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

REPORT TO CONGRESS

Newborn Screening Activities

Fiscal Year 2016

Executive Summary

The newborn screening activities and associated expenditures of funds discussed in this report were 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 Act (Act) (42 U.S.C. §§ 300b-8, 300b-9, and 300b-11 through 300b-14). The programs and activities under this Act were established to enhance, improve, or expand the ability of states and local public health agencies to provide screening, counseling, and health care services to newborns and children having or at risk for having heritable disorders.

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

- Section 1109: Improved Newborn Screening for Heritable Disorders - administered by HRSA;
- Section 1110: Evaluating the Effectiveness of Newborn and Child Screening and Follow-up Programs - administered by HRSA;[1]
- Section 1112: Clearinghouse of Newborn Screening Information - administered by HRSA;
- Section 1113: Laboratory Quality and Surveillance - administered by CDC;
- Section 1114: Interagency Coordinating Committee on Newborn and Child Screening - administered by HRSA and CDC; and
- Section 1115: National Contingency Plan for Newborn Screening - administered by CDC.

This is the second report on newborn screening activities administered by HRSA and CDC. The report covers activities in fiscal year 2016.

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

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Acronym List

AHRQ	Agency for Healthcare Research and Quality
ACHDNC	Advisory Committee on Heritable Disorders in Newborns and Children
CCHD	Critical Congenital Heart Disease
CDC	Centers for Disease Control and Prevention
FDA	Food and Drug Administration
HRSA	Health Resources and Services Administration
ICC	Interagency Coordinating Committee
NBS	Newborn Screening
NBS TA Center	Newborn Screening Data Repository and Technical Assistance Center
NIH	National Institutes of Health
NSQAP	Newborn Screening Quality Assurance Program
RUSP	Recommended Uniform Screening Panel
SCID	Severe Combined Immunodeficiency

Legislative Language

This report is provided to Congress as required by 42 U.S.C. 300b-17, as amended by the Newborn Screening Saves Lives Reauthorization Act of 2014 (P.L. 113-240), which states, in part:

> *(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.*

This is the second report on newborn screening activities administered by the Health Resources and Services Administration (HRSA) and the Centers for Disease Control and Prevention (CDC). The report covers activities in fiscal year 2016.

Introduction

Newborn screening is a vital public health program that identifies newborns with disorders that are not apparent at birth but require immediate intervention. Every year, nearly 4 million infants are born in the United States and nearly all are screened by state newborn screening programs for certain heritable disorders and medical conditions on the Recommended Uniform Screening Panel (RUSP) (see Appendix A).

The RUSP is a list of conditions adopted by the Secretary of Health and Human Services (HHS) and recommended for screening at birth as part of states' newborn screening (NBS) programs. The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) provides recommendations to the Secretary of HHS with regard to which conditions ought to be included on the RUSP. The Secretary then makes the final decision on whether to add, or not add, a recommended condition to the RUSP. The ACHDNC uses a decision matrix that categorizes evidence related to the potential net benefit of screening, the ability of states to screen for the disorder, and the availability of effective treatments to decide whether to recommend the addition of a condition to the RUSP. 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. "Secondary conditions" are conditions that can be identified when screening for a core condition, or as a consequence of confirmatory testing following a positive NBS result (i.e. a result outside of the normal reference range). Although states ultimately determine what disorders their NBS program will screen for, the RUSP establishes a standardized list of

disorders that have undergone a rigorous evidence review and that are supported by the ACHDNC and the Secretary of HHS.

Screening practices and the types of disorders screened for in newborns vary from state to state (see Appendix B). Infants who test positive for one of these disorders receive rapid identification, which may result in early intervention and improved outcomes, including lifesaving treatments. Newborn screening saves or improves the lives of more than 12,000 infants in the United States each year. Federal agencies provide key support to newborn screening programs and the community to help ensure proper and timely screening and intervention.

Overview

This report provides information on ongoing activities authorized by sections 1109, 1110, and 1112 through 1115 of Title XI of the Public Health Service Act (Act), including the amounts expended on such activities and the structure, beneficiaries, and impact of the activities.

- Section 1109: Improved Newborn and Child Screening for Heritable Disorders, administered by HRSA, 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 having or at risk for heritable disorders.

- Section 1110: Evaluating the Effectiveness of Newborn and Child Screening and Follow-up Programs, administered by HRSA,[2] authorizes grants to provide for demonstration programs to evaluate the effectiveness of screening, follow-up, counseling, or health care services in reducing the morbidity and mortality caused by heritable disorders in newborns and children.

- Section 1112: Clearinghouse of Newborn Screening Information, administered by HRSA, authorizes the establishment and maintenance of a central clearinghouse, available on the internet, of current educational and family support and services information, materials, resources, research, and data on newborn screening.

- Section 1113: Laboratory Quality & Surveillance, administered by CDC, authorizes, after taking into consideration the expertise of the ACHDNC, the provision of quality assurance for laboratories involved in screening newborns and children for heritable disorders. This includes quality assurance for newborn screening tests, timeliness for processing such tests, performance evaluation services, technical assistance and technology transfer to newborn screening laboratories to ensure analytic validity and utility of screening tests, and

[2] CDC and HRSA are both authorized to administer programs in under this section. However, CDC does not currently administer any programs under this section.

appropriate quality control and other performance test materials to evaluate the performance of new screening tools. This section also authorizes, after taking into consideration the expertise of the ACHDNC, the coordination of laboratory surveillance activities. Surveillance activities include standardized data collection and reporting, the use of electronic health records, and the promotion of data sharing regarding newborn screening with state-based birth defects and development disabilities monitoring programs.

- Section 1114: Interagency Coordinating Committee on Newborn and Child Screening, administered by HRSA and CDC, authorizes the Interagency Coordinating Committee (ICC) on Newborn and Child Screening to assess existing activities and infrastructure in order to make recommendations for programs to collect, analyze, and make available data on the heritable disorders recommended by the ACHDNC. The ICC is also responsible for making recommendations for the establishment of regional centers for the conduct of applied epidemiological research on effective interventions to promote the prevention of poor health outcomes resulting from heritable disorders, as well as providing information and education to the public on such effective interventions. The ICC is comprised of representatives from CDC, HRSA, the Agency for Healthcare Research and Quality (AHRQ), the Food and Drug Administration (FDA), and the National Institutes of Health (NIH).

- Section 1115: National Contingency Plan for Newborn Screening, administered by CDC, 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.

The programs and activities under this Act 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 having or at risk for having heritable disorders. This report provides information on the programs and activities under the Newborn Screening Saves Lives Reauthorization Act of 2014 as well as funding amounts for newborn screening programs funded by HRSA and CDC.

Part I: Improved Newborn and Child Screening for Heritable Disorders

Section 1109 of the Act, Improved Newborn and Child 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 having or at risk for heritable disorders. These grants 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 having heritable

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, genetic, and metabolic conditions; and
- Improve the timeliness of newborn screening from specimen collection through diagnosis.

Section 1109 of the Act authorizes the programs described below.

Improving the Timeliness of Newborn Screening Diagnosis Initiative[3]

The Improving Timeliness of Newborn Screening Diagnosis Initiative seeks to ensure newborns receive timely screening, diagnosis, and treatment for heritable disorders. To achieve timely diagnosis and treatment of screened conditions and to avoid associated disability, morbidity, and mortality, the ACHDNC recommends the following time frames to communicate results to the newborn's health care provider:

- Presumptive positive results for time-critical conditions should be communicated immediately, but no later than 5 days after birth.
- Presumptive positive results for all other conditions should be communicated as soon as possible, but no later than 7 days after birth.
- All newborn screening tests should be completed within 7 days after birth with results reported as soon as possible.[4]

On September 1, 2015, HRSA awarded a cooperative agreement to the University of Colorado School of Public Health for the Improving the Timeliness of Newborn Screening Diagnosis Initiative. The goal of the Initiative is to increase the number of states that meet the ACHDNC recommendations on screening timeliness and to increase the number of infants receiving timely diagnosis and treatment for heritable disorders. The Improving the Timeliness of Newborn

[3] In December 2016, the U.S. Government Accountability Office (GAO) released a report, Newborn Screening Timeliness: Most States Had Not Met Screening Goals, but Some Are Developing Strategies to Address. The GAO report did not have any recommendations. HRSA provided information to GAO on funded activities that support newborn screening timeliness. HRSA-funded activities conducted during the Fiscal Year 2016 timeframe are described in the Newborn Screening Report to Congress, however, data and findings from the GAO report will not be included as these data were obtained directly by GAO.

[4] See http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/recommendations/timelynewbornscreeninggoalschainletter.pdf

Screening Diagnosis Initiative's activities include:

- Coordinating quality improvement projects using practice-based strategies to improve timeliness of newborn screening, diagnosis, and treatment;
- Developing a strategy for obtaining newborn screening timeliness data;
- Engaging public and private partners to coordinate activities, develop and distribute educational materials, and share best practices and lessons learned; and
- Providing ongoing technical assistance and facilitating collaboration between stakeholders to address the needs of state newborn screening programs and the impact on health disparities within underserved populations such as rural and tribal communities.

Beginning in FY 2015, activities included performing a gap analysis and needs assessment, selecting state participants, developing a communications strategy, assessing gaps and needs of participants, and developing a data collection strategy to reduce reporting burden.

In FY 2016, activities included expanding communication strategies, including communication and education networks to help alleviate lack of follow-up after diagnosis; expanding web-based newborn screening resources for parents and professionals to increase educational outreach; providing continuous quality improvement coaching, education, and financial support to 15 quality improvements teams, representing 20 states.

Newborn Screening Data Repository and Technical Assistance Center

Through a cooperative agreement that began on July 1, 2014, the Association of Public Health Laboratories provides technical assistance on the implementation of state-based public health newborn screening and other genetics programs. Activities include resource development, state education and training, policy initiatives, disorder surveillance, evidence-based data collection, quality improvement activities, and evaluation.

Key objectives of the Newborn Screening Data Repository and Technical Assistance Center (NBS TA Center) include:

- Developing, coordinating, and providing technical assistance through innovative educational and quality improvement activities related to newborn screening.
- Developing and disseminating information that addresses gaps within short-term follow-up[5] identified by providers and public health professionals.

[5] Short-term follow-up is defined as "the process of ensuring that all newborns are screened, that an appropriate follow-up caregiver is informed of results, that confirmatory testing has been completed, that the newborn has received a diagnosis and, if necessary, treatment." (See http://babysfirsttest.org/newborn-screening/glossary#letter_s\)

- Developing a national newborn screening data repository to standardize, maintain, and analyze quantitative quality measures, case definitions, and other data and information in order to evaluate the impact of state and territorial newborn screening programs.
- Supporting activities that strengthen laboratory performance and quality assurance; short- and long-term newborn screening follow-up; and public health interactions at the community, state, regional, and national levels.
- Providing a forum for timely, interactive communication between state and public health stakeholders regarding NBS and supporting training opportunities for public health practitioners.

The NBS TA Center's centralized and secure data repository includes data on state level newborn screening, newborn screening case definitions and newborn screening quality indicators. As of September 2016, 38 states have entered quality indicator data and 22 states have entered case data into the repository. All 50 states, the District of Columbia, and Puerto Rico have entered state profile data. States voluntarily submit data into the repository on an ongoing basis. States generally submit data one to two times a year. These data provide a national picture of newborn screening and can help states and territories evaluate their programs. A snapshot of the number of conditions on the RUSP screened in each state is an example of information generated from the data repository[6] (Appendix B). The NBS TA Center has developed newborn screening case definitions that provide a consistent way to categorize and track newborn screening conditions. This allows for improved public health surveillance by providing a more accurate estimate of the incidence of conditions identified by newborn screening. The NBS TA Center conducts bi-monthly national webinars on topics such as health information technology, implementing newborn screening and data collection for critical congenital heart disease, and short-term follow-up. The NBS TA Center developed pre-evaluation and evaluation site visit tools, and training resources for evaluators for state newborn screening programs that wish to have an objective analysis of their programs.

Regional Genetic and Newborn Screening Service Collaboratives Initiative

The Regional Genetic and Newborn Screening Service Collaboratives initiative, funded from 2004-2016, supported seven grantees in seven regions across the United States to help state and local public health agencies address the challenges of enhancing, improving, or expanding access to screening, counseling, or health care services to newborns and children having or at risk for heritable disorders.

The seven Regional Genetic Service Collaboratives are:

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

- Region 1: New England Genetics Collaborative (awarded to the University System of New Hampshire), states in this region include:
    - Connecticut, Maine, Massachusetts, New Hampshire, Rhode Island, and Vermont
- Region 2: New York Mid-Atlantic Collaborative (awarded to Health Research, Inc.), states in this region include:
    - District of Columbia, Delaware, Maryland, New Jersey, New York, Pennsylvania, Virginia, and West Virginia
- Region 3: Southeast Regional Collaborative (awarded to Emory University), states in this region include:
    - Alabama, Florida, Georgia, Louisiana, Mississippi, North Carolina, Puerto Rico, South Carolina, Tennessee, and U.S. Virgin Islands
- Region 4: Midwest Genetics Collaborative (awarded to Michigan Public Health Institute), states in this region include:
    - Illinois, Indiana, Kentucky, Michigan, Minnesota, Ohio, and Wisconsin
- Region 5: Heartland Genetics and Newborn Screening Collaborative (awarded to Arkansas Children's Hospital Research Institute), including:
    - Arkansas, Iowa, Kansas, Missouri, Nebraska, North Dakota, Oklahoma, and South Dakota
- Region 6: Mountain States Genetics Regional Collaborative (awarded to Texas Health Institute), states in this region include:
    - Arizona, Colorado, Montana, New Mexico, Nevada, Texas, Utah, and Wyoming
- Region 7: Western States Genetic Services Collaborative (awarded to Hawaii Department of Health), states in this region include:
    - Alaska, California, Guam, Hawaii, Idaho, Oregon, and Washington

The goals of the Regional Genetic Service Collaboratives included:

- Ensuring that individuals with genetic conditions and their families have access to genetic services with high quality care, expertise, and resources;
- Applying the translation of genome-based knowledge, genomics best practices, and new technologies to expand services by supporting telemedicine/telegenetics activities; and
- Evaluating project outcomes quantitatively and qualitatively to inform efforts to expand services.

The Regional Genetic Service Collaboratives' activities included:

- Providing education and training on newborn screening and follow-up services to expand the workforce serving individuals with genetic conditions, including those diagnosed via newborn screening;

- Providing guidance on implementation of new conditions added to state newborn screening panels; and
- Conducting regional needs assessment and data collection to improve newborn and child screening and ongoing genetics specialty services.

Building on this work, in 2017 HRSA funded seven Regional Genetic Networks to further improve health outcomes and reduce morbidity and mortality in individuals with genetic conditions. The distribution of regions remain the same.

Severe Combined Immunodeficiency Newborn Screening Implementation Program

Severe combined immunodeficiency (SCID) is a genetic condition that is the result of an immune system so highly compromised that it is almost absent. Infants born with SCID usually die within one year due to severe, recurrent infections unless they have undergone successful stem cell transplantation. In 2010, the Department adopted the ACHDNC's recommendation to add SCID to the RUSP (Appendix A provides a list of conditions on the RUSP).

To better support states in implementing SCID screening, HRSA established the SCID Newborn Screening Implementation Program. HRSA awarded cooperative agreements to the Association of Public Health Laboratories on September 1, 2014, and the Jeffrey Modell Foundation on May 1, 2015. The program's overall goal is to support implementation of universal screening for SCID in every state, with all identified infants receiving appropriate screening and follow up care by:

- Assuring early and accurate laboratory testing and detection of SCID in newborns;
- Expanding laboratory capacity for SCID newborn screening, including funding the costs of bringing a molecular test online;
- Increasing the number of laboratory scientists with knowledge and skill in conducting newborn screening for SCID;
- Providing training for the public health community about newborn screening tests for SCID;
- Integrating SCID screening into newborn screening programs; and
- Developing and distributing appropriate education and training materials for families and public health and health care professionals relevant to SCID screening and treatment.

When the SCID awards first began in September 2014, there were 25 state newborn screening programs (out of the 50 U.S. States, plus the District of Columbia, Puerto Rico, and Guam) screening for SCID. By September 2016, there were 44 (see maps in Appendix C).

Newborn Screening Implementation Program Regarding Conditions Added to the Recommended Uniform Screening Panel

In 2015 and 2016, upon the recommendations of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children, the HHS Secretary added Pompe, Mucopolysaccharidosis I (MPS I), and X-linked Adrenoleukodystrophy (X-ALD) to the RUSP. Adding a new condition to a state newborn screening program can take significant time and investment. Activities may include implementing new laboratory testing procedures and protocols, which could mean a change in existing laboratory infrastructure, workflow, and equipment, developing new expertise and training requirements for laboratory personnel; increasing capacity of newborn screening follow-up personnel, and developing and disseminating educational materials to primary health care providers, the public, and families of newborns identified through newborn screening.

In FY 2016, HRSA awarded funds to the Association of Public Health Laboratories to increase the number of states and/or territories that screen for Pompe disease, MPS I, and X-ALD. At the time these awards began, only five states were screening for Pompe disease, three for MPS-1, and three for X-ALD (as of September 30, 2016). The program aims to:

- Increase state newborn screening programs' capacity to screen for conditions recently added to the RUSP;
- Support implementation, education, and awareness of newborn screening for conditions recently added to the RUSP; and
- Distribute national, regional, and state education and training resources for parents, families, and providers.

Critical Congenital Heart Disease Newborn Screening Demonstration Program

In the United States, about 7,200 infants born every year have critical congenital heart disease (CCHD), a condition that is often undetected during routine clinical exams prior to a newborn's discharge. Some infants born with CCHD appear healthy at first and may be sent home before their heart defect is detected. Newborn screening can identify some of these infants so they can receive prompt care and treatment. Timely care may prevent disability or death early in life. In 2011, the Secretary adopted the ACHDNC's recommendation to add CCHD to the RUSP.

Beginning in June 1, 2012, through May 31, 2015, HRSA supported seven CCHD Newborn Screening Demonstration Program grants to support implementation of CCHD screening. The program provided support to state health departments in Wisconsin, Michigan, New Jersey, New Hampshire, Vermont, Rhode Island, Maine, Utah, and Virginia. Over the course of the awards, six of the seven states added universal CCHD screening and the seventh state (Vermont) added universal CCHD screening in 2016. The CCHD Newborn Screening Demonstration Program was also a catalyst for other states to add CCHD screening. The overall number of states and territories screening for CCHD went from 3 in June 2012 to 50 in September 2016 (see Appendix

D for a map of states screening for CCHD).

The CCHD Newborn Screening Demonstration Program concluded in May 2015 with most states having added CCHD as part of their newborn screening panels. Currently, all but two states offer universal screening for CCHD. The two remaining states, Idaho and Wyoming, are in the process of implementing CCHD screening. The results of this program were not available prior to HHS's submission of the first report to Congress and are shared here to provide continuity between the reports.

Integrating Newborn Screening Long-Term Follow-up into Primary Care Practice

The ACHDNC provides advice to the Secretary on reducing the morbidity and mortality associated with heritable disorders, with particular emphasis on newborn screening. Long-term follow-up is critical to realize the benefit of newborn screening; however, the provision of long-term follow-up care varies. To facilitate efforts to evaluate health outcomes and impact of newborn screening beyond long-term survival, the ACHDNC reviewed the goals and expectations of the type of long-term follow-up that is required to ensure best outcomes and improve access to care for patients identified through newborn screening.

On August 1, 2013, HRSA awarded two demonstration grants through the Integrating Newborn Screening Long-Term Follow-up into Primary Care Practice initiative to the Maryland Department of Health and Mental Hygiene and the Public Health Foundation Enterprises, Inc. in California. Grantees examined the overarching questions outlined in the ACHDNC 2011 report, "*What Questions Should Newborn Screening Long-term Follow-up Be Able to Answer?*,"[7] including questions that follow the central components of long-term follow-up including care coordination, evidence-based treatment, continuous quality improvement, and new knowledge discovery. Grantees also examined the ability of primary care practices to provide accurate and ongoing information on patients identified with conditions detected through newborn screening.

These two demonstration projects concluded in 2015 and were able to make clear some of the challenges associated with implementing long-term follow-up. For example, one demonstration site noted that primary care providers might not have all of the information needed to evaluate long-term follow-up and assess long-term care for NBS conditions. The data needed to assess long-term follow-up will likely need to come from various sources in the future, which poses a logistical challenge with regard to electronic data sharing between public and private sources. The results of this program were not available prior to HHS's submission of the first report to Congress and are shared here to provide continuity between the reports.

Part II: Evaluating the Effectiveness of Newborn and Child Screening and Follow-up Programs

Section 1110, Evaluating the Effectiveness of Newborn and Child Screening and Follow-up

[7] http://www.nature.com/gim/journal/v13/n10/full/gim2011144a.html

Programs, authorizes demonstration programs that evaluate the effectiveness of timely newborn screening, follow-up, counseling, and health care services in order to reduce the morbidity and mortality caused by heritable disorders in newborns and children. CDC has undertaken several activities to understand and evaluate the implementation and effectiveness of newborn screening for critical congenital heart defects (CCHD). CDC continues to work, through technical assistance and evaluation activities, towards improved screening implementation and data collection so that newborns with CCHD are identified and referred to services. HRSA administers the programs discussed below. HRSA requires grantees to evaluate the effectiveness of their programs, to report on their progress, and to adjust their processes in light of evaluation results.

Improving Timeliness of Newborn Screening Diagnosis Initiative

The Improving the Timeliness of Newborn Screening Diagnosis initiative, described in more detail on pages 5-6, is examining the effectiveness of screening in a timely manner. Participating states are working with quality improvement experts to improve the time from collection of specimens for newborn screening to diagnosis and treatment of infants identified with a possible heritable condition. States will implement strategies to improve the timeliness of newborn screening, use real-time data to facilitate improvement, and develop and disseminate best practices. State specific data are collected on the following:

- Percent of invalid dried blood spot specimens/cards due to improper collection and/or transport;
- Percent of dried blood spot specimens/cards missing essential information;
- Percent of initial dried blood spot specimens collected no later than 48 hours after birth; and
- Percent of dried blood spot specimens received at the laboratory within 24 hours of collection.

For example, as of the end of 2016, 11 out of 23 states that reported data on the timing of specimen collection are reaching the goal of 95 percent of specimens collected within 48 hours of birth.

Newborn Screening Data Repository and Technical Assistance Center

As part of its responsibilities, the NBS TA Center, described in more detail on pages 6-7 provides resources to state newborn screening programs to evaluate the effectiveness of their processes and use real time data for quality improvement activities. For example, the Center developed quality indicators in collaboration with state newborn screening programs. State programs are providing data for these quality indicators within a data repository. As of the end of 2016, the data repository has worked with 38 states to collect data for newborn screening quality indicators and case definitions. The NBS TA Center will analyze the data and provide

information on the effectiveness of the many components that make up the newborn screening system.

Regional Genetic and Newborn Screening Service Collaboratives Initiative

Since 2004, the Regional Genetic Service Collaboratives Program, described in more detail on pages 7-8, has provided a regional infrastructure of public health genomics expertise to improve, expand, strengthen, and evaluate access to a system of genetic services and the quality of those services to improve health outcomes for children, youth, and adults across the course of their life. Specifically, they pursue methods to improve quality in the diagnosis, treatment, and disease management of heritable disorders based on gaps in services or care. The Regional Genetic Service Collaboratives quantitatively and qualitatively evaluate project outcomes to ensure that individuals have access to genetic services with high quality care, expertise, and resources.

Part III: Clearinghouse of Newborn Screening Information

The Newborn Screening Clearinghouse maintains a central, online repository of current educational information, materials, and resources on newborn screening. It includes information on family support services, follow-up services, and national and state newborn screening policies. The resources serve to enable parents, family members, and expectant individuals to increase their awareness, knowledge, and understanding of newborn screening and genetic conditions.

Key program activities include:

- Maintaining an interactive, web-based forum (www.babysfirsttest.org) promoting newborn screening information sharing and dissemination that provides culturally sensitive education and decision-making tools regarding newborn screening for heritable disorders;
- Conducting activities to increase awareness, knowledge, and understanding for parents and family members of newborns, health professionals, industry representatives, policy members, and members of the public;
- Conducting activities to increase understanding of newborn screening policies;
- Promoting and supporting community-specific efforts to understand the newborn screening process;
- Promoting national and state level policies and best practices regarding newborn screening;
- Partnering with stakeholders to collaborate, promote, and support and inform them of innovative methods of educational outreach; and
- Evaluating project activities and results.

The Newborn Screening Clearinghouse began in 2009. HRSA provided the second cycle of funding by a cooperative agreement awarded on September 1, 2014, to the Genetic Alliance. Baby's First Test launched in September 2011. Since then, the Clearinghouse website reached more than 1.7 million unique users, accessing the site more than 3.5 million times. Its accompanying Newborn Screening Awareness Campaign has reached 2 million people each year through social media. Recent accomplishments include the launch of the Public Square, an open space for health professionals, parents, policy makers, researchers, and others to share new ideas, knowledge, and bring together a range of communities and experiences in newborn screening to improve education and awareness. The Newborn Screening Clearinghouse has also developed an infographic to inform parents about newborn screening called "Newborn Screening: Get the Facts." Hospitals, community centers, and other organizations can access free printed copies of this educational tool and other resources through the Clearinghouse.

Part IV: Laboratory Quality and Surveillance

CDC operates the nation's only quality assurance program for ensuring the accuracy of newborn screening blood spot tests conducted by public health laboratories. The Newborn Screening Quality Assurance Program (NSQAP) provides unique services directly to laboratories to maintain and improve the quality of their testing so that affected babies in the United States are identified early and correctly. The program:

- Supports all newborn screening laboratories in the United States by providing quality assurance materials and proficiency testing services for tests that detect over 50 congenital conditions in newborns, including all of the laboratory-identified primary disorders on the RUSP;
- Provides training and technical support to state and territorial laboratories to enhance nationwide laboratory capacity and capability;
- Prepares, certifies, and distributes more than 800,000 dried blood spot quality assurance materials that mimic disease samples to participating laboratories each year;
- Develops, conducts, and hosts hands-on training workshops on current and innovative laboratory techniques for state newborn screening programs;
- Helps laboratories add new conditions to their screening panel and implement new screening technologies that improve disease detection and prediction of disease severity;
- Develops new methods for recent and anticipated additions to the RUSP; and
- Evaluates filter paper used to produce blood collection cards for newborn screening to ensure the quality of cards made for screening programs nationwide.

During FY 2016, the program:

- Provided services to more than 650 laboratories covering all states and U.S. territories and 78 countries;
- Developed or improved eight laboratory methods for screening current RUSP conditions or anticipated additions including guanidinoacetate methyltransferase

deficiency, X-linked adrenoleukodystrophy, methylmalonic acidemia, propionic acidemia, homocystinuria and cobalamin disorders, Wilson disease, adenosine deaminase deficiency, spinal muscular atrophy, glucose-6-phosphate dehydrogenase deficiency, and congenital adrenal hyperplasia;

- Provided specialized technical assistance, Molecular Assessment Program site visits, hands-on training, or technology transfer to over 20 state and territorial programs to assist with quality assurance for molecular (DNA-based) testing;
- Provided specialized technical assistance, hands-on training, or technology transfer to 16 state and territorial programs for tandem mass spectrometry for more than 40 conditions;
- Improved SCID newborn screening by funding four states to implement state-wide testing or improve SCID laboratory methods using innovative technology; and
- Collaborated with federal agencies, public health departments, and key partners to improve the newborn screening process, including efforts to develop critical guidance for states in response to new laws.

Part V: Interagency Coordinating Committee on Newborn & Child Screening

The Interagency Coordinating Committee (ICC), co-chaired by HRSA and CDC, is composed of the HRSA Administrator, CDC Director, AHRQ Director, FDA Commissioner, and NIH Director, or their designees. The ICC coordinates collaborative efforts for newborn and child screening among all Department of Health and Human Services agencies. In addition, the ICC assesses existing newborn screening activities and infrastructure to make recommendations on heritable disorders recommended for newborn screening.

At the request of the Secretary, the ICC first met in May 2011. The ICC did not convene in 2016, however prior to 2016 the ICC reviewed the following topics:

- Newborn screening for Critical Congenital Heart Disease (CCHD);
- Use and storage of residual newborn screening blood samples;
- Data quality assurance in newborn screening;
- Newborn screening for Pompe disease;
- Newborn screening for Mucopolysaccharidosis I (MPS I); and
- Newborn screening for X-linked Adrenoleukodystrophy (X-ALD).

Part VI: National Contingency Plan for Newborn Screening

In September 2008, HRSA and CDC held a 2-day workshop to frame the Newborn Screening Contingency Plan with input from subject matter experts and newborn screening community stakeholders. 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. The plan, published in 2010,[8] was distributed to states through HRSA's Regional Genetics and Newborn Screening Service Collaboratives.

Since the development of the initial plan, CDC, HRSA, and the seven funded Regional Genetic and Newborn Screening Service Collaboratives have prioritized emergency preparedness plans and actions to ensure continuity of operations. In 2011, CDC supported updates to the "Guidelines for the Public Health Laboratory Continuity of Operations Plan,"[9] which addresses newborn screening emergency plans. Also in 2011, the CDC published the Public Health Preparedness Capabilities: National Standards for State and Local Planning,[10] which reiterates the need to have state and local plans in place to assure the continuity of newborn screening in public health laboratories. 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. As was done before, input from subject matter experts and NBS community stakeholders was included in the plan. A Public Comment Survey was administered to get feedback, an in-person working meeting with NBS stakeholders was conducted, and NBS experts provided revisions and resource development. Revisions to the plan include: adding a new strategic objective related to communications, adding long-term follow-up language, reordering strategic objectives, expanding the Legal Issues section, incorporating point of care screening, and adding new appendices. A dissemination plan is being developed with the intent to disseminate in 2017. CDC and HRSA are reviewing and finalizing the report.

Part VII: Funding Amounts

(whole dollars)	Fiscal Year 2016 Funding*
HRSA	
Improving the Timeliness of Newborn Screening Diagnosis	$1,800,000
Newborn Screening Data Repository and Technical Assistance	$850,000
Regional Genetic and Newborn Screening Service Collaboratives	$4,200,000
Severe Combined Immunodeficiency Newborn Screening	$2,000,000
Newborn Screening Implementation Program Regarding Conditions Added to the Recommended Uniform Screening Panel	$2,000,000
Clearinghouse of Newborn Screening Information	$725,000
CDC	
Budget Authority – Laboratory Quality and Surveillance	$8,300,000
– Severe Combined Immunodeficiency Disorder	$1,200,000

* HRSA's FY 16 Total Funding Enacted is $13,883,000. The appropriated amount includes funds for the operating costs for the Advisory Committee for Heritable Disorders in Newborns and Children as well as contractual services, salaries and other administrative costs related to the Newborn Screening Programs.

[8] See http://www.cdc.gov/ncbddd/documents/NBS-CONPLAN.pdf
[9] See http://www.aphl.org/MRC/Documents/PHPR_2011Feb_PHL-Continuity-of-Operations-Guidelines.pdf
[10] See https://www.cdc.gov/phpr/readiness/capabilities.htm

Part VIII: Summary and Conclusion

The newborn screening programs and activities, as administered by HRSA and CDC, will continue to ensure that infants born in the United States are properly screened for heritable disorders and referred to appropriate early intervention within an acceptable timeframe in order to achieve the best possible health outcomes. Through evaluation, ongoing assessment, information sharing, and partnership with appropriate experts, HRSA and the CDC were able to:

- Increase the number of states screening for SCID from 25 to 44;
- Increase the overall number of states and territories screening for CCHD from 3 to 50;
- Provide continuous quality improvement, coaching, financial support, or educational opportunities to all states and territories;
- Increase evaluation efforts by states to examine the effectiveness of screening in a timely manner; and
- Provide services to more than 650 laboratories covering all states and U.S. territories and 78 countries to ensure accuracy of newborn screening blood spot tests.

HRSA and CDC will continue to ensure that best practices are identified, shared, and implemented to improve the health of all infants and children in the United States.

Appendix A: Recommended Uniform Screening Panel (RUSP) and Secondary Conditions

RUSP[1] Core[2] Conditions[3]

(As of November 2016)

Core Condition	Metabolic Disorder			Endocrine Disorder	Hemoglobin Disorder	Other Disorder
	Organic acid condition	Fatty acid oxidation disorder	Amino acid disorder			
Propionic Acidemia	X					
Methylmalonic Acidemia (methylmalonyl-CoA mutase)	X					
Methylmalonic Acidemia (Cobalamin disorders)	X					
Isovaleric Acidemia	X					
3-Methylcrotonyl-CoA Carboxylase Deficiency	X					
3-Hydroxy-3-Methyglutaric Aciduria	X					
Holocarboxylase Synthase Deficiency	X					
ß-Ketothiolase Deficiency	X					
Glutaric Acidemia Type I	X					
Carnitine Uptake Defect/Carnitine Transport		X				
Medium-chain Acyl-CoA Dehydrogenase Deficiency		X				
Very Long-chain Acyl-CoA Dehydrogenase Deficiency		X				
Long-chain L-3 Hydroxyacyl-CoA Dehydrogenase		X				
Trifunctional Protein		X				
Argininosuccinic Aciduria			X			
Citrullinemia, Type I			X			
Maple Syrup Urine Disease			X			
Homocystinuria			X			
Classic Phenylketonuria			X			
Tyrosinemia, Type I			X			
Primary Congenital Hypothyroidism				X		
Congenital adrenal hyperplasia				X		
S,S Disease (Sickle Cell					X	
S, βeta-Thalassemia					X	
S,C Disease					X	
Biotinidase Deficiency						X

Core Condition	Metabolic Disorder			Endocrine Disorder	Hemoglobin Disorder	Other Disorder
	Organic acid condition	Fatty acid oxidation disorder	Amino acid disorder			
Critical Congenital Heart Disease						X
Cystic Fibrosis						X
Classic Galactosemia						X
Glycogen Storage Disease Type II (Pompe)						X
Hearing Loss						X
Severe Combined Immunodeficiencies						X
Mucopolysaccharidosis Type						X
X-linked Adrenoleukodystrophy						X

1. Selection of conditions based upon "Newborn Screening: Towards a Uniform Screening Panel and System." *Genetic Med*. 2006; 8(5) Suppl: S12-S252" as authored by the American College of Medical Genetics (ACMG) and commissioned by the Health Resources and Services Administration (HRSA).
2. Disorders that should be included in every Newborn Screening Program.
3. Nomenclature for Conditions based upon "Naming and Counting Disorders (Conditions) Included in Newborn Screening Panels." *Pediatrics*. 2006; 117 (5) Suppl: S308-S314.

Secondary[2] Conditions [3]

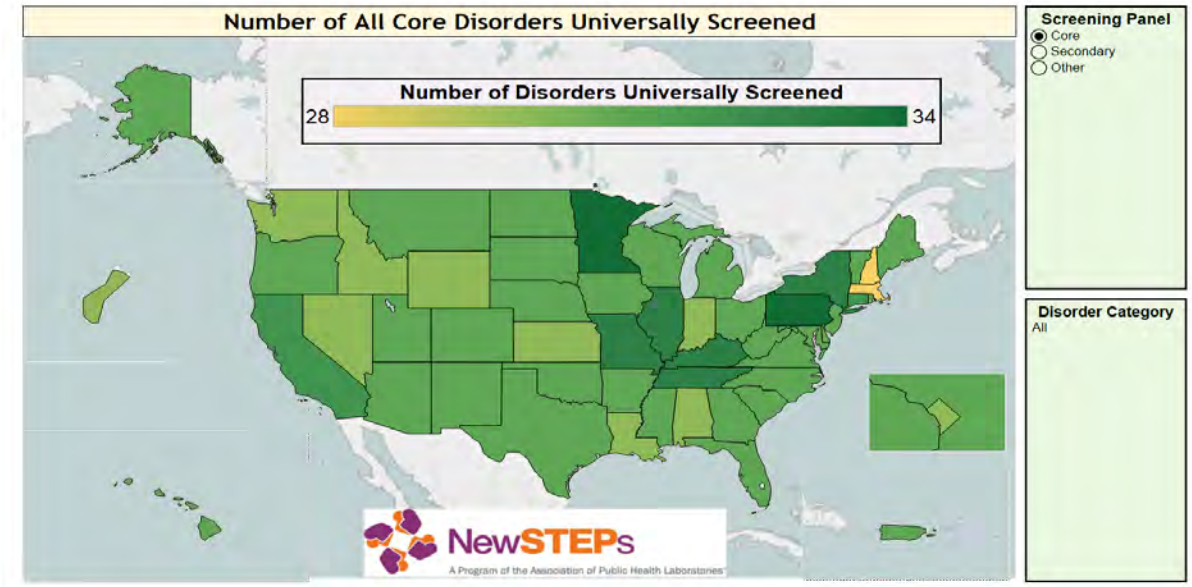
(As of November 2016)

Secondary Condition	Metabolic Disorder			Hemoglobin Disorder	Other Disorder
	Organic acid condition	Fatty acid oxidation disorders	Amino acid disorders		
Methylmalonic acidemia with	X				
Malonic acidemia	X				
Isobutyrylglycinuria	X				
2-Methylbutyrylglycinuria	X				
3-Methylglutaconic aciduria	X				
2-Methyl-3-hydroxybutyric aciduria	X				
Short-chain acyl-CoA dehydrogenase		X			
Medium/short-chain L-3-hydroxyacyl- CoA dehydrogenase deficiency		X			
Glutaric acidemia type II		X			
Medium-chain ketoacyl-CoA thiolase		X			

Secondary Condition	Metabolic Disorder			Hemoglobin Disorder	Other Disorder
	Organic acid condition	Fatty acid oxidation disorders	Amino acid disorders		
2,4 Dienoyl-CoA reductase deficiency		X			
Carnitine palmitoyltransferase type I deficiency		X			
Carnitine palmitoyltransferase type II deficiency		X			
Carnitine acylcarnitine		X			
Argininemia			X		
Citrullinemia, type II			X		
Hypermethioninemia			X		
Benign hyperphenylalaninemia			X		
Biopterin defect in cofactor biosynthesis			X		
Biopterin defect in cofactor regeneration			X		
Tyrosinemia, type II			X		
Tyrosinemia, type III			X		
Various other hemoglobinopathies				X	
Galactoepimerase deficiency					X
Galactokinase deficiency					X
T-cell related lymphocyte deficiencies					X

1. Selection of conditions based upon "Newborn Screening: Towards a Uniform Screening Panel and System." *Genetic Med*. 2006; 8(5) Suppl: S12- S252" as authored by the American College of Medical Genetics (ACMG) and commissioned by the Health Resources and Services Administration (HRSA).
2. Disorders that can be detected in the differential diagnosis of a core disorder.
3. Nomenclature for Conditions based upon "Naming and Counting Disorders (Conditions) Included in Newborn Screening Panels." *Pediatrics*. 2006; 117 (5) Suppl: S308-S314.

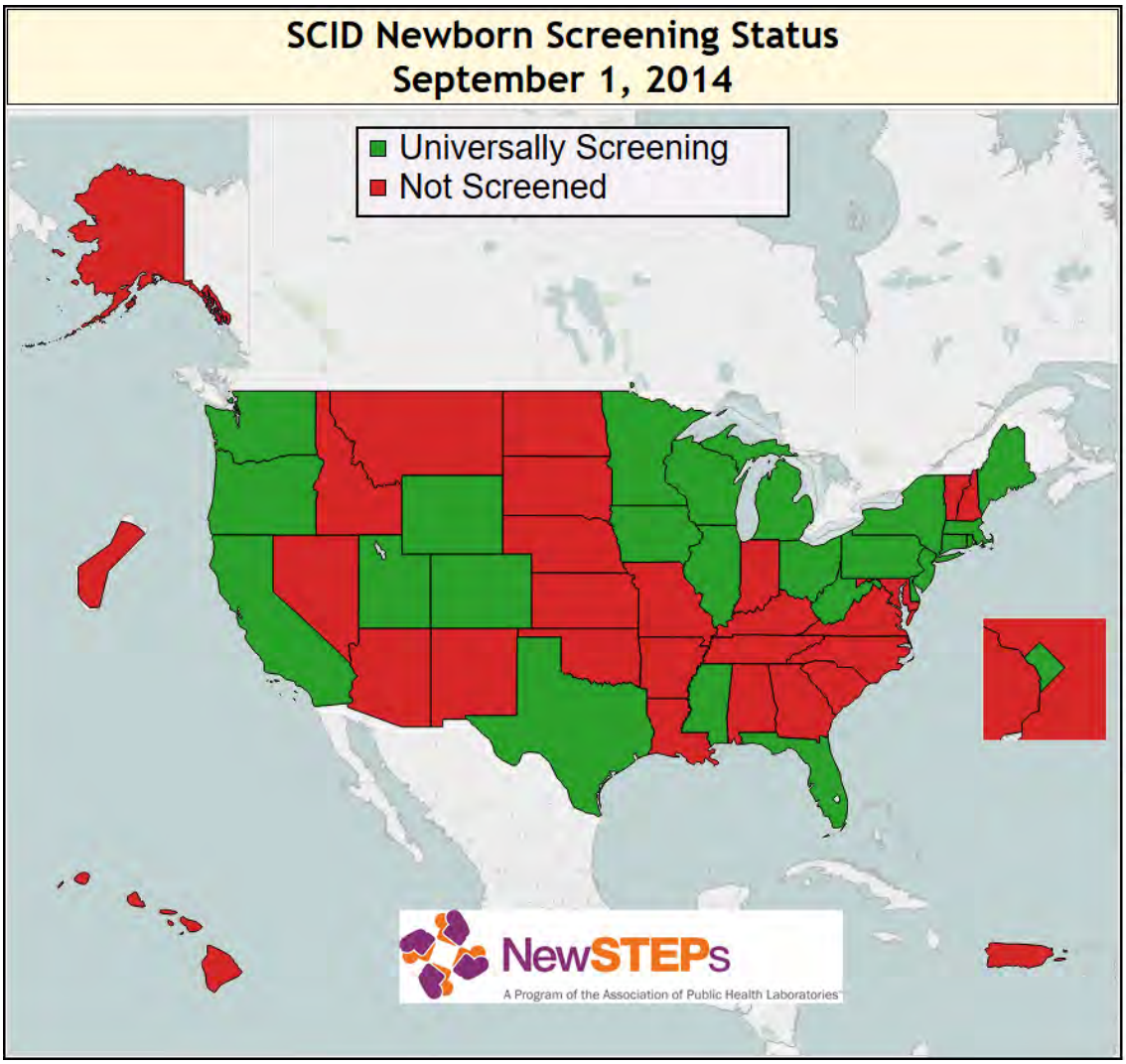
Appendix B: RUSP Conditions Screened by State/Territory as of Sept 30, 2016



Number of Core Disorders Universally Screened			
Alabama	30	Montana	31
Alaska	31	Nebraska	31
Arizona	31	Nevada	30
Arkansas	31	New Hampshire	28
California	32	New Jersey	31
Colorado	31	New Mexico	31
Connecticut	32	New York	33
Delaware	31	North Carolina	31
District of Columbia	30	North Dakota	31
Florida	31	Ohio	31
Georgia	31	Oklahoma	31
Guam	30	Oregon	31
Hawaii	31	Pennsylvania	34
Iowa	31	Puerto Rico	31
Idaho	30	Rhode Island	31
Illinois	33	South Carolina	31
Indiana	30	South Dakota	31
Kansas	30	Tennessee	33
Kentucky	33	Texas	31
Louisiana	30	Utah	31
Massachusetts	28	Vermont	31
Maine	31	Virginia	31
Maryland	31	Washington	30
Michigan	31	West Virginia	31
Minnesota	34	Wisconsin	31
Mississippi	31	Wyoming	30
Missouri	33		

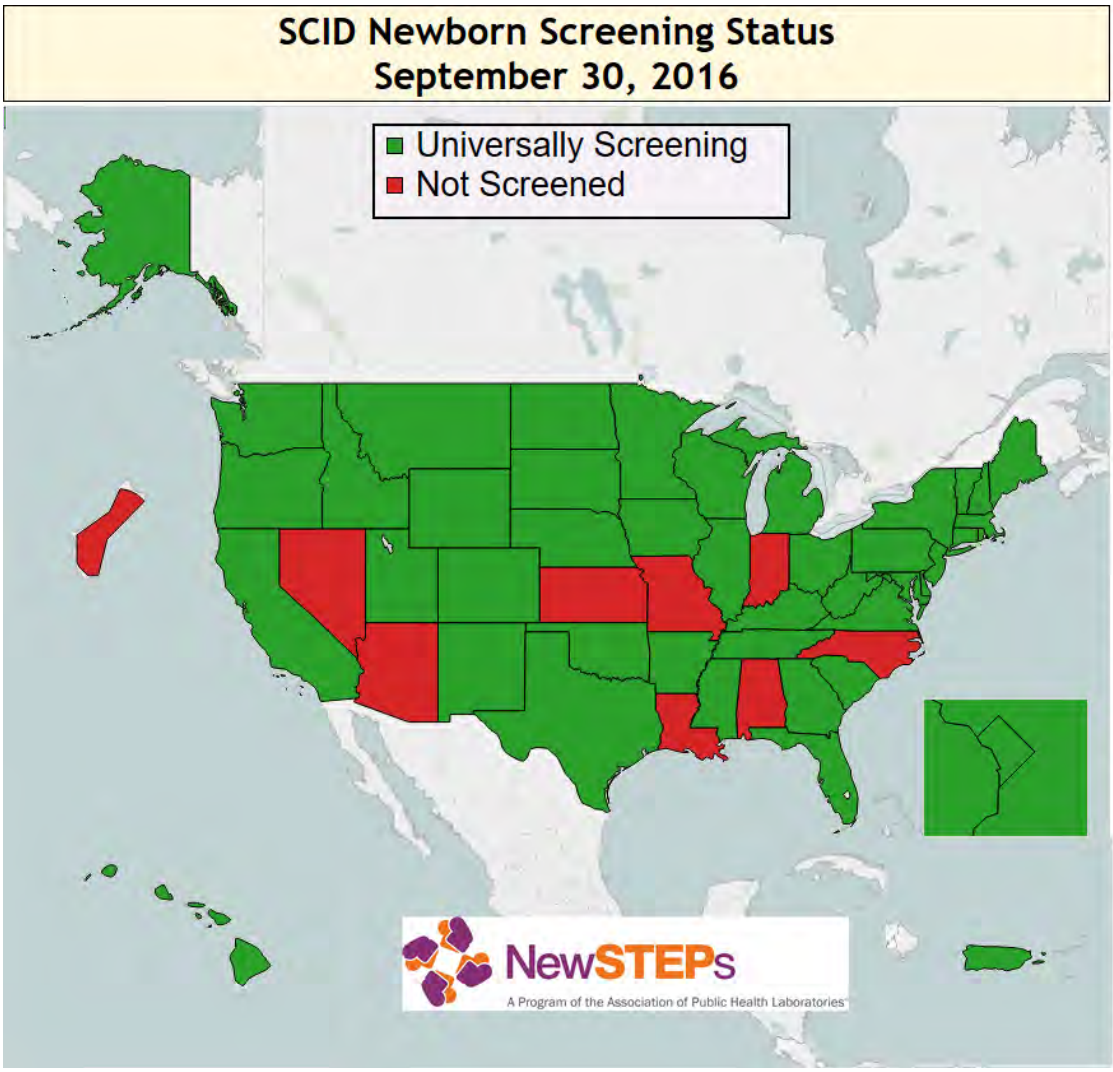
Source: The Newborn Screening Technical assistance and Evaluation Program (NewSTEPs), https://www.newsteps.org/

Appendix C: States and Territories Screening for Severe Combined Immunodeficiencies in 2014 versus 2016



NBS Programs Universally Screening	
California	1
Colorado	1
Connecticut	1
Delaware	1
District of Columbia	1
Florida	1
Illinois	1
Iowa	1
Maine	1
Massachusetts	1
Michigan	1
Minnesota	1
Mississippi	1
New Jersey	1
New York	1
Ohio	1
Oregon	1
Pennsylvania	1
Rhode Island	1
Texas	1
Utah	1
Washington	1
West Virginia	1
Wisconsin	1
Wyoming	1
Grand Total	25

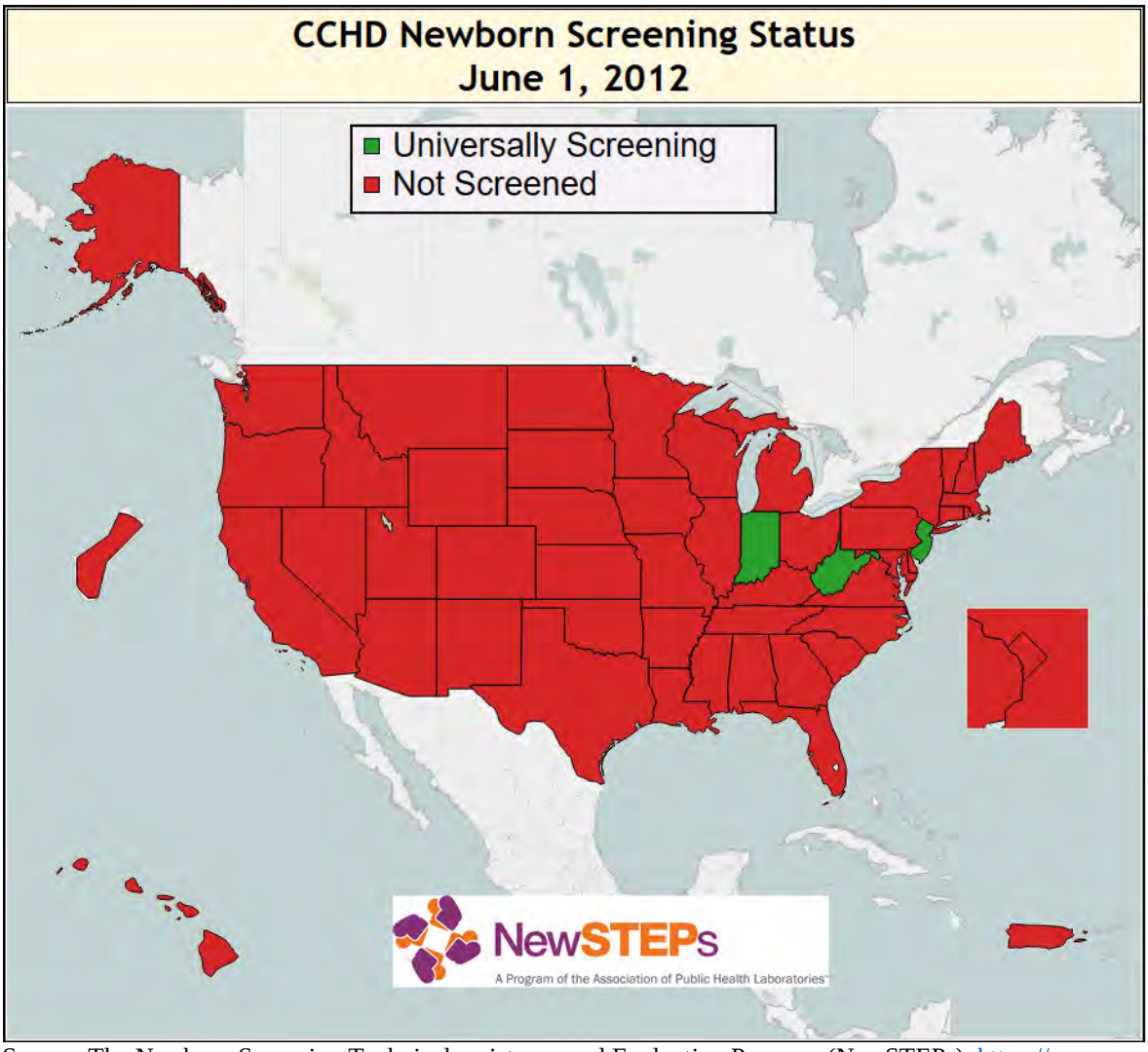
Source: The Newborn Screening Technical assistance and Evaluation Program (NewSTEPs), https://www.newsteps.org/



NBS Programs Universally Screening	
Alaska	1
Arkansas	1
California	1
Colorado	1
Connecticut	1
Delaware	1
District of Columbia	1
Florida	1
Georgia	1
Hawaii	1
Idaho	1
Illinois	1
Iowa	1
Kentucky	1
Maine	1
Maryland	1
Massachusetts	1
Michigan	1
Minnesota	1
Mississippi	1
Montana	1
Nebraska	1
New Hampshire	1
New Jersey	1
New Mexico	1
New York	1
North Dakota	1
Ohio	1
Oklahoma	1
Oregon	1
Pennsylvania	1
Puerto Rico	1
Rhode Island	1
South Carolina	1
South Dakota	1
Tennessee	1
Texas	1
Utah	1
Vermont	1
Virginia	1
Washington	1
West Virginia	1
Wisconsin	1
Wyoming	1
Grand Total	44

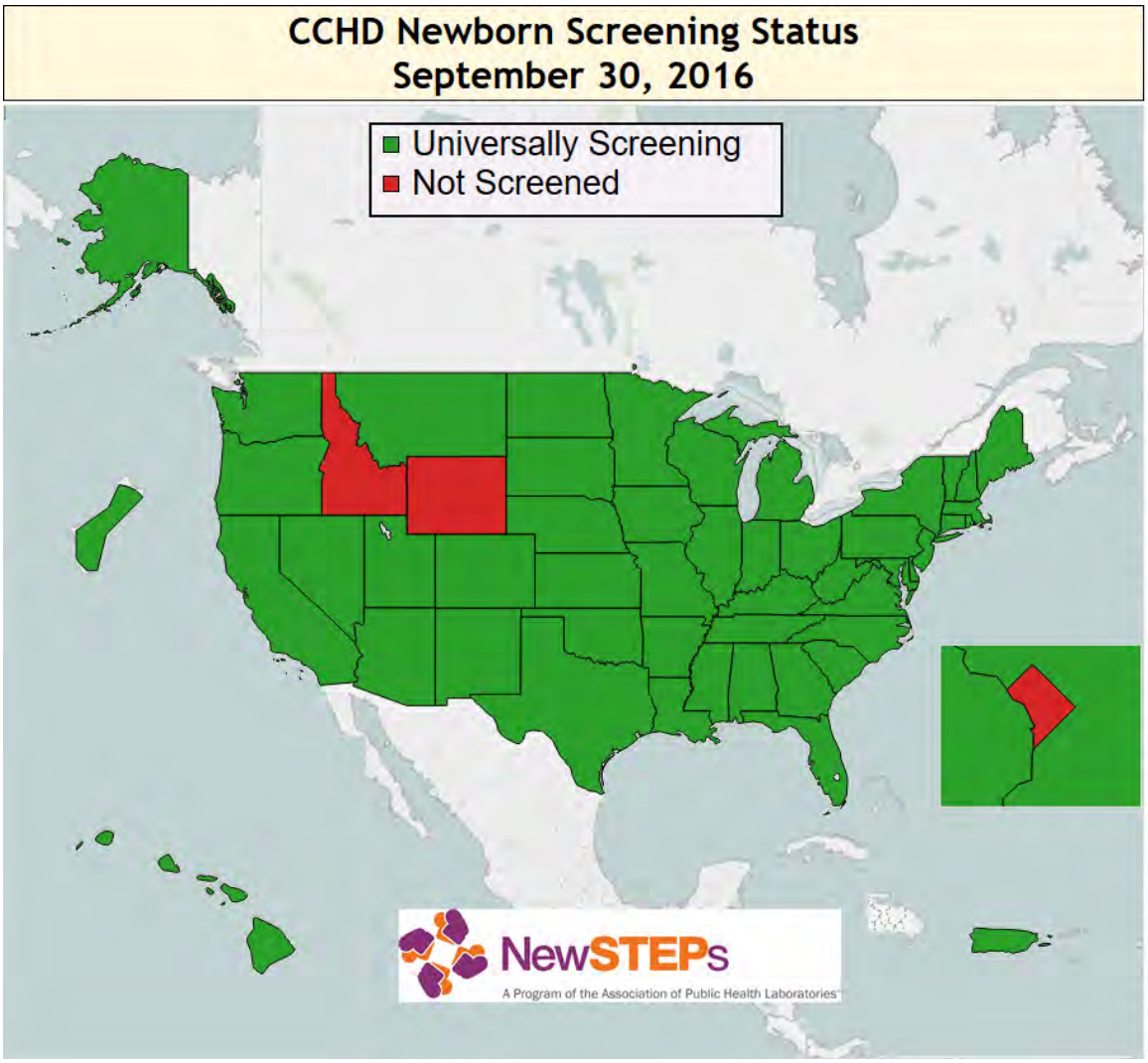
Source: The Newborn Screening Technical assistance and Evaluation Program (NewSTEPs), https://www.newsteps.org/

Appendix D: States and Territories Screening for Critical Congenital Heart Disease in 2012 versus 2016



NBS Programs Universally Screening	
Indiana	1
New Jersey	1
West Virginia	1
Grand Total	3

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



NBS Programs Universally Screening	
Alabama	1
Alaska	1
Arizona	1
Arkansas	1
California	1
Colorado	1
Connecticut	1
Delaware	1
Florida	1
Georgia	1
Guam	1
Hawaii	1
Illinois	1
Indiana	1
Iowa	1
Kansas	1
Kentucky	1
Louisiana	1
Maine	1
Maryland	1
Massachusetts	1
Michigan	1
Minnesota	1
Mississippi	1
Missouri	1
Montana	1
Nebraska	1
Nevada	1
New Hampshire	1
New Jersey	1
New Mexico	1
New York	1
North Carolina	1
North Dakota	1
Ohio	1
Oklahoma	1
Oregon	1
Pennsylvania	1
Puerto Rico	1
Rhode Island	1
South Carolina	1
South Dakota	1
Tennessee	1
Texas	1
Utah	1
Vermont	1
Virginia	1
Washington	1
West Virginia	1
Wisconsin	1
Grand Total	50

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