

Welcome to the National New Disorder Meeting



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Overview of New Disorders

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Vision

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

Mission

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



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The Newborn Screening Technical assistance and Evaluation Program

Providing:

- quality improvement initiatives for newborn screening systems
- innovative data repository
- technical and educational resources for state newborn screening programs

www.newsteps.org

newsteps@aphl.org



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Recommended Uniform Screening Panel

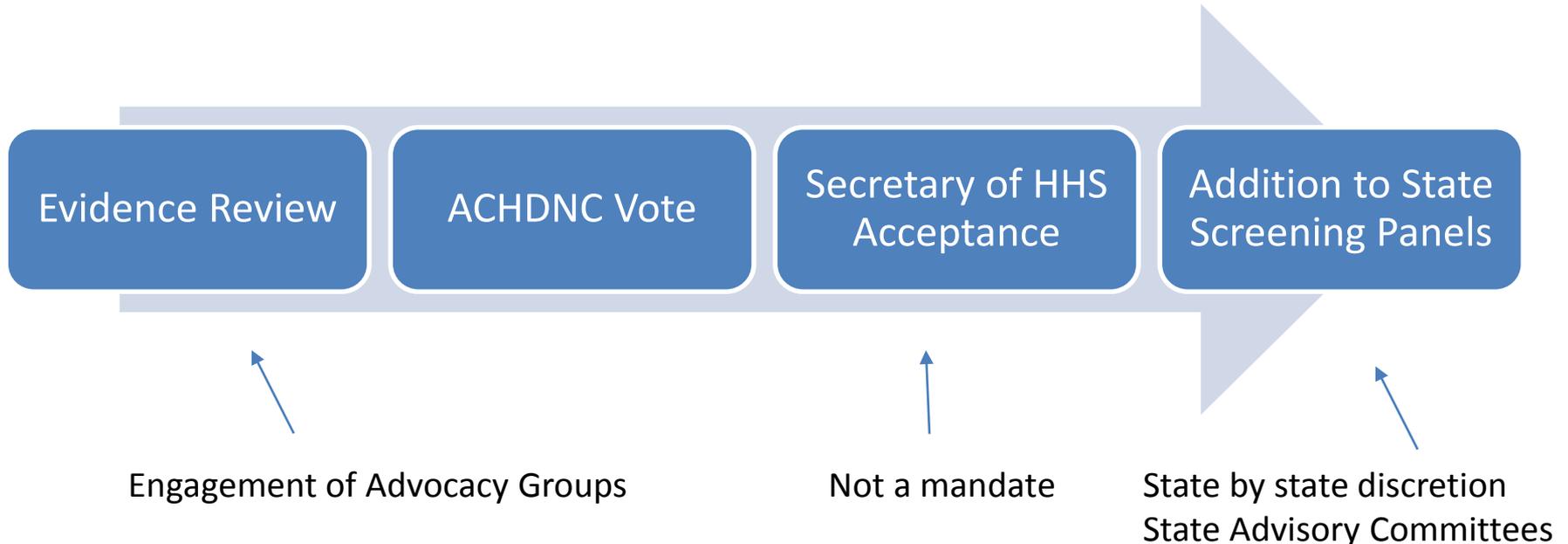


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How Conditions Are Added by the ACHDNC to the RUSP

- **RUSP** = Recommended Uniform Screening Panel
- **ACHDNC** = Advisory Committee on Heritable Disorders in Newborns and Children



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State Considerations for Adding a Disorder to Panels

- Cost to screen
- Technology
- Algorithms
- Fee Increases
- Accepted treatment
- Access to care
- Clinical referral networks
- Late onset versus early onset
- Insurance factors
- Health equity and public health service



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THE SECRETARY OF HEALTH AND HUMAN SERVICES

WASHINGTON, D.C. 20201

MAR 02 2015

Joseph A. Bocchini, Jr., MD
Committee Chairperson
Discretionary Advisory Committee on Heritable Disorders
in Newborns and Children
Professor and Chairperson
Department of Pediatrics
Louisiana State University
1501 Kings Highway
Shreveport, LA 71130

Dear Dr. Bocchini:

As indicated in the January 27, 2014 letter from Secretary Sebelius, the Secretary's Discretionary Advisory Committee on Heritable Disorders in Newborns and Children (DACHDNC) recommendations regarding the addition of Pompe disease to the HHS Recommended Uniform Screening Panel (RUSP) were forwarded to the Interagency Coordinating Committee on Screening in Newborns and Children (ICC) for additional input regarding implementation.

The ICC reviewed the DACHDNC's recommendations as well as evidence from method evaluation studies, information on test quality, national guidance documents, and current state screening activities. In its report to me, the ICC noted challenges associated with the implementation of state newborn screening for Pompe disease including resource limitations for laboratory testing, management of laboratory results, and follow-up systems. However, the ICC's recommendation will help increase the survival and mortality of babies born with the condition.

I would like to commend the DACHDNC's recommendation to add Pompe disease to the RUSP. The information from the objective evidence report, *Newborn Screening for Pompe Disease*, was taken into account as I reviewed the ICC's report.

***I accept the ACHDNC's
recommendation to add Pompe
disease to the RUSP.***

Taking into consideration the information presented in these reports, I accept the DACHDNC recommendation to add Pompe disease to the RUSP. The Affordable Care Act requires that most health plans cover the evidence-informed preventive care and screenings provided for in the comprehensive guidelines supported by Health Resources and Service Administration (HRSA). Because the RUSP is a component of these guidelines, a condition added to the RUSP must be covered. It should be understood that addition of Pompe disease to the RUSP does not constitute a requirement for states to implement screening, only a recommendation. I recognize



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FEB 16 2016

Joseph A. Bocchini, Jr., M.D.
Committee Chairperson
Advisory Committee on Heritable
Disorders in Newborns and Children
5600 Fishers Lane
Room 18W68
Rockville, MD 20857

Dear Dr. Bocchini:

I want to take this opportunity to advise you of my decisions, taking into account the Interagency Coordinating Committee on Newborn and Child Screening's (ICC) review, regarding the Advisory Committee on Heritable Disorders in Newborns and Children's (ACHDNC) recommendations to add Mucopolysaccharidosis type I (MPS I) to the Recommended Uniform Screening Panel (RUSP) and to provide federal funding to state newborn screening programs to implement the screening of MPS I.

The ICC reviewed the ACHDNC's report to me, the ICC noted that screening for MPS I. However, help increase the number of the disorder.

I would like to commend the newborn screening for MPS I. The information from the report, *Newborn Screening for Mucopolysaccharidosis Type 1 (MPS I)*, was taken into reviewed the ICC's report.

Based on the information presented in these reports, I accept the ACHDNC's recommendation to add MPS I to the RUSP. The Affordable Care Act requires that most health plans cover the evidence-based preventive care and screenings provided for in the comprehensive guidelines supported by Health Resources and Service Administration (HRSA). Because the RUSP is a component of these guidelines, a condition added to the RUSP must be covered without cost-sharing. Plans and insurers will have until the first plan year that is one year after the date of adoption of the recommendation to implement coverage. However, it should be understood that addition of MPS I to the RUSP does not constitute a requirement for states to implement screening, only a recommendation.

I accept the ACHDNC's recommendation to add MPS I to the RUSP.



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5600 Fishers Lane
Room 18W68
Rockville, MD 20857

Dear Dr. Bocchini:

I accept the ACHDNC's recommendation to expand the RUSP to include the addition of X-ALD.

I conducted an analysis of the benefits and harms of newborn screening for X-ALD as well as the capability of state newborn screening programs to offer comprehensive screening and follow-up services for infants identified with X-ALD. After reviewing the ACHDNC's report, *Newborn Screening for X-Linked Adrenoleukodystrophy (X-ALD): A Systematic Review of Evidence*, and taking into consideration the utility of current screening technologies, treatment for X-ALD, and the impact on public health systems, I accept the ACHDNC's recommendation to expand the RUSP to include the addition of X-ALD. As you may know the Affordable Care Act requires that most health plans cover without cost-sharing certain children's preventive services. Because the RUSP is a component of preventive services guidelines supported by the Health Resources and Services Administration, a condition added to the RUSP must be covered without cost sharing. I also want to clarify that the addition of X-ALD to the RUSP does not constitute a requirement for states to implement screening and is only a recommendation.

At this time, I am unable to identify new funding consistent with the ACHDNC's second recommendation to provide funding to state newborn screening programs to implement screening of X-ALD. However, I recognize the ongoing challenges that state newborn screening programs are experiencing in maintaining robust quality programs with the increasing demands of adding new conditions. This is why I have asked federal agencies to consider ways within their existing research and technical assistance resources to support state programs as they begin to implement comprehensive population-based screening for X-ALD.

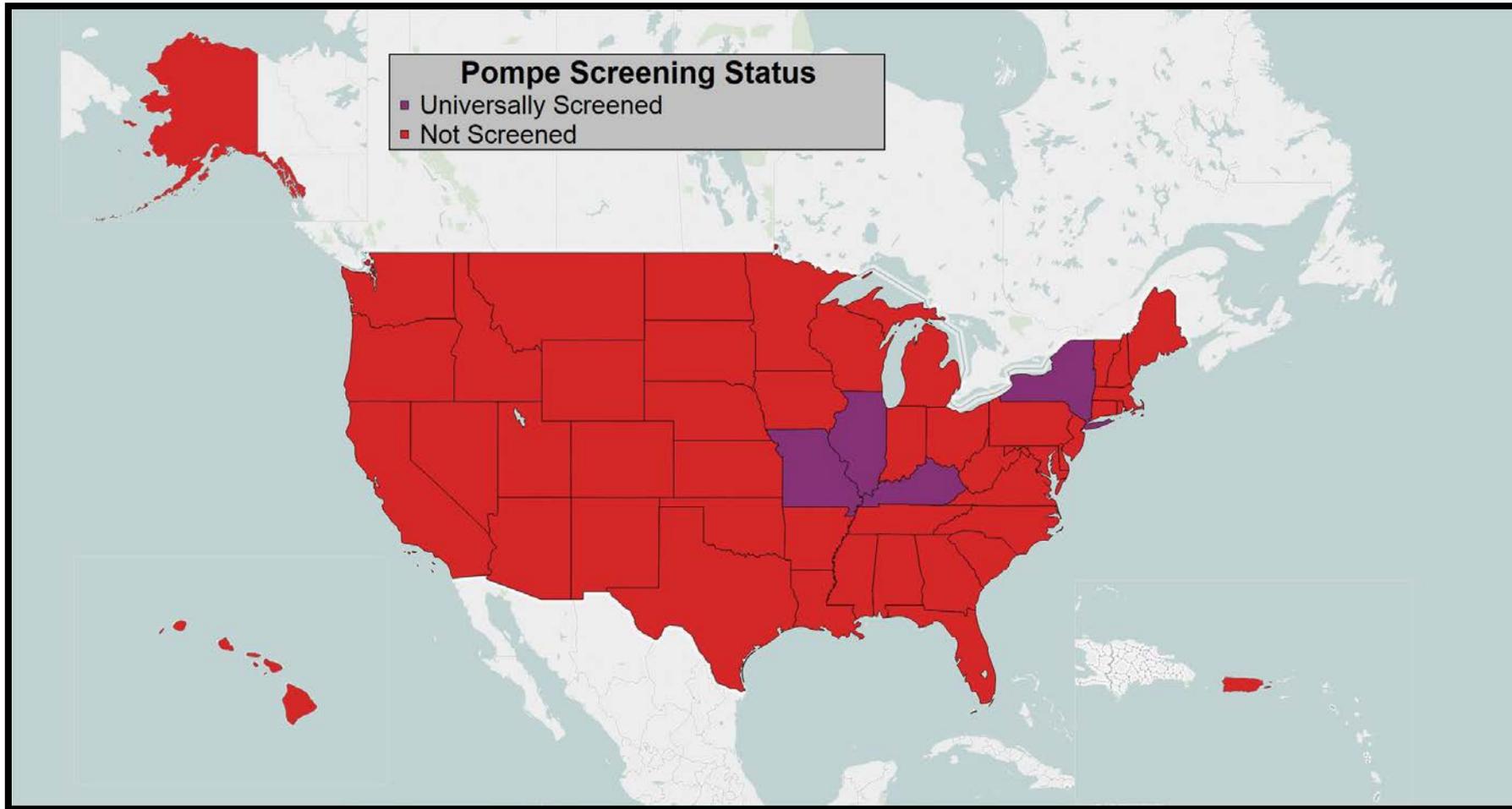
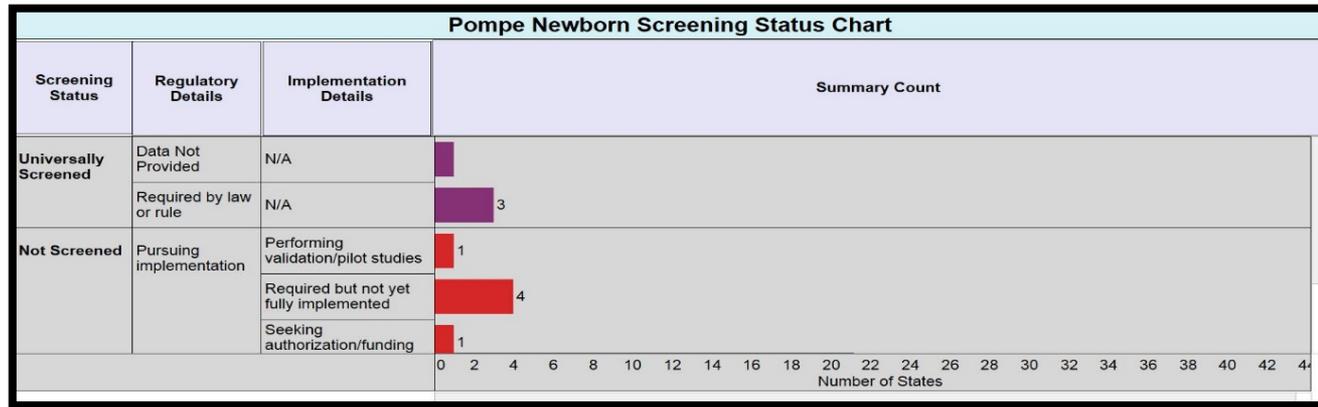
Progress in Pompe NBS Implementation



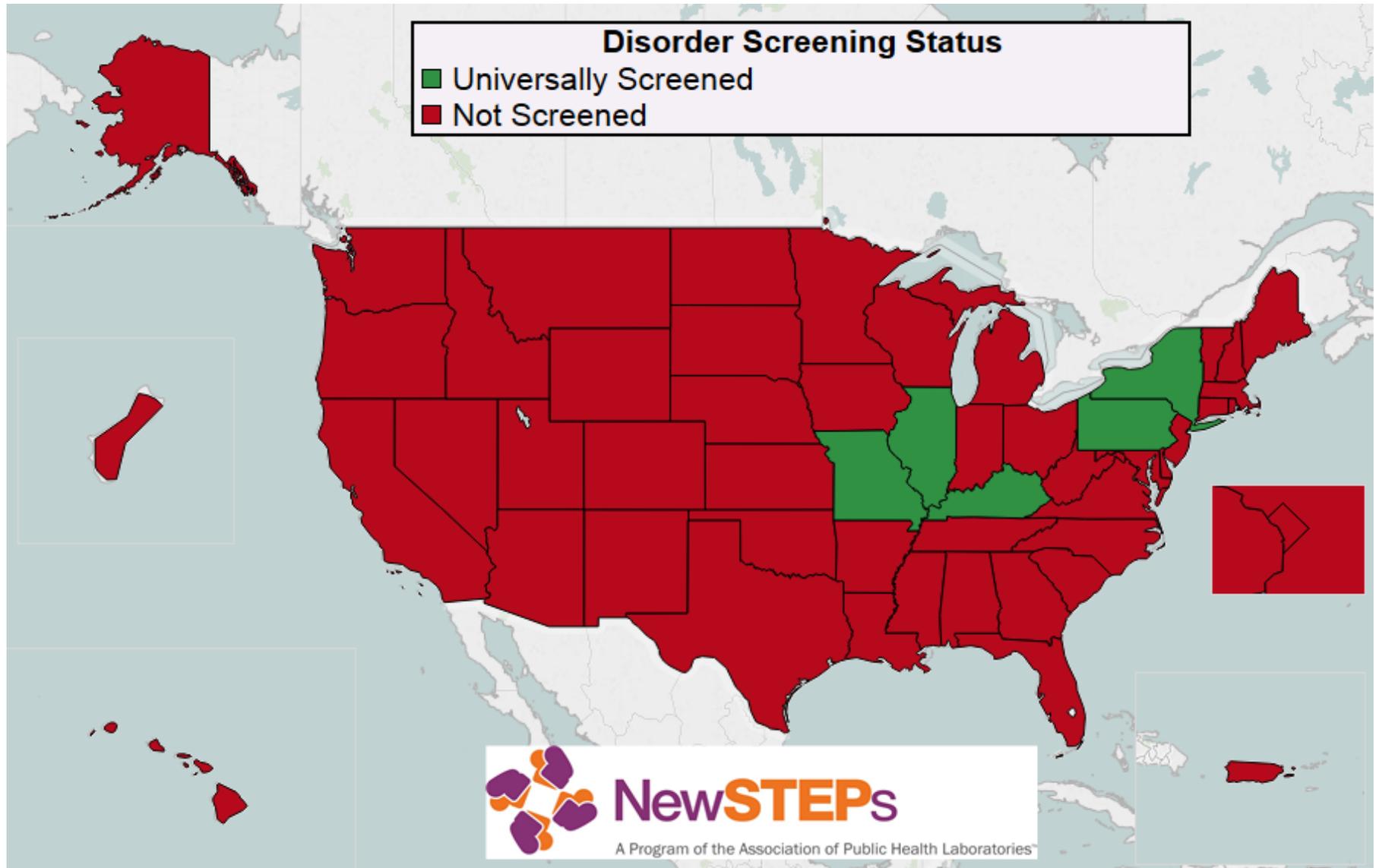
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Pompe: Current Day



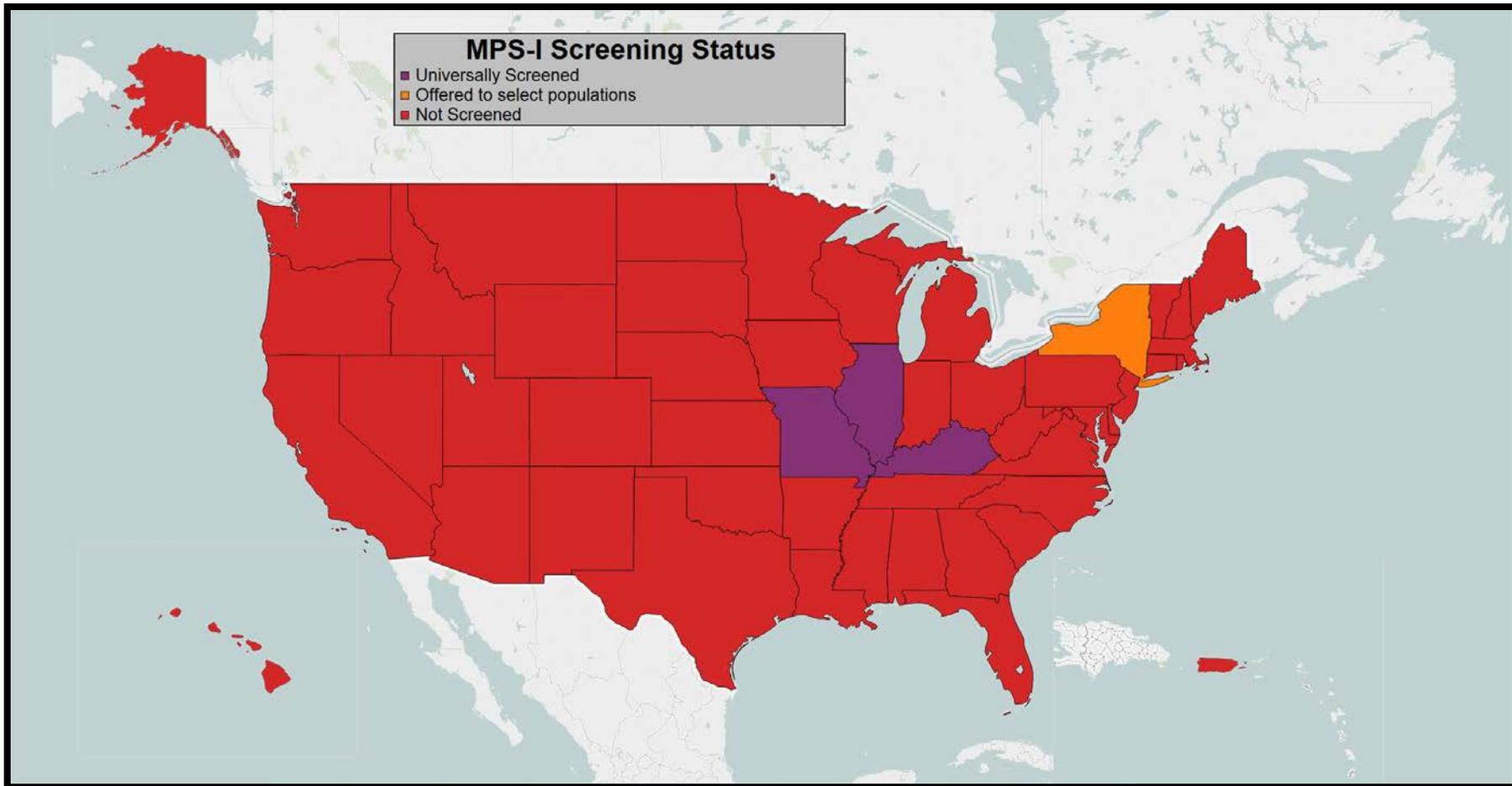
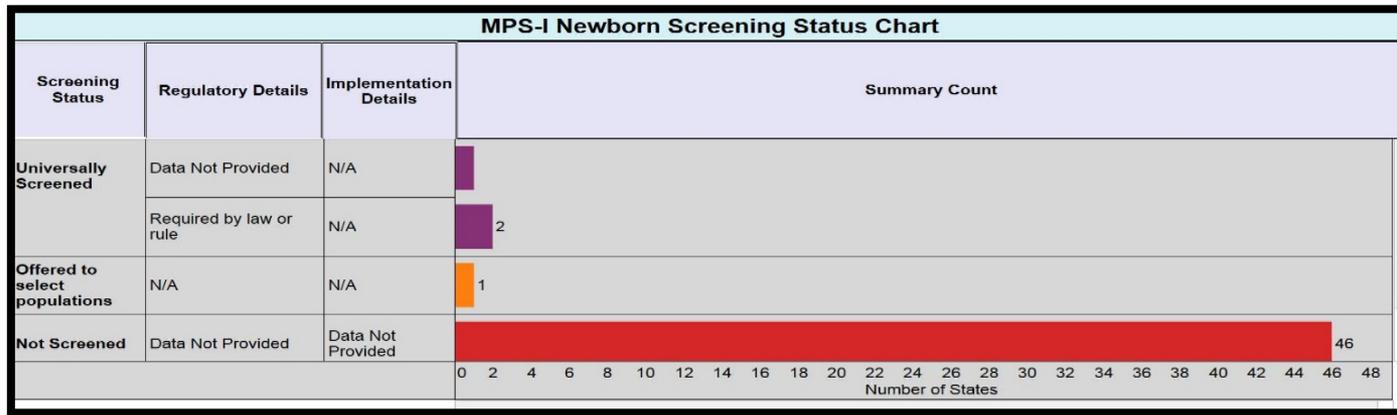
Progress in MPS I NBS Implementation



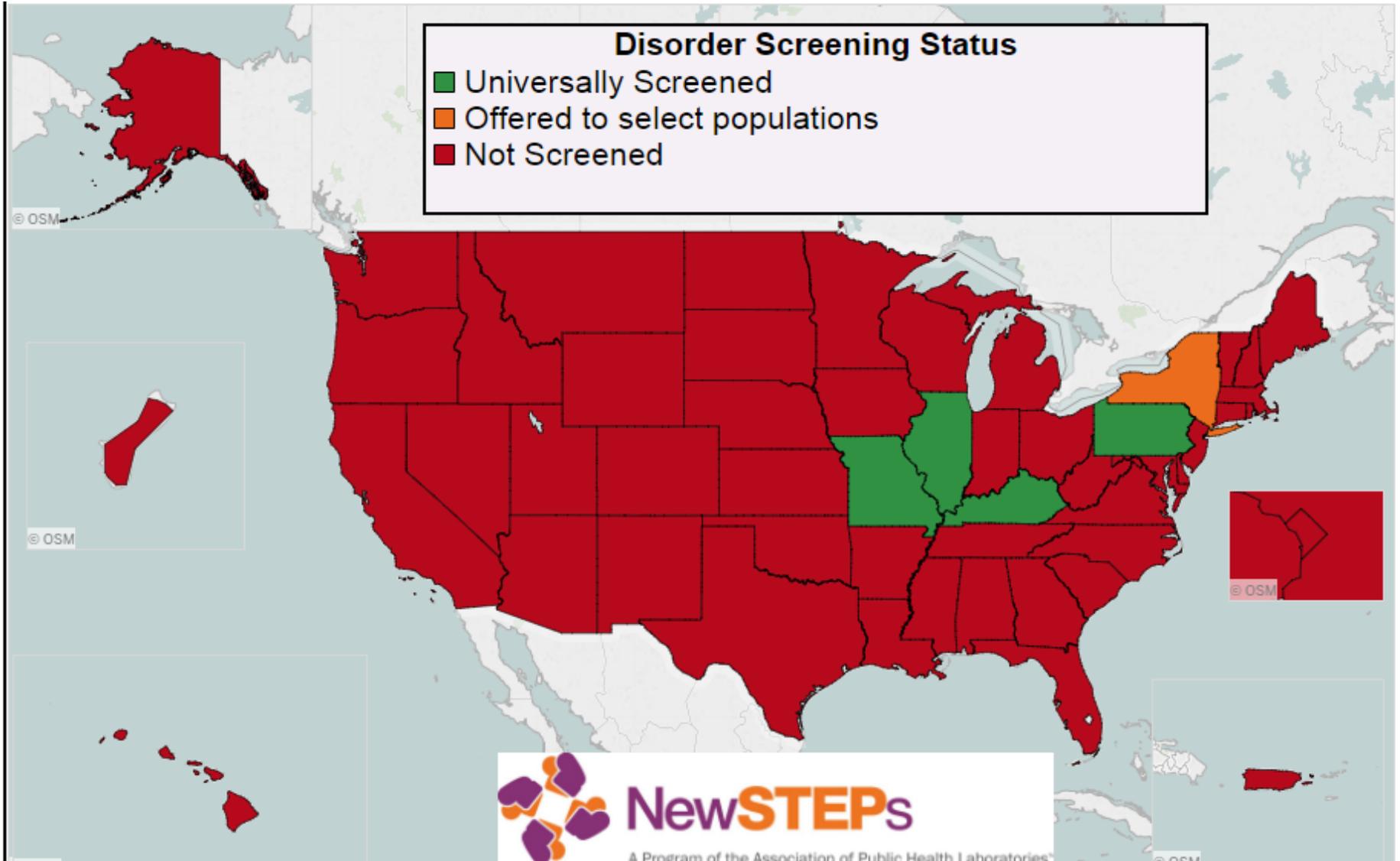
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MPS I: Current Day



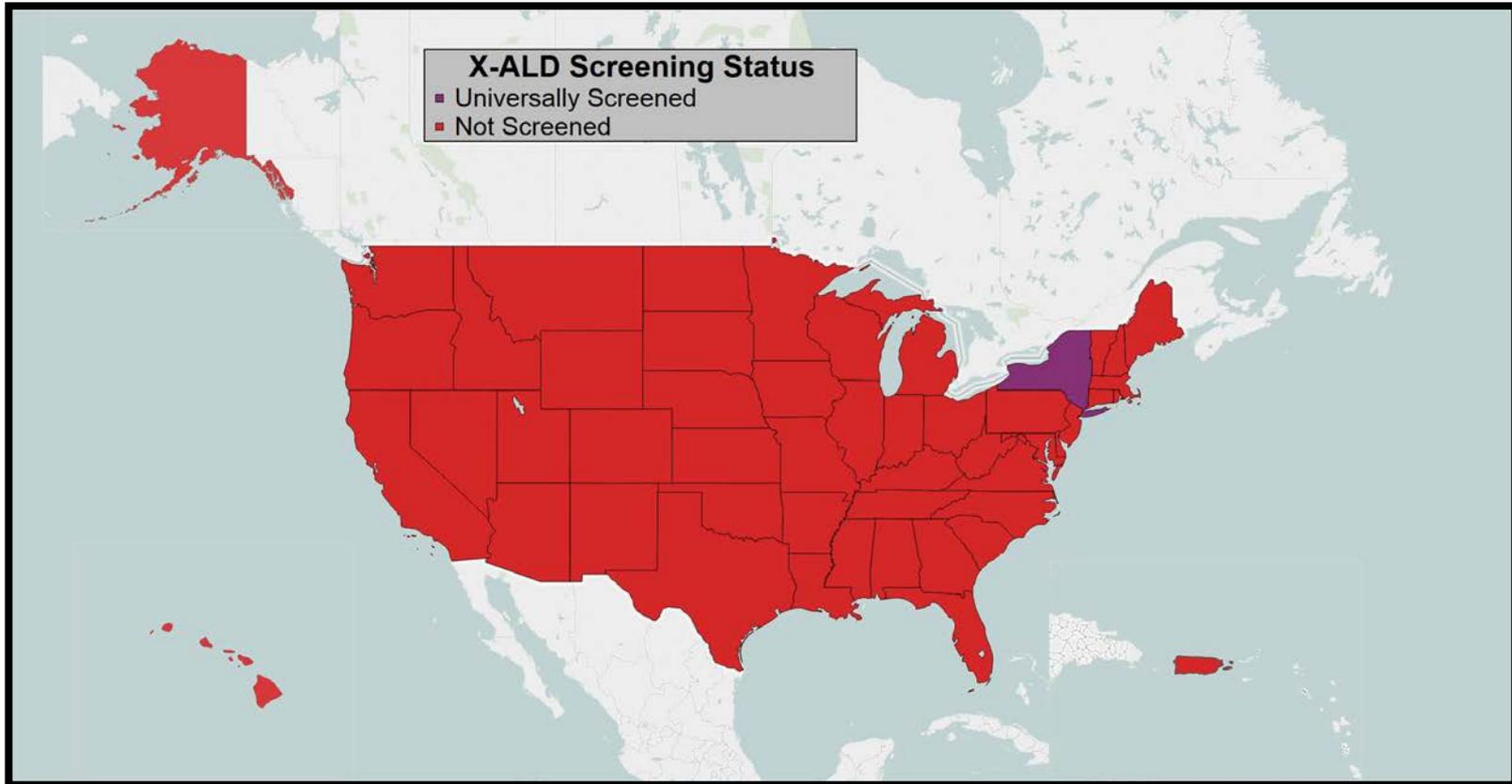
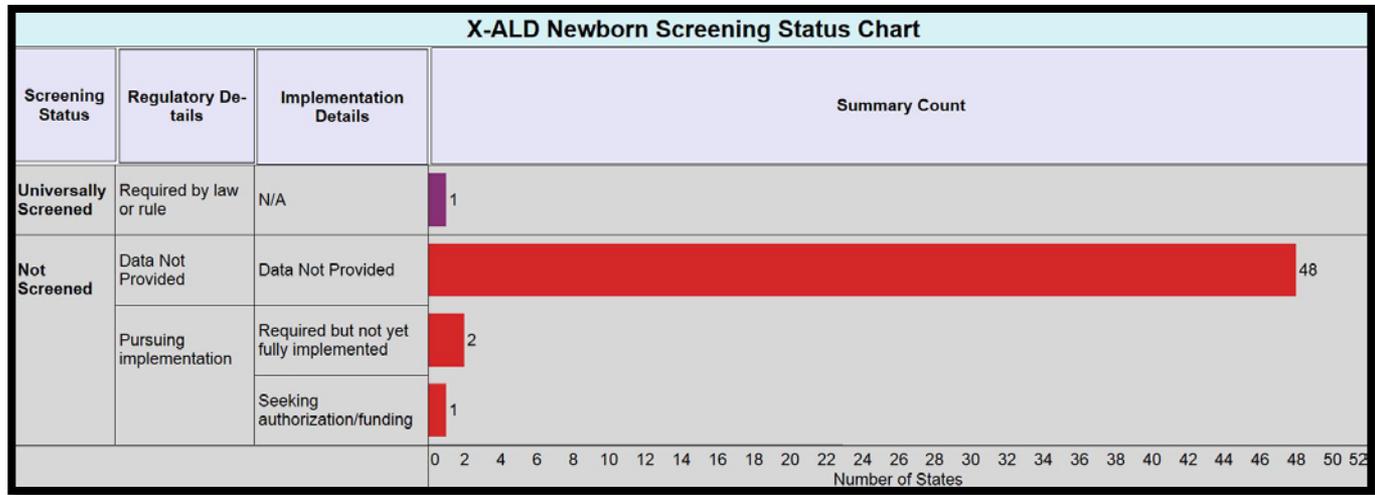
Progress in x-ALD NBS Implementation



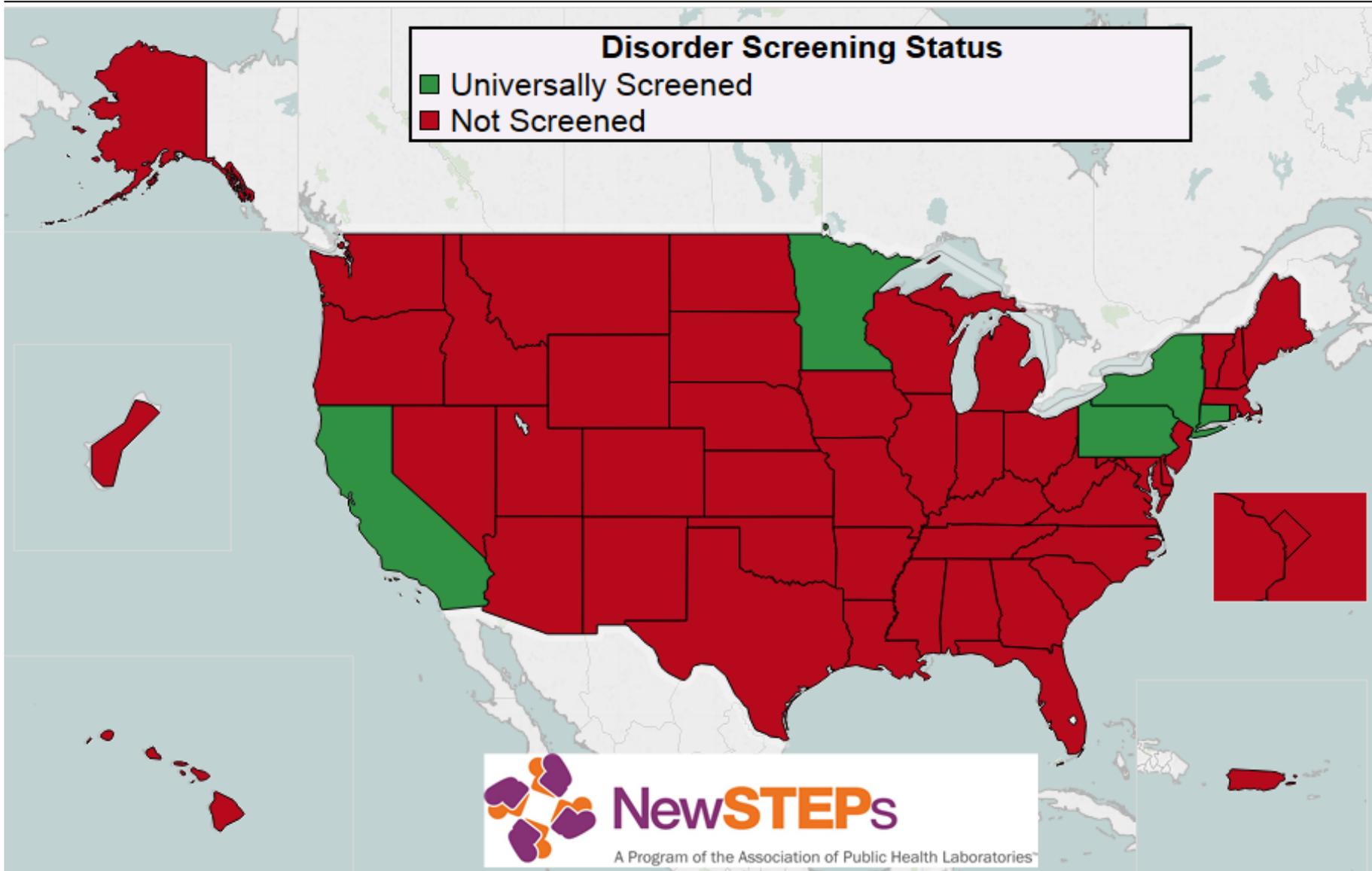
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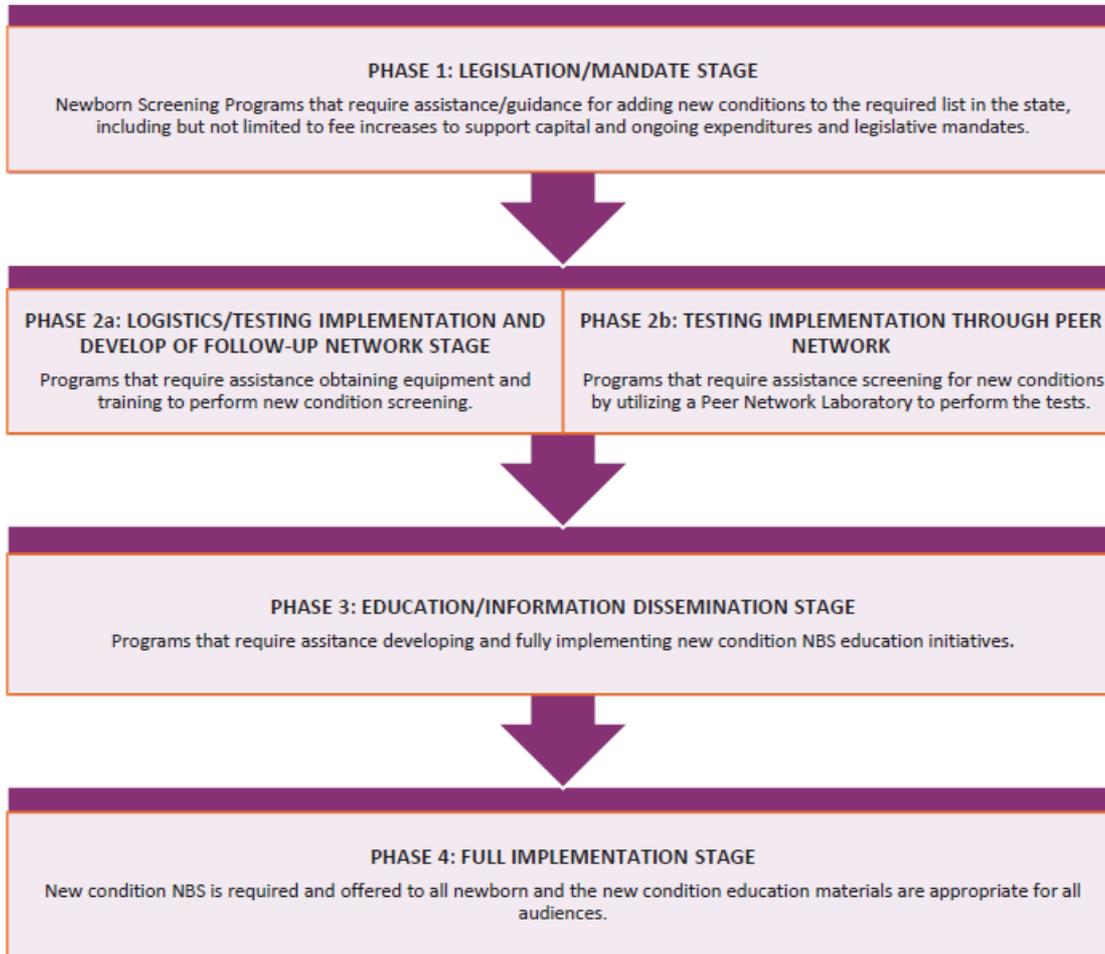


x-ALD: Current Day



Readiness Tool

New Conditions NBS Implementation Stages: Four-Tier Model



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Why?

To gain an understanding as to how long it takes to go from the first step towards beginning screening to statewide implementation.

To look into the timing of the steps and variability across programs.

And. . . to connect states to find solutions to make the implementation process more efficient for all.

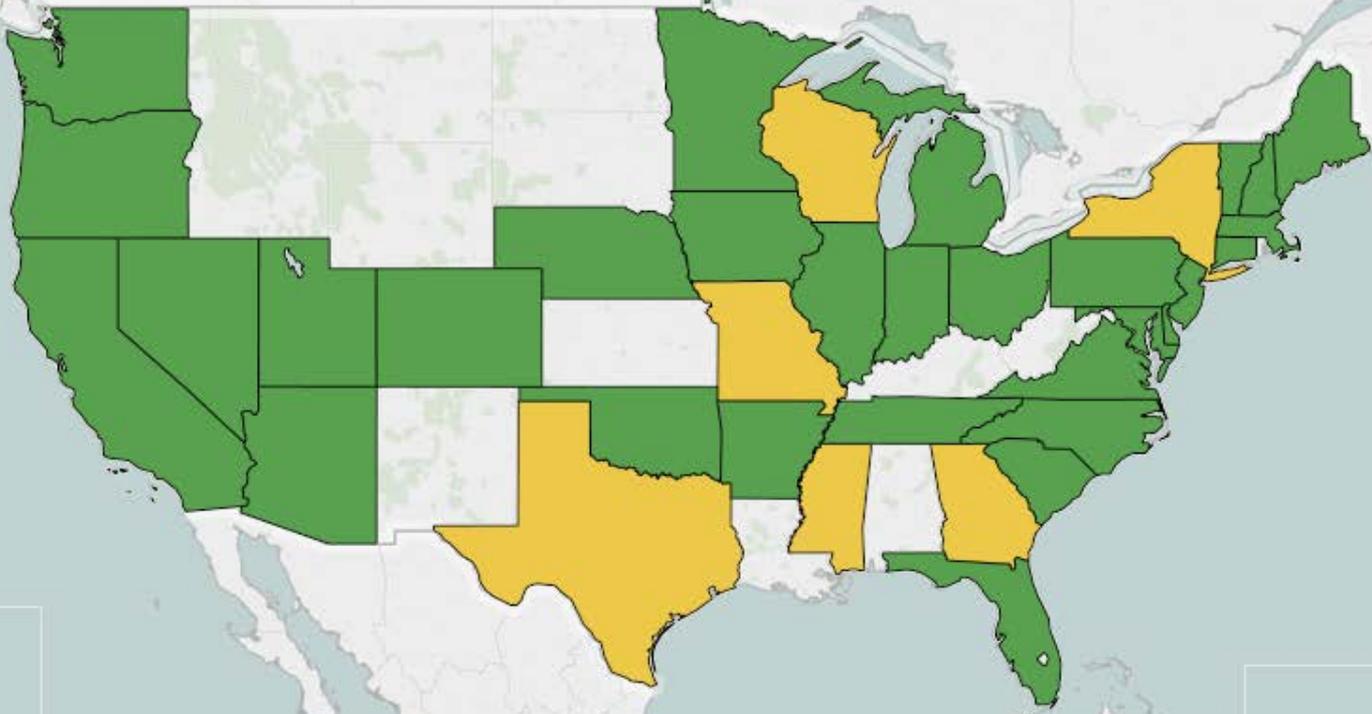
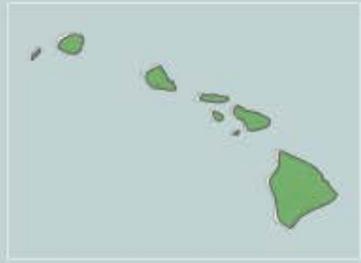


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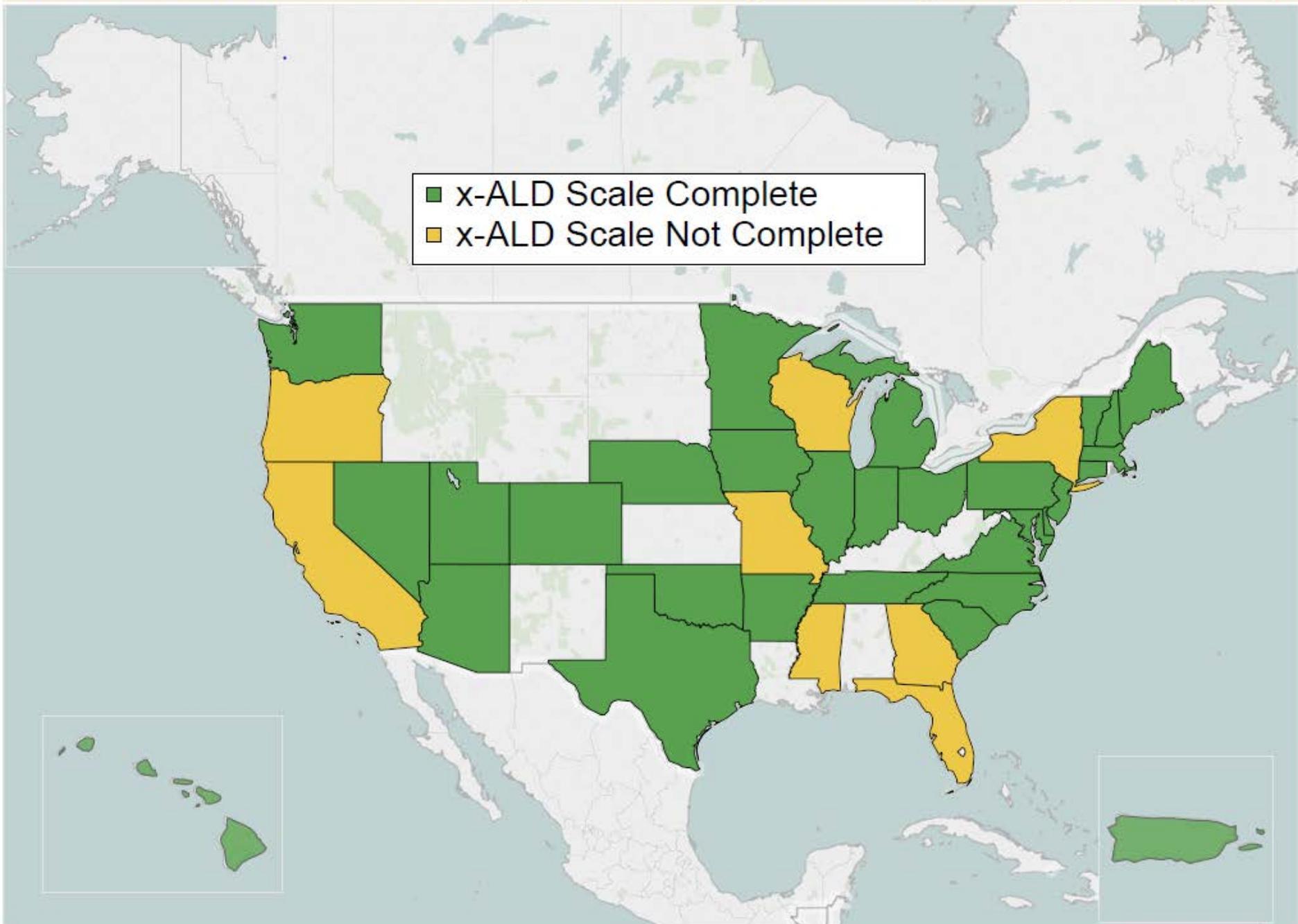
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MPS I: Readiness Scale Completion amongst Meeting Participants (n=32)

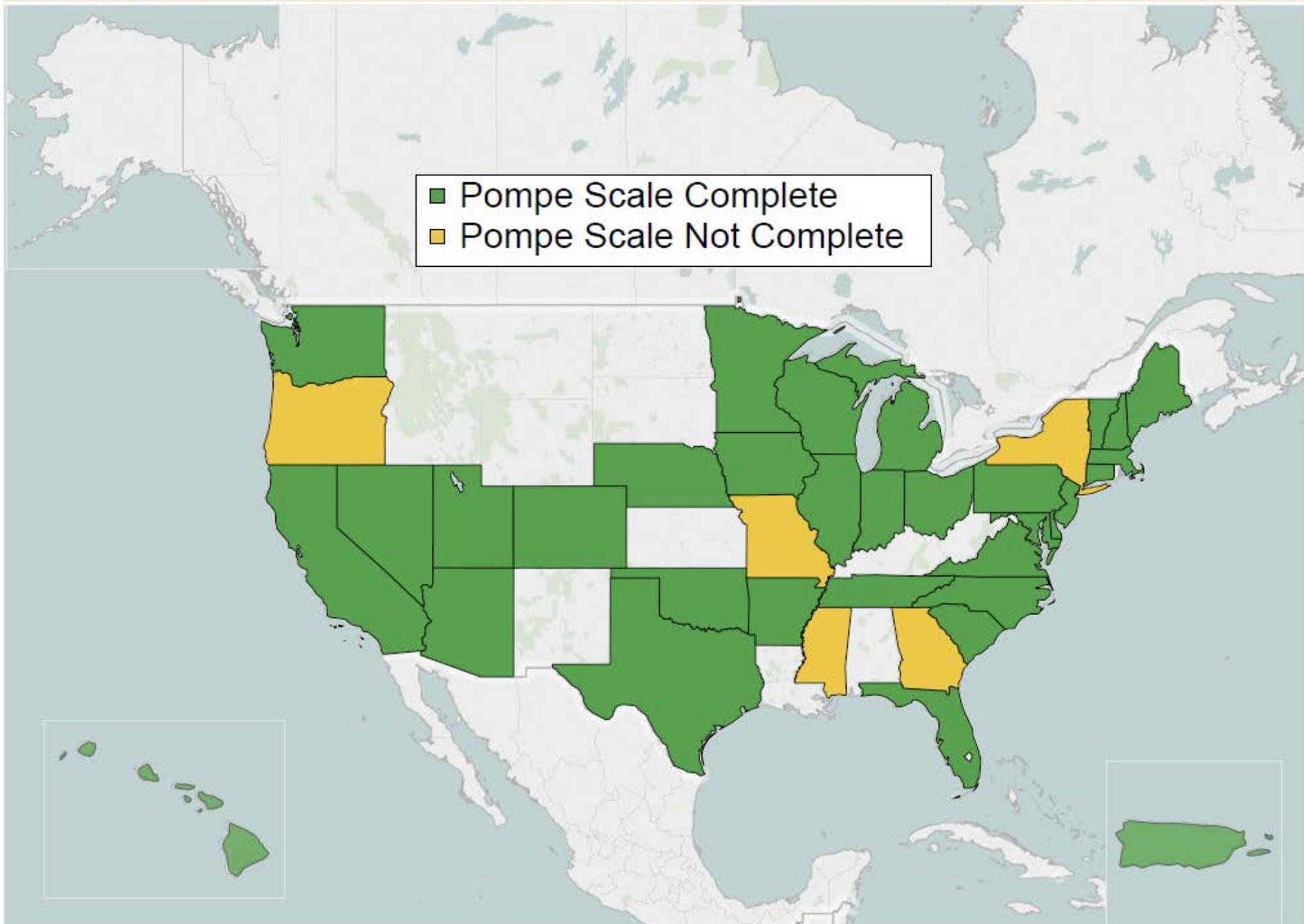
■ MPS I Scale Complete
■ MPS I Scale Not Complete



x-ALD: Readiness Scale Completion amongst Meeting Participants (n=30)



Pompe: Readiness Scale Completion amongst Meeting Participants (n=33)



Goals for this Meeting

- States to gain an understanding of what questions they should ask with regards to screening for MPS I, X-ALD, and Pompe
- NewSTEPs to identify technical assistance needs of states with regards to screening for MPS I, X-ALD, and Pompe
- Understanding how the Readiness Tool can help inform others about how much time is needed to begin screening for a new disorder



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What have we learned from the Readiness Tool so far

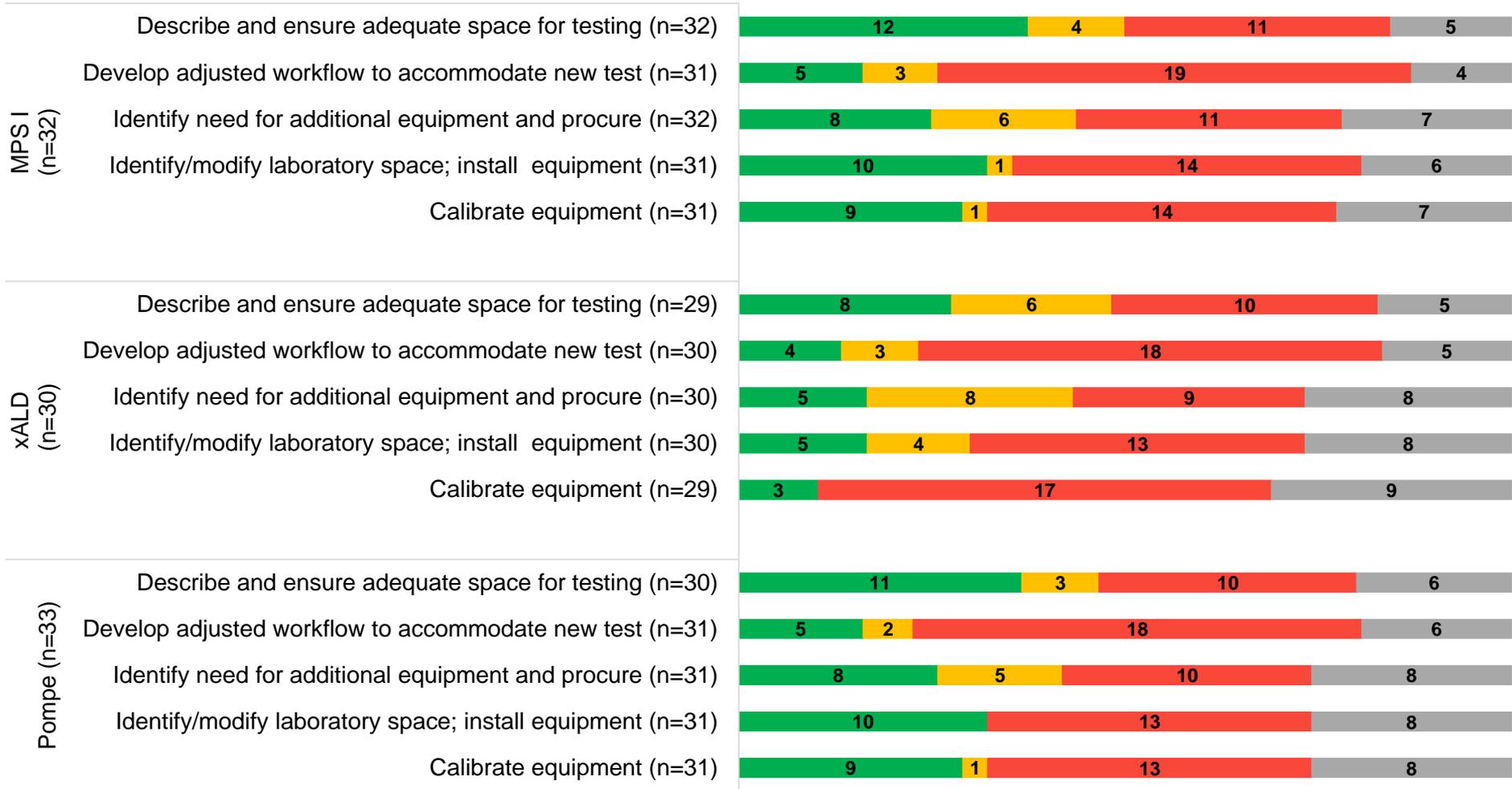


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Phase 2: Lab Facility/Infrastructure Readiness

■ Completed ■ Started ■ Not Started ■ NA

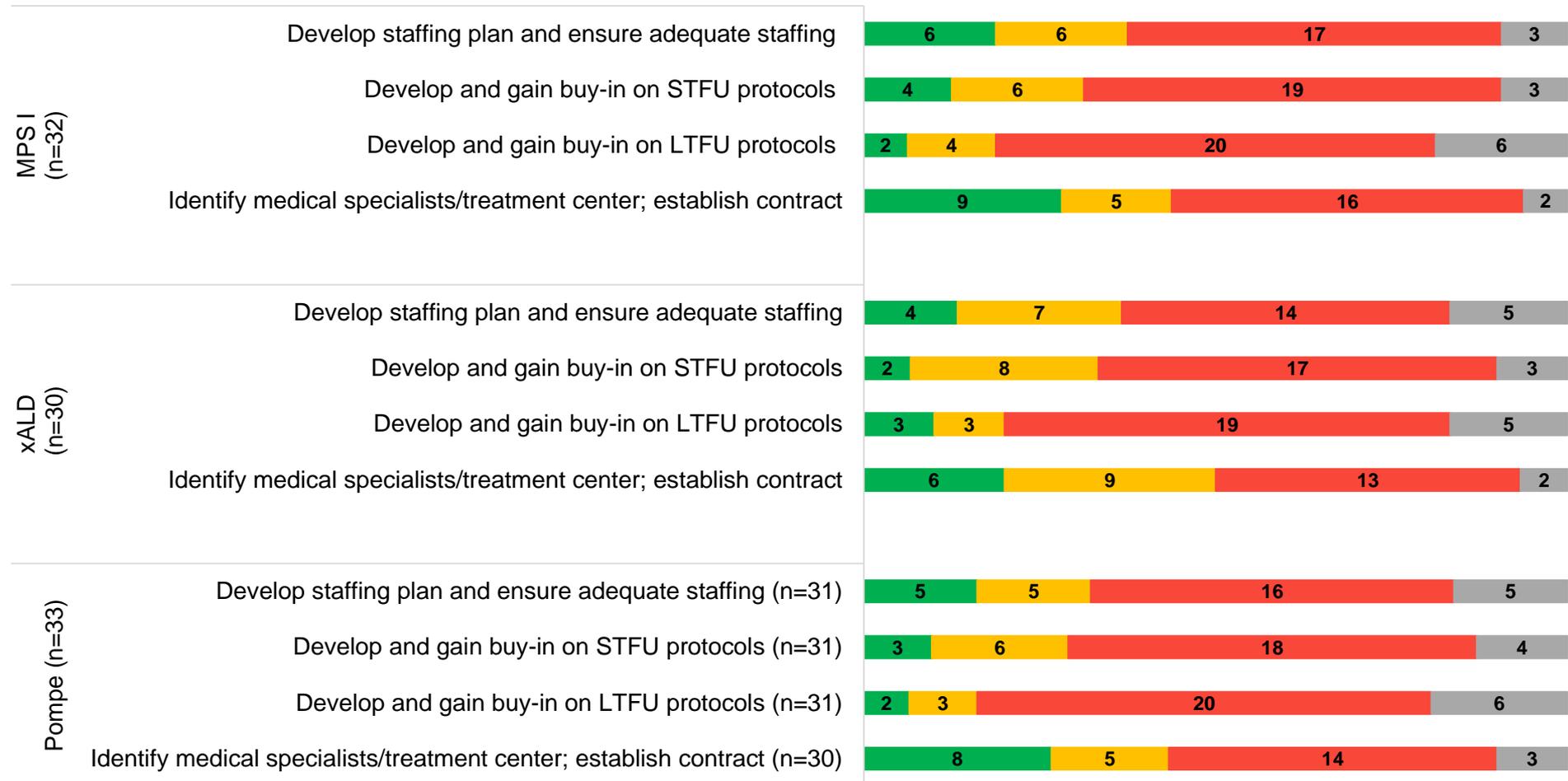


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Phase 2: Follow-up Readiness

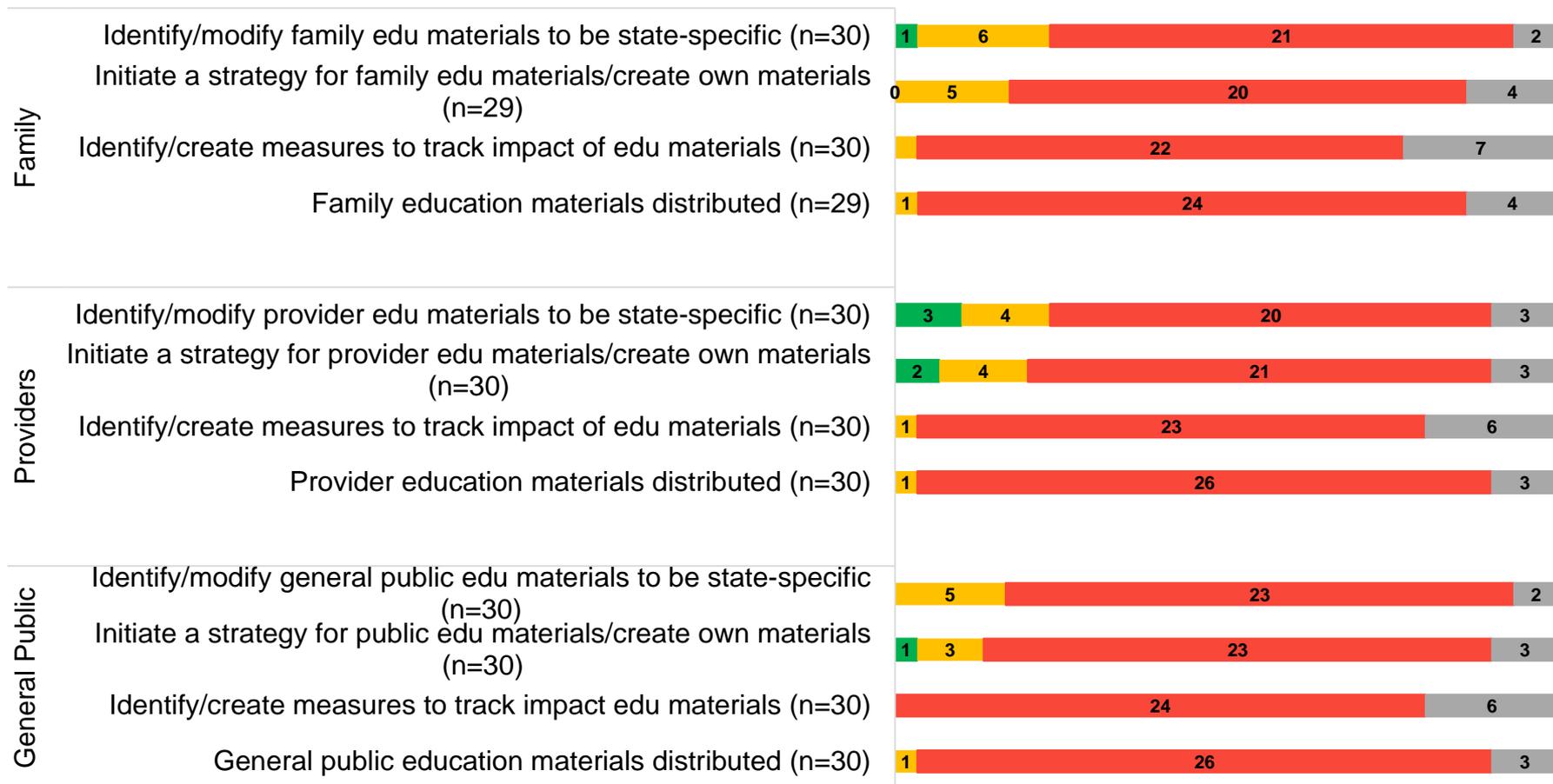
Completed Started Not Started NA



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Pompe-Phase 3: Status of Education Activities (n=33)

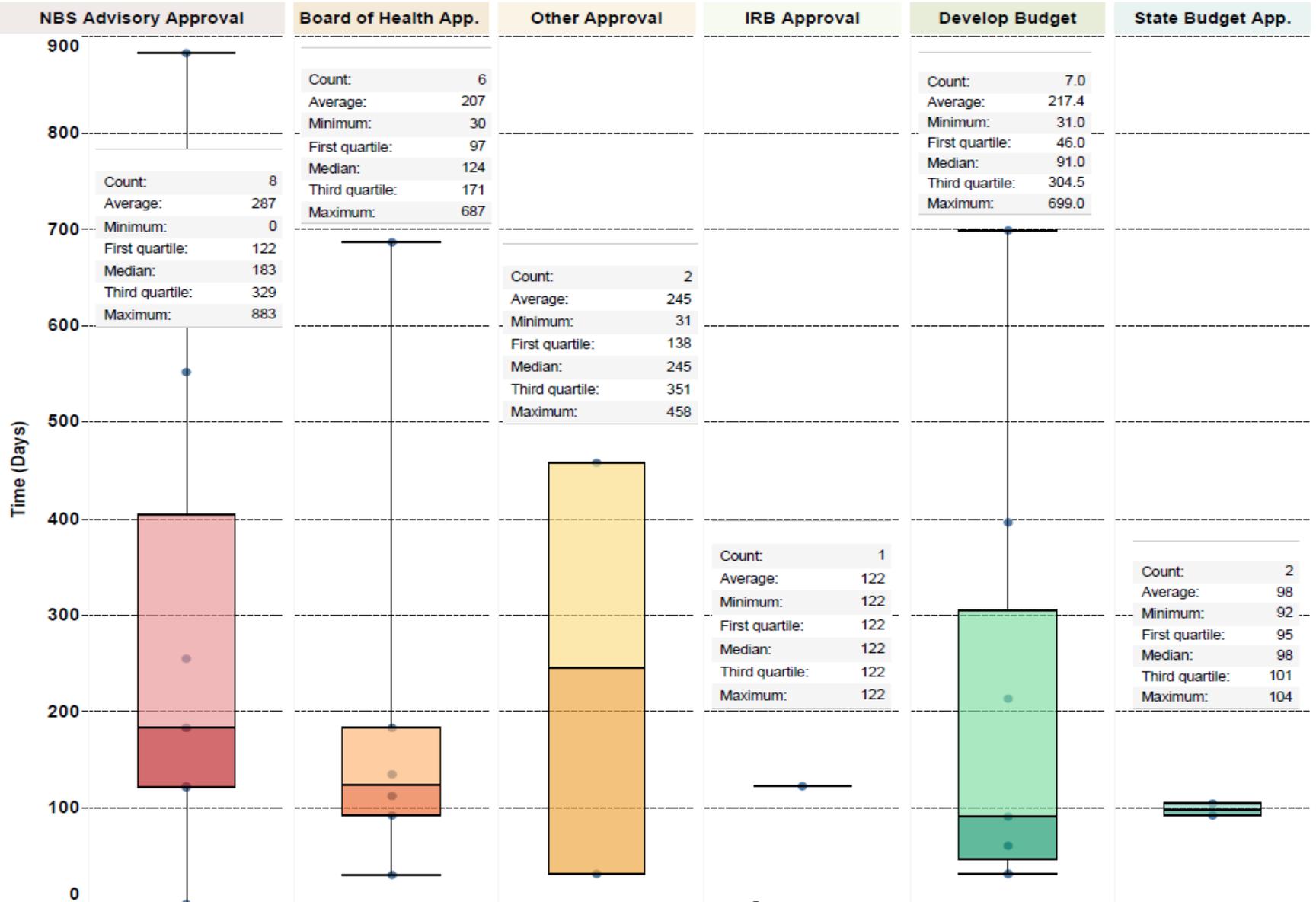


■ Completed
 ■ Started
 ■ Not Started
 ■ NA



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MPS I – Authority to Screen

Phase One

4 states

Approval/Authority To Screen MPS1 (12-36 months)

4 states

Fee MPS1 (5 to 49 months)

4 states

Total Phase 1 MPS1 (5 to 54 months)

2 states

Approval/Authority To Screen Pompe (12 to 18 months)

4 states

Fee Pompe (5 to 21 months)

4 states

Total Phase 1 Pompe (5 to 21 months)

4 states

Approval/Authority to Screen X-ALD (6 to 13 months)

3 states

Fee X-ALD (5 to 21 months)

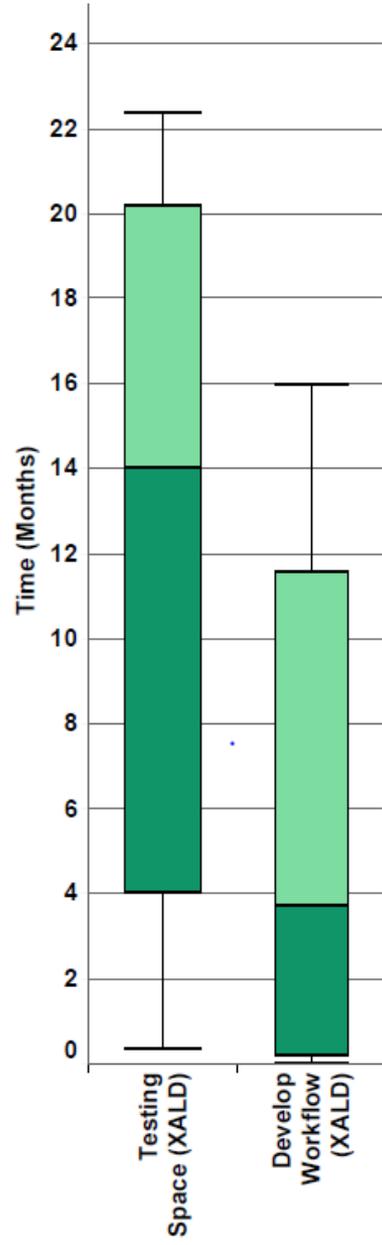
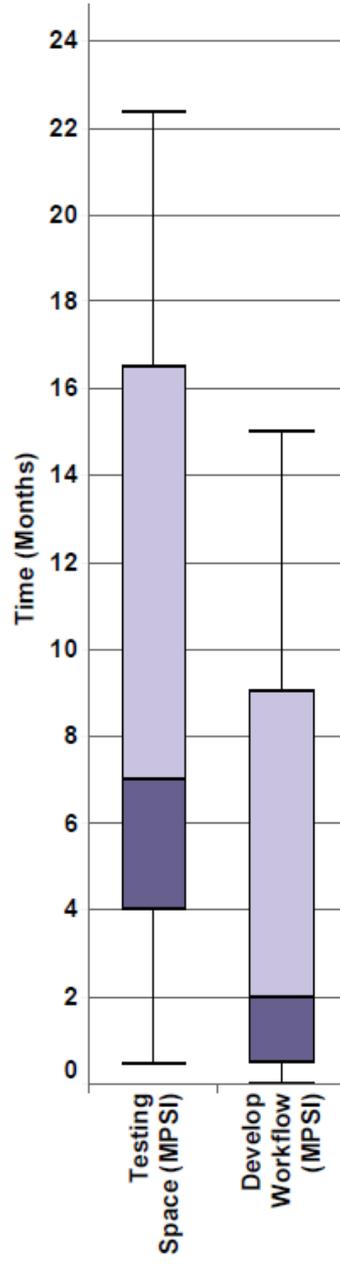
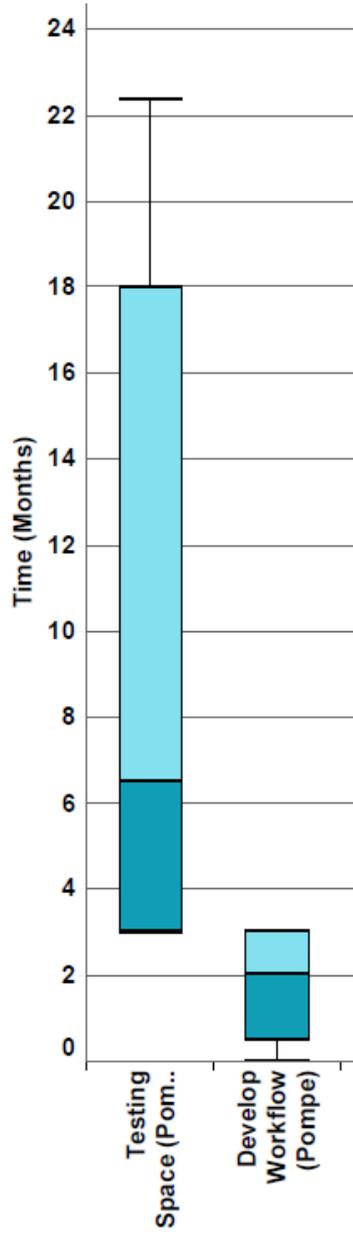
2 states

Total Phase 1 X-ALD (5 to 21 months)



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What is next?

- V
S
-
-



so we can tell the full

set realistic implications for
that are added

explain what is involved
are added with data, not



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Ground Rules for the Meeting

- We need each other
 - We all have expertise in at least one part of the process of screening for new disorders
- Actively participate
 - Ask questions
 - Share experiences
- Share challenges, fears, failures
- Do not assume other's motivations or skills



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Thanks To Our Village

Our Meeting Planning Team

- Michele Caggana
- Patrick Hopkins
- Mei Baker
- Kimberly Piper
- Tony Steyermark
- Amelia Mulford
- Rasoul Koupaei



The Data Team

- Sarah McKasson
- Joshua Miller
- Marci Sontag

The Logistics Team

- Kshea Hale
- Ruthanne Sheller
- Funke Akinsola



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