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

REPORT TO CONGRESS

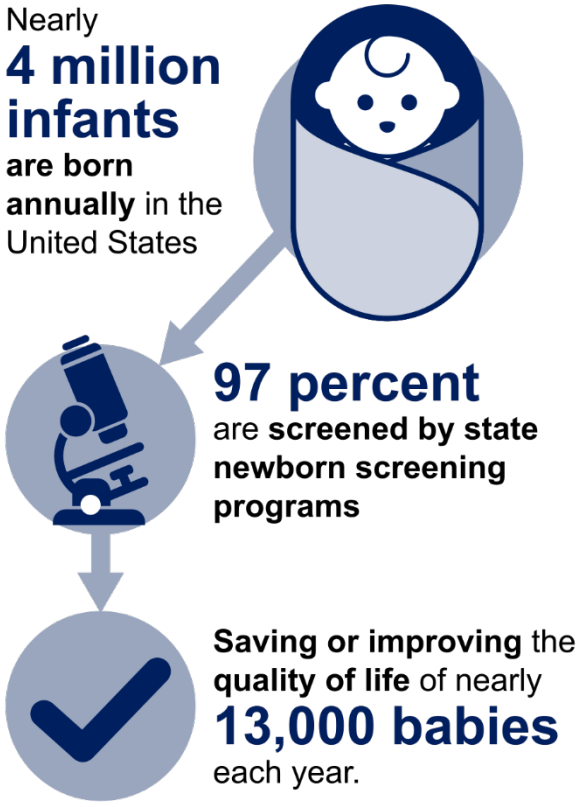
Newborn Screening Activities
Fiscal Year 2019 and Fiscal Year 2020

The Nexight Group prepared this report for the U.S. Department of Health and Human Services, Health Resources and Services Administration under contract number GS10F176AA.

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 at the state level; and the reporting of results to appropriate medical personnel who confirm or rule out a diagnosis and initiate the required 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 conditions recommended by the Secretary for Health and Human Services for newborn screening. Conditions on the RUSP are chosen based on a rigorous evaluation of the evidence supporting the net benefits of screening. (See Appendix A for information on the RUSP and Appendix B for information on state screening practices.)



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

Federal agencies, including the Health Resources and Services Administration (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. This report is required by the Newborn Screening Saves Lives Reauthorization Act of 2014 (the Act), 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 the previous two reports covering fiscal years (FYs) 2015 and 2016 and FYs 2017 and 2018.

The programs authorized by the Act support activities that 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. Highlights from FYs 2019 and 2020 activities include:

- **Technical assistance provided to all state newborn screening programs through the HRSA-funded Newborn Screening Data Repository and Technical Assistance**

[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.

Program:[2] Forty-eight newborn screening programs in states representing 98 percent of births in the United States signed Memoranda of Understanding to participate in 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 Newborn Screening Quality Improvement Program focused on helping newborn screening programs improve several key newborn screening processes: accurate and timely screening, follow-up, and communication of out-of-range results to providers and families, and confirming a diagnosis. Eleven states participated in continuous quality improvement projects beginning in December 2019, and six states began participating in May 2020. Participating states reported decreases in unsatisfactory specimen rates, improved timeliness in collection and reporting, and improvements to follow-up.
- **Funding for states to implement screening for new disorders**: Seven state newborn screening programs received funding from CDC to implement screening for conditions recently added to the RUSP. With this funding, one state added screening for Pompe disease, two states added screening for Mucopolysaccharidosis I (MPS I), three states added screening for X-linked Adrenoleukodystrophy (X-ALD), and eight states added screening for Spinal Muscular Atrophy (SMA). In addition, CDC funding supported SMA implementation activities in Massachusetts, New Hampshire, Rhode Island, Vermont, and Wyoming. HRSA also supported a number of states to implement newborn screening for these conditions through the Newborn Screening Data Repository and Technical Assistance Program. During FYs 2019 and 2020, 10 states implemented screening for MPS I, 10 states for Pompe, 7 states for X-ALD, and 5 states for SMA.
- **Increased number of states screening for SMA**: Twenty-seven states fully implemented newborn screening programs for SMA, the most recent condition added to the RUSP (in 2018), an increase from two states in FY 2018. In July 2020, CDC began a new proficiency testing program for SMA to meet the increasing demand for quality assurance services and to promote accurate screening results.
- **Total number of states screening for recently added conditions:** Support from HRSA and CDC contributed to the total number of states screening for recently added conditions.

[2] Association of Public Health Laboratories, "Newborn Screening Technical assistance and Evaluation Program," Accessed April 26, 2021, https://www.newsteps.org/.

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

Condition	Year added to the RUSP	FY 2019 newborn screening programs offering universal screening	FY 2020 newborn screening programs offering universal screening
Pompe	2015	21	25
X-ALD	2016	16	20
MPS I	2016	19	23
SMA	2018	11	27

Source: Newborn Screening Technical assistance and Evaluation Program (NewSTEPs), *https://www.newsteps.org/*, as of 4/21

COVID-19 Response: To address information needs as well as the staffing and laboratory supply shortages created by the pandemic, the NewSTEPs and Newborn Screening Family Education programs received supplemental funding to support newborn screening programs during this time. The two programs along with the National Center for Hearing Assessment and Management offered a joint webinar on COVID-19 and newborn screening for the blood spot and newborn hearing screening programs. Additionally, the two programs partnered to gather and disseminate pandemic-relevant newborn screening information, provided direct technical assistance to state labs, and developed educational materials. These include two COVID-focused webinars and supplemental resources to states, territories, providers, and family networks, as well as an online course curriculum to help families understand what to expect with newborn screening during the COVID-19 pandemic.

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 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

CDC	Centers for Disease Control and Prevention
FDA	Food and Drug Administration
FY	Fiscal year
GAMT	Guanidinoacetate methyltransferase
HRSA	Health Resources and Services Administration
ICC	Interagency Coordinating Committee
INBSI	Innovations in Newborn Screening Interoperability
MAP	Molecular Assessment Program
MPS I	Mucopolysaccharidosis I
NBSIC	Newborn Screening Information Center
NewSTEPs	Newborn Screening Technical assistance and Evaluation Program
NIH	National Institutes of Health
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 [Public Law 113-240], which added 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) and reauthorized by the Newborn Screening Saves Lives Reauthorization Act of 2014 (P.L. 113-240). The Newborn Screening Saves Lives Reauthorization Act of 2014 requires a 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). Other Department of Health and Human Services activities supporting newborn screening funded under other authorities are not addressed in this report.

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**: This section 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**: This section 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**: This section authorizes the establishment and maintenance of a central, web-based clearinghouse of current newborn screening educational and family support and services information, materials, resources, research, and data. HRSA administers the program.
- **Section 1113 - Laboratory Quality and Surveillance**: This section authorizes the provision of quality assurance for laboratories involved in screening newborns and children for heritable disorders or conditions. CDC administers the program. These

[3] CDC and HRSA are both authorized to administer programs under this section; however, CDC does not currently administer any programs under this section.

activities include quality assurance for conducting newborn screening tests, timeliness of processing such tests, performance evaluation 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.

- **Section 1114 - Interagency Coordinating Committee on Newborn and Child Screening**: This section 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 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 (FDA), and the National Institutes of Health (NIH). HRSA and CDC administer the ICC.
- **Section 1115 - National Contingency Plan for Newborn Screening**: This section 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 report on ongoing activities authorized by the sections listed above is the third report on newborn screening activities administered by HRSA and CDC. The report covers activities in fiscal years (FYs) 2019 and 2020. The agencies structured the report around the sections of the authorizing legislation describing the purpose, goals, and activities for the programs under each section.

Part I: Improved Newborn & 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 screen, identification of out-of-range results, communication with providers and families, confirmation of a diagnosis, and initiation of treatment.

HRSA administers the programs below, under **Section 1109** of the PHS Act. These programs help increase the number of newborns who receive screening, counseling, and health care services and improve the quality of that care. HRSA also administers grants to support activities that:

- 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.
- Establish a system to assess and coordinate follow-up and treatment related to congenital,[5,6] genetic,[7] and metabolic conditions.[8]
- Improve the timeliness of newborn screening from specimen collection[9] through diagnosis.

[4] Health Resources and Services Administration, "About Newborn Screening," Accessed January 11, 2021, https://newbornscreening.hrsa.gov/about-newborn-screening.

[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, Accessed January 11, 2021, https://www.nichd.nih.gov/health/topics/birthdefects/about.)

[6] World Health Organization, "Congenital anomalies," 2016, Accessed January 11, 2021, http://www.who.int/mediacentre/factsheets/fs370/en/.

[7] 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 February 7, 2021, https://newbornscreening.hrsa.gov/about-newborn-screening/glossary.)

[8] 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 February 7, 2021, https://newbornscreening.hrsa.gov/about-newborn-screening/glossary.)

[9] 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, Accessed January 11, 2021, https://www.cdc.gov/nbslabbulletin/bulletin.html.)

Newborn Screening Data Repository and Technical Assistance Program

The Newborn Screening Data Repository and Technical Assistance Program provides technical assistance to state and local public health agencies, health care professionals, newborn screening laboratory personnel, and other newborn screening stakeholders on the implementation of state-based public health newborn screening. The Association of Public Health Laboratories receives funding from HRSA to lead the program known as the Newborn Screening Technical assistance and Evaluation Program (NewSTEPs). In its effort to provide streamlined technical assistance and accurate evaluations, NewSTEPs standardized national newborn screening quality indicators that state newborn screening programs use to determine how their system is performing on the various components of screening and identify areas in need of improvement.

The NewSTEPs Data Repository collects quality indicator data on newborn screening with the goal of supporting quality improvement initiatives and providing 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 plus de-identified, aggregate data from other participating programs. One example of how newborn screening programs use the information to improve performance is an increase in timeliness in screening practices, allowing for earlier diagnosis and intervention for infants with time-critical and non-time-critical disorders.[10]

PURPOSE

- **Reduce morbidity and mortality** caused by heritable disorders in newborns and children by enhancing, improving, and expanding the newborn screening system.

OBJECTIVES

- **Create a national newborn screening data repository** to standardize, maintain, and analyze quantitative newborn screening quality indicators, public health case definitions, and other data related to newborn screening.
- **Collect, analyze, and disseminate newborn screening data and information** to assist states in evaluating the performance of their newborn screening programs.
- **Establish a system to support states' efforts** in implementing new conditions added to the Recommended Uniform Screening Panel (RUSP).
- **Increase the number of states and territories with a data use agreement in place** to submit newborn screening data and information about their newborn screening program into the repository.
- **Increase the number of participating states and territories** contributing data to the repository on all measures.

FYs 2019 AND 2020 UPDATE

During FYs 2019 and 2020, the program:

- Acquired Memoranda of Understanding from 48 newborn screening programs, representing states in which 98 percent of babies are born.

[10] Association of Public Health Laboratories, "NewSTEPs 2019 Annual Report," January 2020, Accessed April 11, 2021, https://www.newsteps.org/sites/default/files/resources/download/nbs-newsteps-2019-annual-report.pdf.

    - These Memoranda of Understanding serve to outline the data security and data sharing parameters inherent within the data repository and delineate data ownership.
- Facilitated continuous quality improvement and data-driven outcome assessments in the newborn screening system by providing a standardized repository (NewSTEPs,[11] a national, secure, and central web-based database) and by supporting the integration of health information infrastructure and systems into newborn screening.
    - As of February 23, 2020:
        - There are 499 registered data repository users.
        - Case definitions,[12] totaling 18,957, were entered by 42 states.
        - State-level quality indicator data[13] on 42 states, enabling comparisons within and across programs, in aggregate.
    - NewSTEPs data include screening status data for all disorders on the RUSP with the data visualization updated weekly.[14]
    - Webinars and other newborn screening resources are available through the NewSTEPs resource library and are filterable by disorder.[15]
    - State profile data reports are publicly available in real time.[16]
    - The 2019 NewSTEPs annual report is available online.[17]
- Provided opportunities for timely, interactive communication and collaboration among national, regional, and state newborn screening programs. Webinar topics included informatics messaging services and electronic data flow, quality improvement, newborn screening challenges and responses, and state-specific program highlights and processes.
- Maintained a national newborn screening technical assistance resource center that proactively identifies newborn screening issues and provides training, addresses challenges, and supports newborn screening systems improvement.
- Fostered the integration of screening for new disorders by offering access to resources and technical assistance.
- Over 1,100 newborn screening community members participated on the NewSTEPs listserv, which issued an average of 35 listserv messages per month. Common messages include requests for lab value cut offs for analytes screened, questions regarding lab supply needs, recruiting for staff, and questions about processes for adding/improving screening.

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[11] Association of Public Health Laboratories, "Newborn Screening Technical assistance and Evaluation Program," Accessed April 26, 2021, https://www.newsteps.org/.
[12] International Journal of Neonatal Screening, "Case Definitions for Conditions Identified by Newborn Screening Public Health Surveillance," May 2018, Accessed February 2, 2021, https://www.mdpi.com/2409-515X/4/2/16/htm.
[13] International Journal of Neonatal Screening, "Development of National Newborn Screening Quality Indicators in the United States," September 2019, Accessed February 2, 2021, https://www.mdpi.com/2409-515X/5/3/34.
[14] Association of Public Health Laboratories, "Data Visualizations," Accessed March 4, 2021, https://www.newsteps.org/resources/data-visualizations?q=data-visualizations&tid=1.
[15] Association of Public Health Laboratories, "Resource Library," Accessed March 4, 2021, https://www.newsteps.org/resources.
[16] Association of Public Health Laboratories, "Reports," Accessed March 4, 2021, https://www.newsteps.org/data-resources/reports?q=data/reports.
[17] Association of Public Health Laboratories, "NewSTEPs 2019 Annual Report," January 2020, https://www.newsteps.org/sites/default/files/nbs-newsteps-2019-annual-report.pdf.

- Supported work groups and hosted national webinars on Critical Congenital Heart Disease, Health Information Technology, New Disorders, and Short-Term Follow-Up.

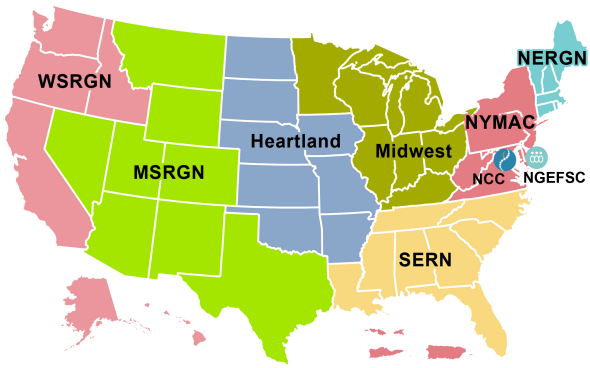
In response to the COVID-19 global pandemic and needs expressed by newborn screening programs across the nation, NewSTEPs has collected and routinely updates frequently asked questions that address challenges, technical assistance opportunities, and contingency planning needs.[18] NewSTEPs began a series of Hot Topics webinars in May 2020 that addressed issues such as virtual engagement, staffing and telework, biosafety of specimens, telehealth in newborn screening, screening unsatisfactory specimens, best practices for remote follow-up work, and strategies and accountability for telework.

Part II lists additional activities and accomplishments during this period.

Regional Genetics Networks Program

Funded since 2017, HRSA supports seven Regional Genetics Networks (RGNs), a National Coordinating Center, and a National Genetics 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.

Figure 1: Regional Genetics Network System



Source: National Coordinating Center for the Regional Genetics Networks, *https://nccrcg.org/rgns/*, as of 4/21

PURPOSE

- **Improve health equity and health outcomes** in individuals with genetic conditions, reduce morbidity and mortality caused by genetic conditions, and improve the quality of coordinated and comprehensive genetic services to children and their families.

OBJECTIVES

- **Link medically underserved populations** (based on poverty, rural geographic location, and/or populations that experience health disparities)[19] to genetic services.

[18] Association of Public Health Laboratories, "COVID-19," Accessed March 4, 2021, https://www.newsteps.org/resources/covid-19?q=covid-19.

[19] Medically underserved areas/populations are areas or populations HRSA has designated as having too few primary care providers (i.e., health professional shortage areas), high infant mortality rates, extreme poverty, or a

- **Implement quality improvement activities** to increase the connection with genetic services for the medically underserved.
- **Facilitate the implementation of telehealth**[20] with a focus on improving access to clinical genetics services.
- **Provide resources** to genetic service providers, public health officials, and families.

FYs 2019 AND 2020 UPDATE

During FYs 2019 and 2020, the RGN program served a total of 20,993 individuals and families through education and training, facilitated connections to genetics services, and facilitated telehealth services. Of those, 8,979 were from medically underserved populations.

One of the RGN program's objectives for the FY 2017-2019 cycle was to increase by 20 percent the number of medically underserved patients served by each RGN by May 2020. During this time, the RGNs increased these numbers by 152 percent, from 2,376 in FY 2017 to 5,977 in FY 2019. Each RGN has identified underserved populations (e.g., rural, Hispanic, Marshallese, Plain Communities) in the region and have engaged them in various ways, including:

- Heartland RGN conducted outreach efforts to Hispanic and Marshallese communities. The Heartland RGN website offers genetics services descriptions in both languages as of 2018.[21]
- The National Coordinating Center, Midwest RGN, and Heartland RGN developed and offered in-person and online healthcare interpreter trainings in genetics. Together these grantees trained more than 500 healthcare interpreters in prenatal and pediatric genetics to improve their ability to communicate genetics information.
- The Western States RGN developed a Minority Genetics Professionals Network to support genetics awareness activities and provide mentorships for students in minority communities interested in becoming genetics providers. As of May 2020, this network had approximately 400 minority participants.

The RGN program has additionally facilitated 3,565 connections to genetic services. The RGNs used a wide range of mechanisms to achieve these connections, such as directly supporting genetics outreach clinics and collaborating with HRSA-funded Family-to-Family Health Information Centers in the regions to provide resources and referrals to genetic services to families. Examples include:

- The Mountain States RGN developed a Genetic Ambassador Program to increase family engagement and provide opportunities for families to get involved with the RGN in a meaningful way, such as reviewing products and serving on workgroups.
- The New England RGN maintains the Genetics Education Materials for School Success website, which provides families, healthcare providers, and other professionals with

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disproportionate elderly population. (See: Health Resources and Services Administration, "Medically Underserved Areas and Populations (MUA/Ps)," Accessed January 21, 2021, https://bhw.hrsa.gov/shortage-designation/muap.)

[20] Telehealth constitutes the use of electronic information and telecommunications technologies to support long-distance clinical health care, patient and professional health-related education, public health, and health administration. (See: Health Resources and Services Administration, "Telehealth Programs," Accessed January 21, 2021, https://www.hrsa.gov/rural-health/telehealth/index.html.)

[21] Heartland Genetics Services Network, "Genetic Services," Accessed February 5, 2021, https://www.heartlandcollaborative.org/for-families/genetics-services/.

health and support information about 37 genetic conditions and receives more than 30,000 visitors a year.[22]

In FYs 2019 and 2020, the RGN program provided education and training to 13,916 professionals, including genetics providers and primary care providers, through webinars and in-person sessions. To better support clinicians, the RGN program offered Continuing Medical Education and Maintenance of Certificate credits to pediatricians for courses such as "Improving Care for Developmental Disabilities and Dysmorphic Features."

During this period, the RGN program has also trained 414 providers in telehealth and supported 2,948 clinical sites that use telehealth modalities through direct support, training, and technical assistance. The number of telehealth clinical and home sites in FY 2020 increased sharply, as the COVID-19 pandemic led to many patients receiving genetics visits from their homes. As a result, the program supported 12,293 patients receiving genetics services through telehealth in both home and clinical settings.

In response to COVID-19, in FY 2020, the program rapidly disseminated telehealth resources for providers and families within their regional networks. For example, the Western States RGN's "What to Expect from a Telehealth Visit" video for families has received more than 10,000 unique views and featured on the telehealth.hhs.gov website as a patient resource.[23]

Severe Combined Immunodeficiency Newborn Screening and Education Program

Severe Combined Immunodeficiency (SCID) is a genetic condition that affects one in 58,000 infants.[24] Infants born with SCID usually die within one year due to severe recurrent infections unless they are identified early and undergo successful stem cell transplantation. In 2010, SCID was added to the RUSP.

Congress provided funding to HRSA in 2014 to better support states in implementing SCID screening.[25] This program successfully led to the implementation of universal SCID screening programs in all 50 states (except for Pennsylvania, which screens for SCID selectively), as well as in Puerto Rico; Guam; and Washington, DC.

However, gaps remained in optimizing outcomes for infants with SCID. To this end, HRSA established the SCID Newborn Screening and Education Program in FY 2018, awarded to the Immune Deficiency Foundation.

PURPOSE

- **Improve outcomes for infants with SCID** detected through newborn screening by:

[22] New England Regional Genetics Network, "GEMSS – Genetics Education Materials for School Success," Accessed February 3, 2021, https://www.gemssforschools.org/.
[23] Health Resources and Services Administration, "Preparing for a virtual visit," Updated January 28, 2021, Accessed February 5, 2021, https://telehealth.hhs.gov/patients/preparing-for-a-video-visit/#preparing-for-your-virtual-visit.
[24] Association of Public Health Laboratories, "Severe Combined Immunodeficiency (SCID)," 2019, Accessed January 21, 2021, https://www.newsteps.org/disorders/scid.
[25] 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)

- Increasing awareness and knowledge about SCID and newborn screening for SCID among parents, families, health care providers, public health professionals, and the public.
- Providing education, training, and support for newborn screening programs.
- Educating families with children diagnosed with SCID and linking them to clinical and other services, especially those in rural and medically underserved areas.
- Improving clinical care through education and training for providers caring for individuals with SCID.

OBJECTIVES

- **Increase the use of a web-based resource on SCID** for families and health care providers.
- **Provide linguistically and culturally appropriate education and awareness materials** on SCID and SCID newborn screening to expectant parents, families, and providers.
- **Provide education on SCID treatment to rural and medically underserved populations** and connect affected families with SCID expert care centers for treatment, management, and follow-up services.
- **Offer increased trainings** (education sessions and/or consultations) to SCID health care providers.

FYs 2019 AND 2020 UPDATE

During this period, the SCID Newborn Screening and Education Program focused on the following activities to make progress toward its objectives:

- Developed an online educational program, accessed more than 23,000 times, which serves as a hub of information and resources, to help families and health care professionals navigate SCID and next steps after an out-of-range screening result.
- Hosted seven lunch and learn webinars for parents and caregivers with 174 participants.
- Developed a short animated video with the 6 stages of the SCID journey that has received 630 views.
- Devoted 2 episodes of the Immune Deficiency Foundation podcast, *Have you Herd?* to SCID-focused topics which have been played more than 1,400 times.
- Collaborated with NewSTEPs to maintain a resource library including SCID resources for newborn screening laboratory and follow-up stakeholders.

Quality Improvement in Newborn Screening Program

The Quality Improvement in Newborn Screening Program began in FY 2018 and is awarded to the Association of Public Health Laboratories. 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.

PURPOSE

- **Improve the outcomes of newborns with conditions identified through newborn screening** by:
  - Improving the **amount of time it takes to identify infants at high risk** for

having one of these conditions.
  - Improving the **processes used for detecting out-of-range results**.
  - Improving the **procedures for reporting out-of-range results** to providers.
  - Improving **methods newborn screening programs use to confirm diagnoses**.
  - **Addressing emerging issues** or any other newborn screening process or procedure that could negatively affect the quality, accuracy, or timeliness of newborn screening.

OBJECTIVES

- **Improve identification and follow-up on out-of-range results** specific to each state's processes.
- **Improve communication of screening results** to providers and families and confirming diagnoses.
- **Meet the recommended screening timeframes** for newborn screens identified by the Advisory Committee on Heritable Disorders in Newborn Children.[26]

FYs 2019 AND 2020 UPDATE

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

- Funded 30 state newborn screening programs to participate in a 5-year continuous quality improvement process. Programs receive continuous quality improvement training and access to resources, technical assistance, and an online database to evaluate their progress in the focus area of their choice.
- Created a model for replication, sharing, and sustainability of newborn screening continuous quality improvement projects. In this model, all state screening programs are able to evaluate progress, develop and share education strategies with other state programs, and utilize the NewSTEPs website for further communication and outreach efforts.

Part II lists additional activities and accomplishments during this period.

Newborn Screening Family Education Program

The Newborn Screening Family Education Program, awarded to the Genetic Alliance, 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.

PURPOSE

- Provide newborn screening education to parents and families to reduce morbidity and mortality caused by heritable conditions in newborns and children.

OBJECTIVES

- **Implement a newborn screening** training and education program for families.

[26] Health and Resources Services Administration, "Newborn Screening Timeliness Goals," September 2017, Accessed January 25, 2021, https://www.hrsa.gov/advisory-committees/heritable-disorders/newborn-screening-timeliness.html.

- **Increase awareness of newborn screening, as well as the dissemination of educational materials**, to parents and families, particularly in medically underserved communities.

FYs 2019 AND 2020 UPDATE

During this period, the Newborn Screening Family Education Program has implemented the following activities to make progress toward its goals:

- Implemented Navigate Newborn Screening, an educational module for families that covers newborn screening information and teaches skills that families can use to serve as leaders on newborn screening systems level teams.
- Recruited 155 total users—81 of whom represented family advocates—to Navigate Newborn Screening, as of June 2020.
- Initiated a family coach and ambassador program to provide peer-to-peer support targeted to medically underserved communities.
- Developed two one-page infographics, the first to convey family perspectives regarding strategies to improve telehealth experiences, and the second to teach the importance of maintaining confirmatory testing.
- Launched an online course curriculum to help families understand what to expect with newborn screening during the COVID-19 pandemic.
- Hosted two COVID-focused webinars and disseminated recordings and supplemental resources to their partner and family networks, with an emphasis on reaching medically underserved communities.

Newborn Screening State Evaluation Program

Since 2012, 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. The Newborn Screening State Evaluation Program supports states 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.

PURPOSE

- **Evaluate the effectiveness of newborn screening programs** in reducing the morbidity and mortality caused by heritable disorders in newborns and children by assessing the timeliness of screening, follow up, and referral to counseling or health care services.

OBJECTIVES

- **Collect and submit all available data on NewSTEPs quality indicators** for all newborns screened in 2019 and 2020 to a national newborn screening data repository.
- **Establish a sustainability plain for all participating states** to support data collection activities beyond the program's period of performance.

FYs 2019 AND 2020 UPDATE

Colorado, Connecticut, Hawaii, New York, Oklahoma, and Rhode Island received funding from HRSA to report data to NewSTEPs. In FY 2019, five of these six states entered data on at least

two quality indicators. To date, two states reported FY 2020 data, with the other states indicating data reporting delays owing to the COVID-19 pandemic. Future funding initiatives will use the lessons learned from this initiative to support state-level informatics and follow-up needs.

Innovations in Newborn Screening Interoperability

In September 2020, HRSA implemented the Innovations in Newborn Screening Interoperability program (INBSI) to enhance data interoperability in the newborn screening system.[27]

Ensuring that every baby is screened, specimens are transported and tested efficiently and results are interpreted and reported in a timely manner, 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 data can be managed, controlled, and shared.

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.

PURPOSE

- **Reduce morbidity and mortality associated with heritable disorders** in newborns and children by enhancing data interoperability in the newborn screening system.
- **Improve the ability of states to conduct screening and report results in a timely manner,** increasing the likelihood that infants with a newborn screening condition will receive timely diagnoses and treatment.

OBJECTIVES

- **Establish a public-facing website** for resources and materials on interoperability for use in newborn screening programs.
- **Provide direct technical assistance and support** to states to develop comprehensive state interoperability plans and timelines for achieving interoperability.
- **Collect and develop data from the states using INBSI's interoperability resources and materials** for the publication of a national report on INBSI's work.

FYs 2019 AND 2020 UPDATE

Because this program began in September 2020, HRSA will report activities and accomplishments in its next report to Congress.

[27] 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 June 24, 2021, https://www.healthit.gov/topic/interoperability.)

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

Section 1110 of the PHS Act focuses on evaluating the effectiveness of newborn screening and follow-up programs and is administered by HRSA. 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 2019 and 2020 as they relate to Section 1110.

Newborn Screening Data Repository and Technical Assistance Program

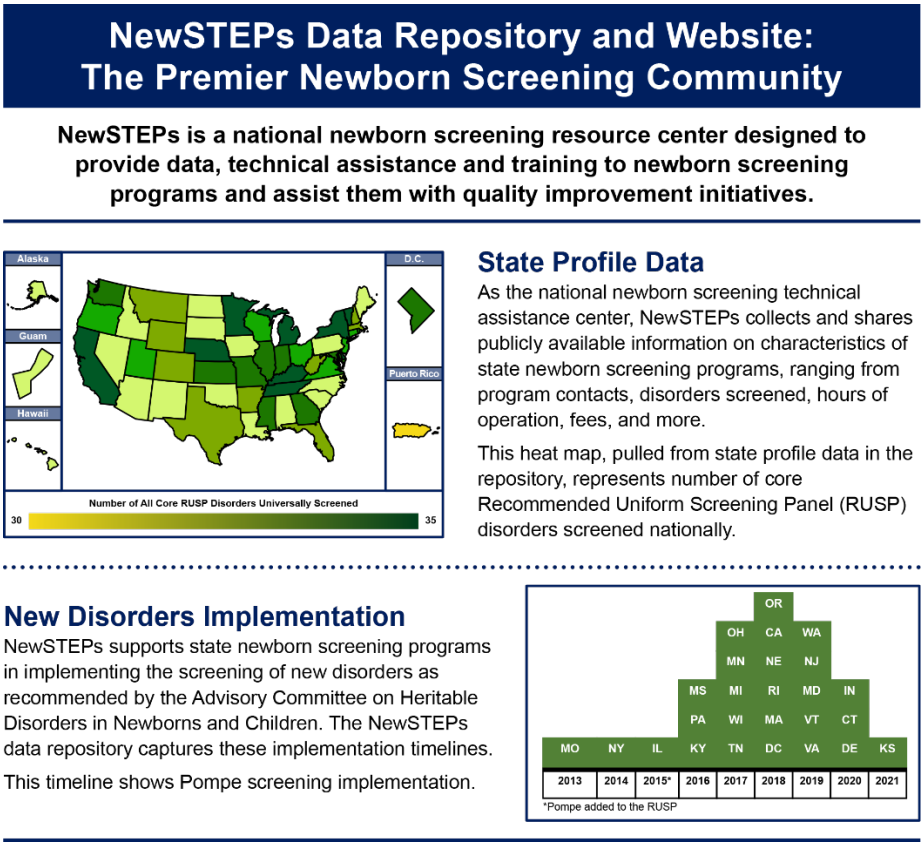
Through the Newborn Screening Data Repository and Technical Assistance Program, NewSTEPs provides resources to state newborn screening programs to evaluate the effectiveness of their processes and use real-time data to inform quality improvements.

KEY ACCOMPLISHMENTS

- At their request, NewSTEPs conducted reviews[28] of the Minnesota and Indiana newborn screening programs in FY 2019 and Tennessee's and New York's programs in FY 2020 to help them address continuity of operations needs and identify newborn screening systems process improvements.
- NewSTEPs provides access to real-time reports and infographics from the data repository, as noted below:

[28] Association of Public Health Laboratories, "Site Review," Accessed February 3, 2021, https://www.newsteps.org/quality-improvement-practices/site-review.

Figure 2: NewSTEPs Data Repository



Source: Newborn Screening Technical assistance and Evaluation Program (NewSTEPs), *https://www.newsteps.org/*, as of 4/21

Quality Improvement in Newborn Screening Program

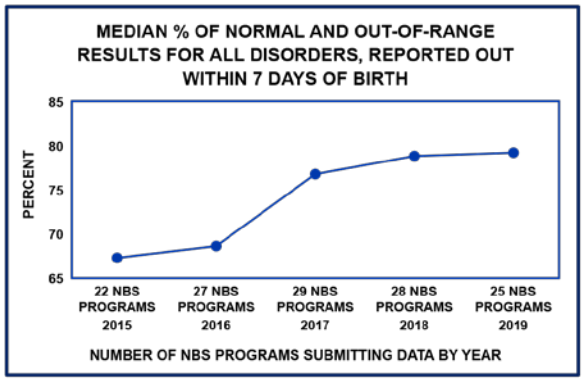
As discussed in Part I, page 4, in FYs 2019 and 2020, this program enhanced the quality of newborn screening across 30 states and territories, focusing on timeliness, detection of out-of-range results, reporting out-of-range results to providers, confirming a diagnosis, and addressing emerging issues or any other newborn screening process or procedure that could negatively affect the quality, accuracy, or timeliness of newborn screening.

KEY ACCOMPLISHMENTS

- Eleven state and territorial newborn screening programs participated in the first Quality Improvement Projects collaborative cohort. The first cohort began in November 2019 and implemented 13 continuous quality improvement projects.
- Six state newborn screening programs are participating in the second cohort, which began in May 2020.
- Webinars were provided on these topics:
    - An Overview of Project Charters and Action Plans
    - Strategies for Beginning a Quality Improvement Project
- Collectively, these programs have reported a decreased number of unsatisfactory specimens and improvements in timeliness of newborn screening results reporting.

- NewSTEPs provided funding opportunities to states to further support newborn screening programs interested in implementing continuous quality improvement projects.
- Quality Improvement Project outcomes and deliverables are tracked via monthly reports and coaching calls.
- The NewSTEPs website and data repository collects and reports on quality improvement.

Figure 3: 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: Newborn Screening Technical assistance and Evaluation Program (NewSTEPs), *https://www.newsteps.org/*, as of 4/21

The COVID-19 pandemic has had a significant impact on program initiatives. These include:

- Delays due to reassignment of screening staff for COVID-19 response.
- Newborn screening project partners (e.g., hospitals, clinicians) and stakeholders' focus on COVID-19 response.
- Inability for some awarded programs to participate at all in the Quality Improvement Project at this time.
- Postponement of staff and member travel to Quality Improvement Project in-person meetings 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 '**Cleaning Methods for Pipette Tips**' webinar, where state newborn screening programs shared experiences in pipette tip washing to address supply limitations as a result of the COVID-19 pandemic and discussed processes to conduct method validation and tests for cross-contamination.
- The **'Staffing and Telework'** webinar, which provided state newborn screening programs with a forum to discuss navigating the sudden shift to teleworking during the pandemic and identifying needs to support robust teleworking (e.g., business phones, secure email, video capabilities).
- The **'Electronic Reporting'** webinar, which provided a platform for several states (i.e., Nevada, Texas, and Tennessee) to discuss their unique efforts to reduce paper reporting and newborn screening messaging.

- The **'Virtual Engagement'** webinar that featured state perspectives from Hawaii, North Dakota, and New York, as well as Baby's First Test, to share best practices for successful virtual engagement of newborn screening stakeholders during the pandemic.

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.

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

PURPOSE

- **Provide clear and up-to-date information, materials, and resources about newborn screening** in the United States to increase awareness, knowledge, and understanding of newborn screening and genetic conditions.

OBJECTIVES

- **Enable parents and family members of newborns, health professionals, industry representatives, and other members of the public** to increase their awareness, knowledge, and understanding of newborn screening.
- **Increase awareness, knowledge, and understanding of newborn screening** for expectant individuals and families.
- **Provide current educational and family support and services information**, materials, resources, research, and information on newborn screening.
- **Provide information organized in a clear format** for multiple audiences that promotes sharing and dissemination of authoritative and/or evidence-based information; lay informational and educational materials; community training initiatives; health care provider educational materials; newborn screening best practices and guidelines; and other information and tools that promote culturally sensitive education and decision-making regarding newborn screening for heritable disorders.
- **Include current information on quality indicators to measure performance of newborn screenings**, such as false-positive rates and other quality indicators, as determined by the Advisory Committee on Heritable Disorders in Newborns and Children.
- **Provide current information on the number of conditions screened** in each state.
- **Contain evidence-based guidelines** related to diagnosis, counseling, and treatment with respect to conditions detected by newborn screening.

FYs 2019 AND 2020 UPDATE

The following content is available at https://newbornscreening.hrsa.gov/:

- A definition of newborn screening, description of the newborn screening process, including follow up, diagnosis, and treatment.
- A description of the conditions on the RUSP.

- Links to connect parents, parents-to-be, and health care providers with state-specific newborn screening resources.
- Resources for readers to learn about updates in newborn screening.

The program is undertaking the following activities for the FY 2020 project period:

- Creating a Spanish-language website.
- Creating four factsheets for newborn screening issues: "Learning the Basics About Newborn Screening," "What Health Care Providers Should Know about Newborn Screening," "Your Guide to Newborn Blood Spot Screening," and "Understanding Newborn Blood Spot Storage and Retention."
- Developing a social media outreach plan.

Since the launch of the NBSIC in September 2020, the website has received over 14,000 views.

Part IV: Laboratory Quality & 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, for ensuring the accuracy of newborn screening blood spot tests conducted by public health laboratories.

PURPOSE

- **Provide unique services directly to laboratories** to improve the detection of newborn disease.

OBJECTIVES

- **Support newborn screening laboratories** by providing 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.
- **Prepare, certify, and distribute** approximately one million dried bloodspot quality assurance materials that mimic disease samples to participating laboratories each year.
- **Provide technical support and training** to state and territorial laboratories to enhance nationwide laboratory capacity, quality, and capability.
- **Develop new methods** for recent and anticipated additions to the RUSP.
- **Improve existing methods** to increase accuracy of detecting newborn diseases.
- **Help laboratories add new conditions to screening panels** and implement new screening technologies to improve disease detection and severity prediction.
- **Develop, conduct, and host hands-on training workshops** on current and innovative laboratory techniques for state newborn screening programs.
- **Evaluate filter paper used to produce blood collection cards** for newborn screening to ensure the quality of the paper complies with set performance criteria.
- **Expand critical infrastructure and support state laboratory staff** to interpret results and minimize false positives.

FYs 2019 AND 2020 UPDATE

During this period, the program focused on the following activities to make progress toward its objectives:

- **Expanded and enhanced quality assurance services:**
  - Provided quality assurance services to nearly 700 laboratories, covering all U.S. states and territories as well as 86 countries.
  - Expanded critical infrastructure and modified in-house procedures to safely create and ship essential quality assurance materials, including the new Spinal Muscular Atrophy (SMA) pilot proficiency testing program, to state newborn screening programs to assure the accuracy of ongoing screening during the COVID-19 pandemic.
  - Modernized database using Microsoft Dynamics CRM to better engage with the nearly 700 newborn screening laboratory participants. This upgrade streamlined workflows for quality assurance material enrollment and shipping, reducing packing errors and increasing efficiency. CDC also updated the web application for reporting results, which now connects to the new CRM and uses Power BI to configure data for better visualization of participant evaluations.
  - Published resources online to support newborn screening program efforts in emergency preparedness, contingency planning, and COVID-19 response.
  - Developed and published procedures for preparing quality assurance materials for the screening and characterization of adenosine deaminase deficiency-SCID.
  - Developed new techniques to produce quality assurance materials using transduced lymphocytes from patient donor families. This new approach provides a renewable source of dried blood spot material and increases CDC's ability to support states using molecular detection of diseases in newborns.
  - Developed a new newborn screening proficiency testing program for SMA in support of its addition to the RUSP.
    Evaluated and published the performance characteristics of nine DNA extraction methods for dried blood spots and their use in next generation sequencing of the cystic fibrosis transmembrane conductance regulator gene.
- **Provided timely technical support and training:**
  - Performed four program-requested Molecular Assessment Program (MAP) site visits in Kansas, Colorado, Tennessee, and California in 2019 and developed customized reports to aid in molecular quality improvements for each program. The newly developed Follow-Up Education module was also successfully piloted during the Kansas MAP site visit. MAP visits were not conducted in 2020 because of COVID-19 restrictions.
  - Provided funding through a cooperative agreement to the New York newborn screening program to host a two-and-a-half-day workshop during the summer of 2019 on laboratory screening for Pompe disease, Mucopolysaccharidosis I (MPS I), SMA, and X-linked Adrenoleukodystrophy (X-ALD). This program developed and maintained laboratory capability by providing a combination of lectures and hands-on

training to ensure quality and consistency in programs across the country. The program was not held in 2020 because of COVID-19 restrictions.
    - Developed on-site laboratory training for state laboratory personnel. These courses addressed training in both biochemical and molecular methods. CDC hosted the 2020 Molecular Methods course in February 2020, prior to COVID-19 restrictions.
- **Developed new methods and improved existing methods:**
    - Developed new molecular newborn screening method to detect SMA. The CDC-developed method has been adopted by 21 states or their contract designees. In addition, CDC provided support to 10 additional states or their contract designees to ensure high quality SMA screening. In all, CDC provided support that helped laboratories to begin or sustain SMA screening for almost 67 percent of newborns, or more than 2.5 million babies born annually in the United States.
    - Developed improved mass spectrometry methods; as a result, Michigan started X-ALD and Guanidinoacetate methyltransferase (GAMT) deficiency screening in October 2019, and Utah and Minnesota began in-house testing of amino acids, acylcarnitines, and succinylacetone-GAMT method to improve high throughput newborn screening protocols.
    - Developed and published a new way to harmonize biochemical mass spectrometry results across states. The method uses CDC-created reference materials and enables states to compare results across labs to increase uniformity in newborn screening data interpretation. States now have a way to establish preliminary cutoffs for new diseases and to reduce potential false positive and negative rates.
    - Improved cystic fibrosis detection in newborns by characterizing eight DNA extraction procedures from dried bloods spots for use in massively parallel next generation sequencing of the gene involved in cystic fibrosis.
    - Provided funding and assistance to states to improve laboratory screening methods through newborn screening cooperative agreements. With this support, New York developed a protocol to optimize multiplex testing for SMA. New York has also developed protocols for additional tests to detect MPS I, Pompe, and X-ALD. Massachusetts has developed tertiary tests that can be used when further clarification is required for SMA screening.
    - Supported interlaboratory harmonization projects using the CDC harmonization approach. These data harmonization projects enable states to compare results, which is particularly critical for rare conditions. New York led the project on X-ALD harmonization, Tennessee led the project on lysosomal storage disorders harmonization, and Michigan and Utah led a project harmonizing data on amino acid, organic acid, and fatty acid oxidation disorders.
- **Assisted state expansion to screen for new conditions:**
    - Expedited nationwide screening of new high-priority conditions on the RUSP by doubling direct support to seven state programs through a 2-year cooperative agreement that continues to provide critical laboratory equipment, staffing, and supplies for population-based testing. The seven state recipients include Colorado, Kansas, Massachusetts, New York, Tennessee, Texas, and Utah. With

CDC funding, Colorado and Massachusetts also supported laboratory testing for newborn screening programs located in Wyoming and states within New England, respectively. CDC funding supported the addition of new tests to state panels; improved screening performance for existing conditions to improve timeliness, increased test sensitivity and reduced false positives; and improved the states' abilities to accurately and quickly interpret screening results. As a result of direct support:

- The following states added SMA: Colorado, Kansas, New York, New Hampshire, Rhode Island, Tennessee, Vermont, and Wyoming.
- The following states added X-ALD: Rhode Island, Texas, Utah, and Vermont.
- The following states added MPS I: New York and Vermont.
- Rhode Island added Pompe Disease.

Part V: Interagency Coordinating Committee on Newborn & 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 FDA Commissioner, and the NIH Director, or their designees. **Section 1114** of the PHS Act authorizes the ICC and its activities.

PURPOSE

Coordinate collaborative efforts for newborn and child screening among all 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 2019 and 2020, the Secretary of Health and Human Services did not convene the ICC, as there were no Committee recommendations to review.

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

The Newborn Screening Contingency Plan takes into account 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 face 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.[29]

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

[29] 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.

Contingency Plan as needed. The 2017 version added point-of-care screening for critical congenital heart defects and newborn hearing and streamlined text into a usable checklist tool for emergency planners at the state and local levels.[30] In 2021-2022, CDC plans in collaboration with HRSA to update the plan in consultation with newborn screening subject matter experts and provide guidance to states based on lessons learned from the impact of COVID-19 on newborn screening systems.

Part VII: Funding Amounts

The FYs 2019 and 2020 funding amounts for HRSA and CDC newborn screening activities are in the table below.

Table 2: Funding Amounts

Program/Initiative	FY 2019 Funding	FY 2020 Funding
HRSA		
Newborn Screening Data Repository and Technical Assistance Program	$1,500,000	$1,700,000
Regional Genetics Networks Program	$4,200,000	$4,200,000
SCID Newborn Screening and Education Program	$1,930,000	$2,970,000
Quality Improvement in Newborn Screening Program[31]	$3,300,000	$3,300,000
Newborn Screening Family Education Program	$400,000	$500,000
Newborn Screening State Evaluation Program	$2,250,000	$2,250,000
Innovations in Newborn Screening Interoperability	n/a	$1,300,000
Clearinghouse of Newborn Screening Information	$353,639.93	$521,516.75
Total HRSA Funding	**$13,933,639.93**	**$16,741,516.75**
CDC		
Laboratory Quality and Surveillance	$16,000,000	$17,000,000
National Contingency Plan for Newborn Screening	n/a	n/a
Total CDC Funding	**$16,000,000**	**$17,000,000**

Data Source: HRSA and CDC, as of 3/17/2021

Part VIII: Summary and Conclusion

The newborn screening programs administered by HRSA and CDC in FYs 2019 and 2020 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 SMA, MPS I, and X-

[30] Ibid.

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

ALD, the three newest conditions added to the RUSP.

- Developed resources, including in-person and remote training and online educational resources, from fact sheets to educational videos, geared toward increasing awareness of the importance of newborn screening.
- Provided opportunities for quality improvement, education, training, and financial support to all state and territory newborn screening programs.
- 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.
- Expanded CDC and state capacity to interpret complex newborn screening tests for better detection of disease.
- Modernized systems for data collection and evaluation and for collaboration and engagement with newborn screening programs and stakeholders.

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 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 chooses disorders on 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 screening of every newborn for all disorders on the RUSP.[32]

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 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 by the Advisory Committee on Heritable Disorders in Newborns and Children and supported by the Secretary.[33]

Recommended Uniform Screening Panel Core Conditions

A condition on the newborn screening panel is classified 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 July 2018)[34]

Core Condition	Metabolic Disorder			Endocrine Disorder	Hemoglobin Disorder	Other Disorder
	Organic acid condition	Fatty acid oxidation disorders	Amino acid disorders			
Propionic acidemia	Included					
Methylmalonic acidemia (methylmalonyl-CoA mutase)	Included					
Methylmalonic acidemia (cobalamin disorders)	Included					
Isovaleric acidemia	Included					
3-Methylcrotonyl-CoA carboxylase deficiency	Included					
3-Hydroxy-3-methylglutaric aciduria	Included					
Holocarboxylase synthetase deficiency	Included					
ß-Ketothiolase deficiency	Included					
Glutaric acidemia type I	Included					
Carnitine uptake defect/carnitine transport defect		Included				
Medium-chain acyl-CoA dehydrogenase deficiency		Included				
Very long-chain acyl-CoA dehydrogenase deficiency		Included				

[32] Health Resources and Services Administration, "Recommended Uniform Screening Panel," February 2019, Accessed February 1, 2021, https://www.hrsa.gov/advisory-committees/heritable-disorders/rusp/index.html.
[33] Ibid.
[34] Ibid.

Core Condition	Metabolic Disorder			Endocrine Disorder	Hemoglobin Disorder	Other Disorder
	Organic acid condition	Fatty acid oxidation disorders	Amino acid disorders			
Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency		Included				
Trifunctional protein deficiency		Included				
Argininosuccinic aciduria			Included			
Citrullinemia, type I			Included			
Maple syrup urine disease			Included			
Homocystinuria			Included			
Classic phenylketonuria			Included			
Tyrosinemia, type I			Included			
Primary congenital hypothyroidism				Included		
Congenital adrenal hyperplasia				Included		
SS disease (Sickle cell anemia)					Included	
S, βeta-thalassemia					Included	
SC disease					Included	
Biotinidase deficiency						Included
Critical congenital heart disease						Included
Cystic fibrosis						Included
Classic galactosemia						Included
Glycogen Storage Disease Type II (Pompe)						Included
Hearing loss						Included
Severe combined immunodeficiencies						Included
Mucopolysaccharidosis Type 1						Included
X-linked Adrenoleukodystrophy						Included
Spinal Muscular Atrophy due to homozygous deletion of exon 7 in SMN1						Included

Recommended Uniform Screening Panel Secondary Conditions

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

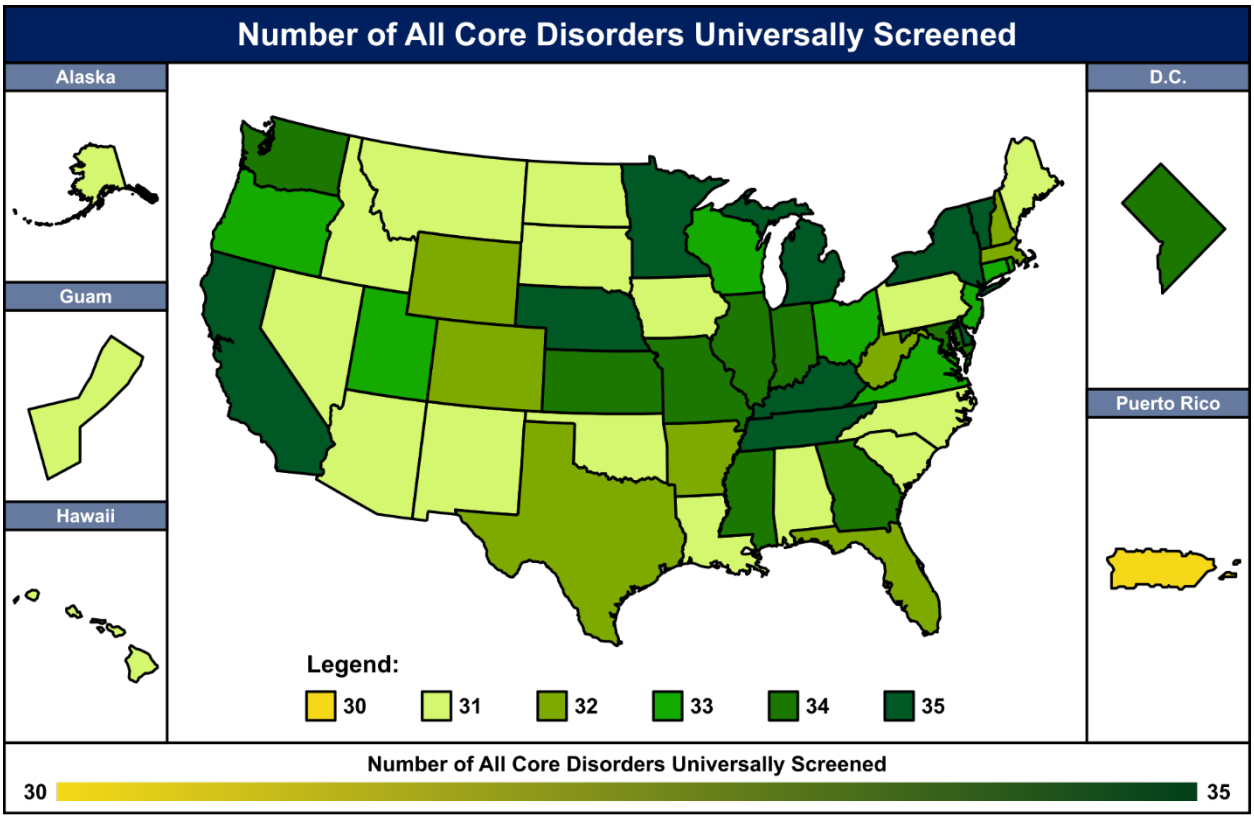
RUSP Secondary Conditions (as of July 2018)[35]

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

[35] Health Resources and Services Administration, "Recommended Uniform Screening Panel," February 2019, Accessed February 1, 2021, https://www.hrsa.gov/advisory-committees/heritable-disorders/rusp/index.html.

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.



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

Number of Core Disorders Universally Screened Per State

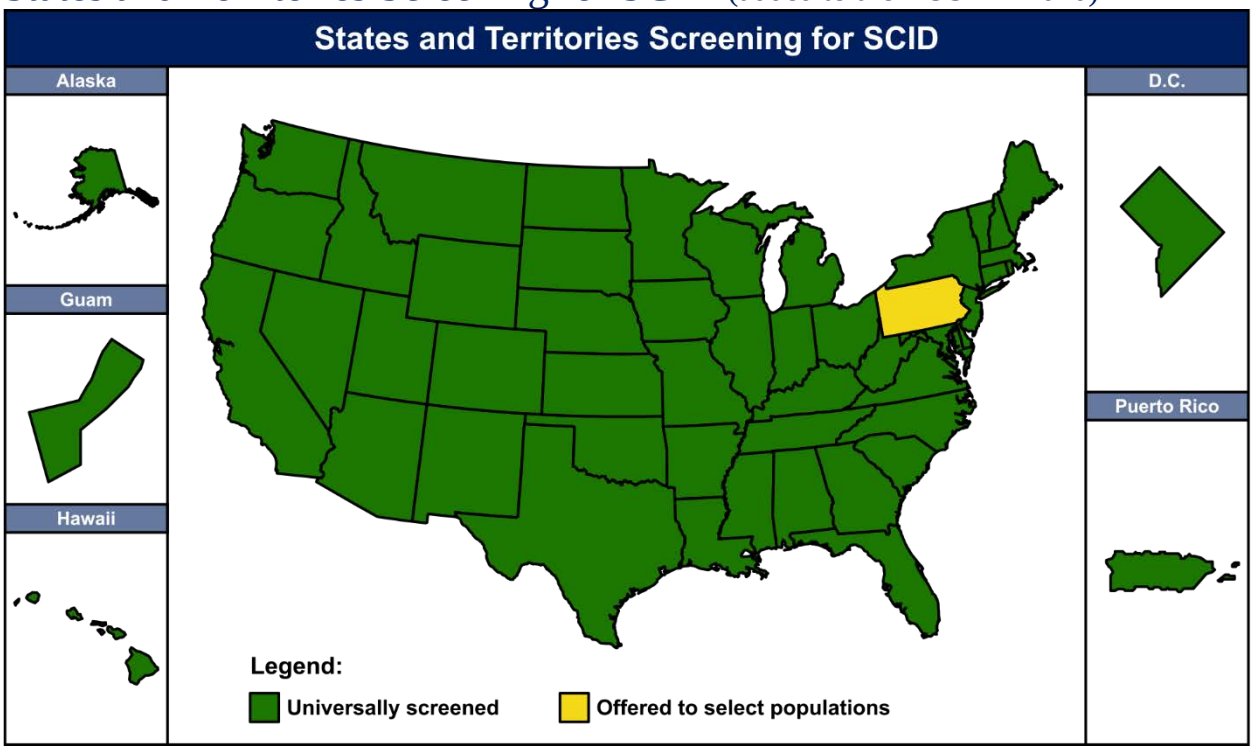
Alabama: 31
Alaska: 31
Arizona: 31
Arkansas: 32
California: 35
Colorado: 32
Connecticut: 33
Delaware: 35
District of Columbia: 34
Florida: 32
Georgia: 33
Guam: 31
Hawaii: 31
Idaho: 31
Illinois: 34
Indiana: 34
Iowa: 31
Kansas: 34
Kentucky: 35
Louisiana: 31
Massachusetts: 32
Maine: 31
Maryland: 34
Michigan: 35
Minnesota: 35
Mississippi: 33
Missouri: 34
Montana: 31
Nebraska: 35
Nevada: 31
New Hampshire: 32
New Jersey: 33
New Mexico: 31
New York: 35
North Carolina: 31
North Dakota: 31
Ohio: 33
Oklahoma: 31
Oregon: 33
Pennsylvania: 31
Puerto Rico: 30
Rhode Island: 34
South Carolina: 31
South Dakota: 31
Tennessee: 35
Texas: 32
Utah: 33
Vermont: 35
Virginia: 33
Washington: 34
West Virginia: 32
Wisconsin: 33
Wyoming: 32

Source: The Newborn Screening Technical assistance and Evaluation Program (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 SCID, Pompe disease, MPS I, X-ALD, and SMA.

States and Territories Screening for SCID (added to the RUSP in 2010)



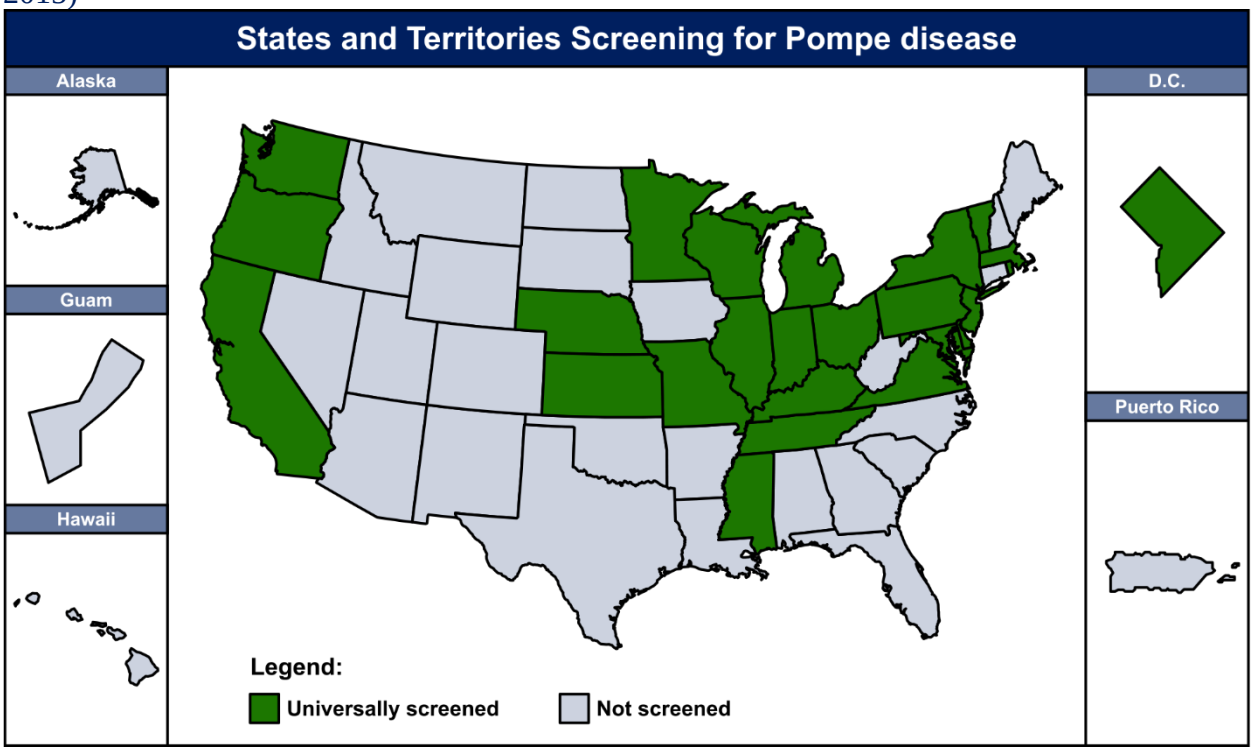
Source: The Newborn Screening Technical assistance and Evaluation Program (NewSTEPs), https://www.newsteps.org.

States and Territories Newborn Screening Programs Universally Screening for SCID

Alabama
Alaska
Arizona
Arkansas
California
Colorado
Connecticut
Delaware
District of Columbia
Florida
Georgia
Guam
Hawaii
Idaho
Illinois
Indiana
Iowa
Kansas
Kentucky
Louisiana
Massachusetts
Maine
Maryland
Michigan
Minnesota
Mississippi
Missouri
Montana
Nebraska
Nevada
New Hampshire
New Jersey
New Mexico
New York
North Carolina
North Dakota
Ohio
Oklahoma
Oregon
Puerto Rico
Rhode Island
South Carolina
South Dakota
Tennessee
Texas
Utah
Vermont
Virginia
Washington
West Virginia
Wisconsin
Wyoming
Total: 52 programs

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

States and Territories Screening for Pompe disease (added to the RUSP in 2015)



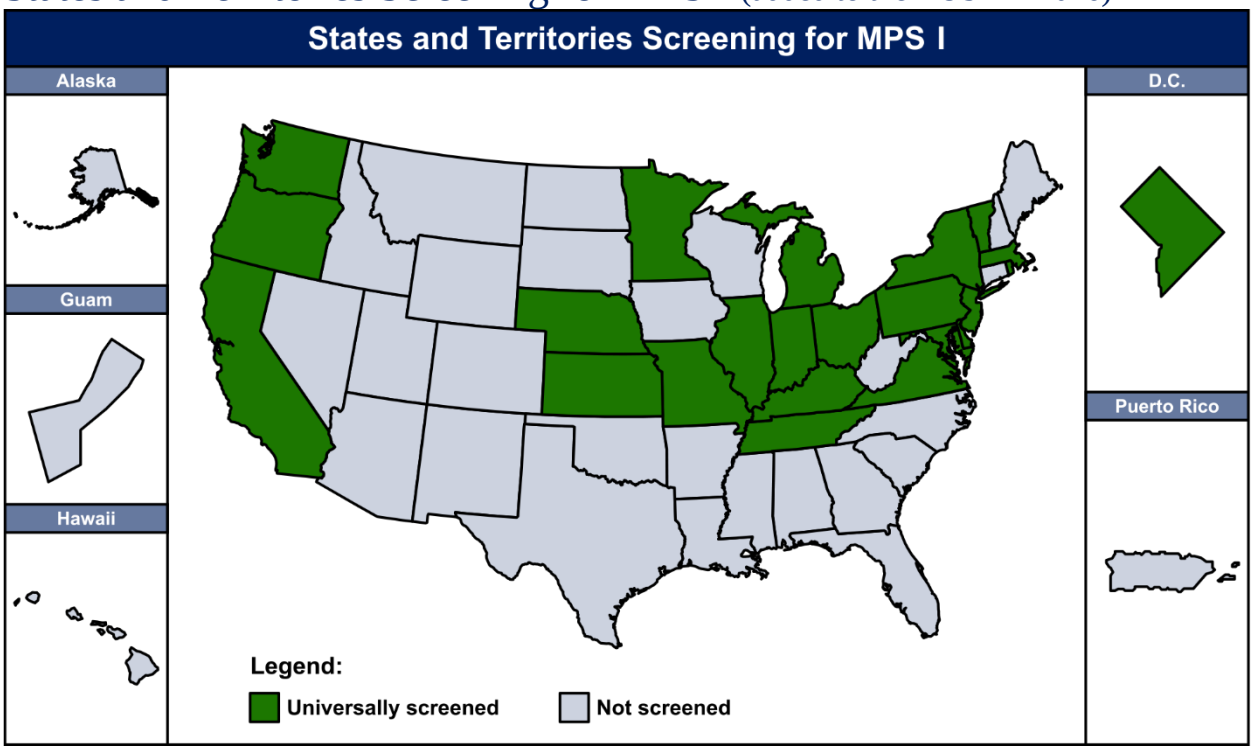
Source: The Newborn Screening Technical assistance and Evaluation Program (NewSTEPs), https://www.newsteps.org.

States and Territories Newborn Screening Programs Universally Screening for Pompe disease

California	Maryland	New Jersey	Vermont
Delaware	Massachusetts	New York	Virginia
District of Columbia	Michigan	Ohio	Washington
Illinois	Minnesota	Oregon	Wisconsin
Indiana	Mississippi	Pennsylvania	**Total: 25 programs**
Kansas	Missouri	Rhode Island	
Kentucky	Nebraska	Tennessee	

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

States and Territories Screening for MPS I (added to the RUSP in 2016)



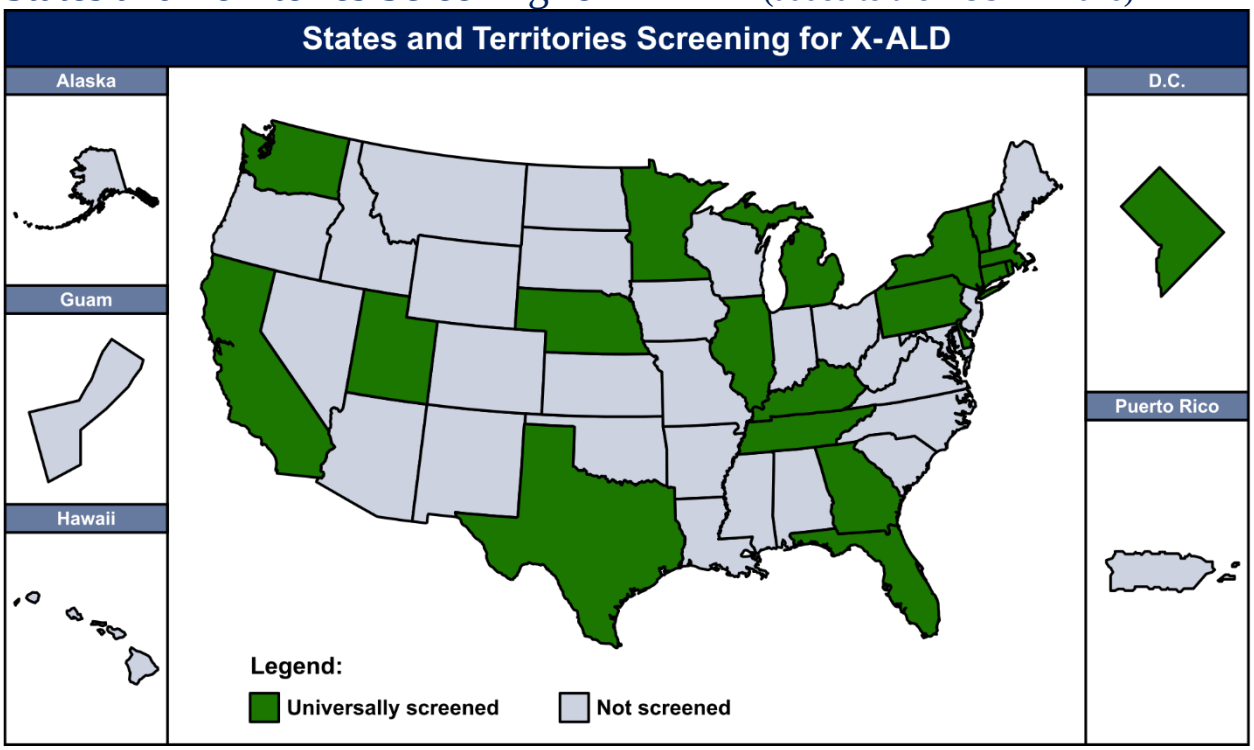
Source: The Newborn Screening Technical assistance and Evaluation Program (NewSTEPs), https://www.newsteps.org.

States and Territories Newborn Screening Programs Universally Screening for MPS I

California	Kentucky	Nebraska	Rhode Island
Delaware	Maryland	New Jersey	Tennessee
District of Columbia	Massachusetts	New York	Vermont
Illinois	Michigan	Ohio	Virginia
Indiana	Minnesota	Oregon	Washington
Kansas	Missouri	Pennsylvania	**Total: 23 programs**

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

States and Territories Screening for X-ALD (added to the RUSP in 2016)



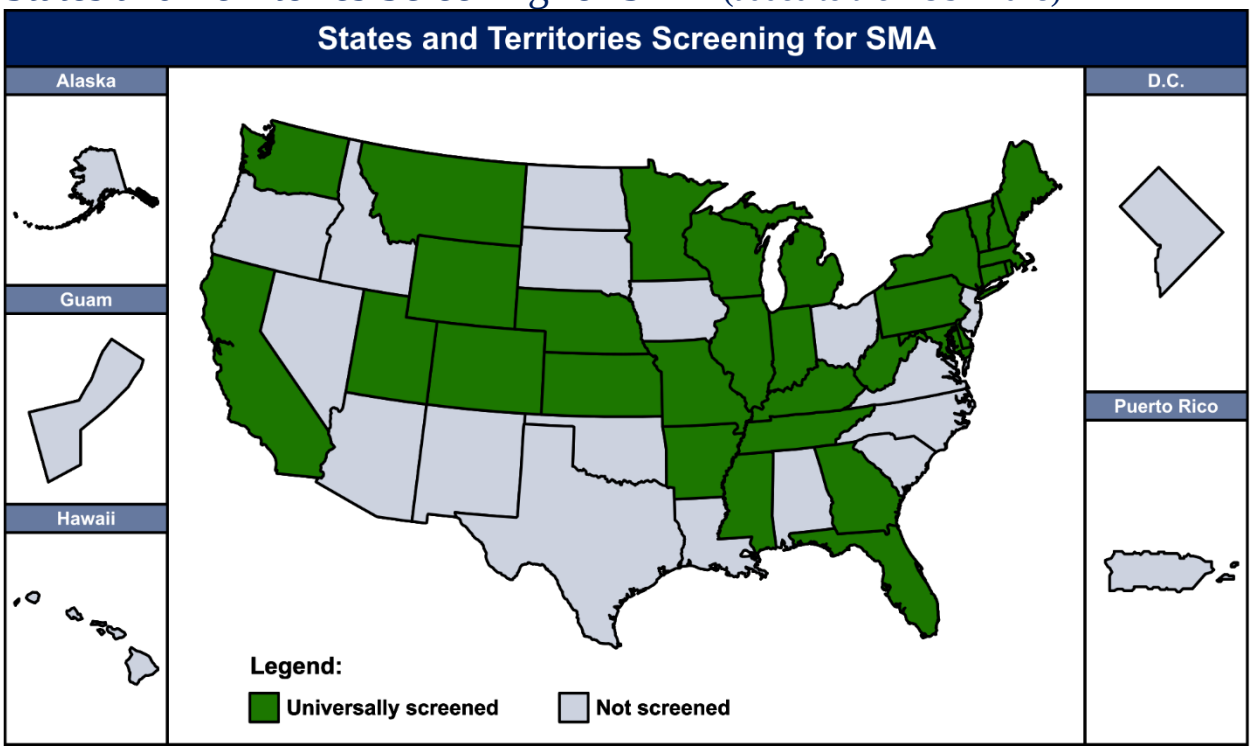
Source: The Newborn Screening Technical assistance and Evaluation Program (NewSTEPs), https://www.newsteps.org.

States and Territories Newborn Screening Programs Universally Screening for X-ALD

California	Illinois	New York	Vermont
Connecticut	Kentucky	Pennsylvania	Washington
Delaware	Massachusetts	Rhode Island	**Total: 20 programs**
District of Columbia	Michigan	Tennessee	
Florida	Minnesota	Texas	
Georgia	Nebraska	Utah	

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

States and Territories Screening for SMA (added to the RUSP 2018)



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

States and Territories Newborn Screening Programs Universally Screening for SMA

Arkansas	Indiana	Mississippi	Tennessee
California	Kansas	Missouri	Utah
Colorado	Kentucky	Montana	Vermont
Connecticut	Maine	Nebraska	Washington
Delaware	Maryland	New Hampshire	West Virginia
Florida	Massachusetts	New York	Wisconsin
Georgia	Michigan	Pennsylvania	Wyoming
Illinois	Minnesota	Rhode Island	**Total: 31 programs**

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