U.S. Department of Health and Human Services
Health Resources and Services Administration

REPORT TO CONGRESS

Newborn Screening Activities
Fiscal Year 2021 and Fiscal Year 2022

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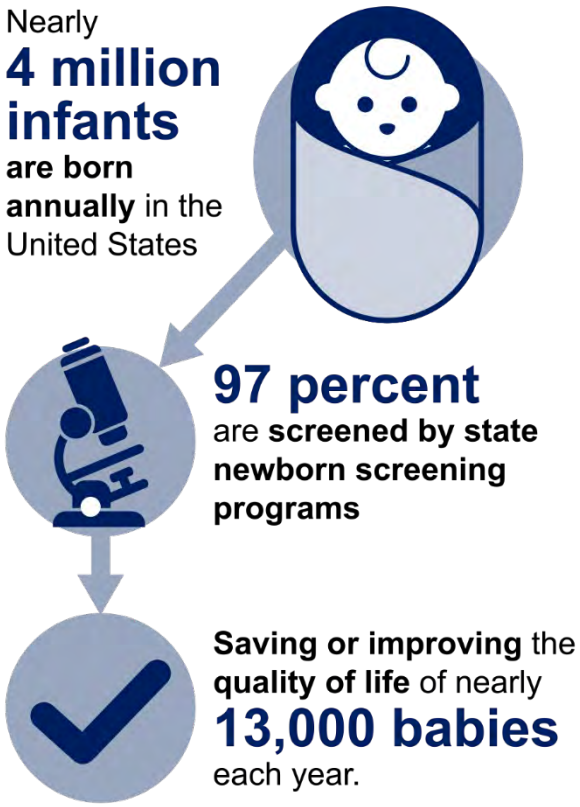
Executive Summary

Newborn screening is a vital public health program that identifies newborns with conditions that may not be apparent at birth but that require immediate intervention to prevent or mitigate permanent disability or death. The newborn screening system comprises multiple components that work in a coordinated and efficient manner in every state. The system includes hospital staff, midwives, and other clinical personnel conducting the screen; laboratory testing; and the reporting of results to appropriate medical personnel who confirm or rule out a diagnosis and initiate treatment, if needed.[1]

The number of conditions screened and screening practices vary by state. The Recommended Uniform Screening Panel (RUSP) is a list of disorders (conditions) adopted by the Secretary of Health and Human Services and recommended to states to be included in newborn screening. Conditions listed on the RUSP are part of the comprehensive preventive health guidelines supported by the Health Resources and Services Administration (HRSA) for infants and children that section 2713 of the Public Health Service Act, 42 U.S.C. § 300gg-13(a)(3) requires to be covered without cost-sharing by certain health insurance plans. (See Appendix A for information on the RUSP and Appendix B for information on state screening practices.)

Figure 1: Newborn Screening Data



Source: Newborn Screening Technical assistance and Evaluation Program (NewSTEPs), *www.newsteps.org*

Federal agencies, including HRSA and the Centers for Disease Control and Prevention (CDC), provide support to newborn screening programs and the newborn screening community to ensure accurate and timely screening, diagnosis, and treatment. Section 11(b) of the Newborn Screening Saves Lives Reauthorization Act of 2014 requires this report, which provides information on activities authorized by Sections 1109, 1110, and 1112 through 1115 of Title XI of the Public Health Service Act and builds on previous reports covering fiscal years (FY) 2015 through 2020.

The programs authorized by the Public Health Service Act support activities that enhance, improve, or expand the ability of state and local public health agencies to provide screening,

[1] American College of Medical Genetics Newborn Screening Expert Group, "Newborn screening: toward a uniform screening panel and system—executive summary." *Pediatrics* 117, no. 5 Pt 2 (May 2006): S296–307. doi: 10.1542/peds.2005-2633I.

counseling, and health care services to newborns and children with or at risk for heritable conditions. Highlights from FYs 2021 and 2022 activities include:

- **HRSA-funded Newborn Screening Data Repository and Technical Assistance:**[2] Forty-nine newborn screening programs in states and territories representing 99 percent of births in the United States signed memoranda of understanding to participate in and contribute data to the data repository. In addition, the program provided technical assistance, training, and national webinars to 53 newborn screening programs across all 50 states as well as in the District of Columbia, Puerto Rico, and Guam.
- **Enhanced quality of newborn screening:** The HRSA-funded Quality Improvement in Newborn Screening Program focused on helping newborn screening programs improve several key newborn screening processes: accurate and timely screening, follow-up, communication of out-of-range results to providers and families, and diagnosis confirmation. The program funded 21 states and territories and 28 project teams for formal quality improvement projects as of May 2022. The program has funded 34 project teams thus far across various newborn screening programs.
- **Funding for states to implement screening for new disorders**: CDC supported five state programs (Iowa, Michigan, New York, North Carolina, and Texas) with cooperative agreement funding. Funded states added new tests to their state panels, enhanced screening performance for improved timeliness, improved test sensitivity for reduced false positives, and improved capability to quickly and accurately interpret screening results.
- **An increase in the total number of states screening for recently added conditions**: Support from HRSA and CDC helped increase the total number of states screening for recently added conditions. Forty-seven states fully implemented universal screening for Spinal Muscular Atrophy (added to the RUSP in 2018).
- **New and improved screening methods:** CDC developed new and improved methods for newborn screenings and provided over 1,600 hours of molecular and biochemical technical assistance to state newborn screening laboratories. This helped laboratories decrease false positives, improve efficiency, and identify newborns with serious conditions early.

[2] Association of Public Health Laboratories. "Newborn Screening Technical assistance and Evaluation Program." Accessed January 4, 2023. https://www.newsteps.org/.

Table 1: Total Number of States Screening for Recently Added Conditions

Condition	Year added to the RUSP	Newborn screening programs offering universal screening – FY 2020	Newborn screening programs offering universal screening – FY 2021	Newborn screening programs offering universal screening – FY 2022
Pompe disease	2015	25	28	36
X-ALD	2016	20	22	28
MPS I	2016	23	26	35
SMA	2018	27	34	47
MPS II	2022	2	2	2

Source: NewSTEPs, *https://www.newsteps.org/*, as of January 2023; NewSTEPs 2020 Annual Report for FY 2021 data (ad hoc request to Association of Public Health Laboratories in September 2021 included updated data), https://www.newsteps.org/resources/newsteps-annual-report. Additional note: the two states screening for MPS II were screening prior to RUSP adoption in 2022.

COVID-19 Impact: Many of the information needs and staffing and laboratory supply shortages created by the COVID-19 pandemic continued into FYs 2021 and 2022. To address these challenges, many newborn screening programs continued to offer extensive virtual services, such as virtual training programs, telehealth services, and educational webinar series. As limited travel resumed in late 2021 and 2022, some events, like industry gatherings and annual meetings, were held in hybrid environments.

CDC assessed the impact of the COVID-19 pandemic on timeliness and receipt of newborn hearing screening and subsequent diagnostic evaluation in four states. In the first year of the pandemic, each participating state reported a decline in the receipt of infant audiological evaluation services and a longer time between not passing the hearing screen and completing the audiological evaluation. Findings from this study will help inform state hearing screening programs should a major public health event occur again.

The newborn screening programs and activities administered by HRSA and CDC help to ensure accurate and timely screening of infants born in the United States and that those identified receive early intervention to achieve the best possible health outcomes. HRSA and CDC are committed to ensuring the early identification, sharing, and implementation of best practices to improve the health of all infants and children in the United States.

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Acronym List

ACHDNC	Advisory Committee on Heritable Disorders in Newborns and Children
APHL	Association of Public Health Laboratories
CDC	Centers for Disease Control and Prevention
CQI	continuous quality improvement
FY	fiscal year
HRSA	Health Resources and Services Administration
ICC	Interagency Coordinating Committee
INBSI	Innovations in Newborn Screening Interoperability
ISO	International Organization for Standardization
MGPN	Minority Genetics Professionals Network
MPS I	Mucopolysaccharidosis I
MPS II	Mucopolysaccharidosis II
NBSIC	Newborn Screening Information Center
NewSTEPs	Newborn Screening Technical Assistance and Evaluation Program
PHS Act	Public Health Service Act
RGN	Regional Genetics Network
RUSP	Recommended Uniform Screening Panel
SCID	Severe Combined Immunodeficiency
SMA	Spinal Muscular Atrophy
X-ALD	X-linked Adrenoleukodystrophy

Legislative Language

Section 11(b) of the Newborn Screening Saves Lives Reauthorization Act of 2014 [P.L. 113-240], which enacted 42 U.S.C. 300b-17, requires that:

> *(b) REPORT BY SECRETARY.— (1) IN GENERAL.—The Secretary of Health and Human Services shall— (A) not later than 1 year after the date of enactment of this Act, submit to the Committee on Health, Education, Labor, and Pensions of the Senate and the Committee on Energy and Commerce of the House of Representatives a report on activities related to— (i) newborn screening; and (ii) screening children who have or are at risk for heritable disorders; and (B) not less than every 2 years, submit to such committees an updated version of such report. (2) CONTENTS.—The report submitted under this subsection shall contain a description of—(A) the ongoing activities under sections 1109, 1110, and 1112 through 1115 of the Public Health Service Act; and (B) the amounts expended on such activities.*

Introduction and Overview

This report discusses the newborn screening activities and associated expenditures of funds for activities authorized by the Newborn Screening Saves Lives Act of 2007 (P.L. 110-204), as amended. This report is prepared pursuant to 42 U.S.C. 300b-17, which requires a biennial report on activities conducted under Sections 1109, 1110, and 1112 through 1115 of Title XI of the Public Health Service (PHS) Act (42 U.S.C. §§ 300b-8, 300b-9, and 300b-11 through 300b-14).

The Health Resources and Services Administration (HRSA) and the Centers for Disease Control and Prevention (CDC) administer these sections of the PHS Act:

- **Section 1109 - Improved Newborn Screening for Heritable Disorders**: Authorizes grants to enhance, improve, or expand the ability of state and local public health agencies to provide screening, counseling, or health care services to newborns and children with or at risk for heritable disorders. HRSA administers this section.
- **Section 1110 - Evaluating the Effectiveness of Newborn and Child Screening and Follow-Up Programs**: Authorizes grants for demonstration programs that evaluate the effectiveness of screening, follow-up, counseling, or health care services in reducing newborn and child morbidity and mortality caused by heritable disorders. HRSA administers this section.[3]
- **Section 1112 - Clearinghouse of Newborn Screening Information**: Authorizes a central, web-based clearinghouse of current newborn screening educational and family support and services information, materials, resources, research, and data. HRSA administers this section.
- **Section 1113 - Laboratory Quality and Surveillance**: Authorizes the provision of quality assurance for laboratories involved in screening newborns and children for heritable disorders (conditions). These activities include quality assurance for conducting newborn screening tests, timeliness of processing such tests, performance evaluation

[3] CDC and HRSA are both authorized to administer programs under this section. CDC did not administer any programs under this section during the time period for this report.

services, technical assistance, technology transfer to newborn screening laboratories to ensure analytic validity and utility of screening tests, and appropriate quality control and other performance test materials to evaluate the performance of new screening tools. This section also authorizes the coordination of laboratory surveillance activities. Surveillance activities include standardizing data collection and reporting, using electronic health records, and promoting newborn screening data sharing with state-based programs related to birth defects and developmental disabilities monitoring. CDC administers this section.

- **Section 1114 - Interagency Coordinating Committee on Newborn and Child Screening**: Authorizes the Interagency Coordinating Committee (ICC) on Newborn and Child Screening to assess existing activities and infrastructure to make recommendations for programs to collect, analyze, and make available data on the heritable disorders recommended by the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) under Section 1111 of the PHS Act. The ICC includes representatives from CDC, HRSA, the Agency for Healthcare Research and Quality, the Food and Drug Administration, and the National Institutes of Health. HRSA and CDC administer the ICC.
- **Section 1115 - National Contingency Plan for Newborn Screening**: Authorizes the development of a national contingency plan for newborn screening for use by a state, region, or consortium of states in the event of a public health emergency. CDC administers the Newborn Screening Contingency Plan.

This is the fifth report on newborn screening activities administered by HRSA and CDC. The report covers activities in fiscal years (FY) 2021 and 2022.

Part I: Improved Newborn and Child Screening for Heritable Disorders (Sec. 1109)

Newborn screening is a public health program that identifies newborns with critical conditions that may be asymptomatic at birth but where early detection and treatment can prevent permanent disability or death.[4] Newborn screening is a complex process involving birthing centers, health care providers, public health programs, and families. The process includes timely and accurate collection of a blood spot sample, transport to a screening laboratory, laboratory testing, identification of out-of-range results, communication with providers and families, confirmation of a diagnosis, and initiation of treatment.

Under **Section 1109** of the PHS Act, HRSA administers programs to help increase the number of newborns screened for heritable disorders. HRSA funding works to:

- Improve the ability of state and local public health agencies to provide screening, counseling, and health care services to newborns and children with these disorders.
- Provide education and training programs about newborn screening counseling, testing, follow-up, treatment, and specialty services for newborn screening stakeholders, including health care professionals, laboratory personnel, parents, families, and support groups.

[4] Health Resources and Services Administration. "About Newborn Screening." Accessed November 17, 2022. https://newbornscreening.hrsa.gov/about-newborn-screening.

- Establish a system to assess and coordinate follow-up and treatment related to congenital,[5] genetic,[6] and metabolic conditions.[7]
- Improve the timeliness of newborn screening from specimen collection[8] through diagnosis.

Newborn Screening Data Repository and Technical Assistance Program

Since 2012, HRSA has funded the Newborn Screening Data Repository and Technical Assistance Program to provide technical assistance to state and local public health agencies, health care professionals, newborn screening laboratory personnel, and other newborn screening stakeholders for the implementation of state-based public health newborn screening. The Association of Public Health Laboratories (APHL) is funded to lead the Newborn Screening Technical assistance and Evaluation Program (NewSTEPs). With this funding, NewSTEPs standardized national newborn screening quality indicators that state screening programs use to analyze their systems' performance and identifies areas in need of improvement.

The NewSTEPs Data Repository collects quality indicator data on newborn screening to support quality improvement initiatives and provide comparative data at the state, regional, and national levels. State newborn screening programs that voluntarily enter quality indicator data into the repository receive access to their own data as well as de-identified, aggregate data from other participating programs. Newborn screening programs use this information to increase their timeliness in screening practices, allowing for earlier diagnosis and intervention for infants with time-critical and non-time-critical disorders.[9]

FYs 2021 AND 2022 UPDATE

During FYs 2021 and 2022, the program:

- Acquired memoranda of understanding from 49 newborn screening programs, representing states, territories, and jurisdictions in which 99 percent of babies are born. These memoranda of understanding served to outline the data security and data sharing parameters inherent within the data repository and delineate data ownership.

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[5] A congenital condition is defined as "structural or functional abnormalities present at birth that can cause physical disability, intellectual and developmental disability, and other health problems. Some may be fatal, especially if not detected and treated early." (See: National Institutes of Health. Eunice Kennedy Shriver National Institute of Child Health and Human Development. "About Birth Defects." 2019. https://www.nichd.nih.gov/health/topics/birthdefects/about.)

[6] A genetic condition is defined as "a condition caused by an error in a gene." This may also be referred to as a hereditary disease or an inherited disorder. (See: Health Resources and Services Administration. Newborn Screening. "Glossary." Accessed November 17, 2022. https://newbornscreening.hrsa.gov/about-newborn-screening/glossary.)

[7] A metabolic condition or disorder constitutes a change to the body's metabolism (the body's process of getting energy from food), making it more difficult for the body to make energy and get rid of toxins. (See: Health Resources and Services Administration. Newborn Screening. "Glossary." Accessed November 17, 2022. https://newbornscreening.hrsa.gov/about-newborn-screening/glossary.)

[8] Specimen collection in newborn screening is defined as when a few drops of blood are obtained from a heel stick within 24 to 48 hours of a child's birth. These blood spots are sent to a laboratory, usually at the state or territorial public health department, for testing. (See: Centers for Disease Control and Prevention. "Newborn Screening Laboratory Bulletin." 2019. https://www.cdc.gov/nbslabbulletin/bulletin.html.)

[9] Association of Public Health Laboratories. "NewSTEPs 2020 Annual Report." September 2021. https://www.newsteps.org/sites/default/files/resources/download/NewSTEPs%20Annual%20Report%209%2022%2021.pdf.

- Facilitated continuous quality improvement (CQI) and data-driven outcome assessments in the newborn screening system by providing a standardized repository (NewSTEPs,[10] a national, secure, and central web-based database) and by supporting the integration of health information infrastructure and systems into newborn screening.
    - As of September 30, 2022:
        - There were 1,022 registered NewSTEPs data repository users.
        - 46 states entered 40,965 case definitions (common classifications for diagnoses across programs),[11] with 52 programs providing aggregate cases.
        - 48 states entered state-level quality indicator data,[12] enabling comparisons within and across programs, in aggregate.
    - NewSTEPs data included screening status data for all disorders on the RUSP, with data visualization updated weekly.[13]
    - There were 634 toolkits, webinars, model practices, videos, policy statements, publications, educational tools, reports, data visualizations, and presentations uploaded on the NewSTEPs resource library.[14] Webinars and other newborn screening resources are filterable by disorder.
    - State profile data reports are publicly available in real time.[15]
    - The 2020 NewSTEPs annual report is available online.[16]
- Reviewed all NewSTEPs quality indicator data elements and reached consensus regarding quality indicator recommendations and modifications suggested by newborn screening programs to ensure program participation in the NewSTEPs Data Repository.
- Provided and maintained discussion forums for newborn screening stakeholders to inform each other of quality improvement, implementation, and data collection.
    - More than 850 newborn screening community members participated in the NewSTEPs Collaborate Community listserv across more than 1,900 discussion posts.
    - More than 210 community members participated in the Newborn Screening Health Information Technology listserv across 75 discussion posts.
    - NewSTEPs hosted more than 12 routine, community-driven topical webinars, which were all recorded and archived on www.newsteps.org. Webinars included the following topics:
        - New condition implementation
        - Health Information Technology
        - Critical Congenital Heart Disease Screening and Follow-up

[10] Association of Public Health Laboratories. "Newborn Screening Technical Assistance and Evaluation Program." Accessed December 7, 2022. https://www.newsteps.org/.
[11] *Ibid*.
[12] *International Journal of Neonatal Screening.* "Development of National Newborn Screening Quality Indicators in the United States." September 2019. https://www.mdpi.com/2409-515X/5/3/34.
[13] Association of Public Health Laboratories. "Dashboards and Reports." Accessed December 7, 2022. https://www.newsteps.org/resources/data-visualizations?q=data-visualizations&tid=1.
[14] Association of Public Health Laboratories. "Resource Library." Accessed December 7, 2022. https://www.newsteps.org/resources.
[15] Association of Public Health Laboratories. "Reports." Accessed December 7, 2022. https://www.newsteps.org/data-resources/reports?q=data/reports.
[16] See footnote 9.

        - COVID-19
            - In May 2020, APHL and the National Center for Hearing Assessment and Management co-hosted the Newborn Screening COVID-19 Challenges and Response webinar.[17] This webinar consisted of a panel of newborn screening experts discussing challenges, barriers, and solutions to dried blood spot and hearing screening and emphasized family perspective during the pandemic.
- Fostered the integration of screening for new disorders into newborn screening systems through NewSTEPs' New Disorders Workgroup by:
    - Offering state newborn screening programs access to resources and technical assistance.
    - Developing resource tools for Spinal Muscular Atrophy (SMA)[18] and Mucopolysaccharidosis Type II (MPS II)[19] for newborn screening programs to use when implementing screening for new disorders and educating stakeholders.
- Assisted states in adding screening programs for conditions on the RUSP, including direct funding to five state newborn screening programs (South Carolina, Wisconsin, Virginia, Texas, and Montana) for new disorder support.

Part II (Section 1110) lists additional activities and accomplishments for the Newborn Screening Data Repository and Technical Assistance Program during this period.

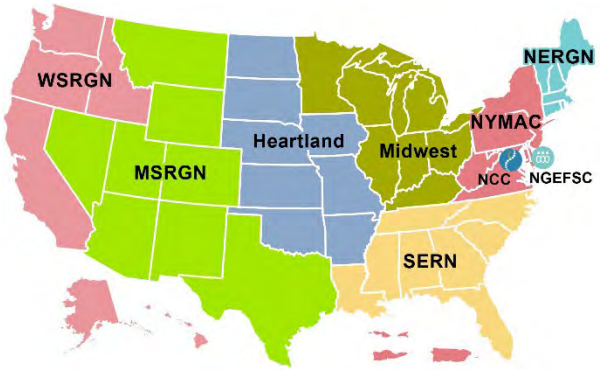
Regional Genetics Networks Program

Through its Regional Genetics Networks (RGN) Program (funded since 2017), HRSA supports seven RGNs, a National Coordinating Center, and a National Genetics Education and Family Support Center. The program focuses on linking patients and families, especially those considered underserved, with clinical genetic services and provides resources to genetic service providers, primary care providers, families, and state public health workers. The seven RGNs are:

- Heartland Regional Genetics Network (Heartland)
- Midwest Genetics Network (Midwest)
- Mountain States Regional Genetics Network (MSRGN)
- New England Regional Genetics Network (NERGN)
- New York Mid-Atlantic Collaborative (NYMAC)
- Southeast Regional Network (SERN)
- Western States Regional Genetics Network (WSRGN)

[17] Association of Public Health Laboratories. "Newborn Screening COVID-19 Challenges and Response Webinar." May 21, 2020. https://www.newsteps.org/resources/webinars-events/newborn-screening-covid-19-challenges-and-response-webinar.
[18] Association of Public Health Laboratories. "Spinal Muscular Atrophy: New Disorder Resources and Tools." June 2021. https://www.aphl.org/aboutAPHL/publications/Documents/NBS-2021-NewSTEPS-SMA-Toolkit.pdf.
[19] Association of Public Health Laboratories. "Mucopolysaccharidosis, Type II: New Disorder Resources and Tools." August 2022. https://www.aphl.org/aboutAPHL/publications/Documents/NBS-NewSTEPS-MPSII-Toolkit.pdf.

Figure 2: Regional Genetics Network System



Source: National Coordinating Center for the Regional Genetics Networks, *https://nccrcg.org/rgns/*.

FYs 2021 AND 2022 UPDATE

During FYs 2021 and 2022, the RGN program served 20,510 individuals and families across the country through education and training, connected parents to genetics services, and facilitated telehealth services, with a focus on aiding medically underserved populations. The RGNs served over 7,600 individuals from medically underserved populations.

By May 2024, HRSA hopes to increase the number of RGN resources accessed by 20 percent. To achieve this objective, the genetic networks have expanded their outreach and education efforts with a focus on underserved communities:

- New York Mid-Atlantic Caribbean RGN established new teams in Puerto Rico and the U.S. Virgin Islands, both of which held their first team meetings in fall 2021 and integrated other HRSA-funded programs, including sickle cell treatment demonstration project groups and Telehealth Resource Centers, into their resource offerings.
- As part of its Access to Care project, Midwest RGN conducted outreach efforts including Community of Practice forums and virtual peer mentoring, to Plain Communities, individuals whose first language is not English, and patients lost to follow-up.
- Heartland RGN established a Marshallese genetics clinic in November 2020.

Many RGNs expanded their education, infrastructure-building, and outreach efforts by strengthening and diversifying their online presence, including:

- RGNs offered education and training to 4,445 health care providers through maintenance of certification, continuing medical education, or continuing education credits.
- Other RGNs began distributing newsletters, including Midwest RGN's bi-monthly e-newsletter, "See What's Unwinding Across the Region" and Southeast RGN's quarterly newsletter, which provides news about telemedicine and regional events. Southeast RGN's first issue was sent to over 488 individuals.
- Western States RGN continued to create, maintain, and update parent fact sheets for genetic disorders online. Their website has over 13,000 visitors per month.[20] So far three fact sheets have been translated into Spanish, while plans for Korean, Ilocano, and Russian are in place and newer disorder fact sheets are being converted into short, animated videos for audiences with visual learning styles.

[20] NewbornScreening.info. "Home." Accessed January 4, 2023. http://www.newbornscreening.info/.

- The Minority Genetics Professionals Network (MGPN), supported by Western States RGN, provided mentorship for students in minority communities interested in becoming genetics professionals. In 2022, MGPN numbered approximately 1,100 members. As part of its mentorship program, MGPN organized its second virtual Genetic Counseling Career Fair in September 2021, with approximately 1,400 students in attendance. Due to its positive response, MGPN plans to hold this fair annually.
- The RGN's National Coordinating Center led a health care interpreter program, which published its interpreter genetics glossary in five new languages (French, Korean, Polish, Portuguese, and Tagalog) and updated the glossary in four languages (Arabic, Chinese, Spanish, and Vietnamese).

Long-Term Follow-Up for Severe Combined Immunodeficiency and Other Newborn Screening Conditions

Severe Combined Immunodeficiency (SCID) is a genetic condition that affects one in 58,000 infants.[21] Infants born with SCID usually die within 1 year due to severe recurrent infections unless screening identifies them early and they undergo successful stem cell transplantation. SCID was added to the RUSP in 2010.

Congress provided funding to HRSA in FY 2014 to better support states in implementing SCID screening.[22] This program successfully led to the implementation of universal SCID screening programs in all 50 states as well as in Puerto Rico, Guam, and the District of Columbia.

Even though there is SCID screening in all states, gaps remain in optimizing outcomes for infants with SCID that are similar to gaps in other newborn screening conditions. The program focused its FY 2021 efforts on long-term follow-up for SCID and other newborn screening conditions to close these gaps and strengthen long-term follow-up models across screening programs. Six grantees worked to develop long-term follow-up systems in their states and institutions.

FYs 2021 AND 2022 UPDATE

During this period, the Long-Term Follow-Up for SCID and Other Newborn Screening Conditions Program focused on the following activities:

- Recruited over 53 clinical sites to conduct long-term follow-up in the program's first year and developed over 70 formal and informal partnerships among primary and specialty care clinicians, state newborn screening and public health programs, families, condition-specific organizations, and relevant community services.
- Improved and expanded the long-term follow-up programs already present in three of the six program awardees by:
    - Adding over seven new conditions to the New York State Department of Health's long-term follow-up patient registries since August 2021.
    - Ensuring that 93 percent of children have a medical home for long-term follow-up through coordination by the North Dakota Department of Health in 2021.

[21] Association of Public Health Laboratories. "Severe Combined Immunodeficiency (SCID)." 2019. https://www.newsteps.org/disorders/scid.

[22] Public Health Service Act, § 1109 (42 U.S.C. 300b-8), as amended by the Newborn Screening Saves Lives Act of 2014 (P.L. 113-240).

- Reducing by 75 percent the number of children and families lost to follow-up in the Connecticut Children's Hospital, from 12 percent in August 2021 to 3 percent in March 2022.

Quality Improvement in Newborn Screening Program

The Quality Improvement in Newborn Screening Program began in FY 2018 and HRSA awarded this to APHL. The program supports state quality improvement activities to address several key components of the newborn screening process: timeliness of newborn screening, identifying and following up on out-of-range results, communicating screening results to providers and families, and confirming diagnoses.

FYs 2021 AND 2022 UPDATE

During this period, the Quality Improvement in Newborn Screening Program:

- Funded 21 states/territories and 28 project teams for formal quality improvement projects as of May 2022. Additionally, six project teams have graduated, successfully completing their improvement projects. HRSA has funded 34 project teams across various newborn screening programs thus far.
  - Programs received CQI training and access to resources, technical assistance, and an online database to evaluate their progress in the focus area of their choice (e.g., timeliness, data entry, and long-term follow-up).
  - All funded and unfunded state and territory programs received access to a centralized APHL collaboration forum called ColLABorate, which allows the newborn screening community to share ideas and best practices related to CQI.
- Funded monthly reports to develop into change strategy documents, which will serve as a tool for other newborn screening programs to replicate successful quality improvement initiatives.

Part II (Section 1110) lists additional activities and accomplishments for the Quality Improvement in Newborn Screening Program during this period.

Newborn Screening Family Education Program

The Newborn Screening Family Education Program, awarded to the Genetic Alliance in 2018, develops and delivers educational programs about a wide range of topics, including newborn screening, counseling, testing, follow-up and treatment, specialty services, and support activities that increase awareness, knowledge, and understanding of newborn screening for parents, families, patient advocacy, and support groups.

FYs 2021 AND 2022 UPDATE

During this period, the Newborn Screening Family Education Program has implemented the following activities:

- Expanded Prenatal Education Efforts to raise awareness and knowledge of newborn screening in prenatal populations (i.e., among all pregnant people). In Year 4, the program successfully completed its second prenatal education pilot program and signed memoranda of understanding with three state newborn screening programs (California, Colorado, and Tennessee) to implement prenatal newborn screening education initiatives in their states.
- Launched the Navigate Newborn Screening Ambassador Program in September 2021.

This program, which includes facilitated quarterly meetings and online training curricula, recruited 12 family leaders representing 10 states to participate in a year-long program to develop leadership skills and increase newborn screening knowledge.

- Developed Spanish-language content for the newborn screening community, including:
  - A translation of the Navigate Newborn Screening online module;
  - A translation of newly developed pilot program materials such as the Navigate Newborn Screening learning book and patient evaluation questions; and
  - New online training curricula, including *Navegando por la Evaluación del Recién Nacido*, an online module for Spanish-speaking families to learn about newborn screening.
- Expanded the program's partnership collaborations, progressing from primarily a network for disseminating program information to collaborative partnerships co-creating, implementing, and providing technical assistance for education initiatives.
- Launched a 6-month social media campaign beginning on March 4, 2022. In its first 60 days, the campaign reached 916,757 individuals from its target audience, including 5,200 visitors who reached the campaign by clicking on its ads.

Newborn Screening State Evaluation Program

Since 2001, HRSA has supported the standardization of national newborn screening quality measures (see the NewSTEPs description on page 4) used by state newborn screening programs to determine how their system is performing on the various components of screening and identify areas in need of improvement. From calendar years 2019 through 2021, the Newborn Screening State Evaluation Program has supported states' efforts to collaborate closely with NewSTEPs to enter quality measure data into the data repository; evaluate their newborn screening system; identify best practices, gaps, and challenges; and develop and implement policies, procedures, and methods to measure their screening performance.

FY 2021 UPDATE

Colorado, Connecticut, Hawaii, New York, Oklahoma, and Rhode Island received funding from HRSA to report data to NewSTEPs. Funding concluded in FY 2021. Despite setbacks due to the COVID-19 pandemic, they achieved the following:

- Reported data to NewSTEPs on at least two quality indicators. Many increased the number of quality indicators they reported to NewSTEPs throughout the funding period.
- Collaborated with other newborn screening stakeholders to assess the newborn screening system for CQI.
- Improved outreach to newborn screening stakeholders by connecting with primary care provider offices. The programs collaborated with stakeholders to facilitate data sharing and assess newborn screening systems for CQI.

Innovations in Newborn Screening Interoperability

In September 2020, HRSA implemented the Innovations in Newborn Screening Interoperability (INBSI) program to enhance data interoperability in the newborn screening system.[23]

Ensuring that every baby is screened, INBSI requires the seamless cooperation and secure transfer of information between hospitals, couriers, state newborn screening labs, follow-up programs, health care providers, and other state databases or national registries. Each entity involved in the newborn screening system must also implement security and privacy policies on how to manage, control, and share data.

Newborn screening systems that can effectively, efficiently, and securely exchange information are more likely to provide timely and accurate screening and lead to early diagnosis, intervention, and treatment. The INBSI program is an important step in achieving these goals.

FYs 2021 AND 2022 UPDATE

During this period, the INBSI program:

- Established and maintained a public-facing Resource Center in July 2021. The website, which has received 9,224 page views from 2,067 unique users as of May 10, 2022, offers:
  - pointers to existing and in-progress newborn screening standards;
  - training materials;
  - announcements and opportunities; and
  - virtual collaborative spaces for INBSI staff and other subject matter experts.
- Completed an environmental scan of interoperability in newborn screening.
- Provided informatics and interoperability training and collaborative learning opportunities.
- Assisted states in forming and training newborn screening interoperability teams, with the goal of developing state-specific implementation plans. INBSI enrolled eight locations from September 2021 to August 2022 (California, the District of Columbia, Indiana, Kentucky, Michigan, Nebraska, Oregon, and Rhode Island) and established eight state interoperability teams.
- Assisted in the development and completion of four individual state interoperability plans (California, the District of Columbia, Kentucky, and Michigan).
- Conducted live monthly educational webinars, which INBSI posted on its INBSI Resource Center. Topics in 2021 and 2022 included:
  - Navigating Change: Change Management and Stakeholder Engagement for Interoperability Efforts in Newborn Screening;
  - Interoperability Stakeholder Engagement and Project Management;
  - How Health Information Exchanges Support Newborn Screening Interoperability;
  - The Future of Newborn Screening: Why Standardization Matters for Interoperability;

[23] The 21st Century Cures Act, Section 4003, defines interoperability with respect to health information technology, as technology that "(A) enables the secure exchange of electronic health information with, and use of electronic health information from, other health information technology without special effort on the part of the user; (B) allows for complete access, exchange, and use of all electronically accessible health information for authorized use under applicable State or Federal law; and (C) does not constitute information blocking as defined in section 3022(a)." (See: Official Website of the Office of the National Coordinator for Health Information Technology. "Interoperability." Accessed January 2, 2023. https://www.healthit.gov/topic/interoperability.)

- Exploring the Unmet Needs; Newborn Screening Data Sharing Uncovered; and
- Hospital Engagement: What Hospitals Need to Engage in Newborn Screening Electronic Data Exchange.

HRSA supported two State Newborn Screening Interoperability Implementation Program grantees in 2021 – the Florida Department of Health and Utah Department of Health and Human Services. This program supported state newborn screening programs in implementing comprehensive data interoperability plans.

- As of May 2022, Florida and Utah both set up steering committees (with 21 and eight reported committee members, respectively), and Florida trained 38 staff members in informatics.
- By August 2022, both Florida and Utah completed a draft of an interoperability plan with a proposed timeline to link with another state public health database.

Part II: Evaluating the Effectiveness of Newborn and Child Screening and Follow-Up Programs (Sec. 1110)

HRSA administers **Section 1110** of the PHS Act, which focuses on evaluating the effectiveness of newborn screening and follow-up programs. Several programs previously described in Part I, including the Newborn Screening Data Repository and Technical Assistance Program and the Quality Improvements in Newborn Screening Program, address these issues. Part II of this report highlights key accomplishments of these programs in FYs 2021 and 2022 as they relate to Section 1110.

Newborn Screening Data Repository and Technical Assistance Program

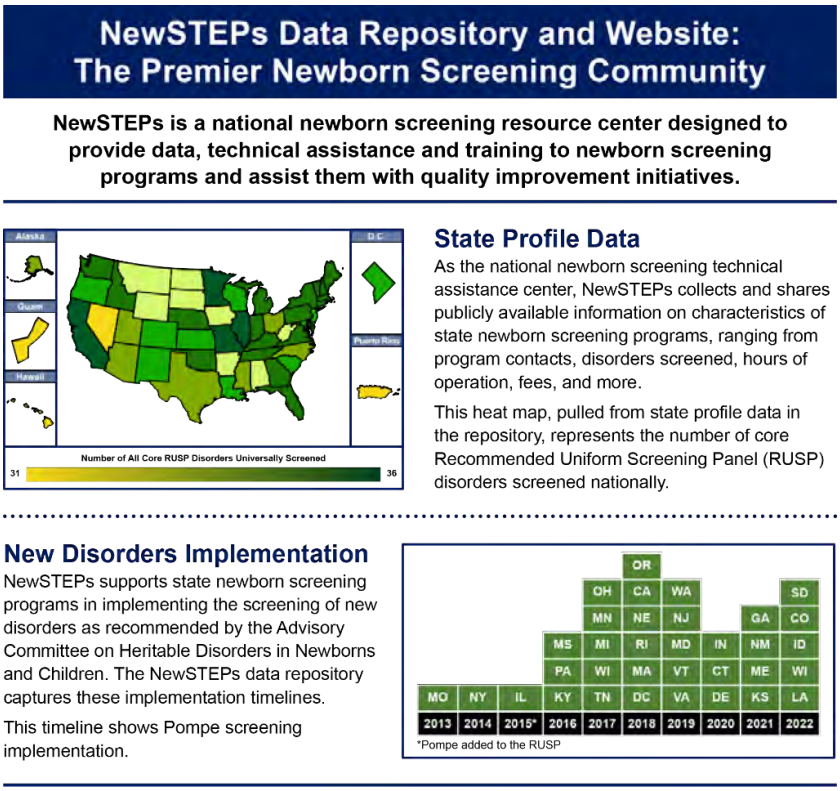
Through the Newborn Screening Data Repository and Technical Assistance Program, NewSTEPs provides resources to state newborn screening programs to evaluate the effectiveness of their processes and use real-time data to inform quality improvements.

KEY ACCOMPLISHMENTS

- NewSTEPs initiated a comprehensive site review[24] of Florida's newborn screening programs in June 2022. NewSTEPs selected a site review team of experts, and initial discussions have begun with the Florida laboratory and follow-up programs to help them address continuity of operations needs and identify newborn screening system process improvements.
- NewSTEPs provides public access to real-time reports and infographics from the data repository (See Figure 3):

[24] Association of Public Health Laboratories. "Site Review." Accessed December 7, 2022. https://www.newsteps.org/quality-improvement-practices/site-review.

Figure 3: NewSTEPs Data Repository



Source: NewSTEPs, https://www.newsteps.org/, as of January 2023.

Quality Improvement in Newborn Screening Program

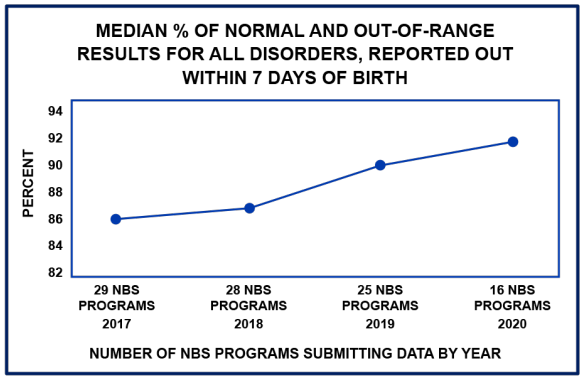
As discussed in Part I, page 11, in FYs 2021 and 2022, this program enhanced the quality of newborn screening across 34 states and territories, focusing on timeliness, detection of out-of-range results, reporting out-of-range results to providers, confirming a diagnosis, and addressing emerging issues or any other newborn screening process or procedure that could negatively affect the quality, accuracy, or timeliness of newborn screening.

KEY ACCOMPLISHMENTS

- APHL announced the fourth cohort of the Quality Improvement Projects collaborative in September 2021, and funded projects began in February 2022. APHL selected 10 states to participate in this cohort via a Request for Proposals process, with focus areas including newborn screening timeliness, implementation of new disorders, and patient and provider education. The cohort is in the midst of activities at the time of this report; therefore, results will be shared in future reports.
- Of the 34 state, territory, or agency programs receiving funding for this collaborative, five have graduated in the following focus areas:
    - Alaska (Follow-Up);
    - Arizona (Satisfactory Specimens);
    - Colorado (Newborn Screening Education);
    - Georgia (Cut-Offs and Follow-up); and
    - Wisconsin (Turn-Around Time).

- As of May 2022, many of the funded state, territory, or agency programs focused on improving identification and follow-up of out-of-range results. In particular:
    - Four programs successfully met their specified goals on improving identification of, and follow-up on, out-of-range results.
    - Three programs successfully met their specified goals on communicating screening results to providers and families and confirming diagnoses.
    - Other programs have made identifying and following up on out-of-range results a project focus.
        - This includes 13 programs that have defined measures to identify and follow up on out-of-range results either by introducing capability to screen, improving algorithms for cut-offs to enhance identification of out-of-range results, or by improving follow-up on presumptive positives.
- The program continued to engage with improvement coaches and subject matter experts to further support the quality improvement needs of newborn screening programs.

Figure 4: NewSTEPs Quality Indicator Data



Quality Indicator Data

NewSTEPs facilitates data-driven continuous quality improvement using community-developed Quality Indicators that serve as national, harmonized metrics to quantify and track quality practices within the newborn screening system from the pre-analytic and post-analytic stages.

Quality Indicator data shows that programs are continuously improving the timeliness of reporting out newborn screening results to meet national recommendations.

Source: NewSTEPs, *https://www.newsteps.org/*, as of January 2023.

The COVID-19 pandemic had a significant impact on program initiatives and capabilities, staff bandwidth and deployments in support of COVID, and interrupted some newborn screening programs' ability to:

- Participate in the Quality Improvement Projects collaborative;
- Respond to current and future quality improvement projects' requests for proposals due to the many competing priorities resulting from the pandemic;
- Complete work in a timely manner due to staffing limitations and an inability to procure resources and supplies; and
- Travel and participate in in-person CQI events, workshops, and trainings.

Despite these impacts, the program has facilitated communication and collaboration between states to address needs as they have arisen during the pandemic, including:

- The program continued to provide webinars on quality improvement to facilitate remote engagement, including webinars on the following topics:
    - Strategies for getting buy-in;

    - Resources, tools, and upcoming programming; and
    - Plan-Do-Study-Act cycles and their implementation and common pitfalls.
- APHL held the NewSTEPs CQI National Meeting on December 7, 2021, as a three-part virtual meeting. The most recent CQI National Meeting took place in August 2022 in a hybrid environment, with session and breakout topics that included "Advancing Equity in Newborn Screening through CQI," "Principles for Sustaining Improvement," and "Knowledge Management Principles for Quality Improvement Purposes." All meeting resources and presentations are available on the NewSTEPs website.[25]

Part III: Clearinghouse of Newborn Screening Information (Sec. 1112)

Launched in September 2020, the current newborn screening clearinghouse, known as the Newborn Screening Information Center (NBSIC), maintains a central, online repository of current educational information, materials, resources, research, and data on newborn screening that is hosted and maintained in a federal web environment. NBSIC was established to educate families of newborns, as well as health professionals, industry representatives, and other members of the public.

HRSA awarded a contract to the National Institute for Children's Health Quality to develop content for NBSIC, found at https://newbornscreening.hrsa.gov/.

FYs 2021 AND 2022 UPDATE

Just as newborn screening is evolving, the NBSIC website is responsive to the needs of families and other stakeholders. The program undertook the following activities:

- Implemented a social media outreach plan to disseminate information from the Newborn Screening Clearinghouse website. Across HRSA's Facebook and Twitter accounts, 85,700 people received NBSIC's social media posts, which they also shared across other users' accounts.
- Launched the Spanish-language NBSIC website in the summer of 2022 (https://newbornscreening.hrsa.gov/es). As with the English-language site, the National Institute for Children's Health Quality will review this site quarterly for consistency, accuracy, and clarity.

From the launch of NBSIC on September 1, 2020, to September 28, 2022, the website has received 335,893 page views.

Part IV: Laboratory Quality and Surveillance (Sec. 1113)

CDC, as authorized under **Section 1113** of the PHS Act, operates the nation's only quality assurance program, the Newborn Screening Quality Assurance Program (NSQAP). This signature program of CDC is responsible for ensuring the accuracy of newborn screening blood

[25] Association of Public Health Laboratories. "2022 APHL NewSTEPs Continuous Quality Improvement (CQI) National Meeting." August 2022. https://www.newsteps.org/index.php/resources/webinars-events/2022-aphl-newsteps-continuous-quality-improvement-cqi-national-meeting-0.

spot tests conducted by public health laboratories. Without accurate testing, the benefits of newborn screening would be severely reduced, putting newborns at serious risk.

PURPOSE

- **Provide unique and essential services directly to state and other laboratories** to improve the early detection of newborns with serious and in some cases fatal diseases. CDC provides expert quality assurance, proficiency testing, and performance evaluation services, and produces quality assurance and proficiency materials for public health dried blood spot newborn screening programs.

OBJECTIVES

- **Advance the addition of new conditions by state program** through in-depth training and technical assistance, and funding via a competitive award process, to update laboratory capacity and capabilities.
- **Improve the performance and interpretation of screening tests within state programs.**
    - CDC's NSQAP provides quality assurance materials and proficiency testing services for tests that detect more than 50 congenital conditions in newborns, including all dried blood spot laboratory-identified primary disorders on the RUSP.
- **Strengthen the performance and interpretation of screening tests within CDC laboratories** by developing new methods and improving existing ones. CDC also supports data interpretation projects across multiple states. These activities improve disease detection and reduce false positives.
- **Enhance CDC's quality assurance programs** by achieving and maintaining external accreditation standards.

FYs 2021 AND 2022 UPDATE

During this period, CDC undertook a range of activities that resulted in progress towards each program objective:

- Supported state newborn screening laboratories with cooperative agreement funding to initiate screening for new conditions and improve screening for current conditions.
    - CDC funded five states (Iowa, Michigan, New York, North Carolina, and Texas) with a 2-year cooperative agreement that provided critical laboratory equipment, staffing, and supplies for state newborn screening programs.
        - These funded states added new tests to their state panels, enhanced screening performance for improved timeliness, improved test sensitivity for reduced false positives, and improved capability to quickly and accurately interpret screening results:
            - Iowa added SMA and began developing benchmarks for system performance.
            - Michigan purchased equipment to decrease the turnaround time to report positive screens for newborn screening disorders. In addition, the state implemented second-tier testing for Pompe disease and Mucopolysaccharidosis Type I (MPS I).
            - North Carolina added SMA and X-linked Adrenoleukodystrophy (X-ALD). CDC funding supported preparation for MPS I and

Pompe screening (still in progress).
    - New York added secondary testing for 17 conditions to enhance screening and reduce the number of newborn babies needing referral for further testing and diagnosis.
    - Texas conducted an in-depth evaluation of its congenital hypothyroidism newborn screening program. This analysis enabled Texas to reduce false positive results and increase screening efficiency.
- Provided quality assurance materials and voluntary assessments of performance via NSQAP:
    - NSQAP is responsible for the administration of proficiency testing and quality control programs to U.S. public health newborn screening laboratories and hundreds of international labs worldwide.
        - Participating laboratories received individualized performance evaluations to support their own laboratory accreditation.
    - NSQAP manufactured and certified dried blood spot materials for 16 proficiency testing programs and 11 quality control programs.
        - A table of proficiency testing and quality control materials is in Appendix D.
    - NSQAP is the only International Organization for Standardization (ISO)-accredited, worldwide provider of comprehensive quality assurance materials that cover all recommended newborn screening disorders in the United States.
        - NSQAP completed an external assessment of compliance to the ISO/International Electrotechnical Commission 17043 standard for Proficiency Testing Providers, and reaffirmed accreditation through September 2025.
        - NSQAP completed a gap analysis of requirements to meet the ISO 17034 standard for Reference Material Producers and anticipates initial accreditation by fall 2023.
    - NSQAP produced quarterly reports summarizing proficiency testing results for 75 biochemical analytes, multiple cystic fibrosis DNA variants, and several abnormal hemoglobin proteins known to cause sickle cell disease.
    - NSQAP summarized quality control results for 67 analytes.
- Expanded and enhanced quality assurance services for state newborn screening laboratories by taking on the following tasks:
    - Expanding the SMA proficiency testing program to allow international participants to join in 2022. Enrollment increased from 30 to 105 participants.
    - Providing new quality assurance services and pathogenic variant panels tailored to the unique needs of next generation sequencing assays used to identify babies at risk for developing cystic fibrosis.
    - Developing a new technique to create sustainable quality assurance materials for SCID detection. These materials, made from cryopreserved nucleated cord blood cells, are used to generate larger lots of dried blood spots in support of creating an external quality control program for public health labs engaged in newborn screening.

    - Increasing participation in the SCID proficiency testing program from 90 newborn screening laboratories in 2021 to 100 in 2022.
    - Determining the minimum cell concentration to optimize robust molecular assay performance of lab-created dried blood spots supplemented with transduced patient white blood cells from patients with cystic fibrosis in support of creating an external quality control program for public health labs engaged in newborn screening.
    - Developing guanidinoacetate methyltransferase proficiency testing materials and organized a successful pilot. The established NSQAP amino acid proficiency testing program will add those materials in 2023.
    - Developing a new technique to create sustainable proficiency testing materials for lysosomal storage disorders using recombinant enzymes. This novel approach was applied to the manufacturing and prototype testing of MPS II proficiency testing materials.
    - Developing quality control materials for adenosine deaminase SCID. An established NSQAP quality control program added the adenosine deaminase SCID biomarkers, adenosine and deoxyadenosine.
    - Developing and testing linearity, quality control, and proficiency testing materials for second-tier screening for multiple metabolic conditions such as maple syrup urine disease, methylmalonic acidemia, propionic acidemia, homocystinuria, etc.
    - Developing and testing first-tier linearity materials containing 43 biomarkers for dozens of metabolic newborn disorders.
- Developed new screening methods and improved existing newborn screening methods:
    - Improved commercial cystic fibrosis next generation sequencing assay by identifying non-coding regions not covered by the assay and then developing a custom panel of Sanger sequencing assays of these regions.
    - Developed and validated a novel method, allowing the multiplexing of homocysteine in first-tier screening, improving the sensitivity and specificity of the assay to detect homocystinuria.
    - Developed and validated a second-tier screening assay that can simultaneously assay 19 biomarkers for 11 disorders. This method will allow public health laboratories to significantly decrease false positives. CDC received several technical assistance requests from public health laboratories looking into adopting the assay.
- Provided direct technical assistance and training for state newborn screening laboratories, including:
    - Over 550 hours of molecular technical support in 2021 to 38 programs for disorders, including: SMA, SCID, cystic fibrosis, congenital adrenal hyperplasia, and galactosemia.
    - Over 450 hours of molecular technical support in 2022 to 66 programs for the same disorders as 2021, as well as congenital cytomegalovirus, hemoglobinopathies, and adrenoleukodystrophy, and provided next-generation sequencing technical advice.
    - Over 700 hours of biochemical technical support in 2021 and 2022 to 27 state and territorial programs. This support ranged from technology transfer to helping

with setting up cutoff, interlaboratory harmonization efforts, and troubleshooting for dozens of metabolic disorders screened by mass spectrometry. Two inter-laboratory harmonization studies concluded in 2022, and one is still in progress.

Part V: Interagency Coordinating Committee on Newborn and Child Screening (Sec. 1114)

The ICC, co-chaired by HRSA and CDC, is composed of the HRSA Administrator, CDC Director, the Agency for Healthcare Research and Quality Director, the Food and Drug Administration Commissioner, and the National Institutes of Health Director, or their designees. **Section 1114** of the PHS Act authorizes the ICC and its activities. The ICC coordinates collaborative efforts for newborn and child screening among all U.S. Department of Health and Human Services agencies and assess existing newborn screening activities and infrastructure to make recommendations on heritable disorders for newborn screening.

In the past, the Secretary of Health and Human Services convened the ICC to review recommendations from the Committee and provide additional information. For FYs 2021 and 2022, the ICC reviewed the ACHDNC's recommendations of adding SMA and MPS I to the RUSP and provided advice to the Secretary.

Part VI: National Contingency Plan for Newborn Screening (Sec. 1115)

The Newborn Screening Contingency Plan considers the variability of state newborn screening resources and processes and provides guidance on the formation of state-specific plans that need to be in place to continue critically important newborn screening and clinical management operations in the event of emergencies. **Section 1115** of the PHS Act authorized the development of the plan and requires updating the plan as needed or at least every 5 years.[26]

CDC originally published the plan in July 2010 and published a revised version in August 2017. In 2015, CDC provided funding to the Association of Maternal and Child Health Programs to assess existing plans and professional literature to update and revise the Newborn Screening Contingency Plan as needed. The August 2017 version added point-of-care screening for critical congenital heart defects and newborn hearing. It also added a checklist tool for emergency planners at the state and local levels.[27] In 2021 and 2022, CDC funded APHL to oversee the latest revision of the plan. Newborn screening subject matter experts with experience in emergency preparedness and response will update the guidance to states to include the impact of COVID-19 and other disruptors on newborn screening systems.

[26] U.S. Department of Health and Human Services. *Newborn Screening Contingency Plan: Version II*. August 2017. https://www.cdc.gov/ncbddd/documents/Screening-Contingency-Plan-Version-II.pdf.
[27] *Ibid*.

Part VII: Funding Amounts

The FYs 2021 and 2022 funding amounts for HRSA and CDC newborn screening activities are in the table below.

Table 2: Funding Amounts

Program/Initiative	FY 2021 Funding	FY 2022 Funding
HRSA		
Newborn Screening Data Repository and Technical Assistance Program	$1,500,000	$1,500,000
RGN Program	$4,261,786	$4,285,116
Long-Term Follow-Up for Severe Combined Immunodeficiency and Other Newborn Screening Conditions Program	$2,773,019	$3,619,319
Quality Improvement in Newborn Screening Program[28]	$3,835,000	$3,300,000
Newborn Screening Family Education Program	$400,000	$400,000
Innovations in Newborn Screening Interoperability	$1,271,627	$1,258,809
Clearinghouse of Newborn Screening Information	$421,854	$420,830
Total HRSA Funding	**$14,463,286**	**$14,784,074**
CDC		
Laboratory Quality and Surveillance	$18,000,000	$19,000,000
National Contingency Plan for Newborn Screening	n/a	$30,000
Total CDC Funding	$18,000,000	$19,030,000

Part VIII: Summary and Conclusion

The newborn screening programs administered by HRSA and CDC in FYs 2021 and 2022 resulted in the screening and treatment for more heritable disorders for infants born in the United States earlier than in previous years, and:

- Increased the number of states implementing screening for conditions previously added to the RUSP, including MPS I, Pompe disease, SCID, SMA, and X-ALD.
- Added MPS II to the RUSP in 2022.
- Increased availability of and access to newborn screening materials through updated program websites, remote training programs, online educational resources, virtual newsletters, an increased social media presence, and online materials translated into Spanish and other languages.
- Provided opportunities for quality improvement, education, training, and financial support to all state and territory newborn screening programs.

[28] Prior to a restructuring in FY 2018, which added additional quality improvement activities and increased funding, HRSA called this program "Improving the Timeliness of Newborn Screening Diagnosis."

- Ensured the accuracy of newborn screening blood spot tests conducted by public health laboratories.
- Funded critical infrastructure and test development in states to reduce barriers to implementing screening for new conditions.

Through expert collaboration, information sharing, resource pooling, and targeted intervention, HRSA and CDC ensure the identification, sharing, and implementation of best practices to improve the health of all infants and children in the United States.

Appendix A: Recommended Uniform Screening Panel

The Recommended Uniform Screening Panel (RUSP) is a list of disorders the Secretary of Health and Human Services recommends for screening at birth as part of states' newborn screening programs. The Secretary adds disorders to the RUSP based on evidence that supports the potential net benefit of screening, the ability of states to screen for the disorder, and the availability of effective treatments. The Secretary recommends that every newborn be screened for all disorders on the RUSP.[29]

Most states screen for the majority of disorders on the RUSP; newer conditions are still in the process of adoption. Some states also screen for additional disorders. Although states ultimately determine what disorders their newborn screening program will screen for, the RUSP establishes a standardized list of disorders that have undergone a rigorous evidence review and are supported by the Advisory Committee on Heritable Disorders in Newborns and Children and the Secretary of Health and Human Services.[30]

RUSP Core Conditions

The RUSP classifies a condition on the newborn screening panel as a "core condition" if there is a specific test available that is sensitive enough to detect the condition, the health outcomes of the condition are well-understood, and there is an available and effective treatment.

RUSP Core Conditions (as of August 2022)[31]

Core Condition	Metabolic Disorder – Organic acid condition	Metabolic Disorder – Fatty acid oxidation disorders	Metabolic Disorder – Amino acid disorders	Endocrine Disorder	Hemoglobin Disorder	Other Disorder
Propionic acidemia	Included	N/A	N/A	N/A	N/A	N/A
Methylmalonic acidemia (methylmalonyl-CoA mutase)	Included	N/A	N/A	N/A	N/A	N/A
Methylmalonic acidemia (cobalamin disorders)	Included	N/A	N/A	N/A	N/A	N/A
Isovaleric acidemia	Included	N/A	N/A	N/A	N/A	N/A
3-Methylcrotonyl-CoA carboxylase deficiency	Included	N/A	N/A	N/A	N/A	N/A
3-Hydroxy-3-methylglutaric aciduria	Included	N/A	N/A	N/A	N/A	N/A

[29] Health Resources and Services Administration. "Recommended Uniform Screening Panel." August 2022. https://www.hrsa.gov/advisory-committees/heritable-disorders/rusp/index.html.
[30] *Ibid*.
[31] *Ibid*.

Core Condition	Metabolic Disorder – Organic acid condition	Metabolic Disorder – Fatty acid oxidation disorders	Metabolic Disorder – Amino acid disorders	Endocrine Disorder	Hemoglobin Disorder	Other Disorder
Holocarboxylase synthetase deficiency	Included	N/A	N/A	N/A	N/A	N/A
ß-Ketothiolase deficiency	Included	N/A	N/A	N/A	N/A	N/A
Glutaric acidemia type I	Included	N/A	N/A	N/A	N/A	N/A
Carnitine uptake defect/carnitine transport defect	N/A	Included	N/A	N/A	N/A	N/A
Medium-chain acyl-CoA dehydrogenase deficiency	N/A	Included	N/A	N/A	N/A	N/A
Very long-chain acyl-CoA dehydrogenase deficiency	N/A	Included	N/A	N/A	N/A	N/A
Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency	N/A	Included	N/A	N/A	N/A	N/A
Trifunctional protein deficiency	N/A	Included	N/A	N/A	N/A	N/A
Argininosuccinic aciduria	N/A	N/A	Included	N/A	N/A	N/A
Citrullinemia, type I	N/A	N/A	Included	N/A	N/A	N/A
Maple syrup urine disease	N/A	N/A	Included	N/A	N/A	N/A
Homocystinuria	N/A	N/A	Included	N/A	N/A	N/A
Classic phenylketonuria	N/A	N/A	Included	N/A	N/A	N/A
Tyrosinemia, type I	N/A	N/A	Included	N/A	N/A	N/A
Primary congenital hypothyroidism	N/A	N/A	N/A	Included	N/A	N/A
Congenital adrenal hyperplasia	N/A	N/A	N/A	Included	N/A	N/A
SS disease (Sickle cell anemia)	N/A	N/A	N/A	N/A	Included	N/A
S, βeta-thalassemia	N/A	N/A	N/A	N/A	Included	N/A
S,C disease	N/A	N/A	N/A	N/A	Included	N/A
Biotinidase deficiency	N/A	N/A	N/A	N/A	N/A	Included
Critical congenital heart disease	N/A	N/A	N/A	N/A	N/A	Included
Cystic fibrosis	N/A	N/A	N/A	N/A	N/A	Included
Classic galactosemia	N/A	N/A	N/A	N/A	N/A	Included

Core Condition	Metabolic Disorder – Organic acid condition	Metabolic Disorder – Fatty acid oxidation disorders	Metabolic Disorder – Amino acid disorders	Endocrine Disorder	Hemoglobin Disorder	Other Disorder
Glycogen Storage Disease Type II (Pompe)	N/A	N/A	N/A	N/A	N/A	Included
Hearing loss	N/A	N/A	N/A	N/A	N/A	Included
Severe combined immunodeficiencies	N/A	N/A	N/A	N/A	N/A	Included
Mucopolysaccharidosis Type 1	N/A	N/A	N/A	N/A	N/A	Included
X-linked Adrenoleukodystrophy	N/A	N/A	N/A	N/A	N/A	Included
Spinal Muscular Atrophy due to homozygous deletion of exon 7 in SMN1	N/A	N/A	N/A	N/A	N/A	Included
Mucopolysaccharidosis Type II	N/A	N/A	N/A	N/A	N/A	Included

RUSP Secondary Conditions

"Secondary conditions" are conditions that can be identified when screening for a core condition, or as a consequence of confirmatory testing following a positive newborn screening result (e.g., a result outside of the normal reference range).

RUSP Secondary Conditions (as of August 2022)[32]

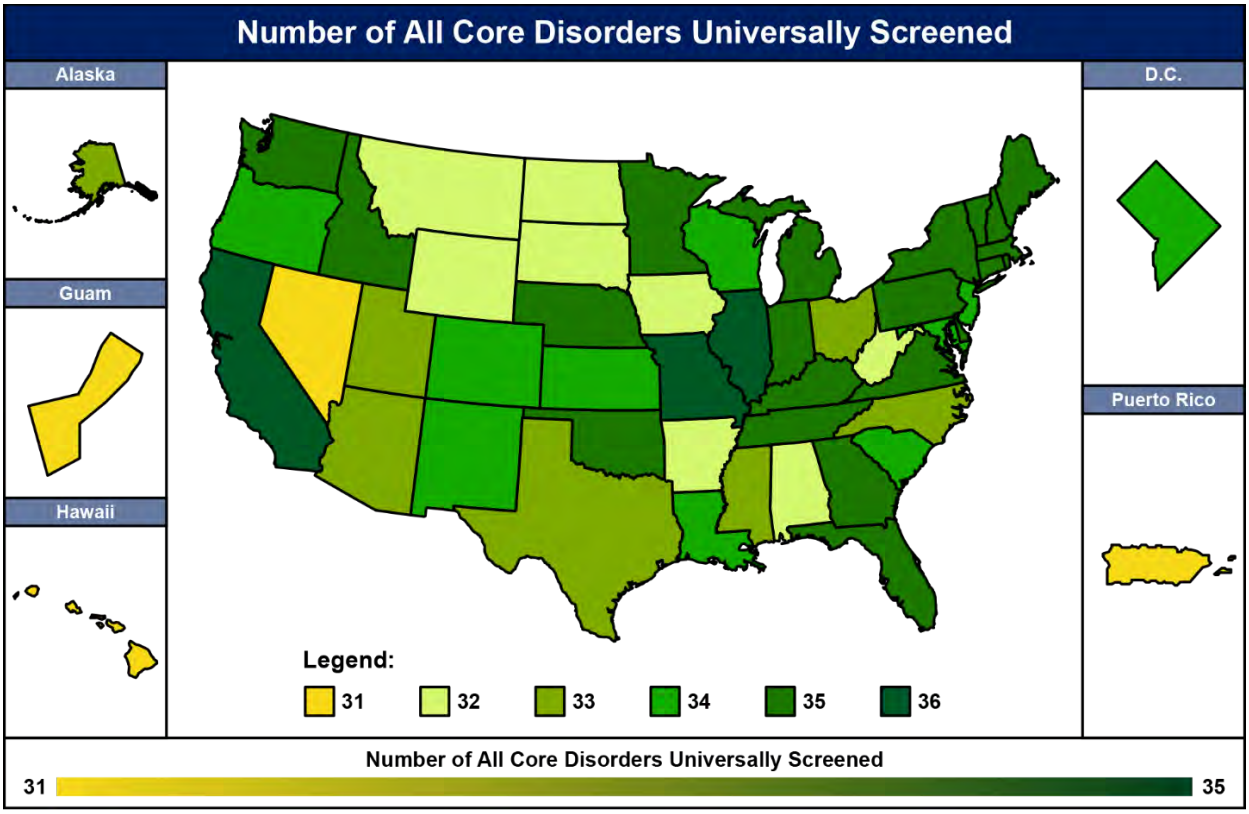
Secondary Condition	Metabolic Disorder – Organic acid condition	Metabolic Disorder – Fatty acid oxidation disorders	Metabolic Disorder – Amino acid disorders	Hemoglobin Disorder	Other Disorder
Methylmalonic acidemia with homocystinuria	Included	N/A	N/A	N/A	N/A
Malonic acidemia	Included	N/A	N/A	N/A	N/A
Isobutyrylglycinuria	Included	N/A	N/A	N/A	N/A
2-Methylbutyrylglycinuria	Included	N/A	N/A	N/A	N/A
3-Methylglutaconic aciduria	Included	N/A	N/A	N/A	N/A
2-Methyl-3-hydroxybutyric aciduria	Included	N/A	N/A	N/A	N/A
Short-chain acyl-CoA dehydrogenase deficiency	N/A	Included	N/A	N/A	N/A
Medium/short-chain L-3-hydroxyacyl-CoA dehydrogenase deficiency	N/A	Included	N/A	N/A	N/A
Glutaric acidemia type II	N/A	Included	N/A	N/A	N/A
Medium-chain ketoacyl-CoA thiolase deficiency	N/A	Included	N/A	N/A	N/A
2,4 Dienoyl-CoA reductase deficiency	N/A	Included	N/A	N/A	N/A
Carnitine palmitoyltransferase type I deficiency	N/A	Included	N/A	N/A	N/A
Carnitine palmitoyltransferase type II deficiency	N/A	Included	N/A	N/A	N/A
Carnitine acylcarnitine translocase deficiency	N/A	Included	N/A	N/A	N/A
Argininemia	N/A	N/A	Included	N/A	N/A
Citrullinemia, type II	N/A	N/A	Included	N/A	N/A
Hypermethioninemia	N/A	N/A	Included	N/A	N/A
Benign hyperphenylalaninemia	N/A	N/A	Included	N/A	N/A
Biopterin defect in cofactor biosynthesis	N/A	N/A	Included	N/A	N/A
Biopterin defect in cofactor regeneration	N/A	N/A	Included	N/A	N/A
Tyrosinemia, type II	N/A	N/A	Included	N/A	N/A
Tyrosinemia, type III	N/A	N/A	Included	N/A	N/A

[32] *Ibid.*

Secondary Condition	Metabolic Disorder – Organic acid condition	Metabolic Disorder – Fatty acid oxidation disorders	Metabolic Disorder – Amino acid disorders	Hemoglobin Disorder	Other Disorder
Various other hemoglobinopathies	N/A	N/A	N/A	Included	N/A
Galactoepimerase deficiency	N/A	N/A	N/A	N/A	Included
Galactokinase deficiency	N/A	N/A	N/A	N/A	Included
T-cell related lymphocyte deficiencies	N/A	N/A	N/A	N/A	Included

Appendix B: Recommended Uniform Screening Panel Conditions Screened by State or Territory

The map and table below show the number of core disorders universally screened per state.



Number of Core Disorders Universally Screened Per State

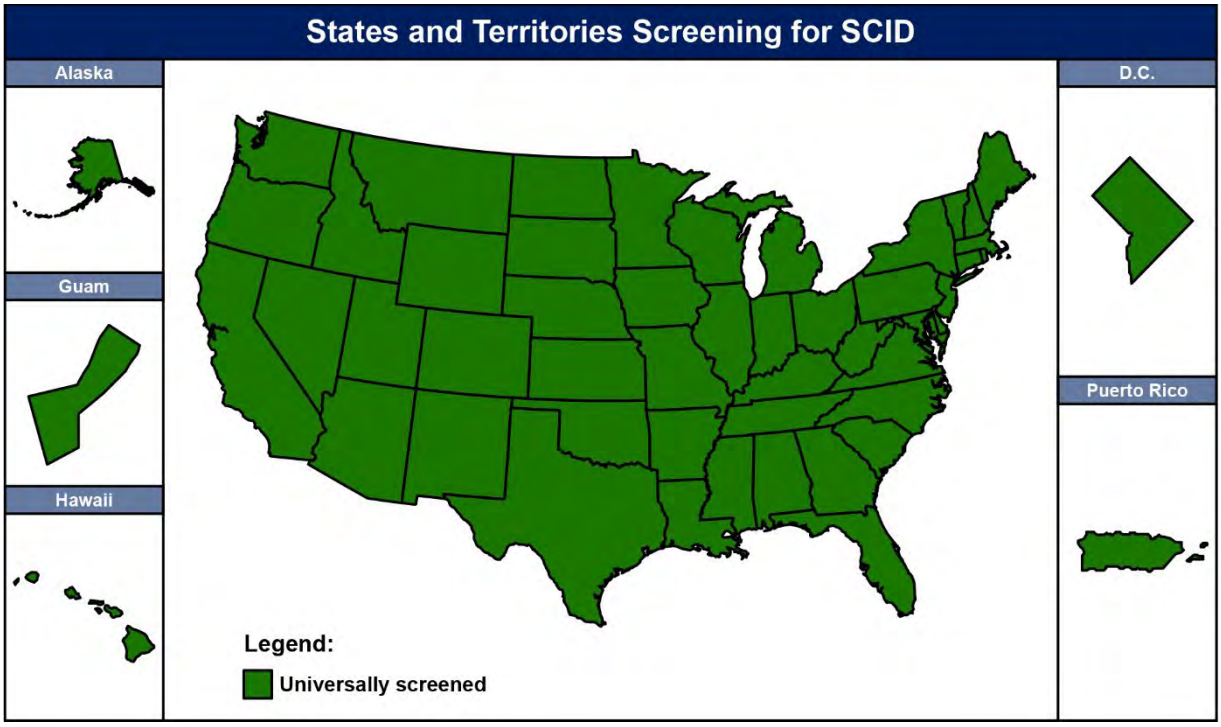
Core Disorders Screened per State			
Alabama: 32	Illinois: 36	Nebraska: 35	South Carolina: 34
Alaska: 32	Indiana: 35	Nevada: 31	South Dakota: 32
Arizona: 33	Iowa: 32	New Hampshire: 35	Tennessee: 35
Arkansas: 32	Kansas: 34	New Jersey: 34	Texas: 33
California: 35	Kentucky: 35	New Mexico: 34	Utah: 33
Colorado: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

Source: NewSTEPs, https://www.newsteps.org.

Appendix C: States and Territories Screening for Specific Conditions

The maps and tables below show the states and territories screening for Severe Combined Immunodeficiency (SCID), Pompe disease, Mucopolysaccharidosis Type I (MPS I), X-linked Adrenoleukodystrophy (X-ALD), Spinal Muscular Atrophy (SMA), and Mucopolysaccharidosis Type II (MPS II) as of September 2022.

States and Territories Screening for SCID

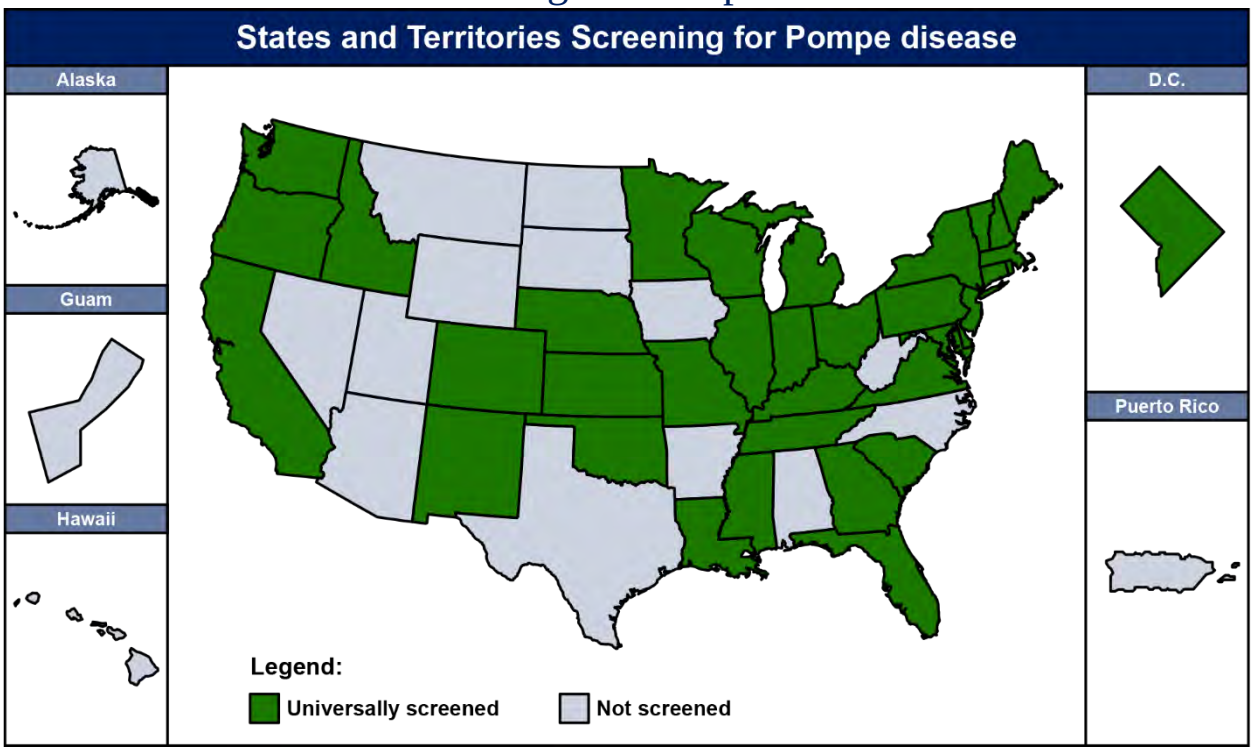


Note: Added to the RUSP in 2010.

States Screening for SCID			
Alabama	Illinois	Nebraska	South Carolina
Alaska	Indiana	Nevada	South Dakota
Arizona	Iowa	New Hampshire	Tennessee
Arkansas	Kansas	New Jersey	Texas
California	Kentucky	New Mexico	Utah
Colorado	Louisiana	New York	Vermont
Connecticut	Maine	North Carolina	Virginia
Delaware	Maryland	North Dakota	Washington
District of Columbia	Massachusetts	Ohio	West Virginia
Florida	Michigan	Oklahoma	Wisconsin
Georgia	Minnesota	Oregon	Wyoming
Guam	Mississippi	Pennsylvania	
Hawaii	Missouri	Puerto Rico	
Idaho	Montana	Rhode Island	
Total programs: 53			

Source: NewSTEPs, https://www.newsteps.org.

States and Territories Screening for Pompe disease

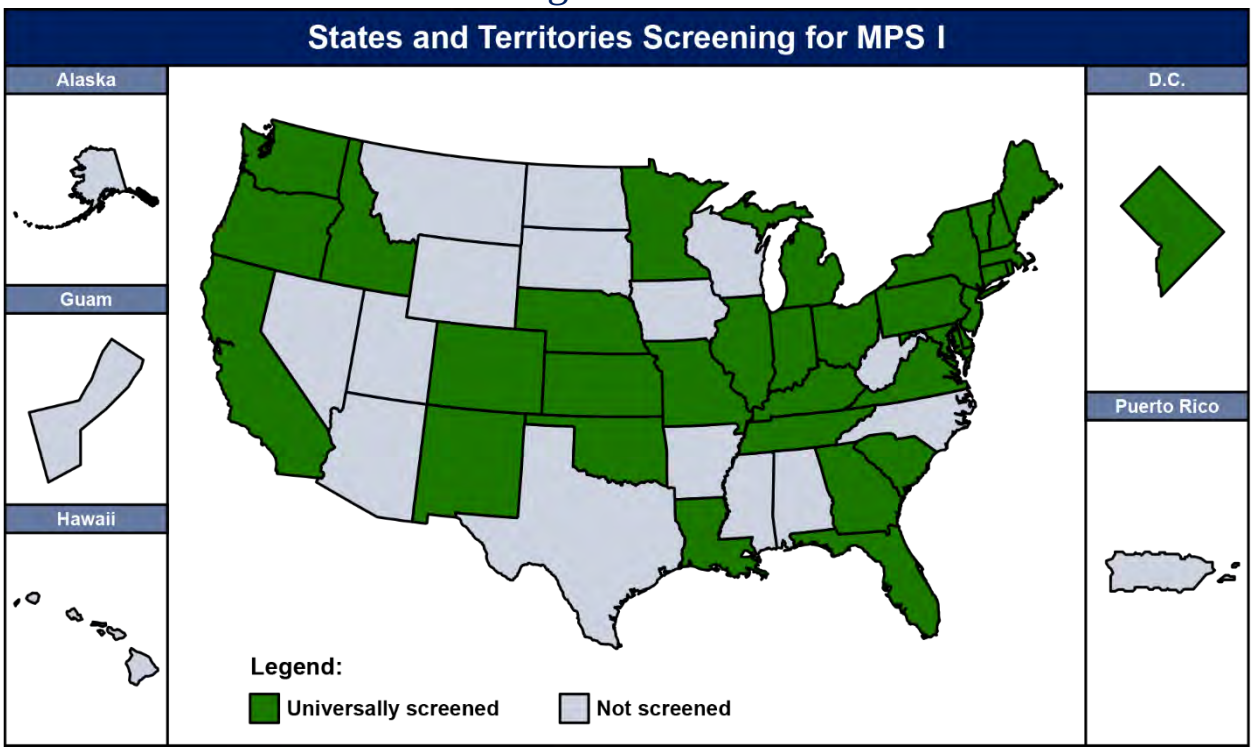


Note: Added to the RUSP in 2015.

States Screening for Pompe disease			
California	Indiana	Mississippi	Oregon
Colorado	Kansas	Missouri	Pennsylvania
Connecticut	Kentucky	Nebraska	Rhode Island
Delaware	Louisiana	New Hampshire	South Carolina
District of Columbia	Maine	New Jersey	Tennessee
Florida	Maryland	New Mexico	Vermont
Georgia	Massachusetts	New York	Virginia
Idaho	Michigan	Ohio	Washington
Illinois	Minnesota	Oklahoma	Wisconsin
Total programs: 36			

Source: NewSTEPs, https://www.newsteps.org.

States and Territories Screening for MPS I

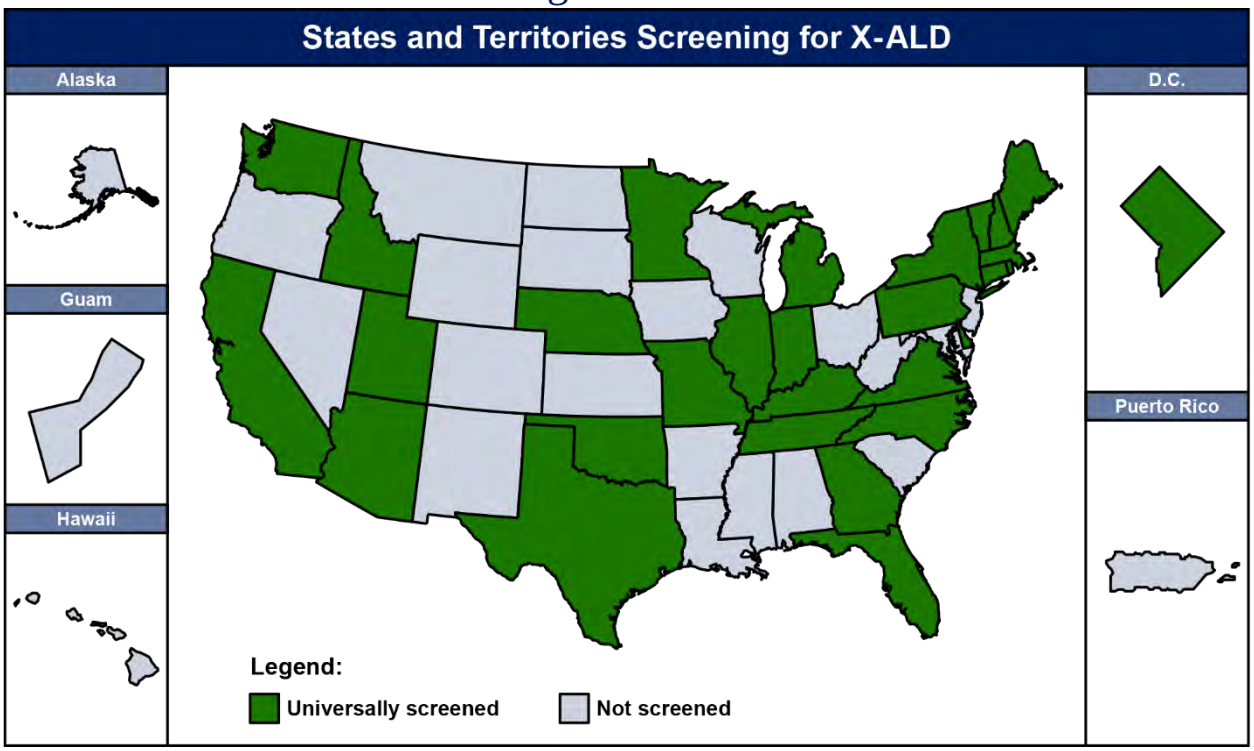


Note: Added to the RUSP in 2016.

States Screening for MPS I				
Arizona	Georgia	Maine	New Hampshire	Pennsylvania
California	Idaho	Maryland	New Jersey	Rhode Island
Colorado	Illinois	Massachusetts	New Mexico	South Carolina
Connecticut	Indiana	Michigan	New York	Tennessee
Delaware	Kansas	Minnesota	Ohio	Vermont
District of Columbia	Kentucky	Missouri	Oklahoma	Virginia
Florida	Louisiana	Nebraska	Oregon	Washington
Total programs: 35				

Source: NewSTEPs, https://www.newsteps.org.

States and Territories Screening for X-ALD

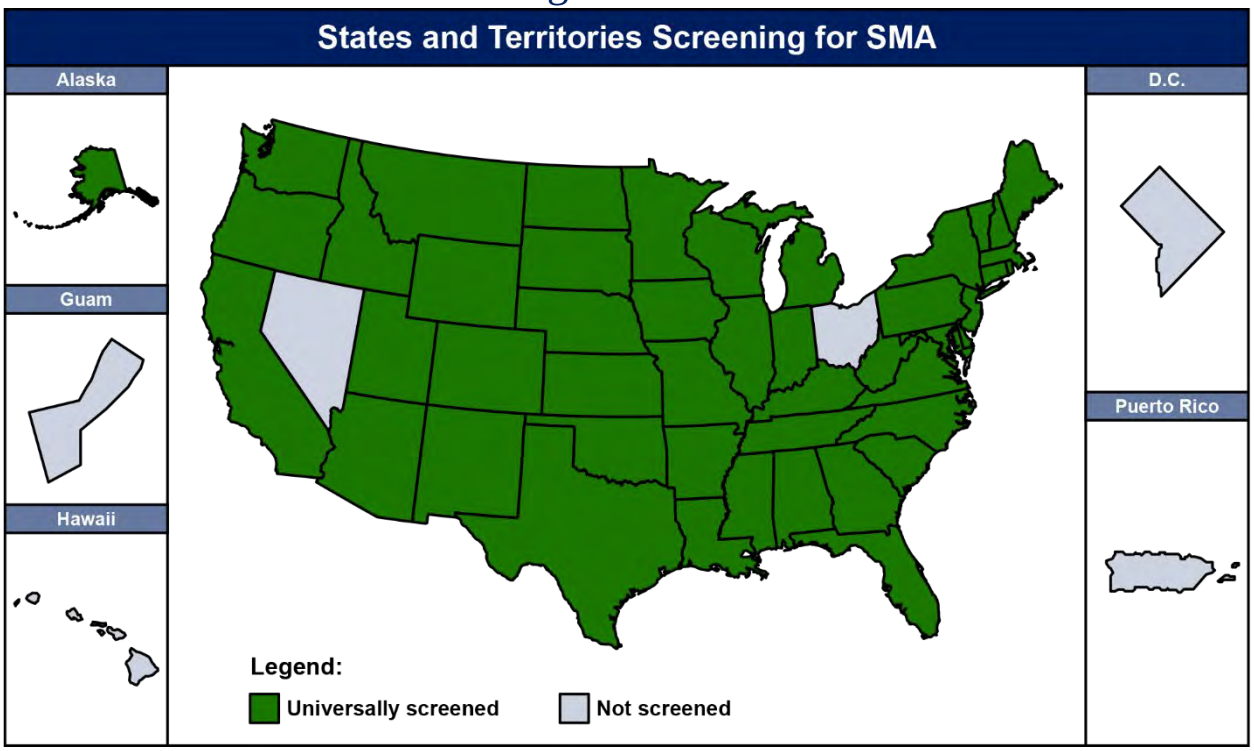


Note: Added to the RUSP in 2016.

States Screening for X-ALD			
Arizona	Idaho	Minnesota	Rhode Island
California	Illinois	Missouri	Tennessee
Connecticut	Indiana	Nebraska	Texas
Delaware	Kentucky	New Hampshire	Utah
District of Columbia	Maine	New York	Vermont
Florida	Massachusetts	North Carolina	Virginia
Georgia	Michigan	Pennsylvania	Washington
Total programs: 28			

Source: NewSTEPs, https://www.newsteps.org.

States and Territories Screening for SMA

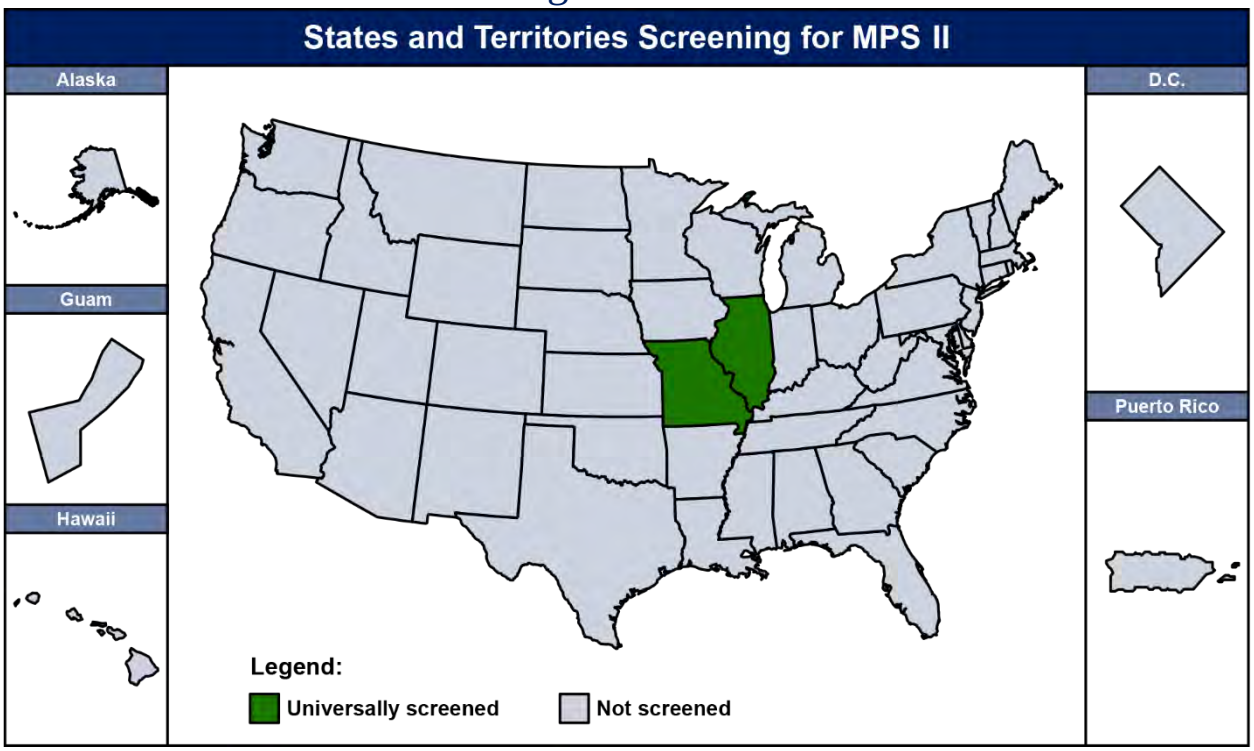


Note: Added to the RUSP in 2018.

States Screening for SMA			
Alabama	Indiana	Montana	South Carolina
Alaska	Iowa	Nebraska	South Dakota
Arizona	Kansas	New Hampshire	Tennessee
Arkansas	Kentucky	New Jersey	Texas
California	Louisiana	New Mexico	Utah
Colorado	Maine	New York	Vermont
Connecticut	Maryland	North Carolina	Virginia
Delaware	Massachusetts	North Dakota	Washington
Florida	Michigan	Oklahoma	West Virginia
Georgia	Minnesota	Oregon	Wisconsin
Idaho	Mississippi	Pennsylvania	Wyoming
Illinois	Missouri	Rhode Island	
Total programs: 47			

Source: NewSTEPs, https://www.newsteps.org.

States and Territories Screening for MPS II



Note: Added to the RUSP in 2022.

States Screening for MPS II	
Illinois	Missouri
Total programs: 2	

Source: NewSTEPs, https://www.newsteps.org.

Appendix D: Quality and Proficiency Programs Administered under the Centers for Disease Control and Prevention's Newborn Screening Quality Assurance Program

Proficiency Testing Programs (Three events per year)	Analyte(s) Included
Acylcarnitines (ACPT)	C0(L), C2(L). C3, C3DC, C3DC+C4OH, C4, C4OH, C5, C5:1, C5DC, C5OH, C6, C8, C10, C10:1, C10:2, C14, C14:1, C16, C16OH, C18, C18:1, C18OH
Adrenoleukodystrophy (ALDPT)	24:0-Lysophosphatidylcholine, 26:0-Lysophosphatidylcholine
Amino Acids and SUAC (AAPT)	Arginine, Citrulline, Leucine, Methionine, Phenylalanine, Succinylacetone, Tyrosine, Valine
Biotinidase Deficiency (BIOTPT)	Biotinidase
Cystic Fibrosis DNA Variant Detection (CFDNA)	Cystic Fibrosis DNA Variants
Galactose-1-phosphate Uridyltransferase Deficiency (GALTPT)	Galactose-1-phosphate Uridyltransferase
Glucose-6-phosphate Dehydrogenase Deficiency (G6PDPT)	Glucose-6-phosphate Dehydrogenase
anti-HIV-1 Antibodies (HIVPT)	anti-HIV-1 Antibodies
Hormone + Total Galactose (HORMPT)	Thyroxine, Thyroid Stimulating Hormone, 17 α-Hydroxyprogesterone, and Total Galactose
Immunoreactive Trypsinogen (IRTPT)	Immunoreactive Trypsinogen
Lysosomal Storage Disorders (LSDPT)	Galactoceramidase, Acid α-Glucosidase, α-L-Iduronidase
Second-tier Congenital Adrenal Hyperplasia (CAHPT) by LC-MS/MS	17 α-hydroxyprogesterone, 4-androstenedione, cortisol, 11-deoxycortisol, 21-deoxycortisol
Sickle Cell Disease and Other Hemoglobinopathies (HbPT)	*Enrollment currently closed. Requests added to waitlist.* Sickle Cell Disease and Other Hemoglobinopathies
Spinal Muscular Atrophy (SMAPT)	Survival Motor Neuron 1 (SMN1) Exon 7
T-cell Receptor Excision Circle (TRECPT)	T-cell Receptor Excision Circle
anti-*Toxoplasma* Antibodies (TOXOPT)	*Toxoplasma gondii* IgM Antibodies

Quality Control Programs (Two events per year)	Analyte(s) Included
17 α-Hydroxyprogesterone + Total Galactose (17OHPQC and TGalQC)	17 α-Hydroxyprogesterone, and Total Galactose
Galactose-1-phosphate Uridyltransferase (GALTQC)	Galactose-1-phosphate Uridyltransferase
anti-HIV-1 Antibodies (HIVQC)	anti-HIV-1 Antibodies
Immunoreactive Trypsinogen (IRTQC)	Immunoreactive Trypsinogen
Lysosomal Storage Disorders (LSDQC)	Galactocerebrosidase, Acid α-Glucosidase, α-L-Iduronidase α-Galactosidase, β-Glucocerebrosidase, Acid Sphingomyelinase
Tandem MS 1 (MSMS1QC)	Arginine, Alanine, Citrulline, Creatine, Creatinine, Guanidinoacetic Acid, Glycine, Leucine, Methionine, Ornithine, Phenylalanine, Succinylacetone, Tyrosine, Valine, C0, C2, C3, C3DC, C3DC+C4OH, C4, C4OH, C5, C5:1, C5DC, C5OH, C6, C8, C10, C12, C14, C14:1, C16, C16OH, C18, C18OH, C20-LPC, C22-LPC, C24-LPC, C26-LPC
Thyroxine (T4QC)	Thyroxine
Thyroid-Stimulating Hormone (TSHQC)	Thyroid Stimulating Hormone

Second-Tier Quality Control Programs (Two events per year)	Analyte(s) Included
Second-tier Congenital Adrenal Hyperplasia by LC-MS/MS (CAHQC)	17 α-Hydroxyprogesterone, 4-Androstenedione, Cortisol, 11-Deoxycortisol, 21- Deoxycortisol
Second-tier Maple Syrup Urine Disease and Phenylketonuria by LC-MS/MS (MSUD-PKUQC)	Alloisoleucine, Isoleucine, Leucine, Phenylalanine, Tyrosine, Valine
Second-tier Methylmalonic/Propionic Acidemia and Homocystinuria by LC-MS/MS (MMA- HCYQC)	Malonic Acid, Methylmalonic Acid, Ethylmalonic Acid, 2-Methylcitric Acid, Total Homocysteine

DEPARTMENT OF HEALTH & HUMAN SERVICES OFFICE OF THE SECRETARY

Assistant Secretary for Legislation
Washington, DC 20201

March 11, 2024

The Honorable Bernie Sanders
Chair
Committee on Health, Education,
Labor and Pensions
United States Senate
Washington, DC 20510

Dear Chair Sanders:

I am pleased to provide you with a report on Newborn Screening Activities for fiscal years (FY) 2021 and 2022. The Health Resources and Services Administration prepared this report, and it is being submitted in accordance with the report requirement in Section 11(b) of the Newborn Screening Saves Lives Reauthorization Act of 2014 [Public Law 113-240], which added 42 U.S.C. 300b-17. It provides information on activities authorized by Sections 1109, 1110, and 1112 through 1115 of Title XI of the Public Health Service Act.

The programs and activities authorized by the Public Health Service Act were established to enhance, improve, or expand the ability of state and local public health agencies to provide screening, counseling, and health care services to newborns and children with or at risk for heritable conditions. The Health Resources and Services Administration and the Centers for Disease Control and Prevention provide key support to newborn screening programs and the newborn screening community to help ensure proper and timely screening and intervention. The report builds on the previous reports covering FY 2015, FY 2016, FYs 2017 and 2018, and FYs 2019 and 2020.

I hope you find this information helpful.

Sincerely,

/Melanie Anne Egorin/

Melanie Anne Egorin, PhD
Assistant Secretary for Legislation

Enclosure

DEPARTMENT OF HEALTH & HUMAN SERVICES OFFICE OF THE SECRETARY

Assistant Secretary for Legislation
Washington, DC 20201

March 11, 2024

The Honorable Bill Cassidy, M.D.
Ranking Member
Committee on Health, Education,
Labor and Pensions
United States Senate
Washington, DC 20510

Dear Senator Cassidy:

I am pleased to provide you with a report on Newborn Screening Activities for fiscal years (FY) 2021 and 2022. The Health Resources and Services Administration prepared this report, and it is being submitted in accordance with the report requirement in Section 11(b) of the Newborn Screening Saves Lives Reauthorization Act of 2014 [Public Law 113-240], which added 42 U.S.C. 300b-17. It provides information on activities authorized by Sections 1109, 1110, and 1112 through 1115 of Title XI of the Public Health Service Act.

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Melanie Anne Egorin, PhD
Assistant Secretary for Legislation

Enclosure

DEPARTMENT OF HEALTH & HUMAN SERVICES OFFICE OF THE SECRETARY

Assistant Secretary for Legislation
Washington, DC 20201

March 11, 2024

The Honorable Cathy McMorris Rodgers
Chair
Committee on Energy and Commerce
U.S. House of Representatives
Washington, DC 20515

Dear Chair Rodgers:

I am pleased to provide you with a report on Newborn Screening Activities for fiscal years (FY) 2021 and 2022. The Health Resources and Services Administration prepared this report, and it is being submitted in accordance with the report requirement in Section 11(b) of the Newborn Screening Saves Lives Reauthorization Act of 2014 [Public Law 113-240], which added 42 U.S.C. 300b-17. It provides information on activities authorized by Sections 1109, 1110, and 1112 through 1115 of Title XI of the Public Health Service Act.

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Melanie Anne Egorin, PhD
Assistant Secretary for Legislation

Enclosure

DEPARTMENT OF HEALTH & HUMAN SERVICES OFFICE OF THE SECRETARY

Assistant Secretary for Legislation
Washington, DC 20201

March 11, 2024

The Honorable Frank Pallone, Jr.
Ranking Member
Committee on Energy and Commerce
U.S. House of Representatives
Washington, DC 20515

Dear Representative Pallone:

I am pleased to provide you with a report on Newborn Screening Activities for fiscal years (FY) 2021 and 2022. The Health Resources and Services Administration prepared this report, and it is being submitted in accordance with the report requirement in Section 11(b) of the Newborn Screening Saves Lives Reauthorization Act of 2014 [Public Law 113-240], which added 42 U.S.C. 300b-17. It provides information on activities authorized by Sections 1109, 1110, and 1112 through 1115 of Title XI of the Public Health Service Act.

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Sincerely,

/Melanie Anne Egorin/

Melanie Anne Egorin, PhD
Assistant Secretary for Legislation

Enclosure