

NEWBORN SCREENING AND GENETICS

NEWBORN SCREENING

As a critical component of the newborn screening community, APHL is pleased with the passage of the Newborn Screening Saves Lives Reauthorization Act — signed into law as P.L. 113-240. This act enhances and assists states in improving state newborn screening programs, including providing education and training in newborn screening technologies, as well as coordinating follow-up care. To assure successful implementation, APHL also encourages Congress to provide \$8.3 million to fund the Centers for Disease Control and Prevention Newborn Screening Quality Assurance Program (NSQAP) to strengthen laboratory quality assurance programs, and \$13.8 million to fund the Health Resources and Services Administration Heritable Disorders Program.

The Newborn Screening Saves Lives Reauthorization Act (P.L. 113-240) is critical in ensuring that the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) continues its activities in support of newborn screening.

BASIC FACTS

- NBS saves lives. Each year, over 12,000 newborn lives are changed by early detection and intervention through newborn screening.
- NBS is used for the early identification of infants affected by certain genetic, metabolic, congenital and/or functional conditions.
- If diagnosed early, many heritable conditions can be cured or successfully treated.
- NBS is one of the largest and most effective programs of public health intervention, saving and improving the lives of children, families and communities.
- 98% of all infants born in the United States undergo newborn screening; however, the number and types of disorders for which newborns are tested varies from state to state. The American College of Medical Genetics, the American Academy of Pediatrics, the March of Dimes and the Secretary's Advisory Committee on Heritable Disorders recommend screening all newborns for 34 treatable disorders and reporting on an additional 26 conditions.



NEWBORN SCREENING SAVES LIVES ACT (P.L. 110-204)

In 2008, Congress and the Administration recognized the need for federal guidance and resources to assist states in improving their newborn screening programs by enacting the Newborn Screening Saves Lives Act (P.L. 110-204). On December 18, 2014, **The Newborn Screening Saves Lives Reauthorization Act (P.L. 113-240) was signed into law by the President, thus ensuring the following:**

- Enhancing state programs to provide screening, counseling and health care services to newborns and children
- Assisting in providing health care professionals with education about screening and training in relevant new technologies
- Developing and delivering educational programs about newborn screening counseling, testing, follow-up, treatment and specialty services to parents, families and patient advocacy and support groups
- Establishing, maintaining and operating a system to assess and coordinate treatment for disorders

CENTERS FOR DISEASE CONTROL AND PREVENTION

CDC's Newborn Screening Quality Assurance Program (NSQAP) is the only comprehensive program in the world devoted to ensuring the accuracy of newborn tests. The Newborn Screening Coalition recommends Congress fund the NSQAP program at \$8.3 million in FY 2017 to:

- Strengthen & enhance laboratory quality assurance programs.
- Enable public health laboratories to develop and refine screening tests, conduct pilot studies and implement new methods to improve detection of treatable disorders.
- Enhance newborn disorder detection through the Innovative Molecular Quality Program.

NEWBORN SCREENING QUALITY ASSURANCE PROGRAM: SUCCESS STORIES

- Ensured accurate newborn testing in more than 500 laboratories nationwide, and assured correct identification of 5,000-6,000 infants with treatable diseases that may have otherwise died or been disabled.
- Conducted laboratory studies and testing improvements that substantially supported the addition of x-linked adrenoleukodystrophy (x-ALD) to the HHS Secretary's Recommended Uniform Screening Panel (RUSP). Mucopolysaccharidosis-I (MPS-I) has also been added to the RUSP by the Advisory Committee on Heritable Disorders in Newborn and Children.
- Provided technical assistance, education, and financial support to NBS programs in Alabama, Arizona, Kansas, Kentucky, Maryland, North Carolina, North Dakota, Puerto Rico, Tennessee and Utah to move all programs towards full implementation of SCID.
- Established the innovative Molecular Assessment Program to provide guidance and technical expertise to newborn screening laboratories as they implemented molecular testing techniques to improve disease detection. This program assessed existing molecular NBS programs during on-site assessment visits to four state NBS laboratories.
- Initiated a program to improve testing for Sickle Cell Disease and Cystic Fibrosis in the US by expanding the diversity of available quality assurance materials in partnership with California, Indiana and Ghana.
- Provided national leadership in the development of guidance documents for newborn screening for Cystic Fibrosis, SCID, and Critical Congenital Heart Defect (CCHD).

FUNDING (in millions)

Heritable Disorders – Newborn Screening Saves Lives Reauthorization Act (HRSA)

FY 2016: \$13.8

FY 2017: \$13.8 (requested)

FY 2017: \$13.8 (required)

Newborn Screening Quality Assurance Program (CDC)

FY 2016: \$8.3

FY 2017: \$8.3 (requested)

FY 2017: \$8.3 (required)

HEALTH RESOURCES AND SERVICES ADMINISTRATION

HRSA's Heritable Disorders Program greatly assists state and local public health agencies in their ability to deliver services for newborns and children affected by heritable disorders. Recently HRSA has supported the Severe Combined Immunodeficiency (SCID) Program to facilitate the implementation of robust and sustainable screening and follow-up for this disorder. HRSA is also engaged in activities to improve the timeliness of newborn screening programs and in the development of data repository and technical assistance center.

In 2016, the SCID Program will support wider implementation, education, and awareness of newborn screening for SCID and continue to assist states in implementing screening for SCID; the Improving Timeliness of Newborn Screening Diagnosis initiative will allow for the continued development and implementation of quality improvement activities to improve the time to diagnosis and treatment for infants undergoing newborn screening who receive a presumptive positive result.

The Newborn Screening Data Repository and Technical Assistance Center will continue to provide technical assistance and programmatic support for the state public health programs, particularly as new conditions for newborn screening are considered and implemented. ■

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