



**U.S. Department of Health and Human Services**

**Report to Congress  
Newborn Screening Activities**

**Submitted to**

**The Committee on Health, Education, Labor and Pensions  
U.S. Senate**

**and**

**The Committee on Energy and Commerce  
U.S. House of Representatives**

## 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 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.

These sections are administered by the Health Resources and Services Administration (HRSA) and the Centers for Disease Control and Prevention (CDC).

- 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;<sup>1</sup>
- 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 initial report on newborn screening activities administered by HRSA and CDC. The report covers activities through the end of fiscal year 2015.

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<sup>1</sup> CDC and HRSA are both authorized to administer programs in 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 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 being 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 initial 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 through the end of fiscal year 2015.

## 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 and recommended for states to screen as part of their state universal newborn screening (NBS) programs. Conditions on the RUSP are initially recommended by the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) based on a decision matrix that categorizes evidence that supports the potential net benefit of screening, the ability of states to screen for the disorder, and the availability of effective treatments. 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 it, the health outcomes of the condition are well understood, there is an available and effective treatment, and identification of the condition could affect the future reproductive decisions of the family. “Secondary conditions” are the genetic conditions that can be identified when screening for a core condition, or as a consequence of confirmatory testing for an out-of-normal-range result for a core condition.

Screening practices and the types of disorders for which newborns are screened may vary from state to state (see Appendix B). Infants who test positive for one of these conditions 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 support to the newborn screening community to help ensure proper and timely screening and intervention.

## Overview

The following is a report on the ongoing activities authorized by sections 1109, 1110, and 1112 through 1115 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,<sup>2</sup> authorizes grants to provide for the conduct of 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 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 by promoting data sharing regarding newborn screening with state-based birth defects and developmental 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

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<sup>2</sup> CDC and HRSA are both authorized to administer programs in under this section. However, CDC does not currently administer any programs under this section.

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 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 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 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.

The programs described below are authorized under section 1109 of the Act.

### **Improving the Timeliness of Newborn Screening Diagnosis**

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.<sup>3</sup>

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 Improving the Timeliness of Newborn Screening Diagnosis 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 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 collection 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 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.

Year 1 activities under the cooperative agreement include 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.

## **Newborn Screening Data Repository and Technical Assistance Center**

The Association of Public Health Laboratories was awarded a cooperative agreement on July 1, 2014, to provide 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 Technical Assistance Center (NBS TA Center) include:

- Developing, coordinating, and providing:
  - technical assistance through innovative educational and quality improvement activities related to newborn screening; and

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<sup>3</sup> See

<http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/recommendations/timelynewbornscreeninggoalschainletter.pdf>



- information that address gaps within short-term follow-up<sup>4</sup> identified by providers and public health professionals.
- Developing a national newborn screening data repository to:
  - standardize, maintain, and analyze quantitative quality measures, case definitions, and other data; and
  - evaluate state and territorial newborn screening programs' impact on newborn screening.
- Supporting activities that strengthen:
  - laboratory performance and quality assurance; and
  - short- and long-term newborn screening follow-up.

The NBS TA Center also established a centralized and secure data repository with information from state newborn screening programs across the nation. The repository includes data on newborn screening case definitions and newborn screening quality indicators. Data is submitted by states into the repository on an annual basis. The NBS TA Center has also developed newborn screening case definitions for public health surveillance. This is the first time newborn screening case definitions for public health surveillance have been created. These definitions provide consistent categorization and tracking of newborn screening conditions and provide a more accurate estimate of the incidence of conditions identified by newborn screening. State programs can use the case definitions tables and data collected on the case definitions worksheets to classify cases in a uniform manner. The NBS TA Center conducts bi-monthly national webinars on topics such as health information technology, critical congenital heart disease (CCHD), and short-term follow-up. It has also developed an evaluation site visit tool, pre-evaluation site visit tool, 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 was awarded on June 1, 2012, and funds seven grants across the United States. The grants were awarded to entities located in seven regions to allow each region to better address the challenges of enhancing, improving, or expanding 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.

The seven Regional Genetic Service Collaboratives are:

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<sup>4</sup> 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](http://babysfirsttest.org/newborn-screening/glossary#letter_s))

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

The goals of the Regional Genetic Service Collaboratives include:

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

The Regional Genetic Service Collaboratives' activities include:

- 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.

In addition, the Regional Genetic Service Collaboratives support the integration of genetics specialty services with medical home practices for patients diagnosed via newborn screening and addresses gaps and barriers to genetic services within the regions, especially for medically underserved populations.

## **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 considered to be 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 Secretary 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. Cooperative agreements were awarded 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.

## **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. Infants with undetected CCHD are at risk of having serious complications within the first few days or weeks of life and often require emergency care. 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.

On June 1, 2012, seven grants were awarded to support the implementation of CCHD screening. The CCHD Newborn Screening Demonstration Program provides support to state health departments in Wisconsin, Michigan, New Jersey, New Hampshire, Vermont, Rhode Island, Maine, Utah, and Virginia in order to do the following activities:

- Create a network, including the state public health department, hospitals, and/or birth facilities to coordinate service delivery;
- Develop a plan to incorporate CCHD screening and reporting at the provider and state levels, including a process for educating and training health care professionals on technologies for CCHD screening (such as the use of pulse oximetry monitoring);
- Develop guidelines for collecting and reporting results of CCHD screening and follow-up;
- Develop and deliver educational programs about CCHD newborn screening, counseling, testing, follow-up, treatment, and specialty services to parents, families, and patient advocacy and support groups;
- Document and track the number of infants screened, number of infants with abnormal screens who were referred for additional evaluations, cost per infant with an abnormal screen, number of infants diagnosed with CCHD before discharge, and number of infants diagnosed with CCHD by one year of age and the health outcomes of those infants;
- Establish guidelines for follow-up methodology and reporting; and
- Maintain a state level electronic system to collect information from hospitals and specialty care facilities in order to standardize, maintain, and analyze quantitative and qualitative data to monitor and evaluate the impact of CCHD screening.

## **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. Efforts to systematically evaluate health outcomes and impact beyond long-term survival, with a few exceptions, are just beginning. To facilitate these promising efforts, 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.

The Integrating Newborn Screening Long-Term Follow-up into Primary Care Practice initiative was awarded on August 1, 2013, and provided two grants to the Maryland Department of Health and Mental Hygiene and the Public Health Foundation Enterprises, Inc. in California. Grantees are examining the overarching questions outlined in the ACHDNC report for primary care practices, and the ability of primary care practices to provide accurate and ongoing information on patients identified with disorders detected through newborn screening in order to answer the following questions:

- Are children/adolescents receiving coordinated care through a medical home?
- How are the children/adolescents doing clinically?
- Are children identified through newborn screening and enrolled in care doing better than those identified clinically?
- As the primary care provider, am I doing the best for my patients?
- Do children in my care have the opportunity to enroll in clinical research studies?

Data from these demonstration projects will help determine the feasibility of accomplishing long-term follow-up in the primary care setting.

## **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 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 currently does not administer any programs under this section. The programs discussed below are administered by HRSA. HRSA grantees are required 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 7-8, is examining the effectiveness of screening in a timely manner. Participating states will be 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 will be 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; and
- Timing of newborn screening activities through categorization of the number of samples/screens collected within specific time intervals from birth through confirmation of diagnosis.

## **Newborn Screening Data Repository and Technical Assistance Center**

As part of its responsibilities, the NBS TA Center, described in more detail on pages 8-9, provides resources to state newborn screening programs so that they can evaluate the effectiveness of their processes and use real time data for quality improvement activities. For example, quality indicators were developed in collaboration with state newborn screening programs. State programs are in the process of providing the data for these quality indicators within a data repository. 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 9-11, 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](http://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 communities in 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 efforts and to inform them of innovative methods of educational outreach; and
- Evaluating project activities and results.

The Newborn Screening Clearinghouse began in 2009. The second cycle of funding was provided by a HRSA cooperative agreement awarded on September 1, 2014, to the Genetic Alliance. Recent accomplishments include launching the Spanish Newborn Screening Clearinghouse in April 2015. The Spanish site was created by focusing on the needs of Spanish-speaking families and includes information on health care access and costs. The Newborn Screening Clearinghouse has also developed a motion graphic to explain the newborn screening process called “What to Expect from Your Baby’s First Test.” The motion graphic is available on social media outlets including Facebook and YouTube and was displayed in Time Square in New York City as part of an evaluation to assess how large-scale visibility impacts people accessing the Newborn Screening Clearinghouse.

## **Part IV: Laboratory Quality and Surveillance**

CDC operates the nation's only newborn screening quality assurance program to ensure the accuracy of laboratory tests for heritable disorders in newborns. The Newborn Screening Quality Assurance Program (NSQAP) provides unique services directly to laboratories to maintain and enhance the quality of newborn dried blood spot screening. The program:

- Provides proficiency testing and quality assurance services for more than 50 conditions, including all primary disorders on the RUSP;
- Provides training and technical support to state laboratories to enhance nationwide laboratory capacity and capability;
- Prepares, certifies, and distributes more than 880,000 dried blood spot quality assurance materials to participating laboratories each year;
- Develops and hosts yearly hands-on training on laboratory techniques;
- Helps laboratories implement new screening technologies or add new conditions to their screening panel;
- 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.

NSQAP also helps public health laboratories implement new testing techniques as the field of newborn screening expands to include new technologies. NSQAP provides critical services to laboratories performing molecular (DNA-based) testing, which improves disease detection through increased test specificity and prediction of disease severity. Specifically, NSQAP:

- Provides molecular proficiency testing programs and quality assurance materials;
- Provides specialized technical assistance, laboratory development and design, and training on specific molecular tools for newborn screening laboratories;
- Develops and improves methods for molecular newborn screening tests;
- Facilitates the dissemination and implementation of recommended molecular lab practices; and
- Evaluates existing and newly implemented molecular programs through the innovative Molecular Assessment Program.

Program highlights during 2014-2015 included:

- Providing services to more than 600 laboratories, covering all 50 states and territories as well as 77 countries;
- Developing or improving five laboratory methods for screening current RUSP conditions or anticipated additions;
- Developing a newborn screening method for detecting spinal muscular atrophy, a common fatal defect of the nervous system, and combining the new method with an established test for detecting SCID, substantially reducing the additional cost of the new test; and



- Transferring and providing technical support to 12 states for molecular diagnostic methods for conditions, including galactosemia, hemoglobinopathies, and congenital adrenal hyperplasia.

## **Part V: Interagency Coordinating Committee on Newborn & Child Screening**

The ICC, co-chaired by HRSA and CDC, is composed of the HRSA Administrator, CDC Director, AHRQ Director, FDA Commissioner, 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 in order to make recommendations on heritable disorders that are recommended for newborn screening.

At the request of the Secretary, the ICC first met in May 2011 and has reviewed the following topics:

- Newborn screening for CCHD;
- Use and storage of residual newborn screening blood samples;
- Data quality assurance in newborn screening; and
- Newborn screening for Pompe disease.

## **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 was published in 2010<sup>5</sup> and distributed to the 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,"<sup>6</sup> which addresses newborn screening emergency plans. Also in 2011, the CDC's Office of Public Health Preparedness and Response published the Public Health Preparedness Capabilities: National Standards for State and Local Planning,<sup>7</sup> 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,

<sup>5</sup> See <http://www.cdc.gov/ncbddd/documents/NBS-CONPLAN.pdf>

<sup>6</sup> See [http://www.aphl.org/MRC/Documents/PHPR\\_2011Feb\\_PHL-Continuity-of-Operations-Guidelines.pdf](http://www.aphl.org/MRC/Documents/PHPR_2011Feb_PHL-Continuity-of-Operations-Guidelines.pdf)

<sup>7</sup> See [http://www.cdc.gov/phpr/capabilities/dslr\\_capabilities\\_july.pdf](http://www.cdc.gov/phpr/capabilities/dslr_capabilities_july.pdf)

CDC’s National Center on Birth Defects and Developmental Disabilities funded a non-governmental organization to assess existing plans and professional literature to update and revise the plan as needed.

## Part VII: Funding Amounts

(whole dollars)	Fiscal Year 2015 Enacted
<b>HRSA</b>	
Budget Authority – Heritable Disorders Program	\$13,883,000
Number of Awards	14
Average Award	\$842,400
Range of Awards	\$110,000 - \$2,000,000
<b>CDC</b>	
Budget Authority – Laboratory Quality and Surveillance	\$8,243,000
<b>TOTAL</b>	<b>\$22,126,000</b>

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

**Report to Congress**  
**Newborn Screening Activities**  
**Appendix A**

**Recommended Uniform Screening Panel - Core Conditions**

ACMG Code	Core Condition	Metabolic Disorder			Endocrine Disorder	Hemoglobin Disorder	Other Disorder
		Organic acid condition	Fatty acid oxidation disorder	Amino acid disorder			
PROP	Propionic Acidemia	X					
MUT	Methylmalonic Acidemia (methylmalonyl-CoA mutase)	X					
Cbl A,B	Methylmalonic Acidemia (Cobalamin disorders)	X					
IVA	Isovaleric Acidemia	X					
3-MCC	3-Methylcrotonyl-CoA Carboxylase Deficiency	X					
HMG	3-Hydroxy-3-Methylglutaric Aciduria	X					
MCD	Holocarboxylase Synthase Deficiency	X					
BKT	β-Ketothiolase Deficiency	X					
GA1	Glutaric Acidemia Type I	X					
CUD	Carnitine Uptake Defect/Carnitine Transport Defect		X				
MCAD	Medium-chain Acyl-CoA Dehydrogenase Deficiency		X				
VLCAD	Very Long-chain Acyl-CoA Dehydrogenase Deficiency		X				
LCHAD	Long-chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency		X				
TFP	Trifunctional Protein Deficiency		X				
ASA	Argininosuccinic Aciduria			X			
CIT	Citrullinemia, Type I			X			
MSUD	Maple Syrup Urine Disease			X			
HCY	Homocystinuria			X			
PKU	Classic Phenylketonuria			X			
TYR I	Tyrosinemia, Type I			X			
CH	Primary Congenital Hypothyroidism				X		
CAH	Congenital adrenal hyperplasia				X		
Hb SS	S,S Disease (Sickle Cell Anemia)					X	
Hb S/BTh	S, β-Thalassemia					X	
Hb S/C	S,C Disease					X	
BIOT	Biotinidase Deficiency						X
CCHD	Critical Congenital Heart Disease						X
CF	Cystic Fibrosis						X
GALT	Classic Galactosemia						X
GSD II	Glycogen Storage Disease Type II (Pompe)						X
HEAR	Hearing Loss						X
SCID	Severe Combined Immunodeficiencies						X

**Recommended Uniform Screening Panel<sup>1</sup>**  
**SECONDARY<sup>2</sup> CONDITIONS<sup>3</sup>**  
(As of March 2015)

ACMG Code	Secondary Condition	Metabolic Disorder			Hemoglobin Disorder	Other Disorder
		Organic acid condition	Fatty acid oxidation disorders	Amino acid disorders		
Cbl C,D	Methylmalonic acidemia with homocystinuria	X				
MAL	Malonic acidemia	X				
IBG	Isobutyrylglycinuria	X				
2MBG	2-Methylbutyrylglycinuria	X				
3MGA	3-Methylglutaconic aciduria	X				
2M3HBA	2-Methyl-3-hydroxybutyric aciduria	X				
SCAD	Short-chain acyl-CoA dehydrogenase deficiency		X			
M/SCHAD	Medium/short-chain L-3-hydroxyacyl-CoA dehydrogenase deficiency		X			
GA2	Glutaric acidemia type II		X			
MCAT	Medium-chain ketoacyl-CoA thiolase deficiency		X			
DE RED	2,4 Dienoyl-CoA reductase deficiency		X			
CPT IA	Carnitine palmitoyltransferase type I deficiency		X			
CPT II	Carnitine palmitoyltransferase type II deficiency		X			
CACT	Carnitine acylcarnitine translocase deficiency		X			
ARG	Argininemia			X		
CIT II	Citrullinemia, type II			X		
MET	Hypermethioninemia			X		
H-PHE	Benign hyperphenylalaninemia			X		
BIOPT (BS)	Biopterin defect in cofactor biosynthesis			X		
BIOPT (REG)	Biopterin defect in cofactor regeneration			X		
TYR II	Tyrosinemia, type II			X		
TYR III	Tyrosinemia, type III			X		
Var Hb	Various other hemoglobinopathies				X	
GALE	Galactosepimerase deficiency					X
GALK	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.

**Report to Congress**  
**Newborn Screening Activities**  
**Appendix B**

# Number of Core Disorders Screened

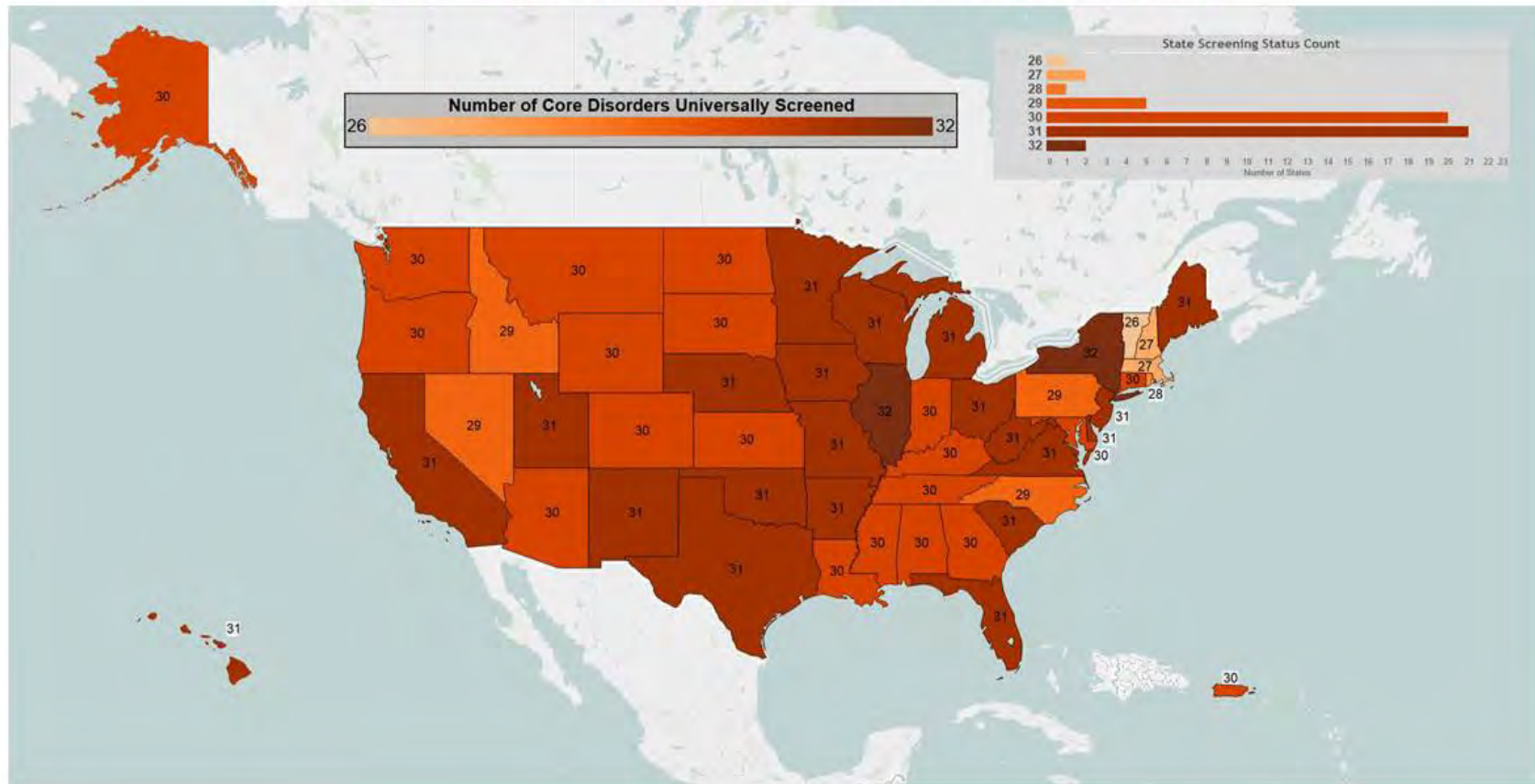


Figure 1: A map of the United States that shows the number of core RUSP disorders that are screened for within each state and territory as of September 2015. (From NewSTEPs data repository ([www.newsteps.org](http://www.newsteps.org)))