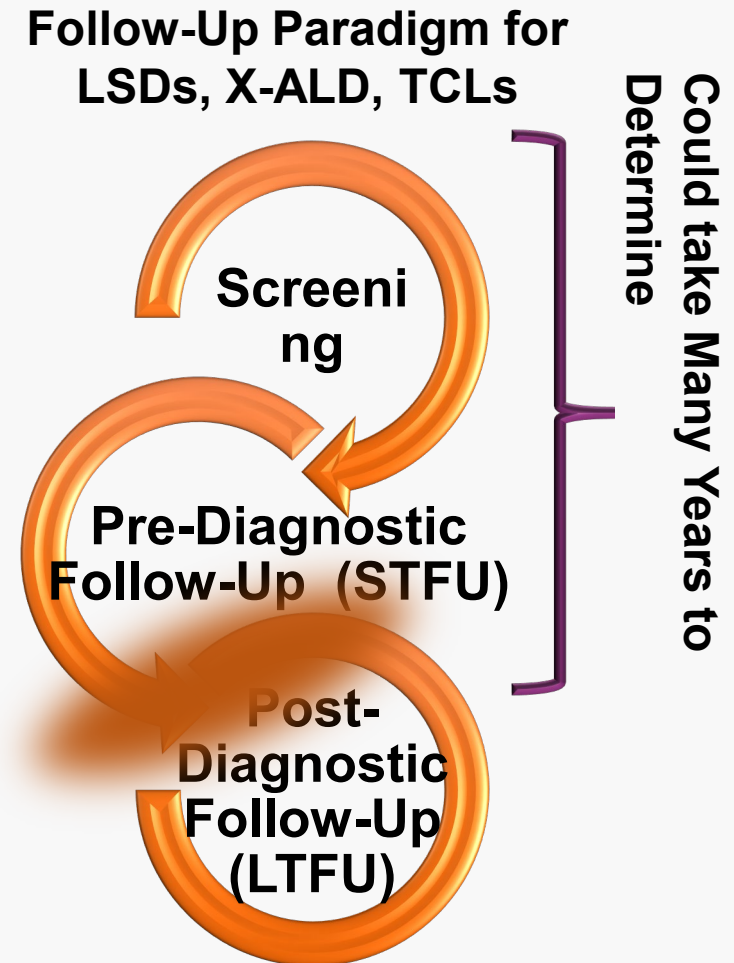
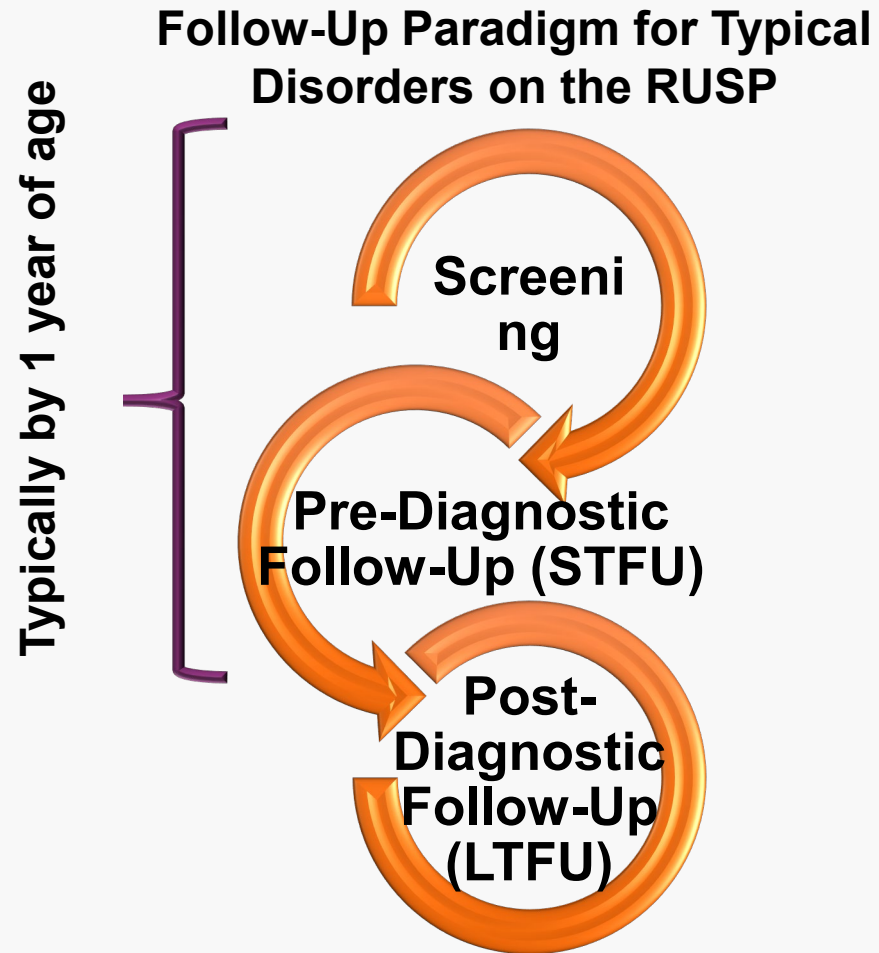
LTFU sends survey link to appropriate specialty clinic annually

NBS reimburses specialty clinic for data submission



The Future of Long-term Follow-up

Stretching Follow-Up: When is a Case a Case?



Late-Onset Variants and Follow-Up

- How does public health best intersect with clinical team? And for how long?
- How does public health best interface with the family/patient? And for how long?
- Reported drop off rate for adherence to monitoring – how do we ameliorate this?
 - Motivation for families presenting clinically varies from asymptomatic families detected via newborn screening



How can I support this work if I'm not a member of the LTFU workgroup?



Thank you!

Jennifer Baysinger
JenniferXA@health.ok.gov

Jennifer Hauser
jennifer.hauser@state.mn.us

Discussion Groups

Discussion Prompts

Breakout Rooms

1. How do the long-term follow-up models presented align with the needs of the populations served by your newborn screening program?
 - a. How does public health best interface with the family/patient? And for how long?
 - b. How does public health best intersect with clinical team? And for how long?
2. What performance metrics and outcome measures should be prioritized to assess the effectiveness and impact of long-term follow-up efforts?
3. How can technology and data systems be leveraged to streamline communication and information sharing between healthcare providers, families, and the newborn screening program during long-term follow-up?
4. What challenges or barriers might arise in implementing the long-term follow-up models presented, and how can they be addressed proactively?
5. How can partnerships with community organizations, advocacy groups, and healthcare providers strengthen long-term follow-up services?
6. Create your own discussion prompt (choose your own adventure 😊)

NewSTEPs Quality Indicators and Improvement Strategies




New**STEP**s

NewSTEPs Quality Indicators and Improvement Strategies

Ashley Comer and Amanda Jenkins, APHL

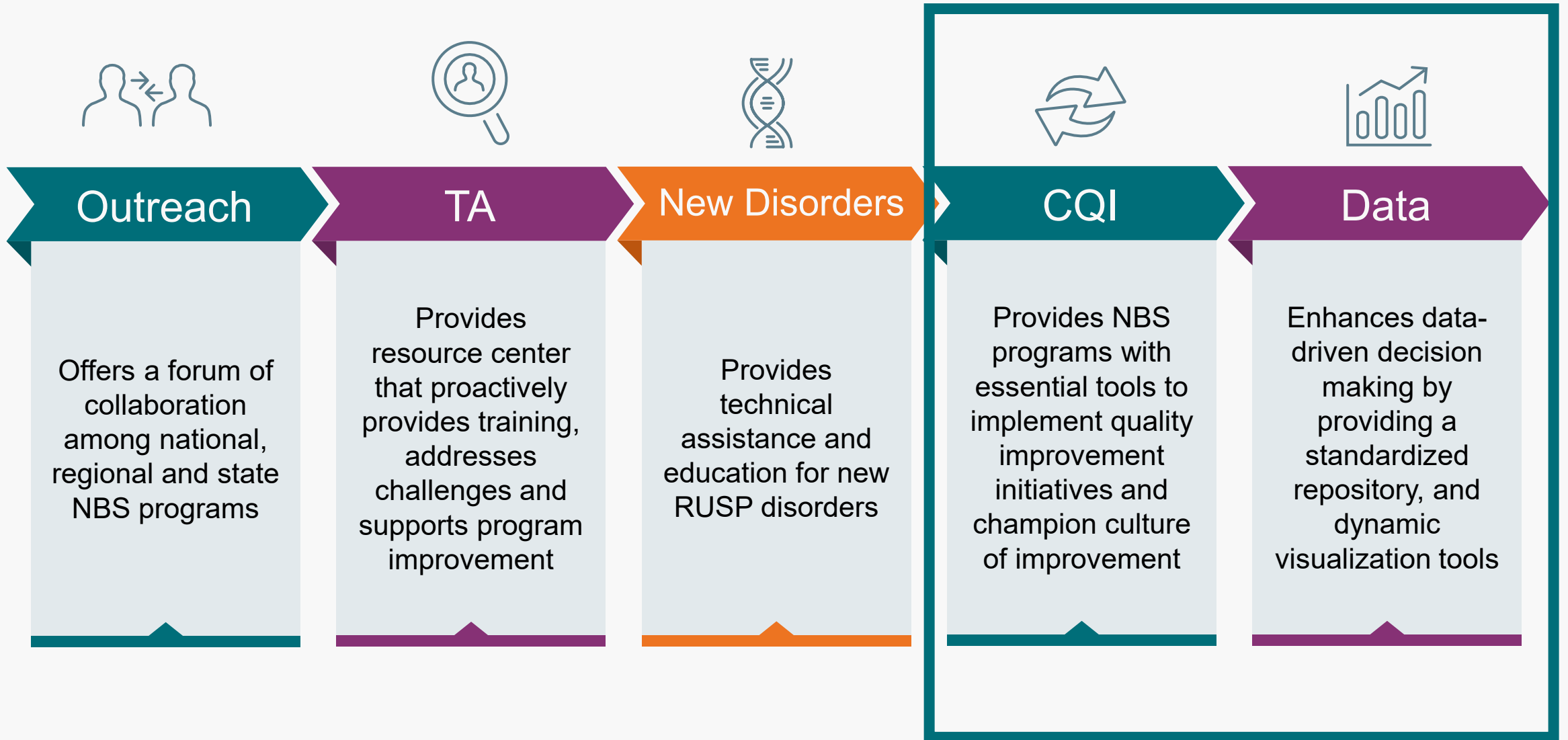
June 12, 2024

National Center for Newborn Screening System Excellence (NBS Excel)

 **NewSTEPS** is a national newborn screening resource center designed to provide data, technical assistance to newborn screening programs and assist with quality improvement initiatives.

This presentation is supported by the Health Resources and Services Administration (HRSA) under Cooperative Agreement #U22MC24078 for \$2,750,000. This content is those of the authors and should not be construed as the official position or policy of, nor should any endorsements be inferred by HRSA, HHS or the US Government.

What We Do



Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) Timeliness Recommendations

Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) Timeliness

Recommendations: To achieve the goals of timely diagnosis and treatment of screened conditions and to avoid associated disability, morbidity and mortality, the following time frames should be achieved by NBS systems for the initial newborn screening specimen:

- Presumptive positive results for time-critical conditions should be communicated immediately to the newborn's healthcare provider but no later than five days of life. **(Q15d.i)**
- Presumptive positive results for all other conditions should be communicated to the newborn's healthcare provider as soon as possible but no later than seven days of life. **(Q15d.iii)**
- All NBS tests should be completed within seven days of life with results reported to the healthcare provider as soon as possible. **(Q15d.iii)**

In order to achieve the above goals:

- Initial NBS specimens should be collected in the appropriate time frame for the newborn's condition but no later than 48 hours after birth **(Q15a.i)**, and
- NBS specimens should be received at the laboratory as soon as possible; ideally within 24 hours of collection. (Q15b.i)

Quality Indicators

Overview

Data Submission Process



[NewSTEPs Quality Indicator Webinar](#)

Why are the Quality Indicators Important?

- Performance metrics
- Tracks trends over time
- Informs quality improvement initiatives
- Standardization and harmonization → see [QI definitions](#)



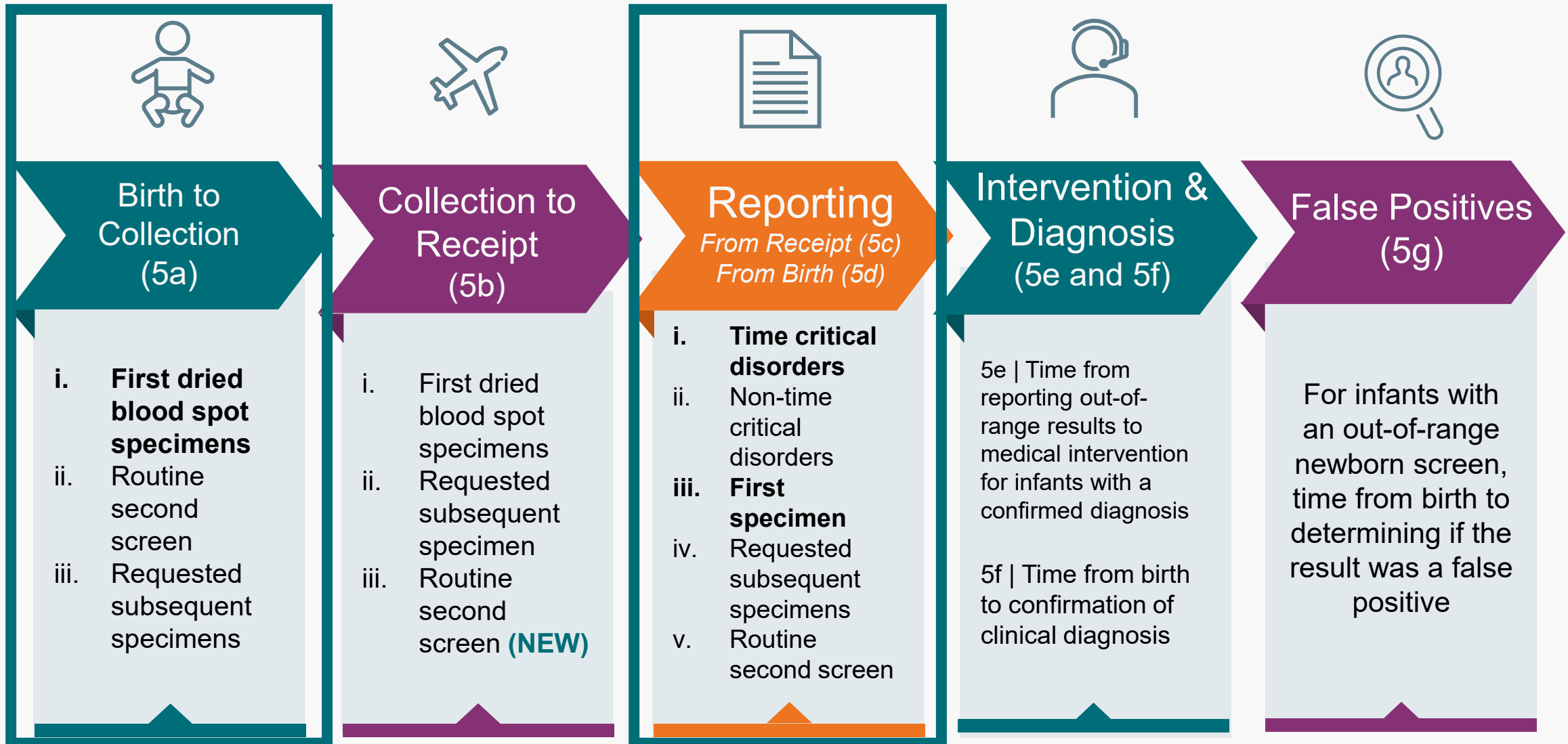
What are the Quality Indicators?

Quality Indicator	Level	Submission
1-Unsatisfactory Specimens	Specimen	Monthly and Annual
2- Missing Essential Information	Specimen	Monthly and Annual
3- Unscreened Newborns	Baby	Annual
4-Lost to Follow-up	Baby	Annual
5-Timeliness	Specimen and Baby	Monthly and Annual
6-Screen Positives	Baby	Annual
7-Confirmed Cases	Baby	Annual
8-Missed Cases	Baby	Annual

Quality Indicators

Timeliness

Timeliness Quality Indicators



Overview of the QI 5 | Timeliness

To identify the components of the newborn screening system that can be shortened to decrease the risk of potential harm to infants who may be identified with a disorder on the newborn screening panel

QI5a.i

Time from birth to specimen collection for the **first** dried blood spot specimens

Goal: within 48 hours of life

QI5d.i

Time from birth to reporting **out-of-range** results to an appropriate medical provider, for **time-critical disorders**

Goal: within 5 days of life

QI5d.iii

Time from birth to reporting out for **all** results to an appropriate medical provider, for **first** dried blood spot specimens

Goal: within 7 days of life

QI5a.i

Time from birth
to specimen collection for
the **first** dried blood spot
specimens

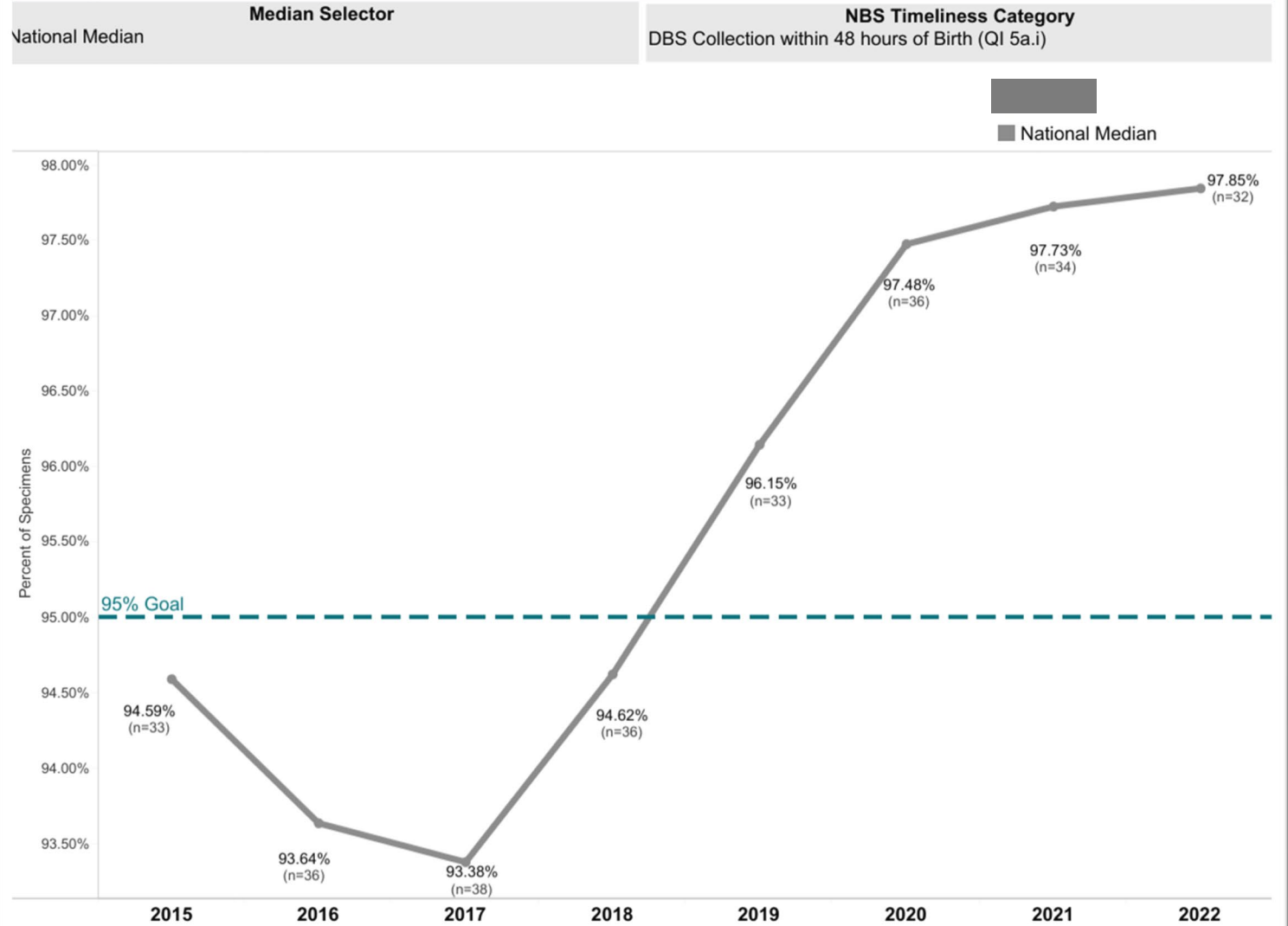
Goal: within 48 hours of life

Q15a.i

Time from birth to specimen collection for the **first** dried blood spot specimens

Goal: within 48 hours of life

DBS Collection within 48 hours of Birth (Q1 5a.i)



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Back



More Information

Q15a.i

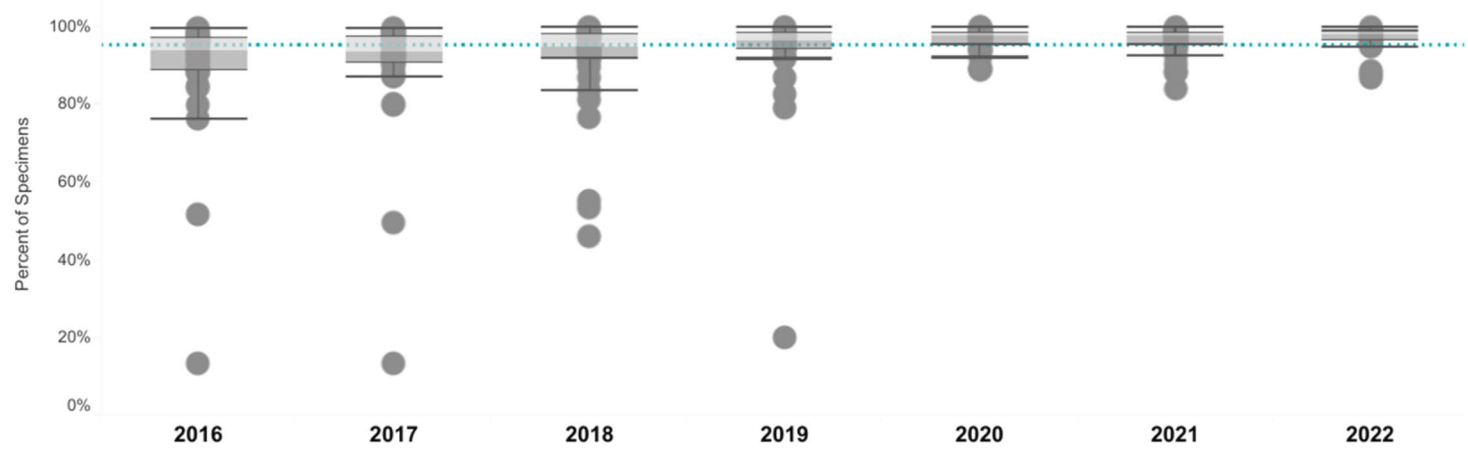
Time from birth to specimen collection for the **first** dried blood spot specimens

Goal: within 48 hours of life

DBS Collection within 48 hours of Birth (Q1 5a.i)

NBS Timeliness Measure

DBS Collection within 48 hours of Birth (Q1 5a.i)

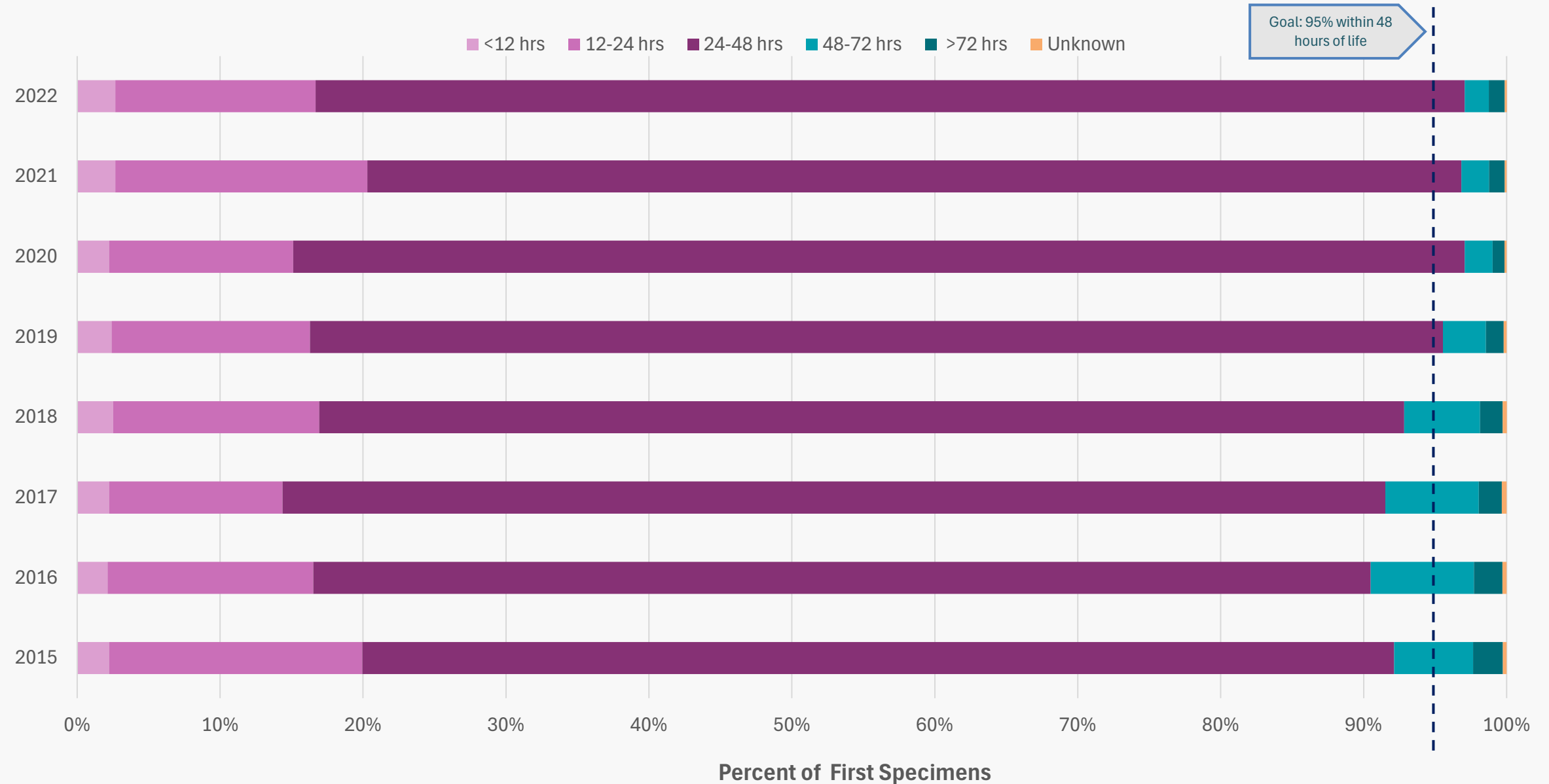


Year	Number of Programs Reporting	No. of Programs Above 95%	% of Programs Above 95%	Minimum	Maximum	National Median	Standard Deviation
2022	32	27	84.4%	86.8%	99.7%	97.8%	3.2%
2021	34	27	79.4%	83.8%	99.6%	97.7%	3.7%
2020	36	28	77.8%	88.9%	99.7%	97.5%	2.9%
2019	33	21	63.6%	20.1%	99.6%	96.1%	13.9%
2018	36	17	47.2%	46.1%	99.6%	94.6%	13.1%
2017	38	16	42.1%	13.2%	99.5%	93.4%	15.5%
2016	36	16	44.4%	13.3%	99.4%	93.6%	15.7%

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Time from birth to specimen collection for the first dried blood spot specimens (QI 5a.i)



Q15a.i

Time from birth to specimen collection for the **first** dried blood spot specimens

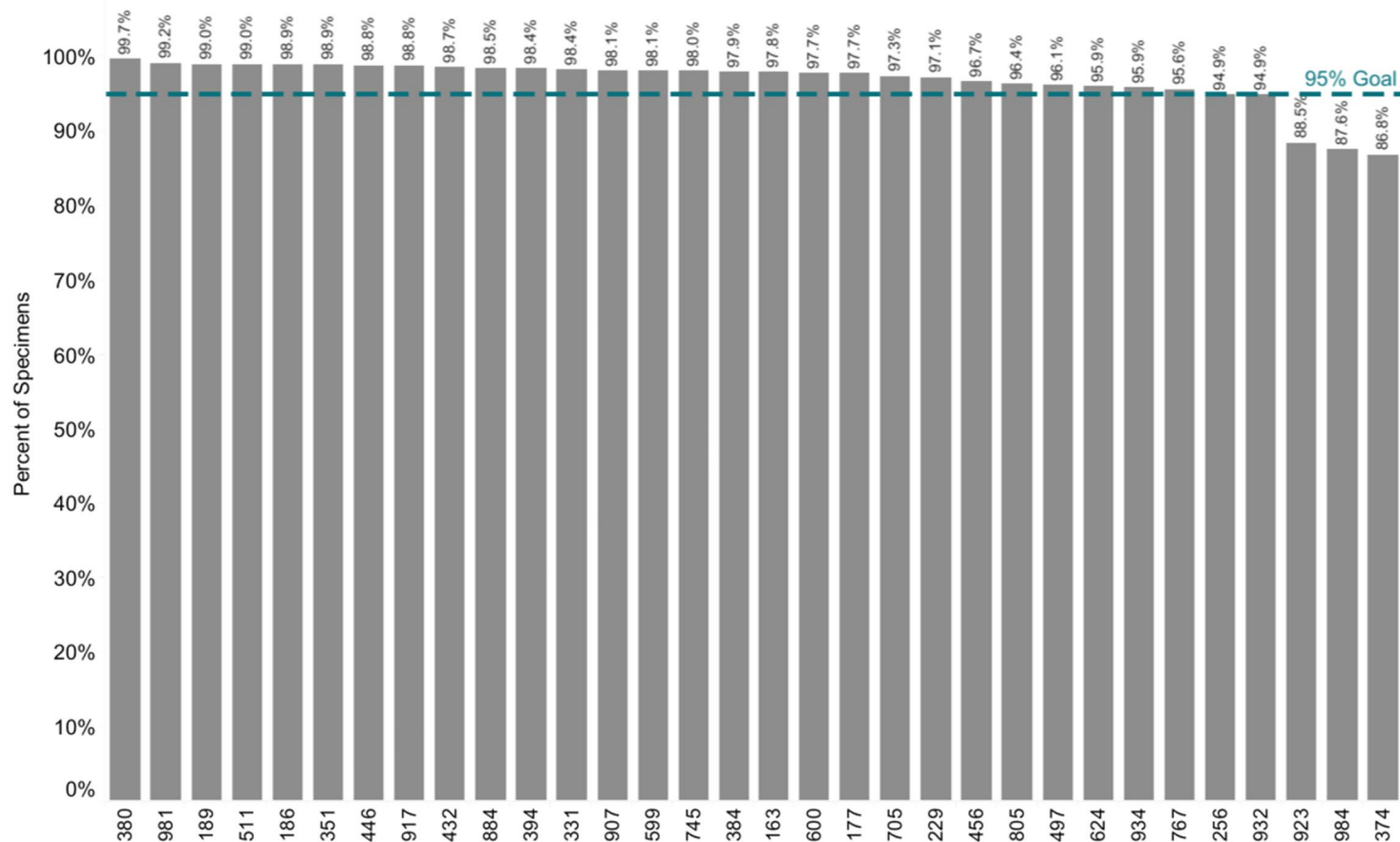
Goal: within 48 hours of life

DBS Collection within 48 hours of Birth (Q1 5a.i)

NBS Timeliness Activity
DBS Collection within 48 hours of Birth (Q1 5a.i)

2022

Year



Program ID

Last Updated: 5/21/2024 3:09:10 PM



Back



A Program of the Association of Public Health Laboratories



More Information

Quality Improvement Strategies

QI5a.i Goal: Collect the **first** dried blood spot specimens within **48 hours** from birth.

Quality Improvement Tools:

- Swimlane diagram
- Map workflow
- Gemba walk

Considerations:

- Outreach efforts
 - Safety and quality language with submitters
 - Visits (in-person or virtual)
 - Trends on certain days of the week (weekend package, holidays) impacting collection for individual collectors
 - Time of birth impacts on collection time (routine sweeps in hospital)
 - Determine barriers and strengths for community births
- Policy and regulations

Resources

NewSTEPS Toolkit

Timeliness Toolkit for Expanding Newborn Screening Services

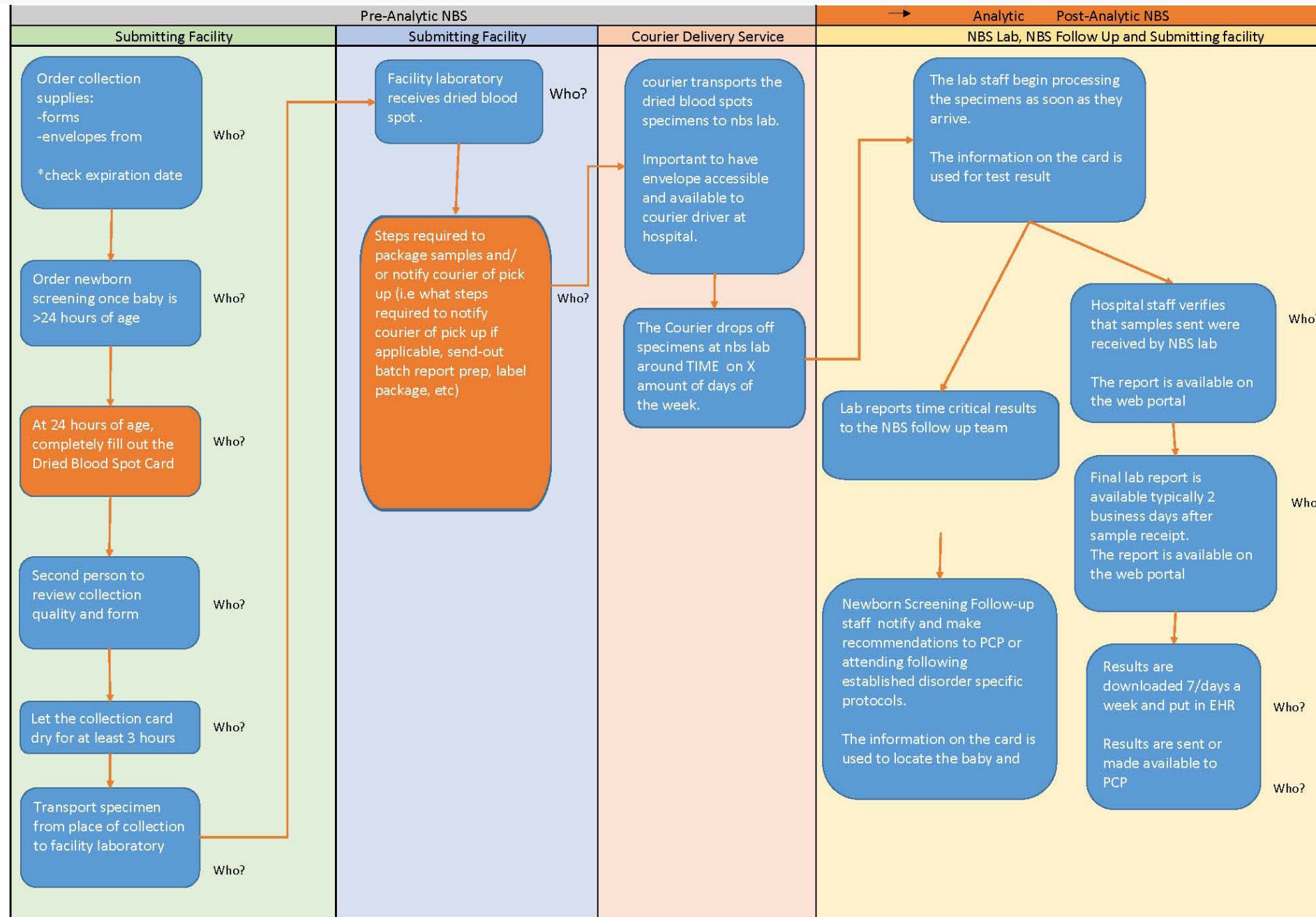
- Policy Guide
- Overview of NBS Weekend and Holiday Operations
- Steps and Considerations for Newborn Screening Staffing
- Communication Materials

Newborn Screening Program Features

- [New York's Poster on Reducing Late Collections](#)
- [ND NBS Online Educational Modules](#)
- [Virginia Online Newborn Screening Education for Healthcare Professionals](#)
- [CO Educational activities](#)

Quality Improvement Tool Templates

- [Root Cause Analysis \(RCA\) Tools](#)
- [What is Process Analysis? Definition, Tools, Maps & Flow Charts](#)



QI5d.i

Time from birth to reporting **out-of-range** results to an appropriate medical provider, for **time-critical disorders**

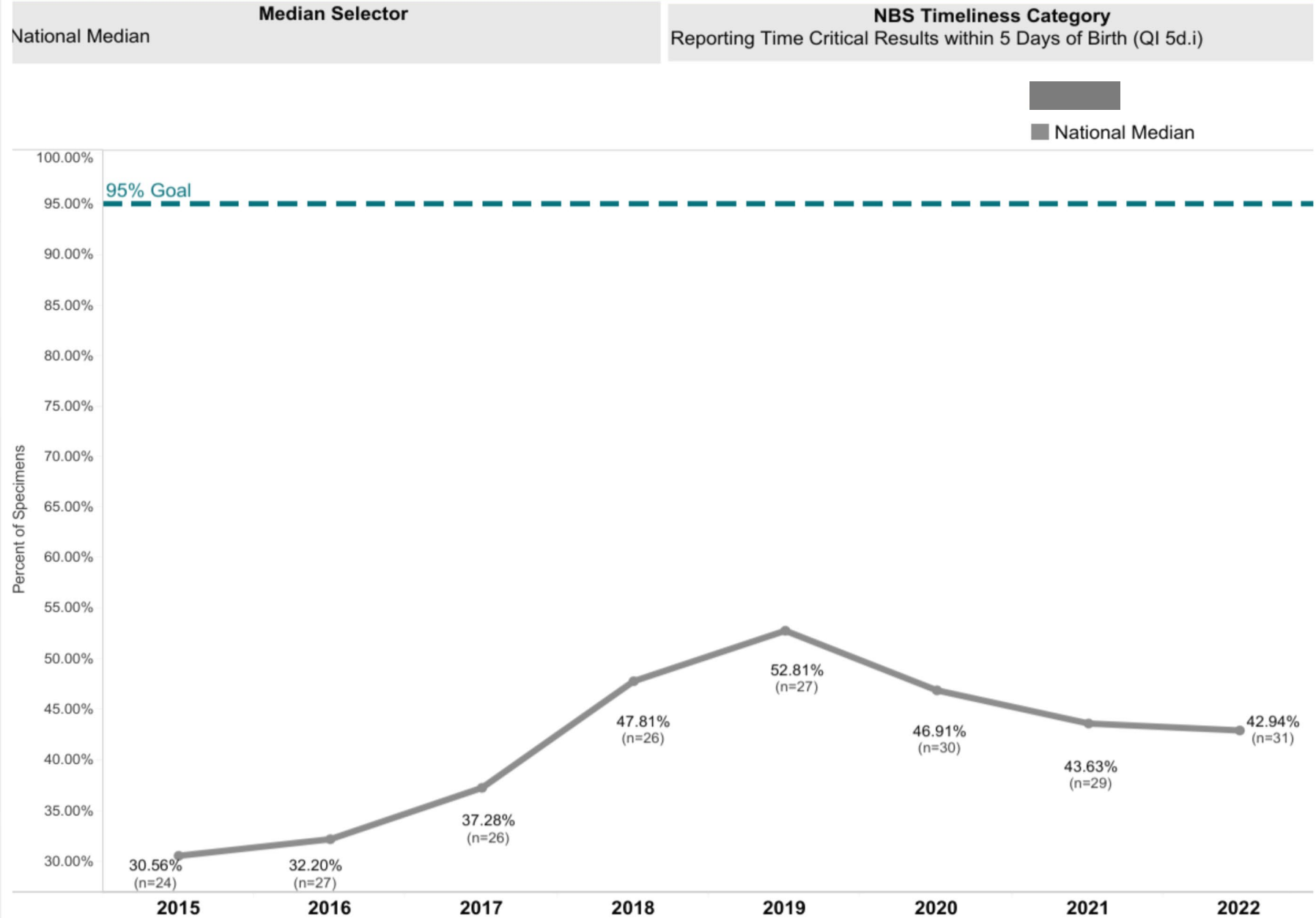
Goal: within 5 days of life

QI5d.i

Time from birth to reporting **out-of-range** results to an appropriate medical provider, for **time-critical disorders**

Goal: within 5 days of life

Reporting Time Critical Results within 5 Days of Birth (QI 5d.i)



QI5d.i

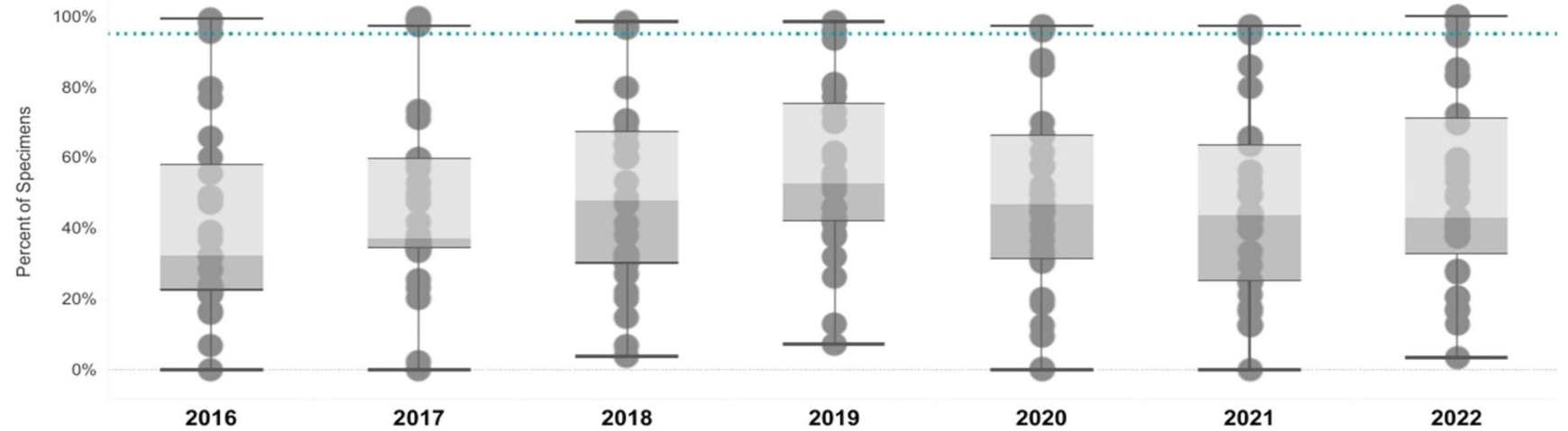
Time from birth to reporting **out-of-range** results to an appropriate medical provider, for **time-critical disorders**

Goal: within 5 days of life

Reporting Time Critical Results within 5 Days of Birth (QI 5d.i)

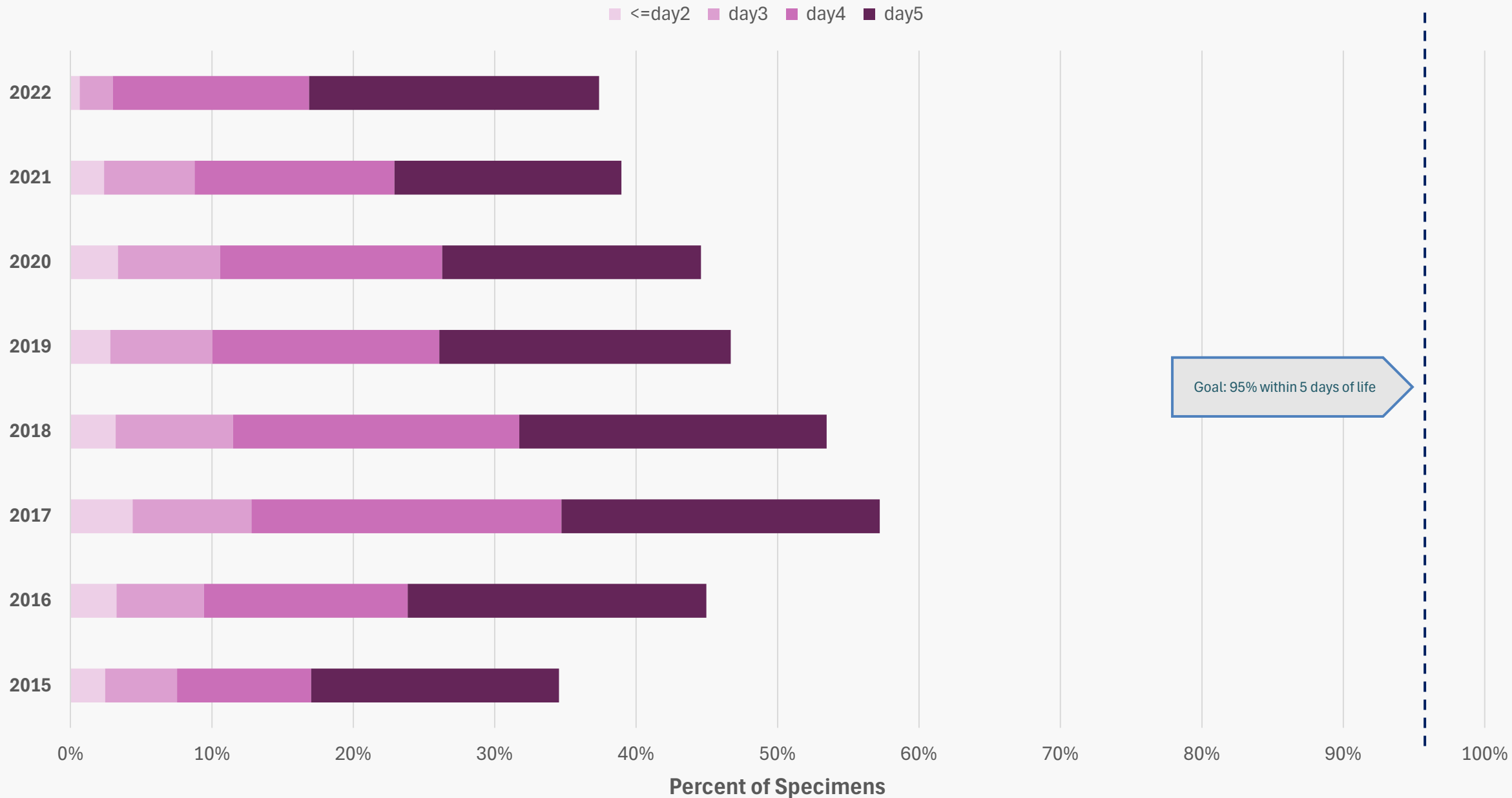
NBS Timeliness Measure

Reporting Time Critical Results within 5 Days of Birth (QI 5d.i)



Year	Number of Programs Reporting	No. of Programs Above 95%	% of Programs Above 95%	Minimum	Maximum	National Median	Standard Deviation
2022	31	3	9.7%	3.3%	100.0%	42.9%	27.7%
2021	29	3	10.3%	0.0%	97.4%	43.6%	26.5%
2020	30	3	10.0%	0.0%	97.3%	46.9%	27.3%
2019	27	3	11.1%	7.2%	98.4%	52.8%	24.7%
2018	26	3	11.5%	3.9%	98.6%	47.8%	27.1%
2017	26	3	11.5%	0.0%	99.5%	37.3%	26.0%
2016	27	3	11.1%	0.0%	99.2%	32.2%	27.9%

Time from birth to reporting out-of-range results to an appropriate medical provider, for time-critical disorders (QI 5d.i)



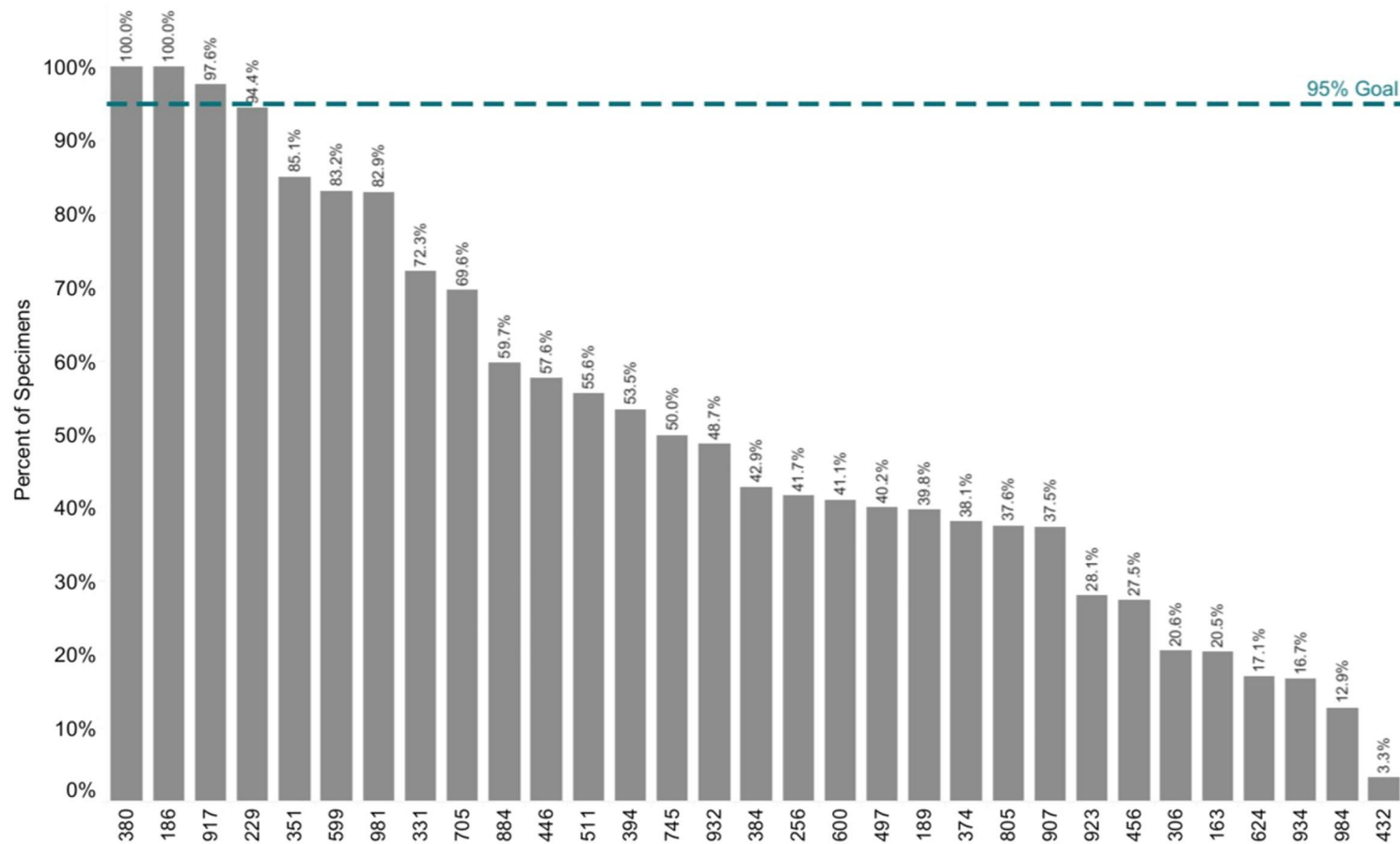
QI5d.i

Time from birth to reporting **out-of-range** results to an appropriate medical provider, for **time-critical disorders**

Goal: within 5 days of life

Reporting Time Critical Results within 5 Days of Birth (QI 5d.i)

NBS Timeliness Activity	Year
Reporting Time Critical Results within 5 Days of Birth (QI 5d.i)	2022



Program ID

Last Updated: 5/21/2024 3:31:27 PM



Rank

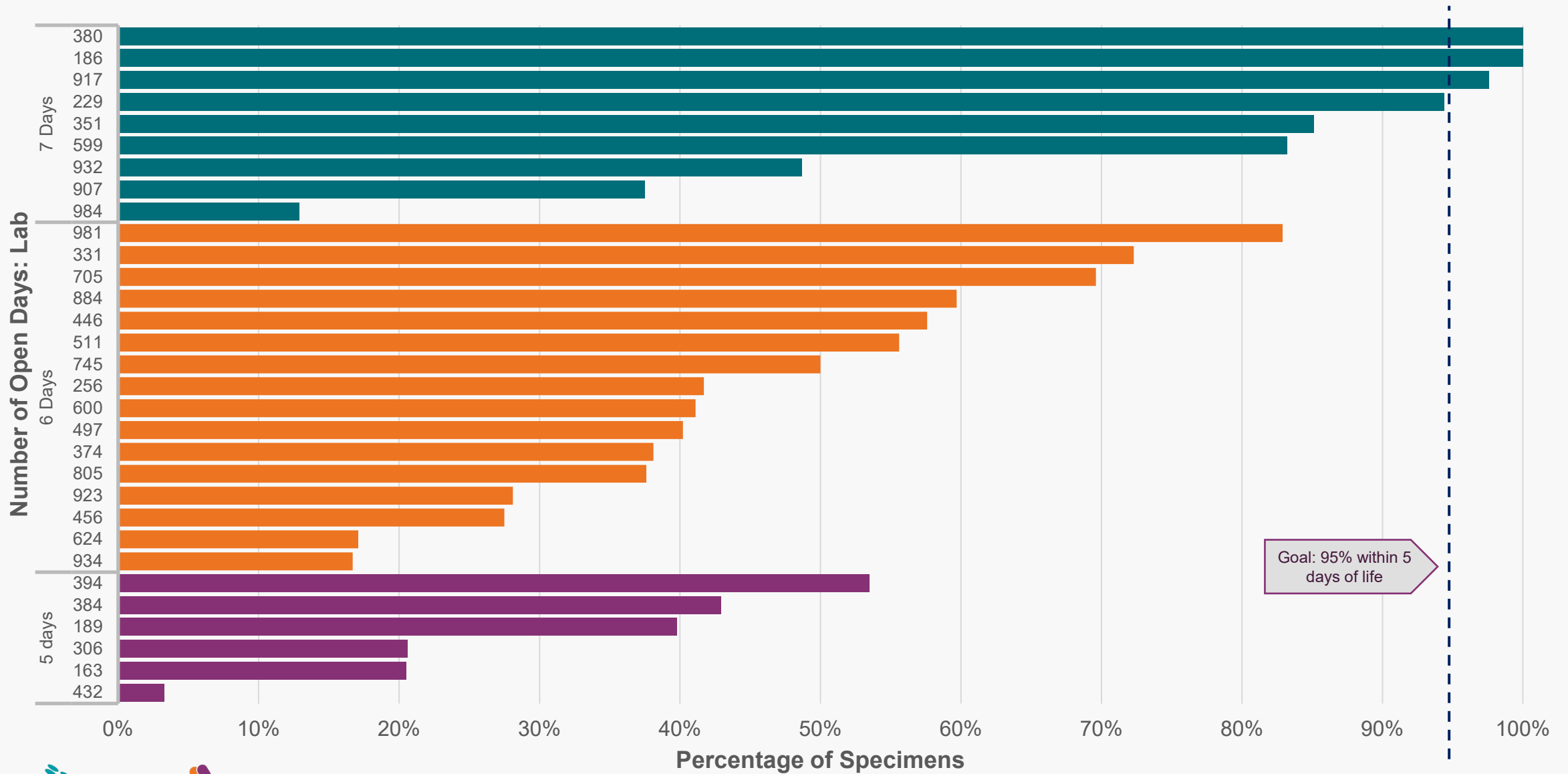


A Program of the Association



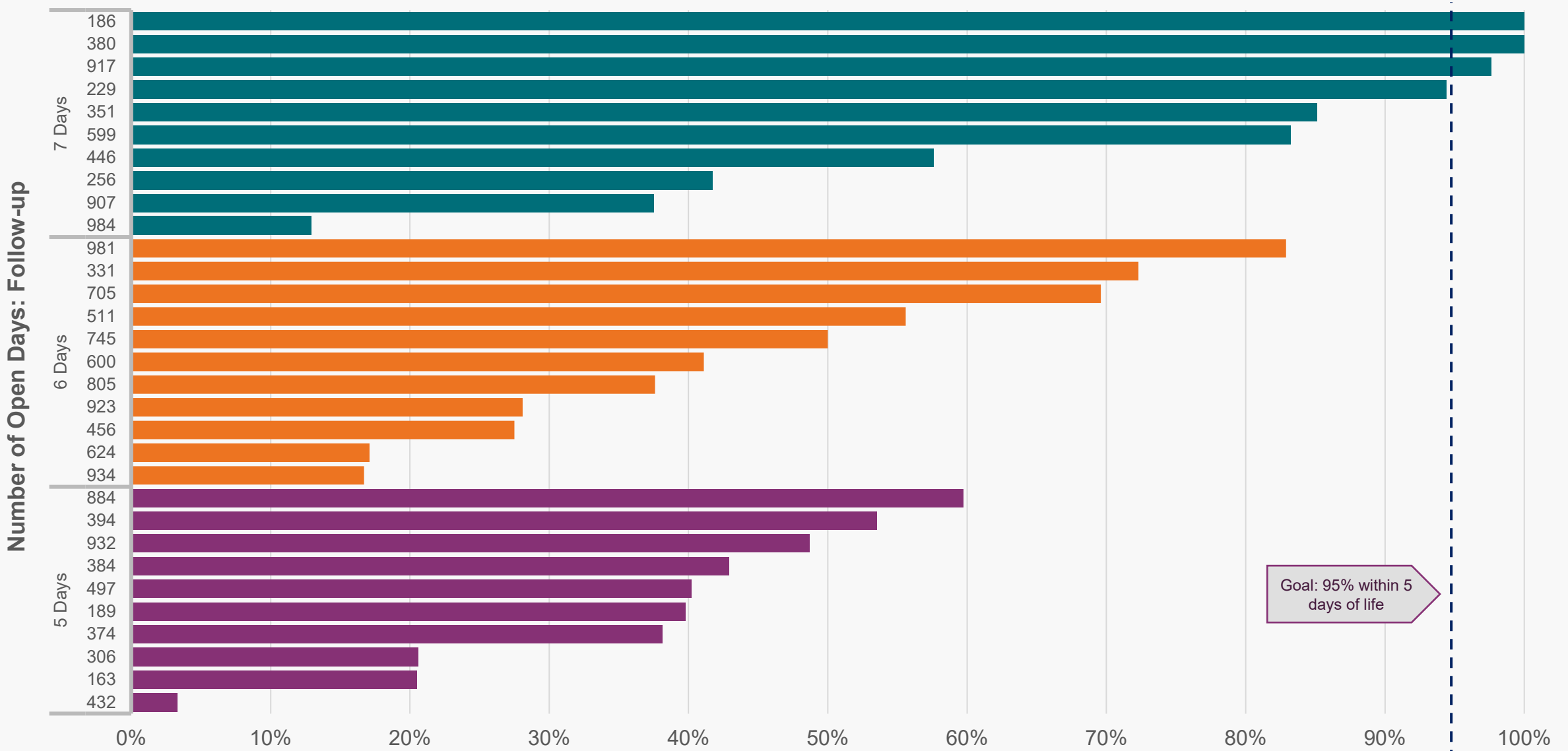
More Information

Impact of Laboratory Operational Days on the Percent of Specimens Reported for Out-of-range Results within Five (5) Days for Time-Critical Disorders



Goal: 95% within 5 days of life

Impact of Follow-up Operational Days on the Percent of Specimens Reported for Out-of-range Results within Five (5) Days for Time-Critical Disorders



Goal: 95% within 5 days of life



QI5d.iii

Time from birth to reporting **all** results to an appropriate medical provider, for **first** dried blood spot specimens

Goal: within 7 days of life

Q15d.iii

Time from birth to reporting **all** results to an appropriate medical provider, for **first** dried blood spot specimens

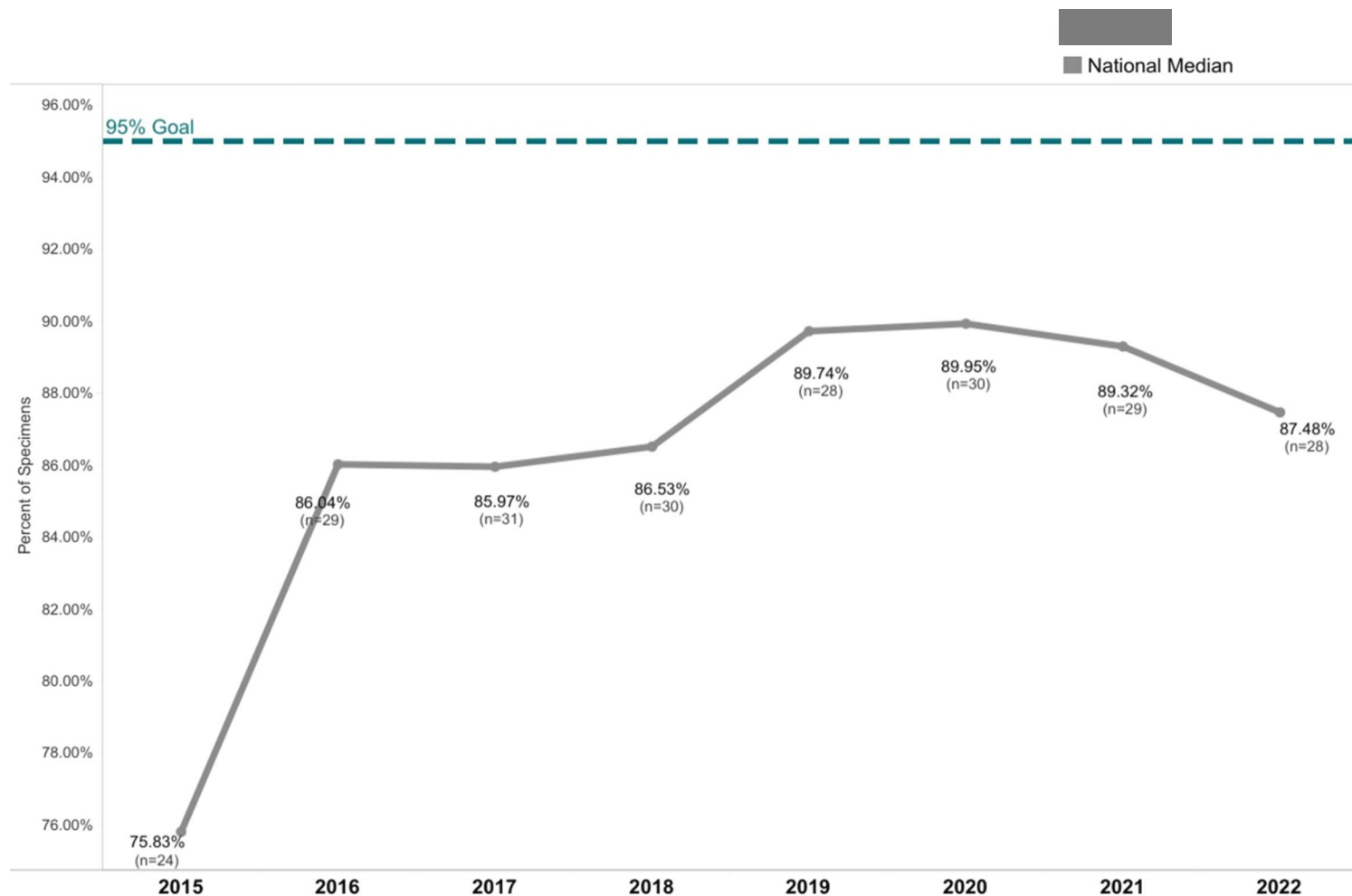
Goal: within 7 days of life

Reporting All NBS Results within 7 Days of Birth (QI 5d.iii)

National Median

Median Selector

NBS Timeliness Category
Reporting All NBS Results within 7 Days of Birth (QI 5d.iii)



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Q15d.iii

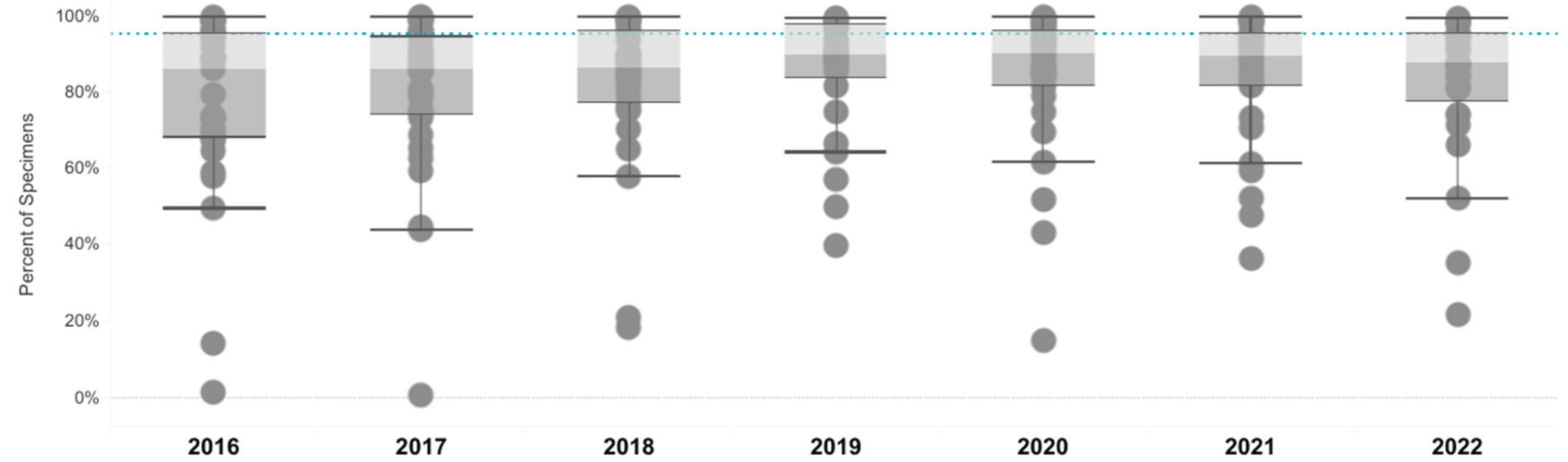
Time from birth to reporting **all** results to an appropriate medical provider, for **first** dried blood spot specimens

Goal: within 7 days of life

Reporting All NBS Results within 7 Days of Birth (Q1 5d.iii)

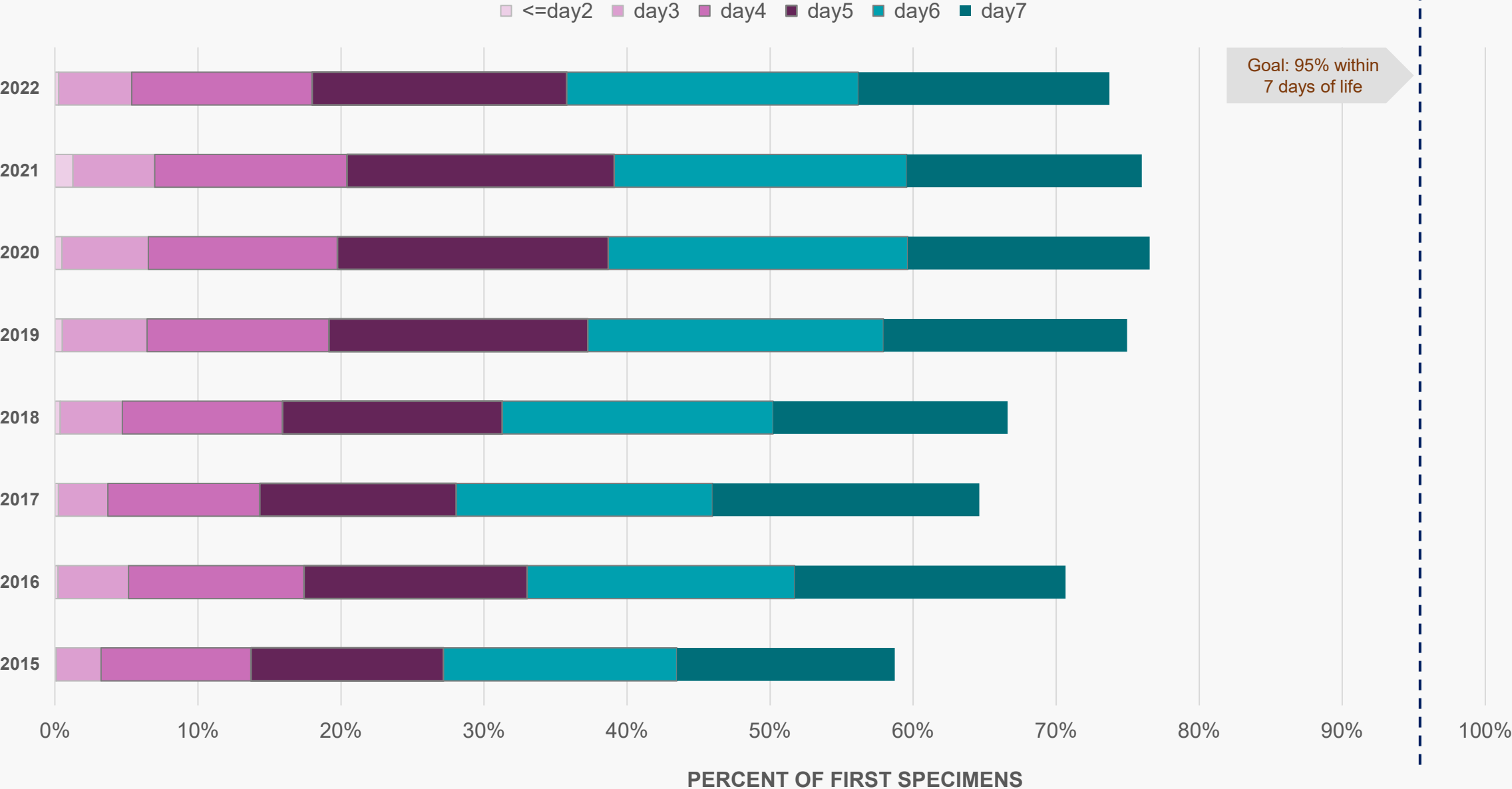
NBS Timeliness Measure

Reporting All NBS Results within 7 Days of Birth (Q1 5d.iii)



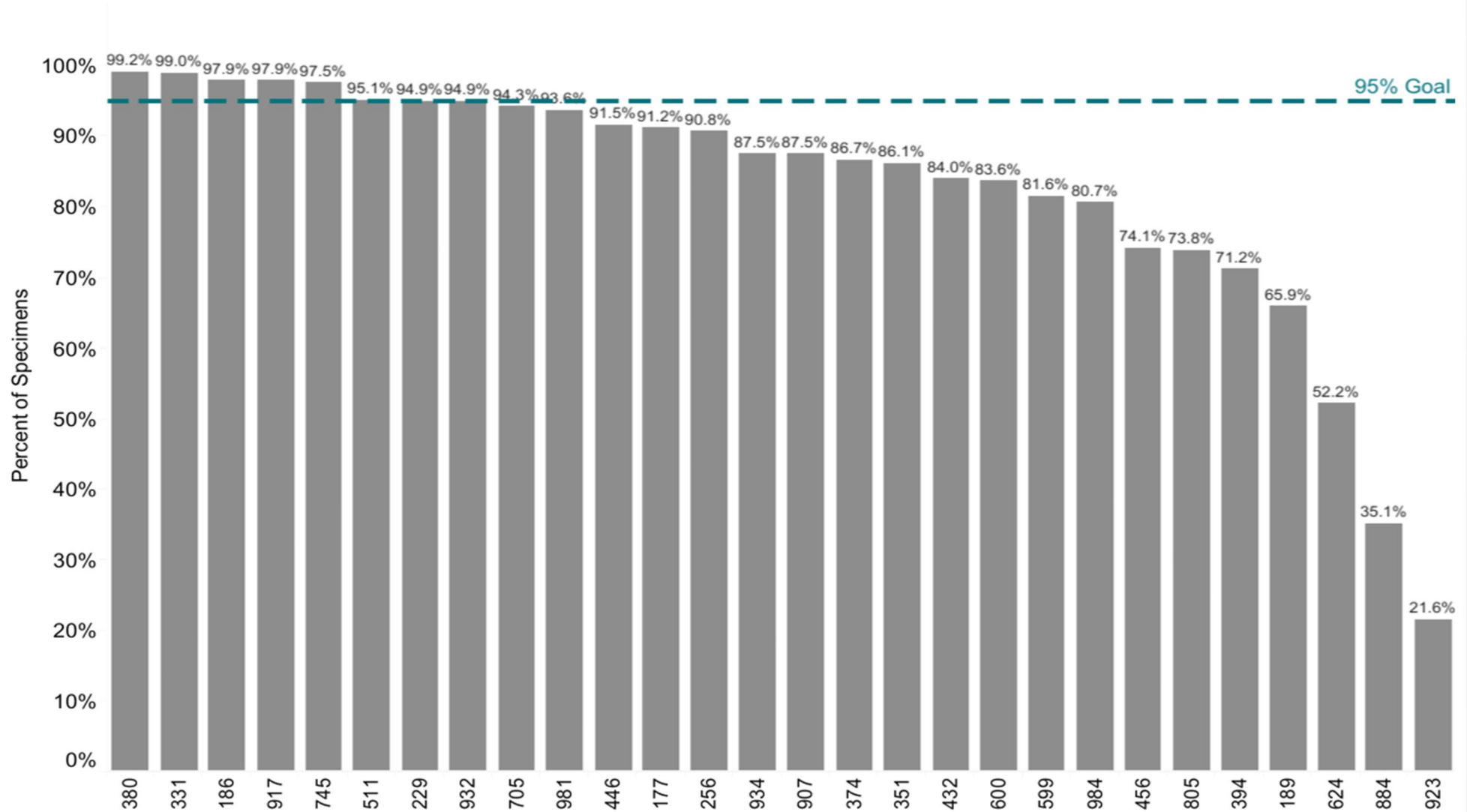
Year	Number of Programs Reporting	No. of Programs Above 95%	% of Programs Above 95%	Minimum	Maximum	National Median	Standard Deviation
2022	28	6	21.4%	21.6%	99.2%	87.5%	19.0%
2021	29	7	24.1%	36.5%	99.5%	89.3%	17.0%
2020	30	8	26.7%	14.8%	99.4%	89.9%	19.0%
2019	28	9	32.1%	39.7%	99.3%	89.7%	15.9%
2018	30	8	26.7%	18.1%	99.3%	86.5%	19.9%
2017	31	8	25.8%	0.6%	99.6%	86.0%	21.1%
2016	29	8	27.6%	1.4%	99.4%	86.0%	24.0%

Time from birth to reporting all results to an appropriate medical provider, for first dried blood spot specimens



Reporting All NBS Results within 7 Days of Birth (QI 5d.iii)

NBS Timeliness Activity	Year
Reporting All NBS Results within 7 Days of Birth (QI 5d.iii)	2022

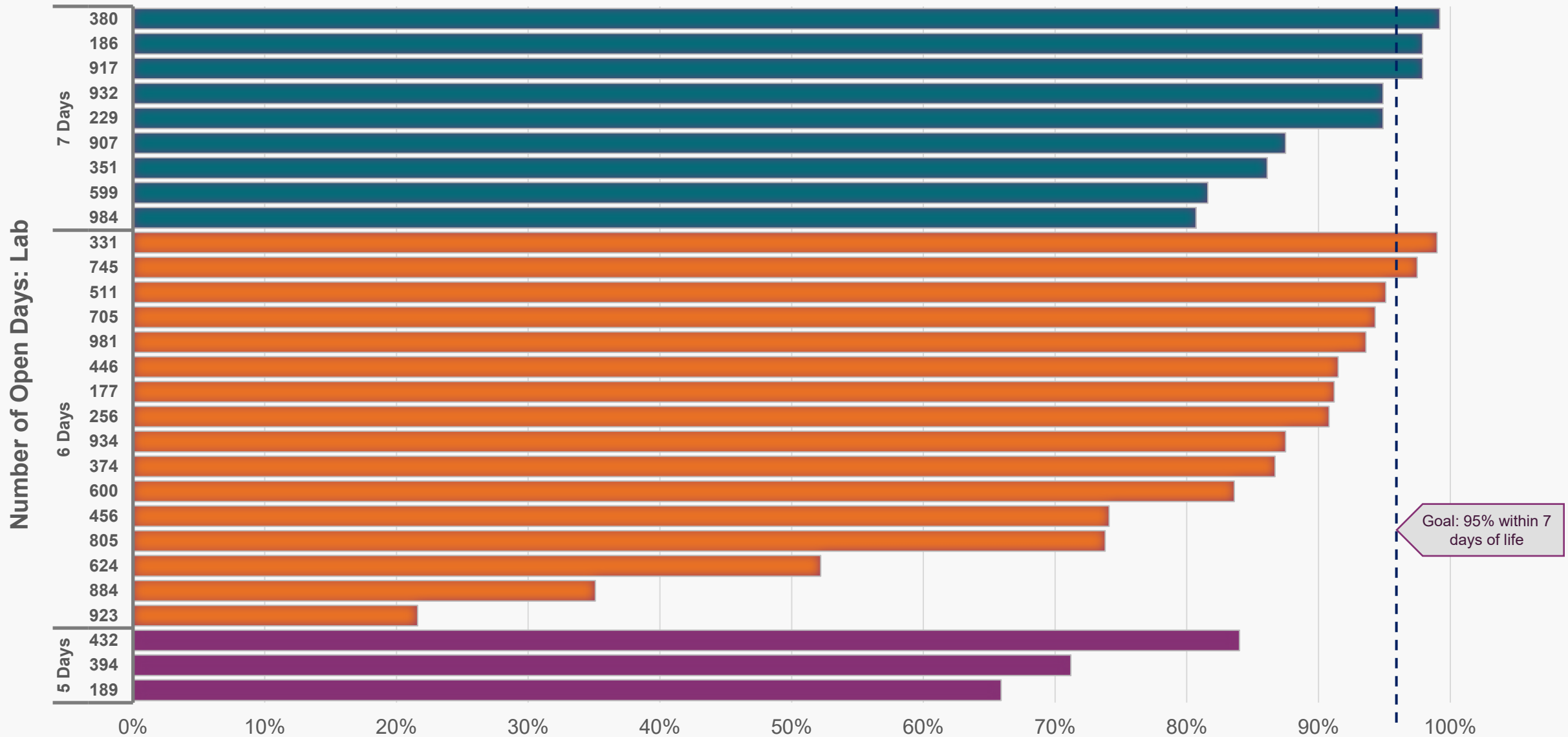


QI5d.iii

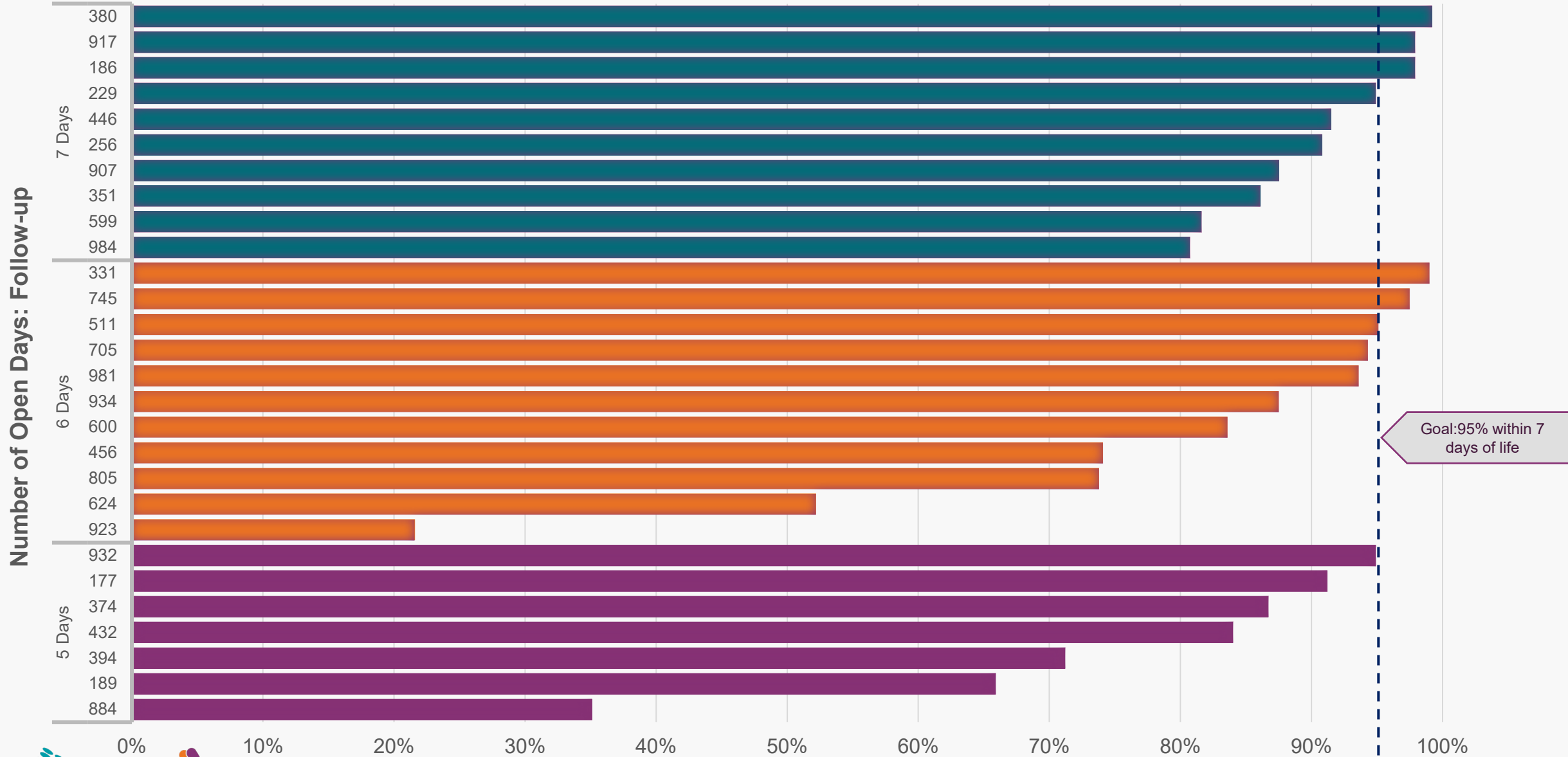
Time from birth to reporting **all** results to an appropriate medical provider, for **first** dried blood spot specimens

Goal: within 7 days of life

Impact of Laboratory Operational Days on the Percent of Specimens Reported for All Results within Seven (7) Days for First Specimens



Impact of Follow-up Operational Days on the Percent of Specimens Reported for All Results within Seven (7) Days for First Specimens



Quality Improvement Ideas

QI5d.i Goal: Communicate out-of-range results to an appropriate medical provider, for time-critical disorders within 5 days from birth.

QI5d.iii Goal: Communicate all results to an appropriate medical provider, for all disorders within 7 days from birth

To achieve this and reduce delays, initial specimens should be:

- collected no later than 48 hours of life and
- should be received at the laboratory within one or two days for testing.

[Newborn Screening Timeliness Goals | HRSA](#)

Quality Improvement Tools:

- Map workflow
- Gemba walk
- Cause and effect diagram
- Force field analysis
- Interrelationships Diagram

Considerations:

- Hours of operations
- Staffing levels
- Specimen transportation
- Receiving process
- Disorder specific workflows
- Policy or regulations
- Feedback to submitters

Resources

NewSTEPS Toolkit

Timeliness Toolkit for Expanding Newborn Screening Services

- Policy Guide
- Overview of NBS Weekend and Holiday Operations
- Steps and Considerations for Newborn Screening Staffing
- Communication Materials

Newborn Screening Program Features

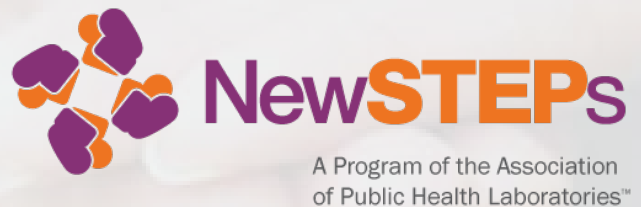
- [Using CQI to Improve Newborn Screening Timeliness](#)
- [California's IDEAL project using the Gemba Walk](#)
- [NewSTEPS Quality Improvement Projects Collaborative Showcase Series](#)
- [NBS Courier Fact Sheet from Oklahoma](#)

Quality Improvement Tool Templates

- [Root Cause Analysis \(RCA\) Tools](#)
- [What is Process Analysis? Definition, Tools, Maps & Flow Charts](#)
- [Force Field Analysis](#)
- [Interrelationship Digraph](#)

Thank you!

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Discussion Groups

Discussion Questions

Breakout Rooms

1. What promising practices or lessons learned can you share to assist other programs to enhance timeliness?
2. Share how your program uses performance metrics and data analysis to identify trends, root causes of delays, and opportunities for improvement in your newborn screening program?
3. What specific challenges or bottlenecks contribute to delays in the newborn screening processes, and how can they be addressed effectively?
4. What were your takeaways from the data and information shared during this session?
5. Create your own discussion prompt (choose your own adventure 😊)

Navigating New Disorder Implementation

Navigating New Disorder Implementation

Propel Awardee Forum

June 12, 2024

Patrick V. Hopkins; Project Specialist and NBS Consultant

Newer Disorders on the Federal RUSP

Disorder/ Disorder Category	Time-Critical	When Added to RUSP
Severe Combined Immunodeficiency	N	2011
Pompe disease	Y/N	2015
Mucopolysaccharidosis type I	N	2016
X-linked Adrenoleukodystrophy	N	2016
Spinal Muscular Atrophy	Y/N	2018
Mucopolysaccharidosis type II	N	2022
Guanidinoacetate Methyltransferase	N	2023
<i>Krabbe (pending approval by Secretary of HHS)</i>	Y?	<i>Passed by ACHDNC 2024</i>

A Few Challenges to Implementation

- State and Program Internal Approvals
- Prioritizing
- Funding
- Deciding on the testing method, including 2nd tiers. LDT vs: FDA cleared?
- Procurement process
- Staffing
- Limited experience with the disorder around the country
- Laboratory Space
- LIMS changes
- Validation of new method and/or the marker analyte.
- Cutoffs and decisions schemes
- Follow-up preparations and follow-up materials

You Are Not Alone

There are resources and technical assistance that you can utilize:

1. The New Disorder Subcommittee Resources
2. APHL subject matter experts
3. Colleagues in other states

New Disorders Subcommittee Members

George Dizikes | Co-chair

Fizza Gulamali-Majid

Michelle Mills

Miriam Schachter

Suzanne Cordovado | CDC

Kshea Hale | NewSTEPs Staff Liaison

Amy Gaviglio | Co-chair

Patrick Hopkins

Paul Orchard

Erica Wright

Kim Morrison | HRSA

Kenneth Coursey

Tate Kapell

Joseph Orsini

Pawan Vohra

Christine Dorley

Rose Maase

Andreas Rohrwasser

Melissa Parisi | NIH

New Disorders Resources

Publications

NewSTEPS
NEW DISORDER CHECKLIST

Phase 1

- Obtain approval
- Determine testing methodology and tiered testing strategy
- Identify lab and follow-up staffing needs
- Develop budget
- Procure vendor contracts for equipment

Phase 2

- Obtain equipment
- Perform validation(s)
- Identify and meet with sub-specialists to discuss notification strategy and follow-up algorithms
- Gain understanding of possible incidental findings
- Consider sub-populations that may affect results

Phase 3


- Integrate testing into current workflow
- Notify submitters of NBS report changes
- Identify website/brochure changes needed
- Develop fact sheets and follow-up letters
- Develop follow-up data needs (short and long)

Phase 4


- Build and test cut-offs/logic into LIMS (Lab and Follow-Up)
- Press release
- Notify health care practitioners of new disorder with expectations

Ongoing Internal Communication (biweekly or weekly)

Go Live / Post Go Live
www.newsteps.org



Spinal Muscular Atrophy
New Disorder Resources and Tools




NewSTEPS
A Program of the Association of Public Health Laboratories™

JUNE 2021

SMA Toolkit

Mucopolysaccharidosis, Type II
New Disorder Resources and Tools




NewSTEPS
A Program of the Association of Public Health Laboratories™

AUGUST 2022

MPS II Toolkit

Guanidinoacetate Methyltransferase Deficiency
New Disorder Resources and Tools



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A Program of the Association of Public Health Laboratories™

AUGUST 2023

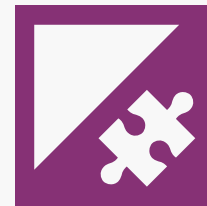
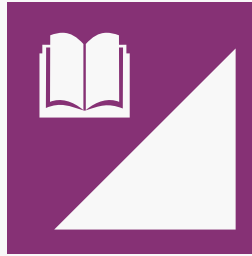
GAMT Toolkit

New Disorders Checklist

New Disorders Resources

Archived Webinars and Meetings

- **Adrenoleukodystrophy Open Office Webinar, December 2023**
- **Exploring the Use of Biochemical & Sequencing Methods in Newborn Screening: Future of Biochemical & Sequencing Testing, September 2023**
- **Exploring the Use of Biochemical & Sequencing Methods in Newborn Screening: State Newborn Screening Program Experiences, September 2023**
- **Exploring the use of Biochemical and Sequencing Methods in Newborn Screening: Overview of Current Sequencing Projects, August 2023**
- **Newborn Screening Considerations for Additional Lysosomal Disorders and GAMT, January 2021**
- **Considerations for Infectious Disease Screening or Monitoring in the Newborn Screening Program, February 2021**
- **On the Horizon: A Review of Up and Coming Disorders and the Future of the RUSP, February 2021**
- **New Disorders & Short-Term Follow-up Virtual Meeting, August 2021**
- **New Disorders Workgroup: Ornithine transcarbamylase (OTC) deficiency, December 2021**
- **Newborn Screening and Recommended Uniform Screening Panel (RUSP) Alignment, July 2022**



Please visit NewSTEPS.org for additional webinars.

Upcoming Webinars and Publications

Webinars:

- The Subcommittee will host a webinar in July 2024 that introduces multiple upcoming conditions that are being considered for addition to the RUSP or could be considered. Disorders to be featured include:
 - Pyruvate dehydrogenase complex deficiency (PDCD)
 - Cerebrotendinous xanthomatosis (CTX)
 - Pyridoxine Dependent Epilepsy
 - Congenital cytomegalovirus (cCMV)
 - Metachromatic leukodystrophy (MLD)
 - Other Mucopolysaccharidosis (MPS) conditions (MPS types IV, VI, and VII)
 - Biliary Atresia
- The Subcommittee will host a webinar to address the status of artificial intelligence and deep learning in predicting the impact of sequence changes (VOUS) on enzyme function and disease severity. Date TBD.
 - Potential collaboration with the APHL Molecular Subcommittee

Publications:

- Krabbe Resource Toolkit, Summer 2024
- Duchenne muscular dystrophy (DMD) Resource Toolkit, TBD

Assistance from APHL

- APHL can connect you with subject matter experts to provide advice and technical assistance.
- APHL can connect you with NewSTEPs data and reports that are available that may assist you in making informed decisions.
- NewSTEPs site reviews are available for non-regulatory evaluation and guidance from a team of experts. Their recommendations could help you overcome administrative roadblocks.
- The NewSTEPs listserv is available for communication with the NBS community regarding any questions that you may have about new disorders.
- The APHL NBS Symposium highlights new disorders (and several legacy disorders) sharing experiences and successes from other state programs.

Assistance from Other NBS Colleagues

Reach out to states that have been screening for the disorder to share:

1. Experiences, do's and don'ts and answers to frequently asked questions
2. SOP's and validation write-ups
3. Validation samples for confirmed true positives and false positives
4. Cutoffs and decision schemes
5. Example lab reports and results comments
6. Follow-up educational materials they have produced
7. Ongoing networking and moral support

Laboratory Implementation Plan Considerations

- Form a Task Force for implementing the new disorder
- Choose a screening method
- Obtain IRB approval or exemption for pilot study?
- Initiate procurement steps and establish contract with vendor
- Installation, training and method familiarization
- Validations, Cutoffs and Decision Schemes
- Communication with follow-up on positive screens
- Pre-pilot phase (data collection on de-identified samples)
- Pilot Phase (statewide testing with referrals)
- Live Testing with reporting on all NBS lab reports

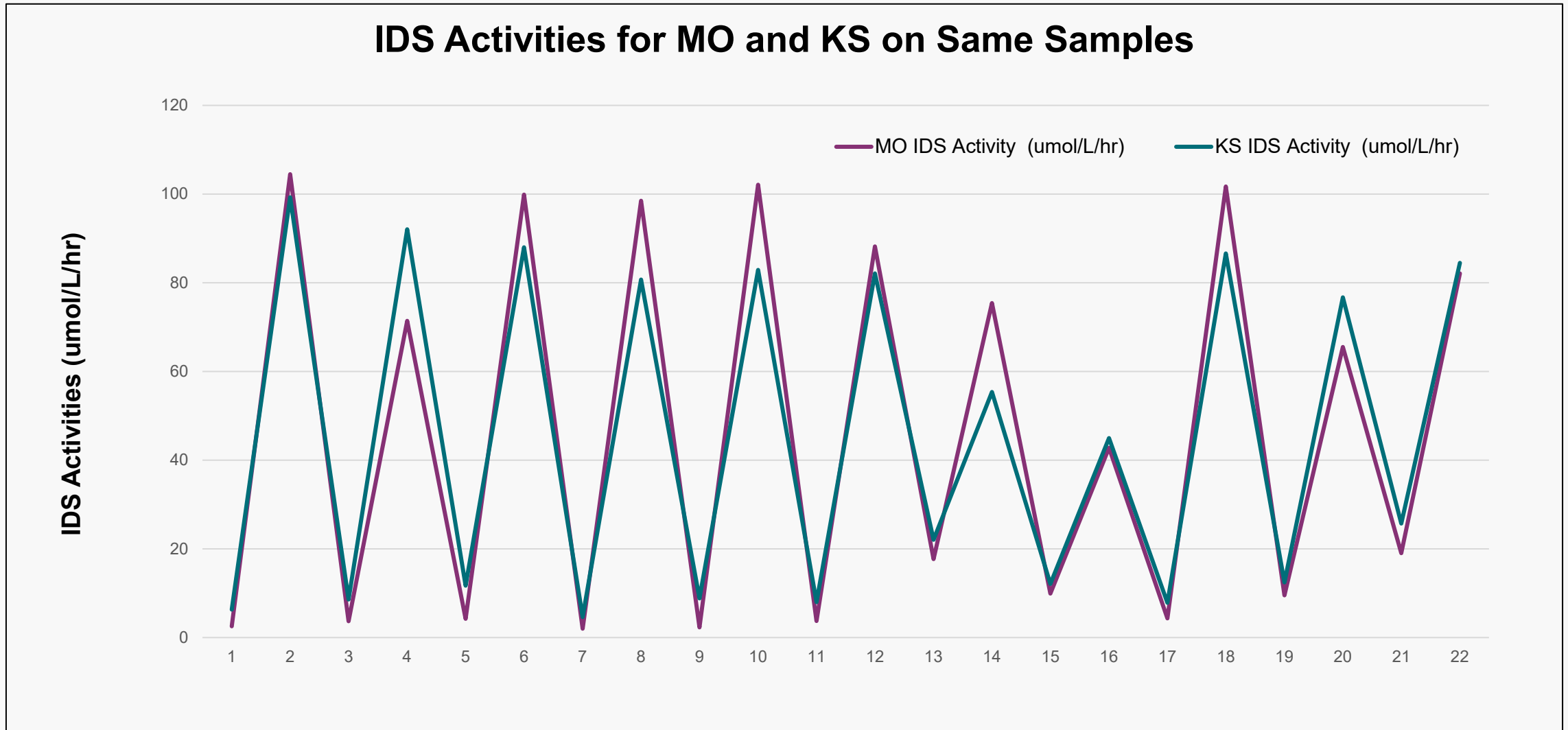
Follow-up Implementation Plan Considerations

- Form a Task Force for implementing the new disorder.
- Secure follow-up referral and confirmatory protocols.
- Communication plan with the NBS lab on out-of-range results and final confirmatory results on all referred screens.
- Education on the new disorder for birthing hospitals and PCP's. Convey any time-critical action information or reminders that may be needed.
- Provide 2nd tier testing education if indicated.
- Pilot Phase (announce that screening will be done on all newborns and that they may be notified about a few presumptive positive screens)
- Live Testing with reporting on all NBS lab reports (make an announcement and that normal and/or out-of-range results will be provided on the NBS reports with actions requires if necessary).

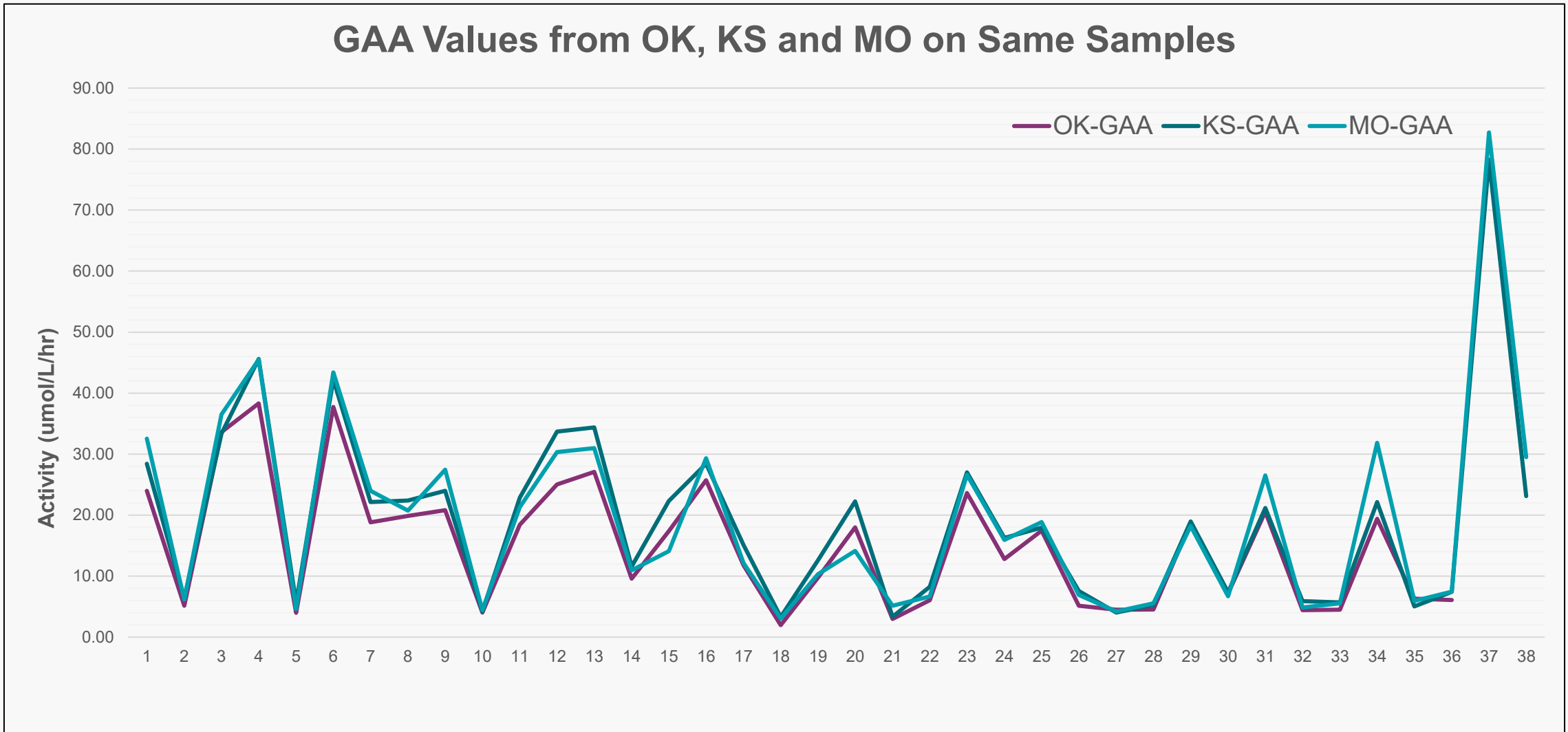
Validation of New Methods

- Accuracy/Sensitivity – Using known positives, quality control and proficiency test samples.
- Precision/Specificity – Within run, between runs, between different reagent lots.
- Linearity/Limit of detection – Consistency from high to low levels of the detection range.
- Instrument matching – to maintain same cutoffs for all instruments.
- Carryover
- Testing interferences – Health status, age of baby, transfusions, etc.

Confirmed Abnormal and Normal Samples



Confirmed Abnormal and Normal Samples



Materials That May Be Shared by Other States

- Laboratory SOP's and Validation Write-ups
- Cutoffs and Decision Schemes
- Example NBS lab reports
- Example Follow-up guidelines
- Example Letters to PCPs
- Example Confirmatory Data Reports
- Example Fact Sheets
- Example Flow Charts for Screening and Follow-up
- Data showing Incidence, PPVs, FPRs and other performance metrics
- Interesting cases and Tough Calls

The Babies Thank You for All You Do!



Discussion Groups

Discussion Questions

Breakout Rooms

1. How can we leverage the experiences and insights from other programs to inform our approach to implementing new disorders in our newborn screening program?
2. How can performance metrics and data analysis be utilized to monitor the effectiveness of new disorder implementation and identify areas for improvement in our newborn screening program?
3. Discuss ways programs could share materials more widely (e.g., validation plans, SOPs, example reports, algorithms, data showing incidence and assay performance, etc.)
4. What were your takeaways from this session? What resource(s) or support do you plan to use for future new disorder implementation?
5. Create your own discussion prompt (choose your own adventure 😊)



NEWSTEPS NATIONAL MEETING

ON
POST-ANALYTICAL NEWBORN SCREENING

YOU'RE INVITED
REGISTRATION OPENS SOON



February 11 - 12, 2025
Rockville, MD

FOR MORE INFORMATION

Join the NewSTEPs National Meeting Teams Site [here](#)

Contact Noah Sinangil at Noah.Sinangil@aphl.org

This meeting is supported by the Health Resources and Services Administration (HRSA) under Cooperative Agreement #U22MC24078 for \$2,725,000. This content is those of the authors and should not be construed as the official position or policy of, nor should any endorsements be inferred by HRSA, HHS or the US Government.



Thank
you

[Please complete the event evaluation](#)