

Public Health Impact of SCID Newborn Screening

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in Newborns and Children

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NewSTEPs Vision

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

NewSTEPs 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.

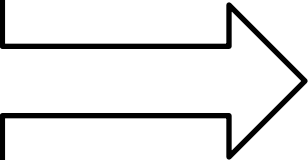


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SCID Funding Opportunities:

- ✓ NIH
- ✓ CDC
- ✓ HRSA

- ✓ Technical Assistance
 - ✓ Education
 - ✓ Implementation Support
- 



September 24, 2014 16:05 ET

Newborn Screening Cooperative Agreement Awarded to APHL for Implementation of SCID Testing

SILVER SPRING, MD--(Marketwired - Sep 24, 2014) - The [Association of Public Health Laboratories \(APHL\)](#) has been awarded \$4 million through a two-year cooperative agreement with the Genetic Services Branch of the U.S. Health and Human Services [Health Resources and Services Administration \(HRSA\)](#). In close partnership with the [Colorado School of Public Health \(ColoradoSPH\)](#), APHL will provide technical assistance, education, and financial support to state newborn screening programs in order fully implement testing for [Severe Combined Immunodeficiency \(SCID\)](#).

Babies born with SCID (also known as "Bubble Boy Disease") lack a functioning immune system and often die within their first year of life. Early detection through [newborn screening](#) allows for life-saving medical intervention. To date, less than half of US states [screen for SCID](#). While many other states have approved the addition of SCID to their newborn screening panel, they have not yet been able to implement screening due to considerable challenges such as integration of new technology, the need for greater laboratory capacity and necessary training within the broader newborn screening system.

"Because SCID testing requires different technology and different clinical follow-up than the other conditions on [HRSA's Recommended Uniform Screening Panel \(RUSP\)](#), implementation has been out of reach for many states," said Dr. Marci Sontag, associate professor of epidemiology at the Colorado School of Public Health. "We aim to provide the support necessary to make SCID testing a reality around the country."

Jelili Ojodu, director of the [Newborn Screening and Genetics](#) program at APHL added, "Supporting state newborn screening programs in their effort to implement SCID testing means detecting this disorder in more babies and allowing them to get critical treatment. Our goals come back to one simple objective: saving more babies' lives."

The Association of Public Health Laboratories (APHL) works to strengthen laboratory systems serving the public's health in the US and globally. APHL's member laboratories protect the public's health by monitoring and detecting infectious and foodborne diseases, environmental contaminants, terrorist agents, genetic disorders in newborns and other diverse health threats.

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APHL Announces Recipients of SCID Implementation Awards

SILVER SPRING, MD--(Marketwired - Feb 2, 2015) - The Association of Public Health Laboratories (APHL) is pleased to announce the recipients of the Severe Combined Immunodeficiency (SCID) Implementation Awards, funding to support full implementation of SCID newborn screening in states which have not been able to do so. Successful use of these awards will result in approximately 450,000 additional newborns receiving this critical testing.

The following agencies have been selected to receive financial support, training and technical assistance for SCID related activities:

- Alabama Department of Health Bureau of Clinical Laboratories
- Arizona Department of Health Services
- Hawaii Department of Health
- Immune Deficiency Foundation
- Kansas Health and Environmental Laboratories
- Kentucky Department for Public Health
- Maryland Department of Health and Mental Hygiene
- North Carolina State Laboratory of Public Health
- North Dakota Department of Health
- Puerto Rico Newborn Screening Program, University of Puerto Rico
- Tennessee Department of Health Division of Laboratory Services
- Utah Department of Health

Babies born with SCID (also known as "Bubble Boy Disease") lack a functioning immune system and often die within their first year of life if untreated. Early detection through newborn screening allows for life-saving medical intervention. To date, just over half of US states screen for SCID. While many other states have approved the addition of SCID to their newborn screening panel, they have not yet been able to implement screening due to considerable challenges such as integration of new technology, the need for greater laboratory capacity and necessary training within the broader newborn screening system. This additional support will result in 75% of US babies being tested, an increase of 15%.

Funded through a \$4 million two-year cooperative agreement with the Maternal and Child Health Bureau of the Genetic Services Branch of the U.S. Health and Human Services Health Resources and Services Administration (HRSA) and in close partnership with the Colorado School of Public Health (ColoradoSPH), the selected agencies will work closely with APHL's Newborn Screening Technical assistance and Evaluation Program (NewSTEPS) for successful implementation of their awards.

This program is 100% funded through Cooperative Agreement # UG5MC27837 from the Health Resources and Services Administration. All publications and presentations are solely the responsibility of the authors and do not necessarily represent the official views of HRSA.

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TIER 1

- **LEGISLATION/MANDATE STAGE:** Newborn Screening Programs that require assistance/guidance for adding SCID to the required list in the state (up to 30 states)

TIER 2

- **LOGISTICS/TESTING IMPLEMENTATION AND DEVELOPMENT OF FOLLOW-UP NETWORK STAGE:** Programs that require assistance obtaining Equipment or Contracting with an entity to perform SCID Testing (up to 25 states)

TIER 3

- **EDUCATION/INFORMATION DISSEMINATION STAGE:** Programs that require assistance developing and fully implementing SCID NBS education initiatives (potentially all states).

TIER 4

- **FULL IMPLEMENTATION STAGE:** SCID NBS is required and offered to all newborns and the SCID education materials are appropriate for all audiences.

Funding for 11 states (2014-2017)

- SCID Implementation
- Technical Assistance
- Training
- Education
- Network Building

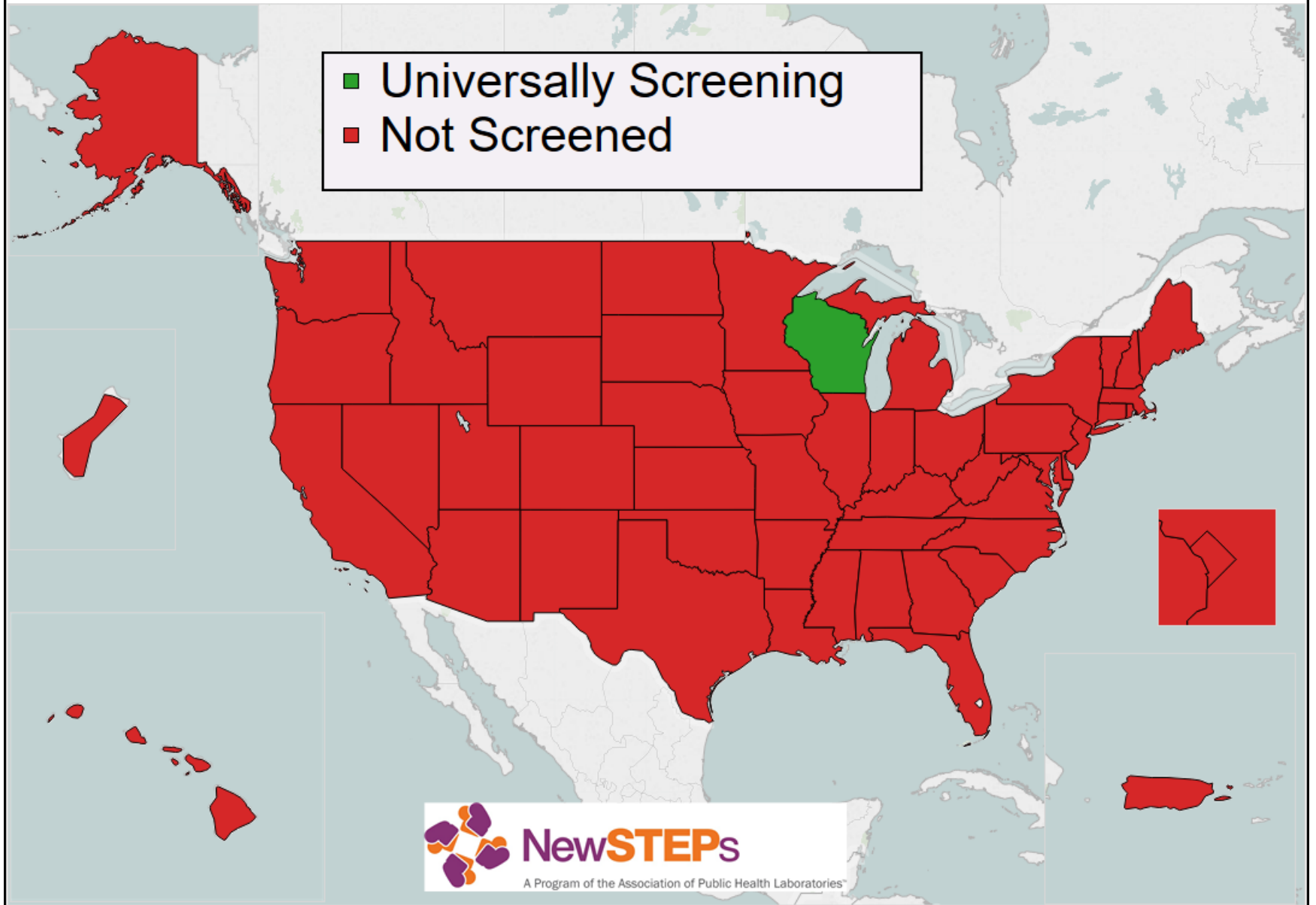


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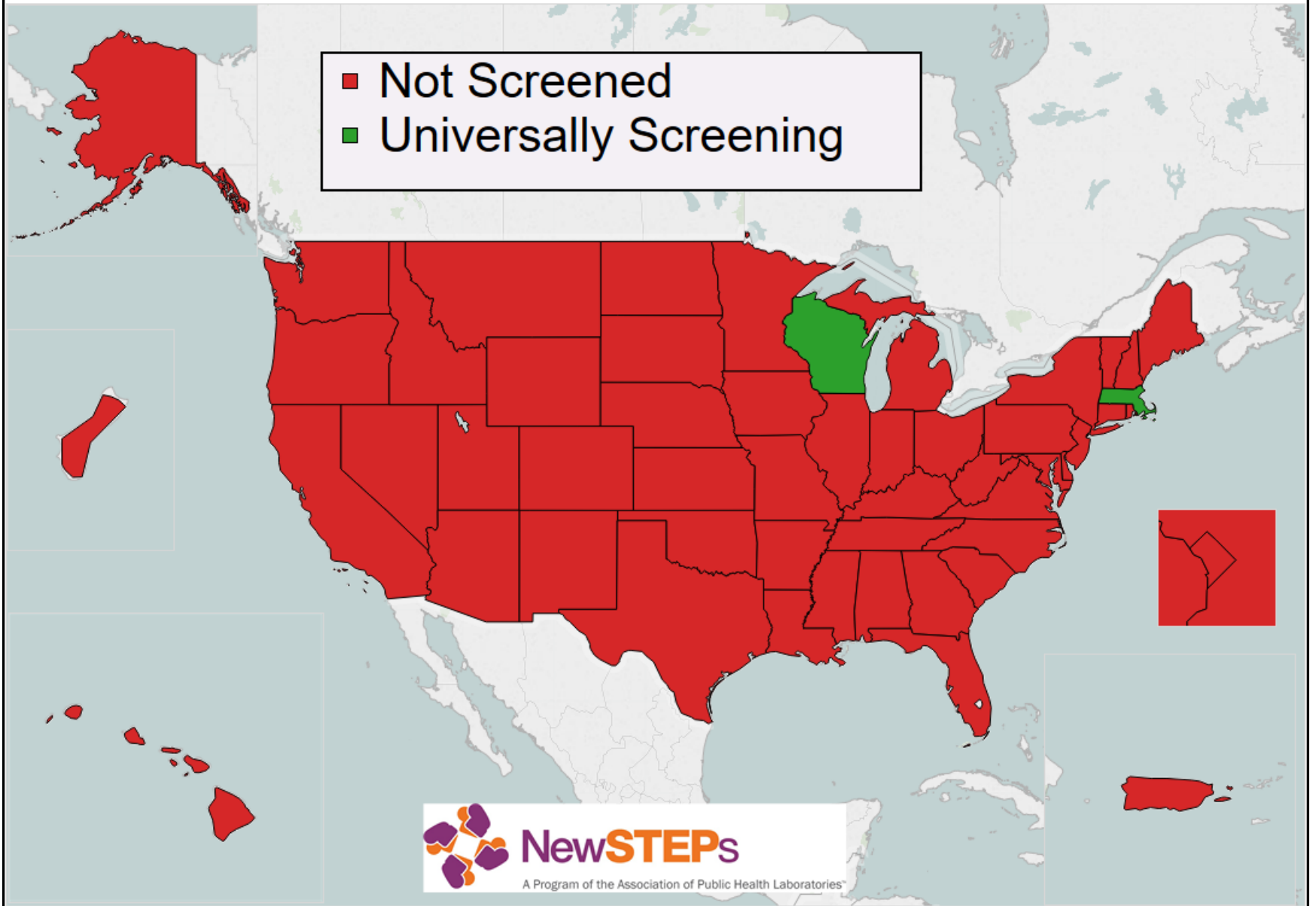
SCID Newborn Screening Status 2008

- Universally Screening
- Not Screened



SCID Newborn Screening Status 2009

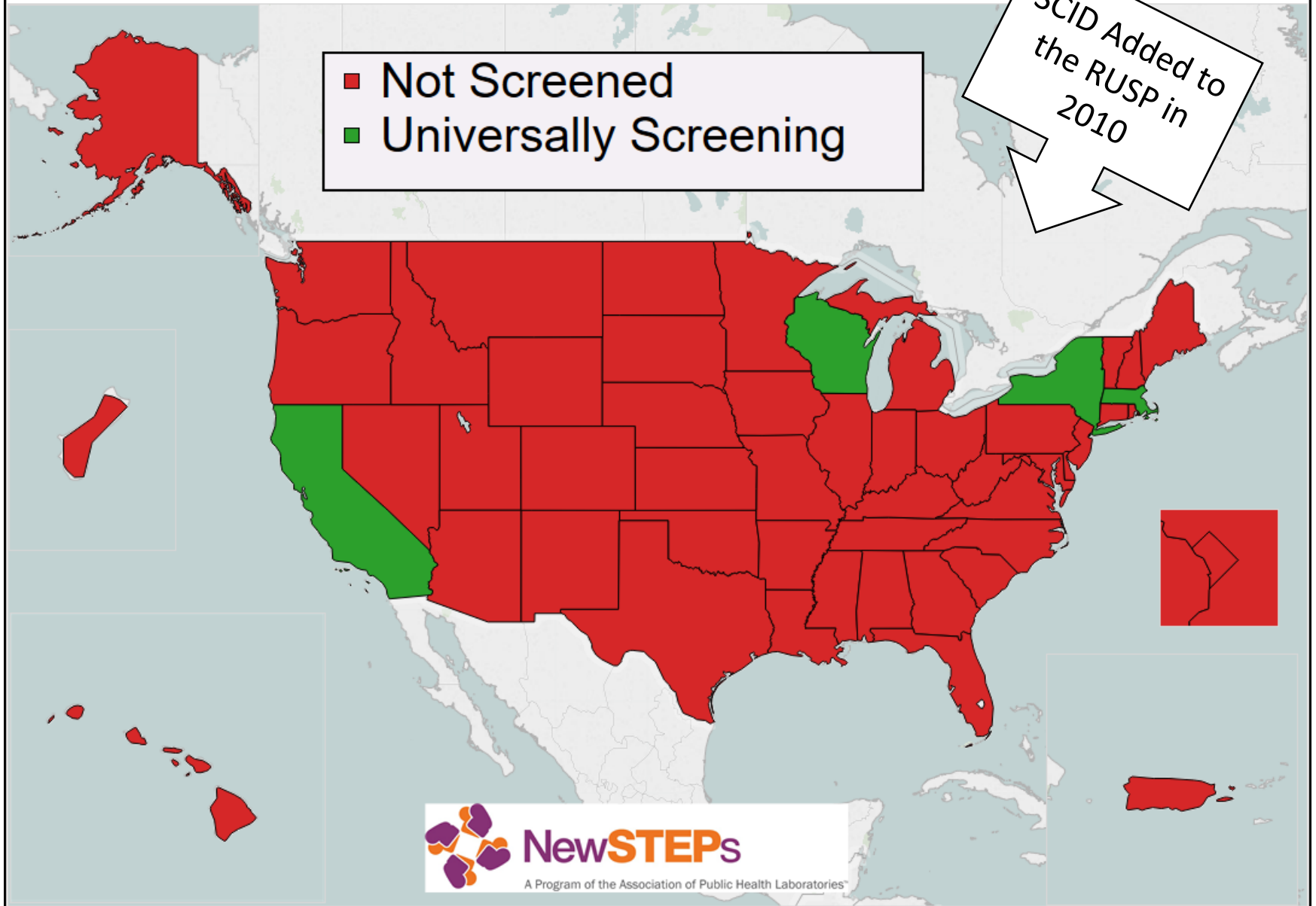
- Not Screened
- Universally Screening



SCID Newborn Screening Status 2010

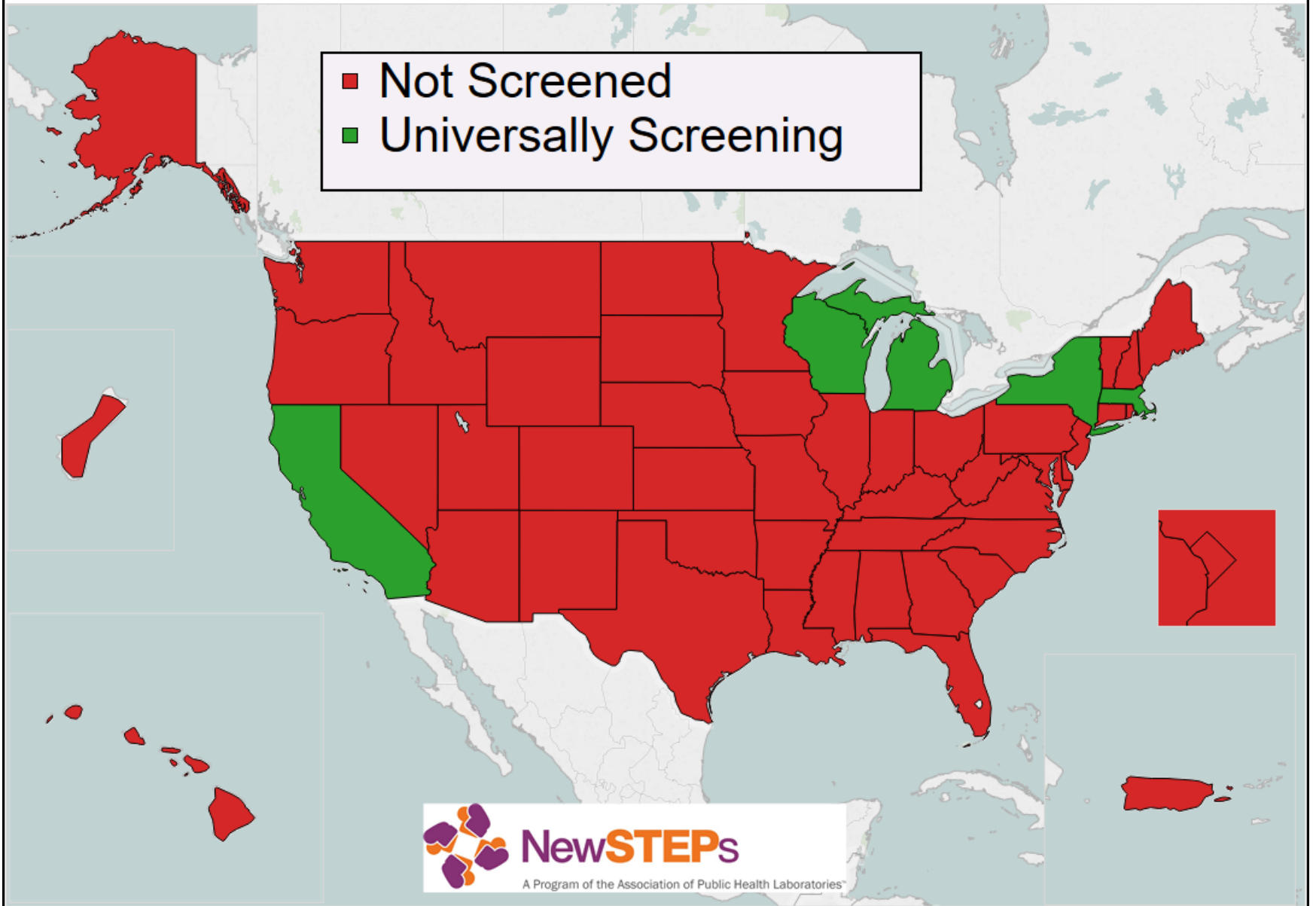
- Not Screened
- Universally Screening

SCID Added to
the RUSP in
2010



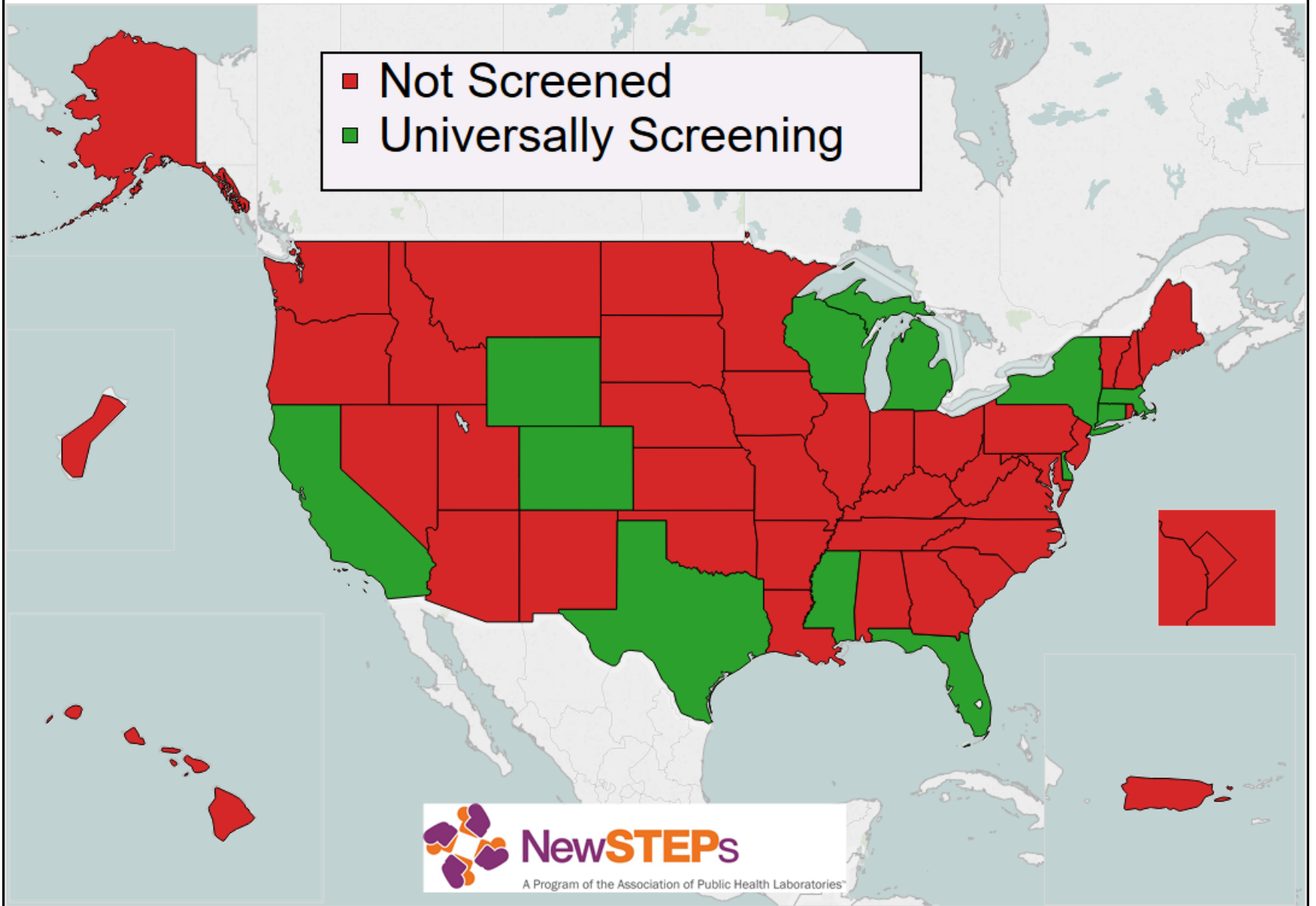
SCID Newborn Screening Status 2011

- Not Screened
- Universally Screening



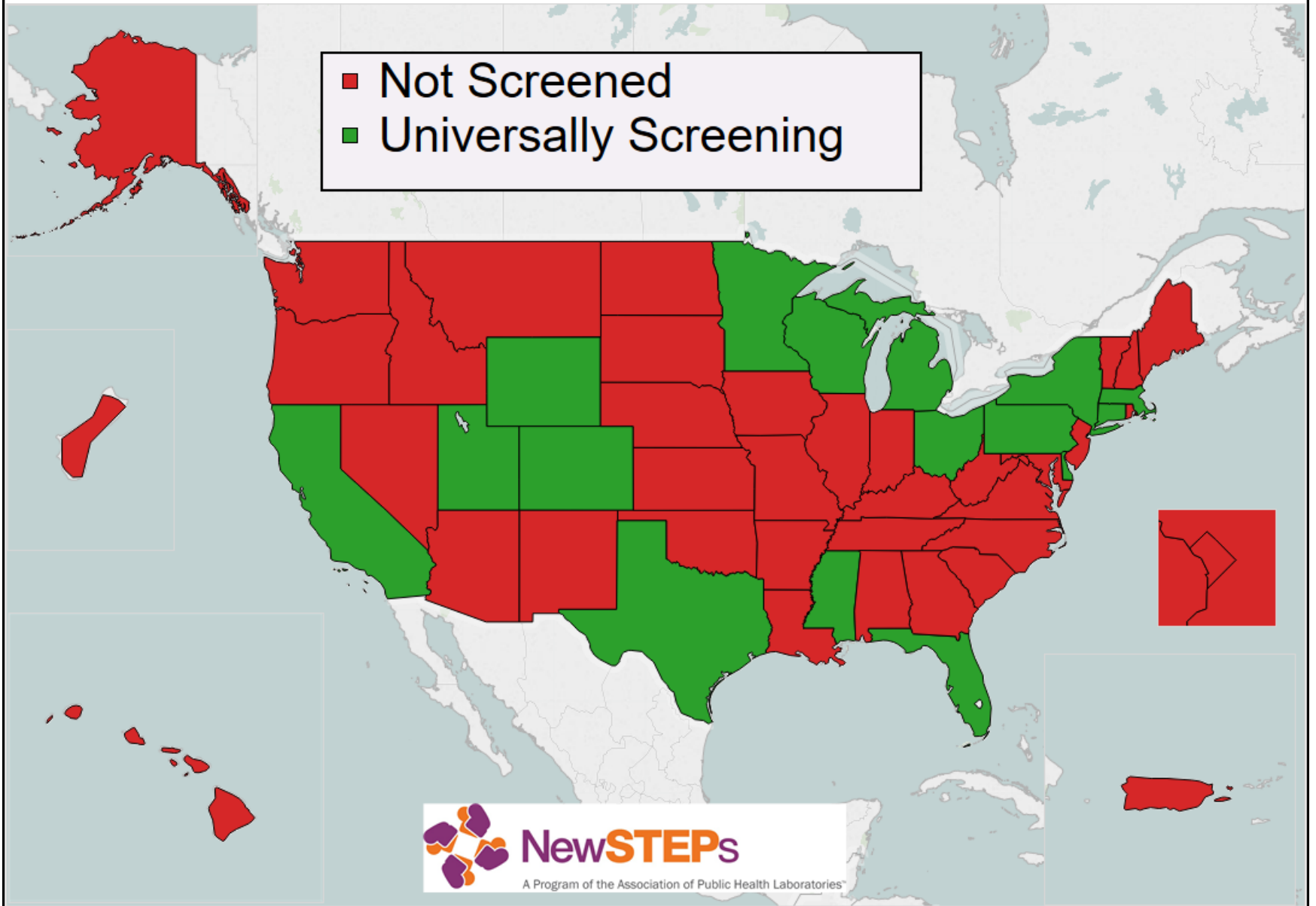
SCID Newborn Screening Status 2012

- Not Screened
- Universally Screening



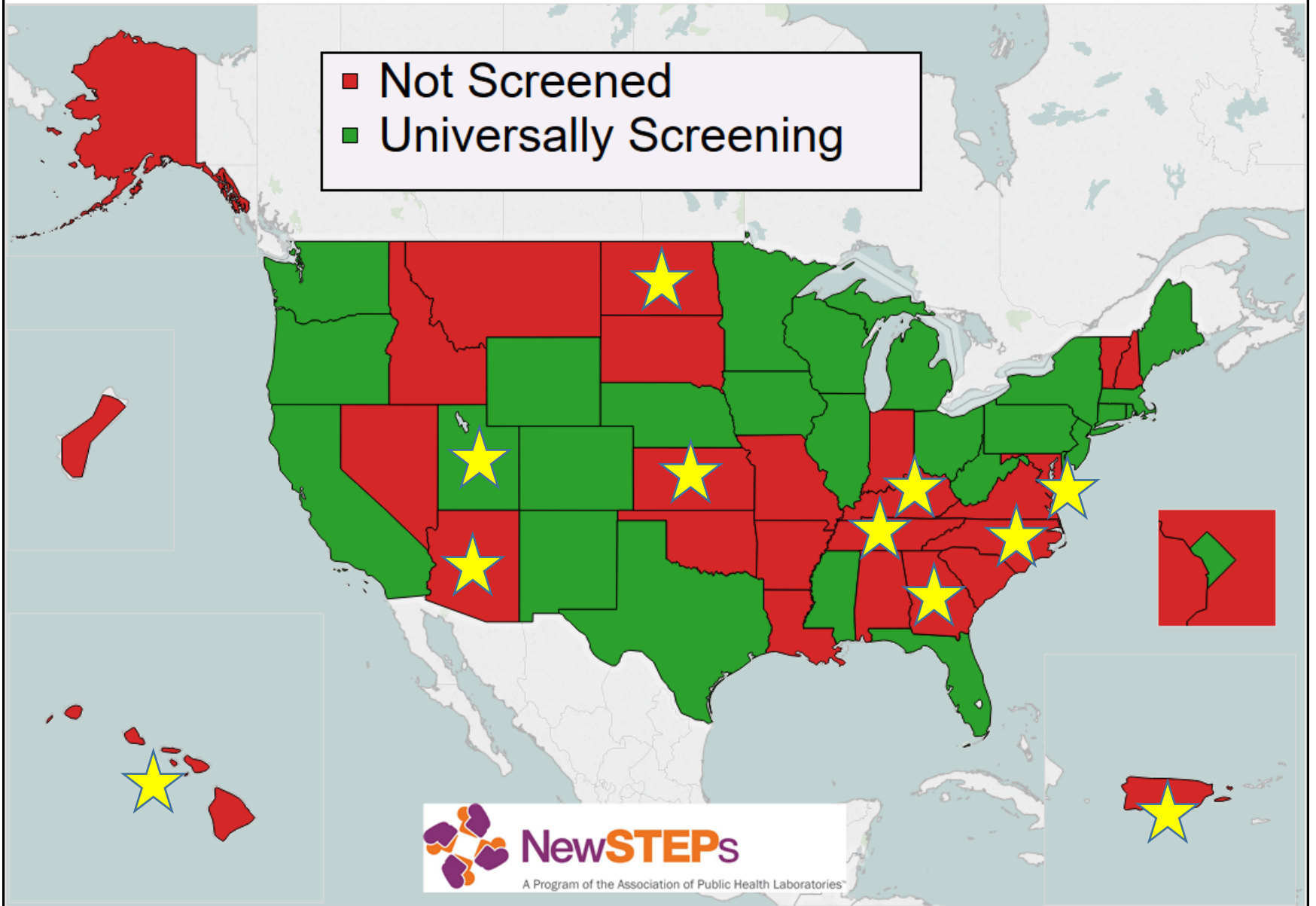
SCID Newborn Screening Status 2013

- Not Screened
- Universally Screening



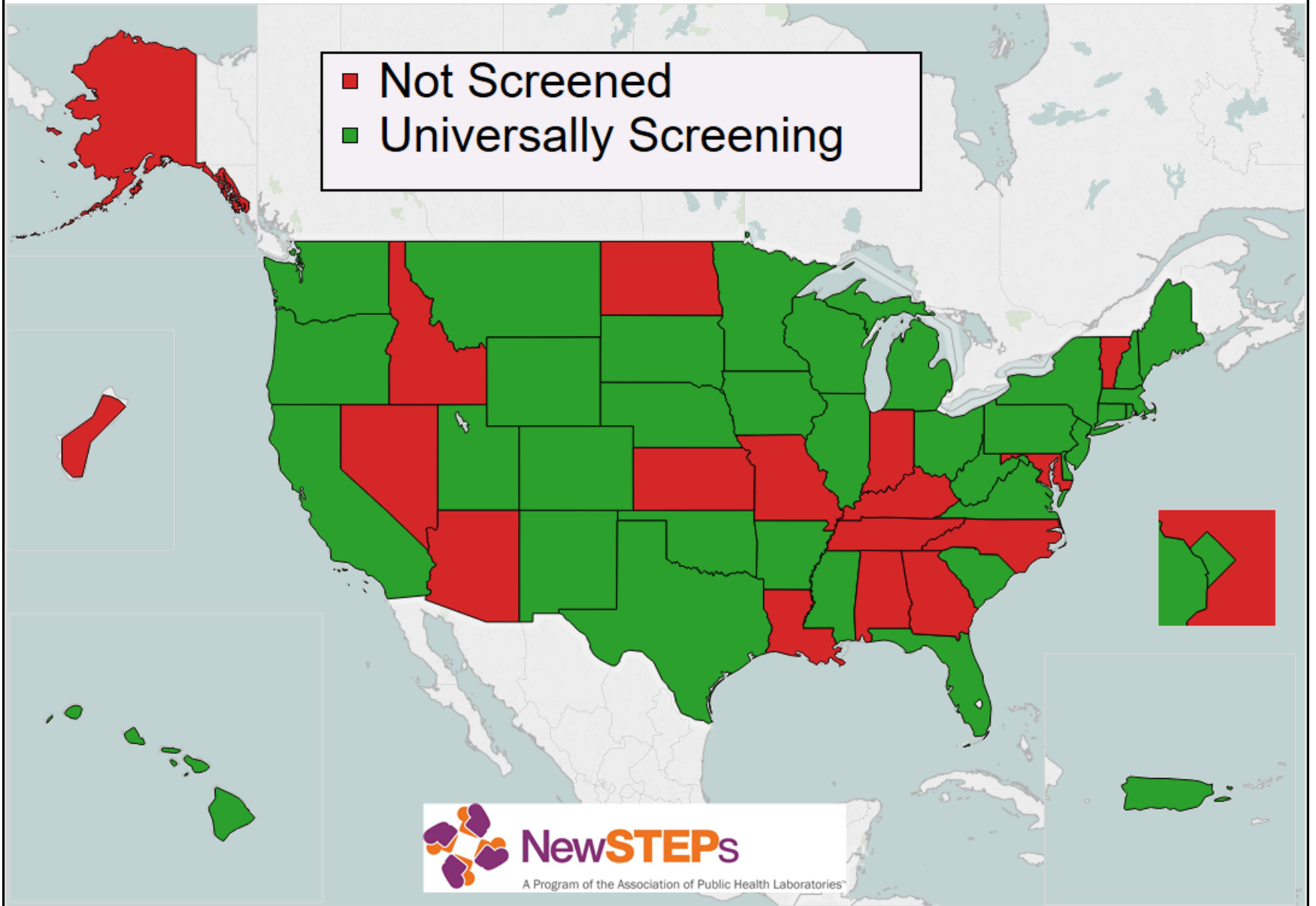
SCID Newborn Screening Status 2014

- Not Screened
- Universally Screening



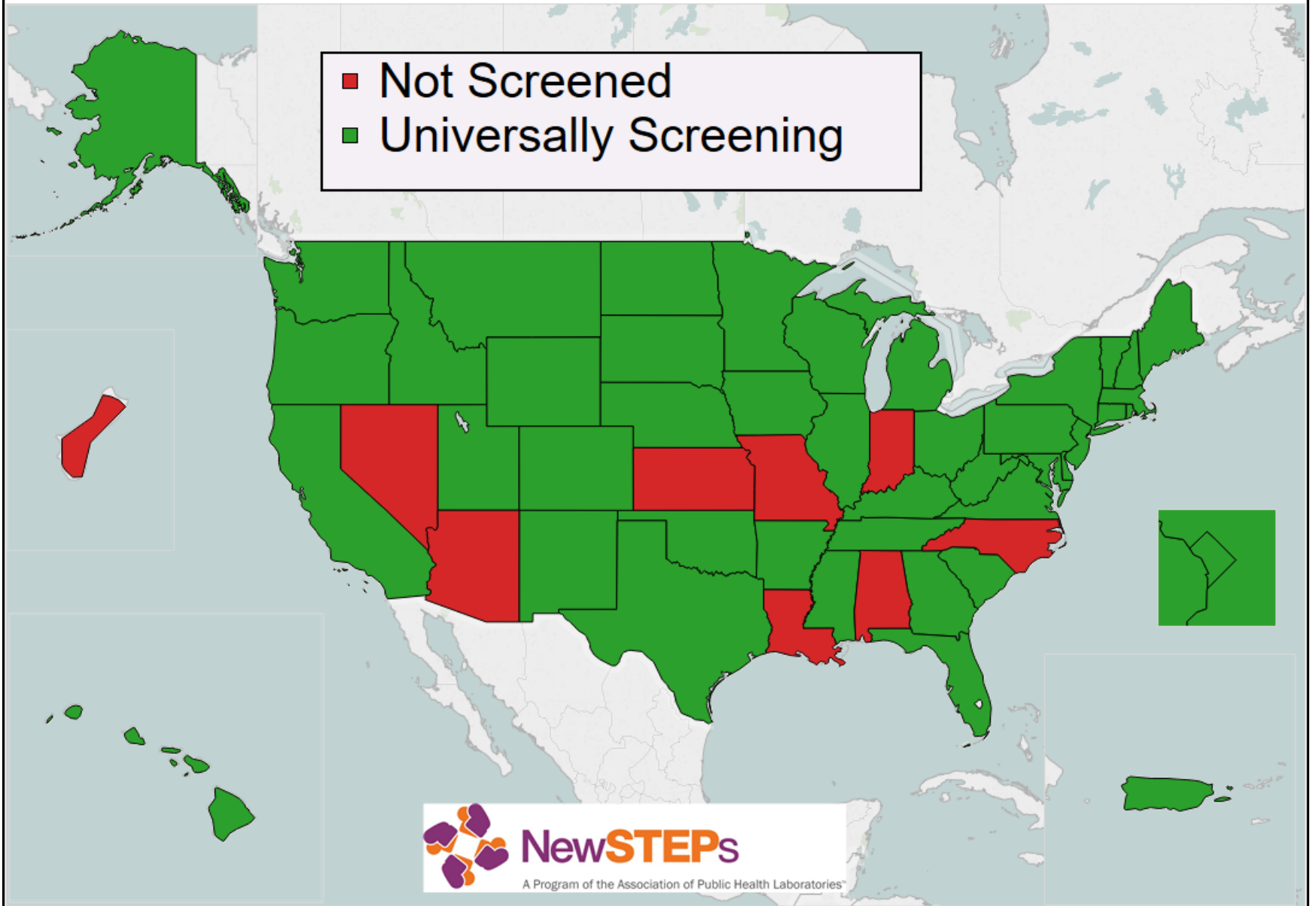
SCID Newborn Screening Status 2015

- Not Screened
- Universally Screening

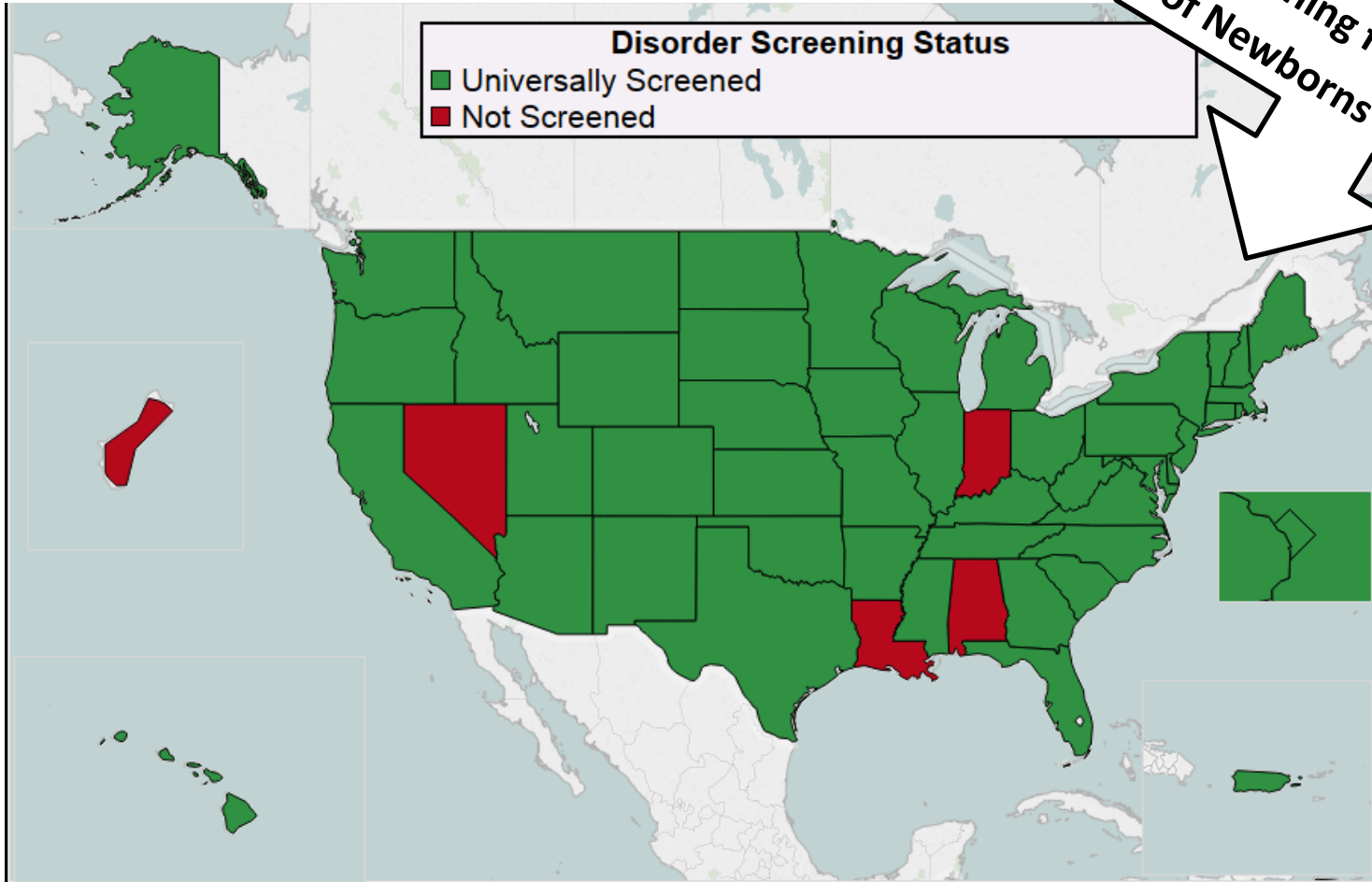


SCID Newborn Screening Status 2016

- Not Screened
- Universally Screening



SCID Screening Status: November 2017



PRESENT DAY:
SCID Universal
Screening for **94%**
of Newborns in the

SCID National Meetings

July 2015

Purpose: Convene state newborn screening programs to discuss **challenges** in and **solutions** to integrating new technology, laboratory, follow-up, personnel, education, and policy needs for SCID.

Participants: Over **100** newborn screening stakeholders representing **laboratory** and **follow-up** programs from over **40** states.

August 2017

Purpose: Convene SCID newborn screening stakeholders to facilitate the **strengthening of relationships** between the SCID **clinical network** and the **newborn screening community** within each state

Participants: Over **107 newborn screening program** staff and **clinicians** representing **38** newborn screening programs.



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Challenges/Barriers/Opportunities



Image source: <https://imgur.com/reRjv>



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Challenges/Barriers/Opportunities

POLICY

- Legislative Mandates
- Fee Increases
- Insurance

FOLLOW-UP

- Interpretation of results
- Relationship between program and clinicians
- Short- and Long- Term Follow-Up

TESTING

- Molecular methodology
- Training staff
- Algorithm variations
- New assays
- Timeliness

EDUCATION

- Providers and parents
- Newborn should receive the rest care possible in as timely a manner as possible
- National guidelines



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Current/Emerging Priorities

Newborn Screening

- Sharing of secondary algorithms
- Educating healthcare providers of NBS process through treatment
- Formal recommendations for long-term follow-up
- Establishment of networks of communications between screeners and pediatric immunology consultants – ongoing conversation

Clinical

- Better guidelines for treatment of asymptomatic patients
- Information shared with parents
- Integration with Infectious Disease specialists
- Published evidence-based articles regarding transfusion

Legislative

- Insurance/policy coverage (treatments, insurance coverage for costs associated with NBS)
- Invite legislative representatives from states to conversations/meetings



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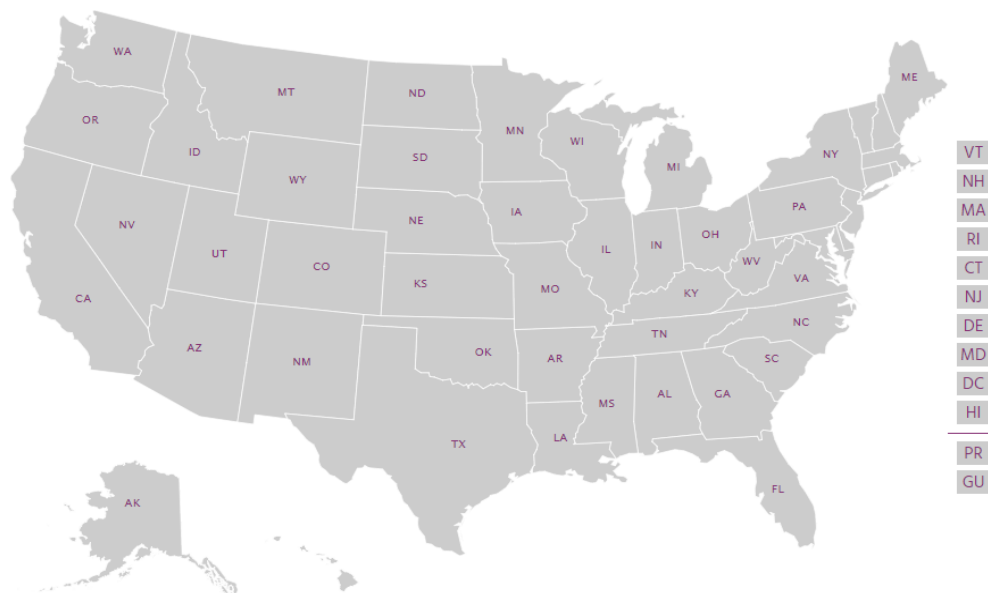
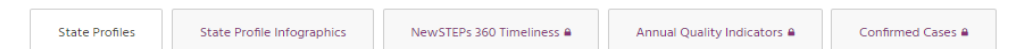
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Contact Us

Data Visualizations

These interactive data dashboards utilize data entered into the NewSTEPs data repository including state profile data, monthly NewSTEPs 360 quality indicator data, annual quality indicator data and case data. These are currently being developed and the list will continue to grow incrementally.

State profile infographics are public facing and accessible to anyone. Infographics for monthly NewSTEPs 360 quality indicator data, annual quality indicator data and case level data are de-identified and can only be accessed using a secure login obtained separately from your general NewSTEPs user account. Infographic user accounts are currently limited. Please contact [Joshua Miller](#) or [Careema Yusuf](#) if you are interested in obtaining an infographic user account, or if you have any questions or suggestions concerning any current or future data dashboards. Enjoy!



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