

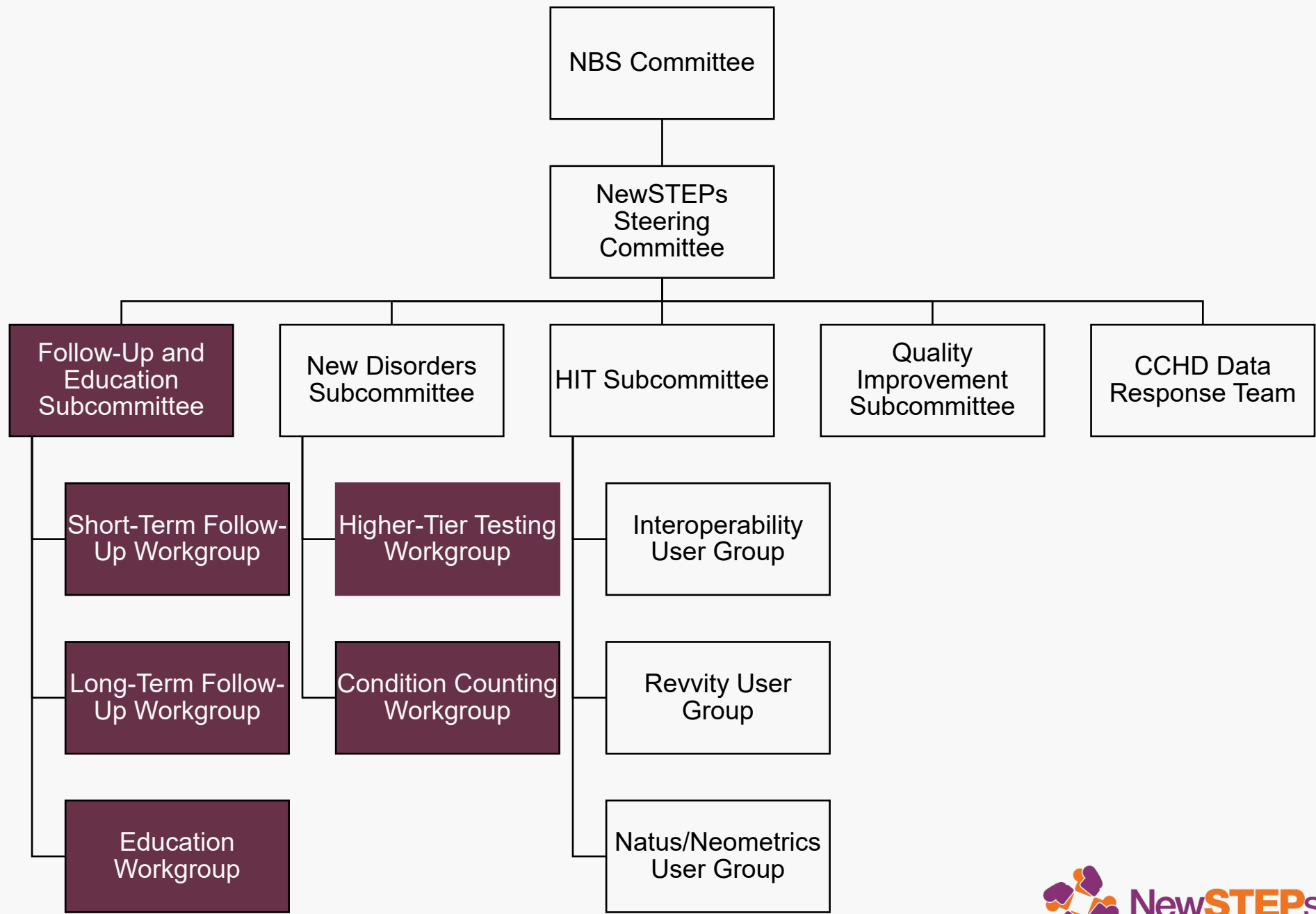


NewSTEPs Ad Hoc Newborn Screening Topic Groups: UPDATE

*Newborn Screening Technical assistance and Evaluation Program (NewSTEPs), a
program of the Association of Public Health Laboratories*

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Advancing Health Equity in Newborn Screening: Community of Practice

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BACKGROUND

While NBS has allowed for nearly universal access to testing (at least within a specific state or jurisdiction), it is evident that disparities exist throughout the newborn screening system.

CHARGE

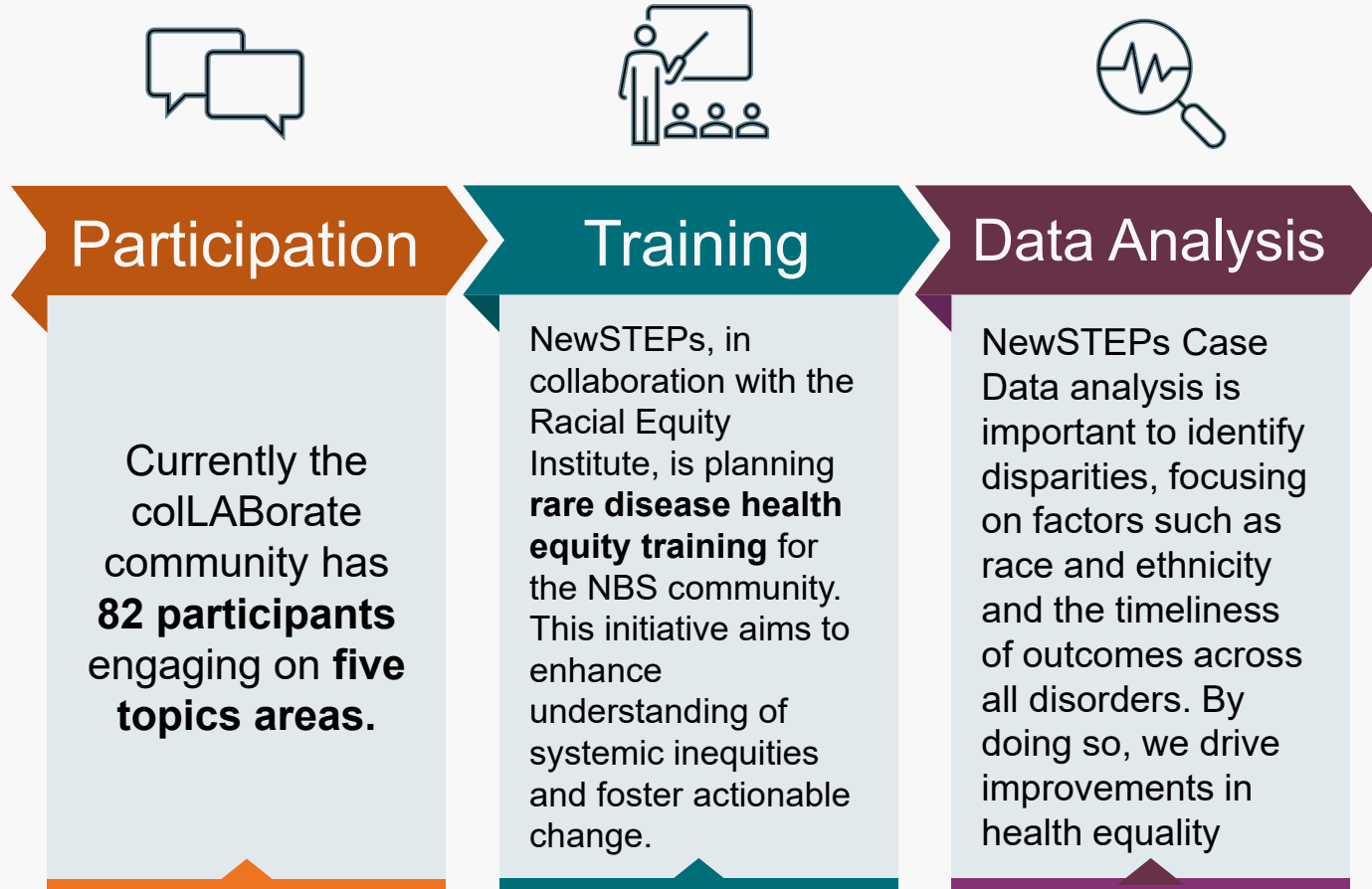
NewSTEPS is pursuing a multipronged approach to establish a community of practice that allows NBS partners to discuss issues of health disparities and proposed solutions within the NBS system. The Community of Practice includes a “collABorate community” for real-time discussions, webinars, and trainings.

DESIRED OUTCOMES

Fostering discussions and projects addressing issues of health disparities will help all newborn screening programs work towards their mission of improving health outcomes for all newborns and families affected with newborn screening conditions.

NewSTEPS will provide summary reports to HRSA and presentations to the ACHDNC detailing this project on an as needed/as requested basis.

Advancing Health Equity in Newborn Screening: Community of Practice



Committee Discussion



Follow-Up and Education Subcommittee: Workgroups

Follow-Up and Education Subcommittee

Short Term Follow-Up Workgroup

- Strengthen the NBS system by providing input, guidance, and technical assistance on follow-up in newborn screening.
- Offer a forum for communication in which follow-up staff from regional and state newborn screening follow-up programs can network and collaborate on quality improvement efforts.
- Identifying needs and offer NBS programs technical assistance related to short-term follow-up.

Long Term Follow-Up Workgroup

- Develop a LTFU landscape survey.
- Define LTFU and the essential elements of a LTFU program.
- Develop a LTFU fact sheet for programs to demonstrate the value of LTFU to their leadership.
- Develop LTFU Quality Indicators.

Education Workgroup

- Develop state and family needs assessments.
- Engage families and individuals with lived experience in NBS.
- Evaluate ongoing needs of NBS community and perform outreach to providers serving medically underserved and historically excluded communities.



Participants

Condition Counting	Higher Tier	Follow Up & Education	Short Term Follow Up	Long Term Follow Up	Education
Amy Gaviglio Bradford Therrell Dianne Webster Erica Wright George Dizikes Inderneel Sahai Jeremy Penn Joseph Orsini Kelsey Medrano Mei Baker Michele Caggana Rachel Lee Scott Shone Shawn McCandless Stanton Berberich Sue Berry Susan Tanksley Tory Kaye	Amy Calhoun Amy Gaviglio Erica Wright George Dizikes Jennifer Baysinger Jennifer Lewis Joseph Orsini Loraine Swanson Mei Baker Patrick Hopkins Rachel Lee Shawn Moloney Taraka Donti Tricia Hall	Allison Forkner Angela Wittenauer Carol Johnson Fran Altmaier Jennifer Smith Kathy Inkhamfong Kim Morrison Marianna Raia Michael Cellucci Olivia Giglio Sara Etienne Sondra Gilbert Twaina Williams	Alexis Chavez Amanda Peterson Amy Magyar Donna Holstein Emily Reeves Fran Altmaier Gloria Rochester Heidi Elsinger Jodi Philippon Kathryn McLaughlin Leslie McKenzie Lisa Song Michelle Bargren Nicole Guysi Niki Lowrie Shelby Heppe Vincent R. Bonagura Virginia Sack	Amanda Ingram Amy Burke Amy Gaviglio Angela Shepard Ashish Gupta Catharine Riley Celia I. Kaye Cristina Novoa Jamie Matteson Jennifer Baysinger Jennifer Hauser Jeremy Penn Kathy Chou Kristi Bentler Kristy Karasinski Lisa Song Maria M. Baranyai Marianna Raia Mollie Minear Olivia Giglio Ryan Colburn Susan Mays	Alaina Harris Alison Breitbarth Debra Ellis Desiree Naibauer Emily Hasser Emma Mertens Hilary Pamperin Jacob Ginter Jacqui Umstead Katherine Fullerton Lara Percenti Lisa Marie Shook-Chiles Lisa Song Liz Carter Marianna Raia Mary Lowe Meg Callahan Shari Arceneaux

Long-Term Follow-Up

- 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
- Establish national impact measures (Quality Indicators) for LTFU

Committee Discussion



Higher-Tier Testing Workgroup

Higher Tier Testing ad hoc Topic Group

BACKGROUND

Several new or recent disorders (e.g., GAMT, MPS I, MPS II, x-ALD, Pompe) and others recently nominated for addition to the RUSP (e.g., Krabbe, DMD) require second-tier testing, either to confirm an abnormal result on first-tier or to provide variant information needed to distinguish early from late-onset forms. For some core conditions (e.g., x-ALD), second-tier or higher-tier testing may lead to identification of a secondary condition (e.g., Zellweger spectrum disorder). Given the rarity of these newborn screening disorders, states with a lower birth rate may not have the resources to perform or implement second-tier testing within their state.

CHARGE

The NewSTEPs Higher-tier Testing topic group, under the guidance of the NewSTEPs New Disorders Subcommittee, will propose model practices to build a higher-tier testing cross-program model of collaboration.

The ad hoc topic group will identify and discuss the considerations to implementing tiered testing in-house or through outsourcing and whether those barriers differ between new and legacy RUSP disorders.

Higher Tier Testing ad hoc Topic Group

OBJECTIVES

- Examine the current use of higher-tier testing in NBS programs
- Describe and prioritize the systematic utility of tiered testing across newborn screening conditions
- Explore existing models of collaboration for higher-tier testing and examine the strengths and weaknesses of each model
- Identify the barriers to building a model of cross-program collaboration for higher-tier testing
- Define priorities, scope of work, and model practices for higher-tier cross-program collaboration
- Identify the newborn screening programs most at-need for support to implement higher-tier testing
- Share model practices and resources with NBS programs determined to be most at-need and further refine practices if needed

DESIRED OUTCOMES

Determine barriers to sustainable cross-state collaboration for higher tier testing, identify solutions to improve positive predictive value. Provide summary reports to HRSA and ACHDNC.

Current Activities To Date

Updates

- Developed working definitions on tiered testing
- Scheduled the Higher-Tier Testing Workgroup in-person meeting for July 23-24, 2024

Next Steps

- Finalize and disseminate a barrier survey to understand the barriers and challenges that NBS programs encounter when implementing higher-tier testing, whether in-house or outsourcing to other laboratories and identify the factors that prevent or delay them from initiating tiered screening
- Host a listening session webinar in the Summer of 2024 to gain a better understanding of the challenges of outsourcing testing to any lab in general, whether it is private or public, with regards to higher-tiered testing and how we can address those challenges

Committee Discussion



Condition Naming & Counting Workgroup

Condition Counting ad hoc Topic Group

BACKGROUND

Recent media articles have called national attention to the differences that exist across state NBS panels. Yet in many cases, these differences are owing to the lack of uniformity in how states *count* the conditions on their panel, and not true differences in the conditions screened (“state” refers to both states and territories). This lack of uniformity in counting leads to confusion and misunderstanding among parents, policy-makers, and the public about what is meant by the term ***screening***.

APHL convened a taskforce in summer 2021 to develop a framework for standardization of counting conditions. This Condition Counting Taskforce comprised 17 members representing NBS laboratories and follow-up programs, clinicians, parents, and international partners and met monthly over the course of two years to develop the framework, which also involved defining the term *screening*.

Condition Counting ad hoc Topic Group

CHARGE

HRSA charged APHL with convening a new Condition Counting group as an ad hoc topic group of the ACHDNC in September 2023. The group is comprised of subject matter experts (inclusive of NBS laboratory and follow-up staff, as well as federal partners, clinicians, ACHDNC members, parents, and international partners). This ad hoc topic group is considering the recommendations of the original taskforce (pre-ACHDNC involvement) and will formalize them as national suggestions. Once the group formalizes/finalizes the suggestions, and receives recommendations from the ACHDNC, we will work on a communications plan and messaging to explain the changes.

Because condition counting and condition naming are interrelated, this ad hoc topic group will also discuss the naming and nomenclature of certain conditions as listed on the RUSP and make suggestions to update condition names to those more widely recognized, accepted, and used by the medical community.

DESIRED OUTCOMES

Nationally standardized condition counting as well as revised condition names/nomenclature reflected on the RUSP.

Condition Counting ad hoc Topic Group

OBJECTIVES

- Meet monthly via teleconference between October 2023 and July 2024
- Develop talking points; for example:
 - Why is standardization of counting necessary?
 - How should we address pushback regarding the apparent decrease in the number of disorders on our state panel?
 - How will this help all NBS programs?
- Present final suggestions to the ACHDNC for endorsement as national recommendations for condition counting
 - Recommendations will be presented step-wise / as they are formulated, as well as presented in final format at the end.
- At least one national webinar to provide sample message pallets or toolkits to NBS programs and their system partners (e.g., parents, clinicians, advocacy groups, legislators)

Next Steps for Condition Counting ad hoc Topic Group



June 2024

Workgroup will meet in person



HRSA website

Present areas of discrepancy between HRSA website and 2006 ACMG paper (future ACHDNC meeting)



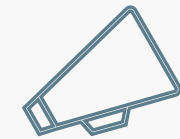
Align

Suggest alternate wording (future ACHDNC meeting)



Secondary Conditions

Address confusion around Secondary Conditions (future ACHDNC meeting)



Get the word out

Implement communication and dissemination plan

Committee Discussion



Other NBS Related Updates

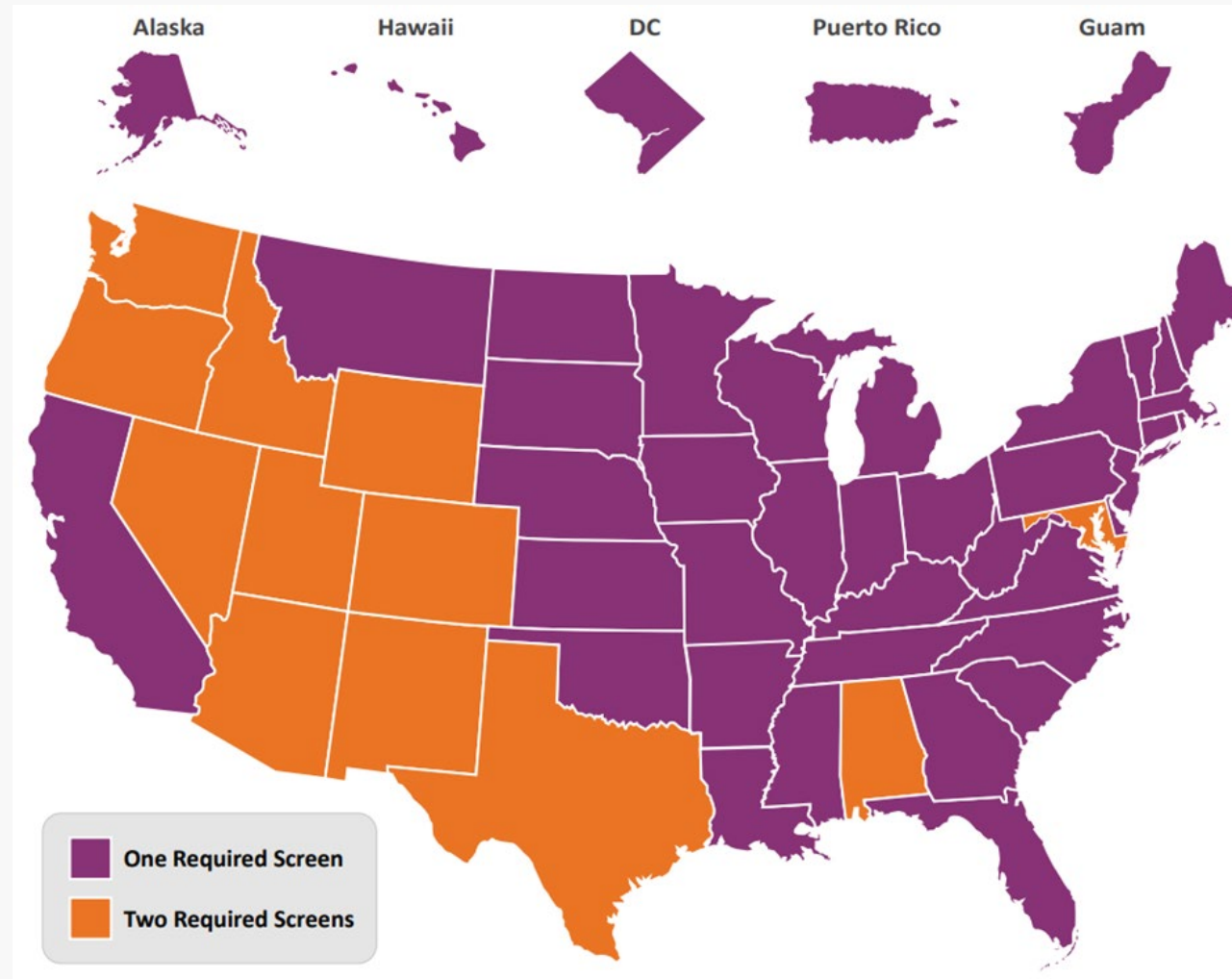
Brief State of the States

United States Newborn Screening Programs

- **53 NBS programs**

- **36** NBS laboratories
- Regional labs serve **13** states/territories
- Each (**53**) state/territory has its own follow-up program, with **17** programs reporting that they perform long term follow-up activities
- **7** programs outsource some or all of their screening to Revvity Omics (a private laboratory)

Number of Screens



Number of screens required by state/territory (N = 53)

Number of Core RUSP disorders screened by state/territory as of March 2024

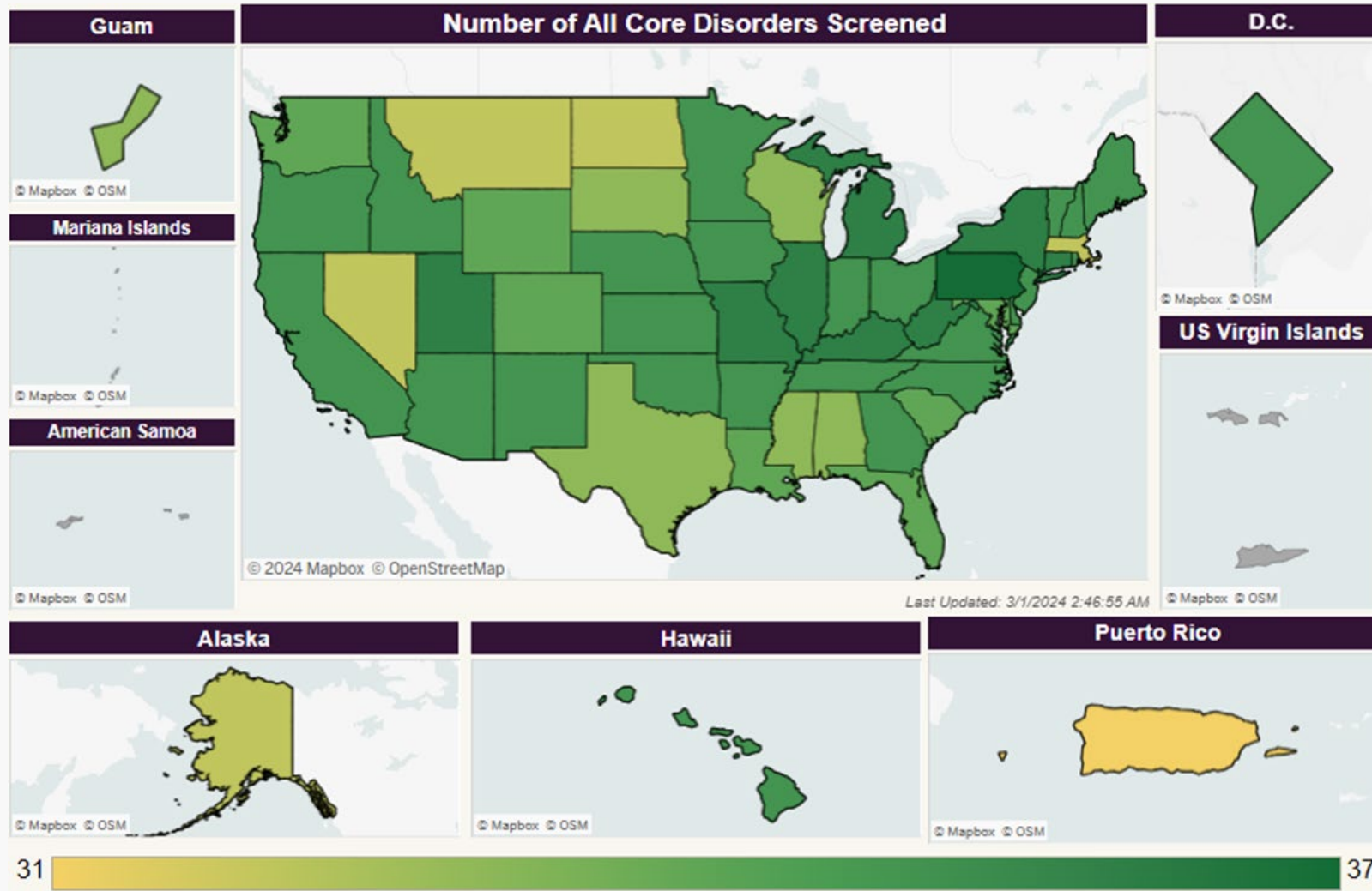
# Core RUSP Disorders Screened	States /Territories
31	Puerto Rico
32	Alaska, Montana, Nevada, North Dakota
33	Alabama, Guam, Mississippi, South Dakota, Texas, Wisconsin
34	Colorado, Louisiana, Maryland, South Carolina, Wyoming
35	Arizona, Arkansas, California, Delaware, District of Columbia, Florida, Georgia, Hawaii, Idaho, Indiana, Iowa, Kansas, Maine, Massachusetts, Minnesota, Nebraska, New Hampshire, New Jersey, New Mexico, North Carolina, Ohio, Oklahoma, Oregon, Rhode Island, Tennessee, Vermont, Virginia, Washington
36	Connecticut, Illinois, Kentucky, Michigan, Missouri, New York, Utah, West Virginia
37	Pennsylvania

Recommended Uniform Screening Panel Core Disorders (37)

Year	Disorders Added to RUSP						
2006*	3-MCC	Cbl A, B	GA1	HCY	MCAD		PROP
	ASA	CF	GALT	HEAR	MCD		TFP
	BIOT	CIT	Hb S/B+ Th	HMG	MSUD		TYR I
	BKT	CH	Hb S/C	IVA	MUT		VLCAD
	CAH	CUD	Hb SS	LCHAD	PKU		
2010	Severe Combined Immunodeficiency (SCID)						
2011	Critical Congenital Heart Disease (CCHD)						
2015	Pompe						
2016	Mucopolysaccharidosis Type I (MPS I)						
	X-linked Adrenoleukodystrophy (XALD)						
2018	Spinal Muscular Atrophy (SMA)						
2022	Mucopolysaccharidosis Type II (MPS II)						
2023	Guanidinoacetate methyltransferase deficiency (GAMT)						

* The first 29 disorders added to the RUSP are displayed here in their abbreviated form. For the full list please visit the NewSTEPS website.

Core RUSP Disorders | March 2024



Implementation summary for newest RUSP disorders, as of March 2024 (N = 53)

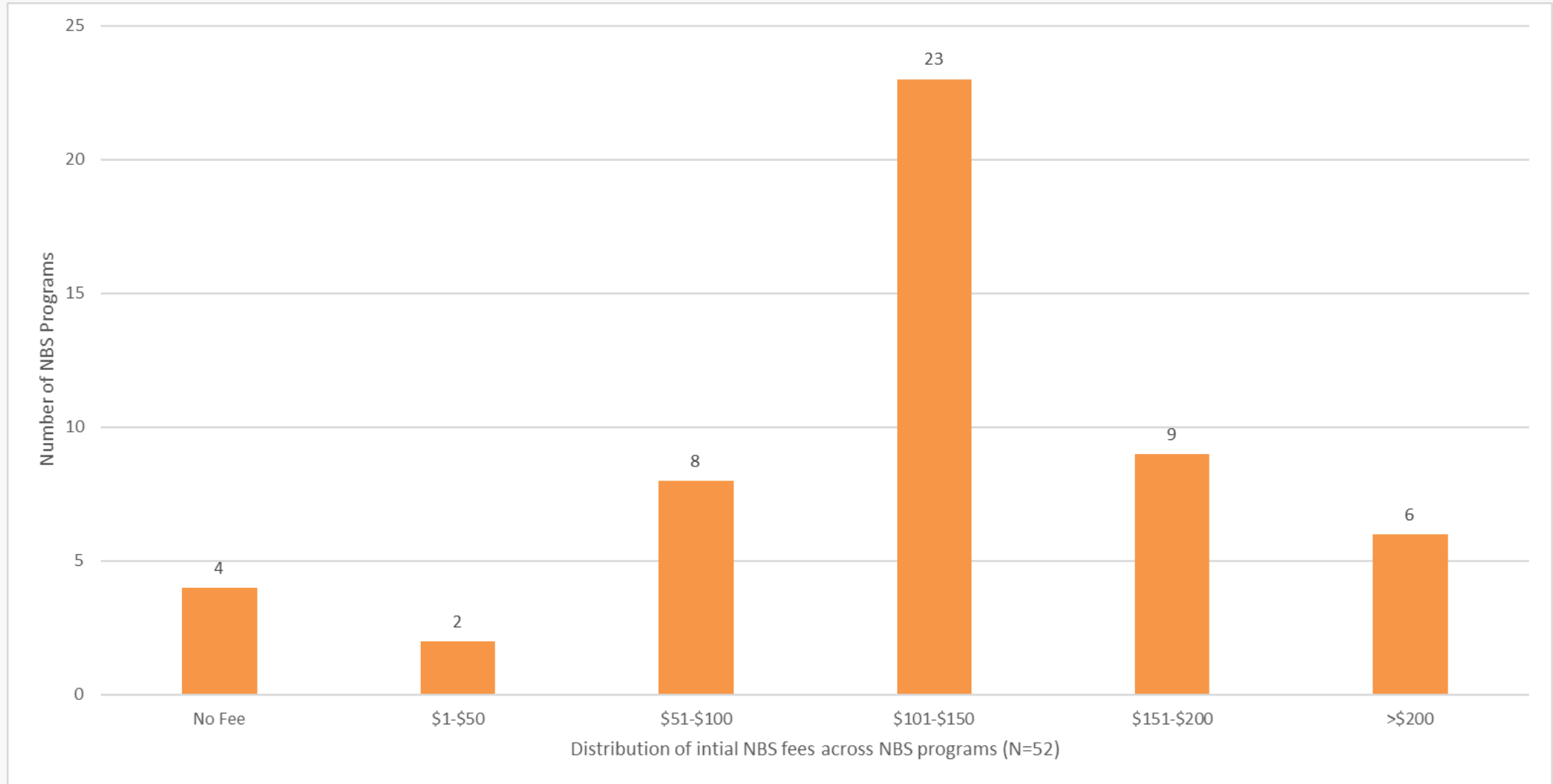
Conditions	SCID	CCHD	POMPE	MPS I	x-ALD	SMA	MPS II	GAMT
Year Added to RUSP	2010	2011	2015	2016	2016	2018	2022	2023
Number of States/Territories Performing Population Screening	53	53	46	43	40	51	5	5
Percent of newborns with access to universal screening*	100%	100%	86%	83%	90%	99%	10%	14%
Average number of years to implement after addition to the RUSP	4.3	2.7	4.8**	4.0**	4.2**	2.2**	***	***

* Calculated using 2022 provisional births

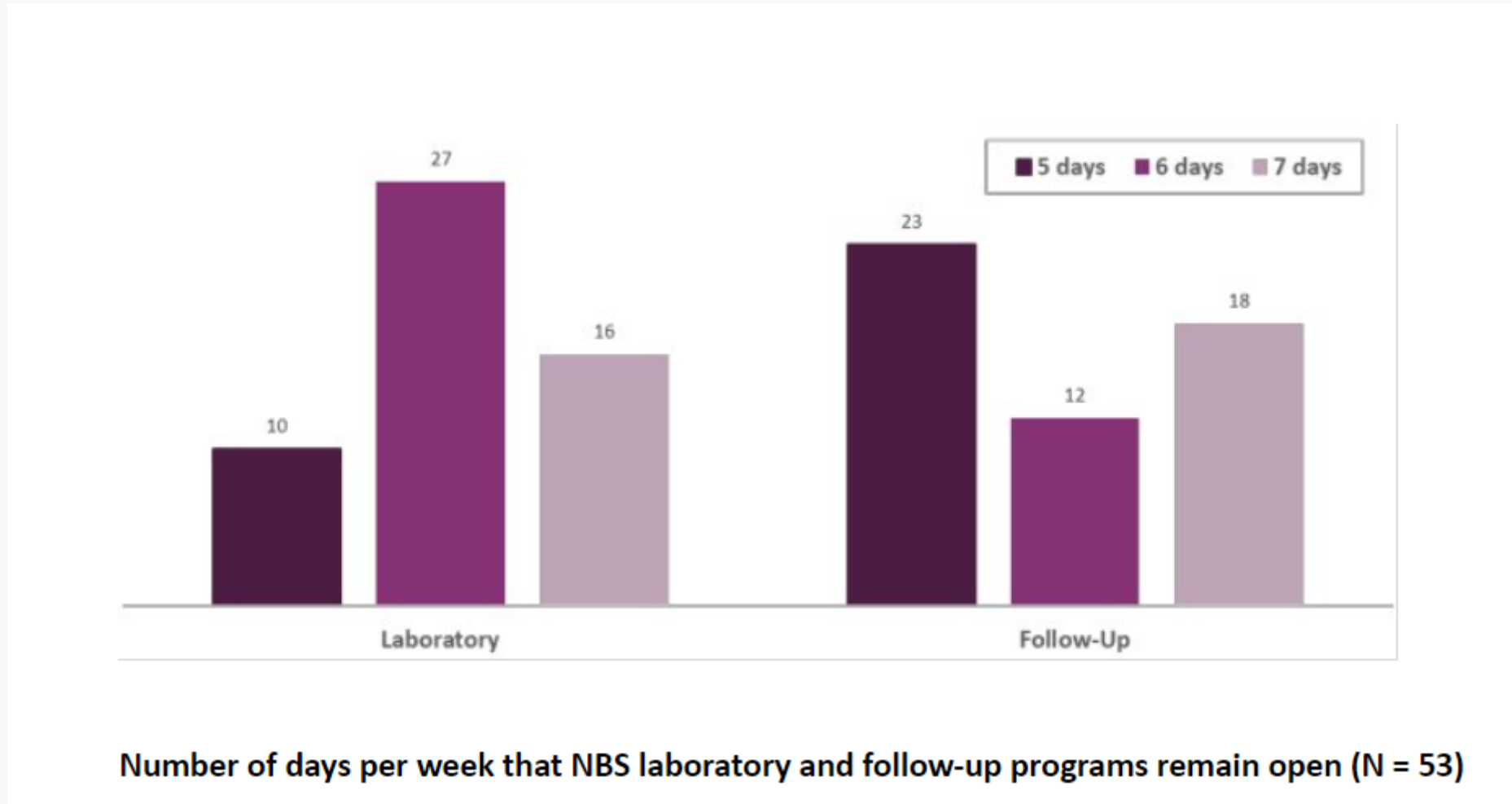
** Of states/territories offering universal screening

*** Insufficient data

Newborn Screening Fees



Newborn Screening Program Operating Hours



Other NBS Related Updates

- Lab Developed Tests – FDA regulations
- Bearder v State of Minnesota
- Lovaglio and Jedynak v. Baton and Scotto-Rosato
- 2024 Newborn Screening Symposium



Questions?

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