NEWBORN SCREENING & GENETICS

UNMET NEEDS:

- Reduce delay of implementation of newly added disorders
- Funding for equipment, qualified staff and infrastructure changes to accommodate new testing
- Funding for test development and validation
- Quality assurance materials that reflect increased complexity of disease markers and address state's expanding needs are not available
- Coordinated efforts nationwide in leading novel advances in public health laboratories for newborn screening

NEWBORN SCREENING BASIC FACTS

- Newborn screening (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 public health interventions, 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 and Genomics, the American Academy of Pediatrics, the March of Dimes and the Advisory Committee on Heritable Disorders in Newborns and Children 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.

ASSOCIATION OF PUBLIC HEALTH LABORATORIES
8515 GEORGIA AVENUE, SUITE 700, SILVER SPRING, MD 20910 | WWW.APHL.ORG
240.485.2745 (P) | 240.485.2700 (F)

CDC EFFORTS TO SUPPORT NBS

The US 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 and 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 Improvement Program (MQIP).

SUCCESSES OF THE NSQAP AND MQIP:

- Ensured accurate newborn testing in more than 500 laboratories nationwide, and assured correct identification of 5,000–6,000 infants with treatable diseases, who may have otherwise died or been disabled.
- Conducted laboratory studies and testing improvements that supported the addition of x-linked adrenoleukodystrophy (x-ALD) to the HHS Secretary's Recommended Uniform Screening Panel.
- Provided technical assistance, education and financial support to move 10 state NBS programs towards full implementation of Severe Combined Immunodeficiency (SCID) screening, and supported the implementation of Mucopolysaccharidosis-I (MPS-1), Pompe and X-ALD newborn screening in 11 states.
- Established the innovative Molecular Assessment Program to provide guidance and technical expertise on molecular testing techniques, improving disease detection.
- 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, through partnership with California, Indiana and Ghana.
- Lead the development of national guidance documents for Cystic Fibrosis, SCID, and Critical Congenital Heart Defect screening.

CDC FUNDING

NEWBORN SCREENING QUALITY ASSURANCE PROGRAM

FY 2016:	**$8.3**
FY 2017:	**$8.3 (requested)**
FY 2017:	**$8.3 (necessary)**
FY 2018:	**$29.8 (necessary)**

HRSA FUNDING

HERITABLE DISORDERS—NEWBORN SCREENING SAVES LIVES REAUTHORIZATION ACT

FY 2016:	**$13.8**
FY 2017:	**$13.8 (requested)**
FY 2017/18:	**$13.8 (necessary)**

HRSA EFFORTS TO SUPPORT NBS

The US 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 NBS programs to facilitate the implementation of robust and sustainable screening and follow-up for SCID, MPS-1, X-ALD and Pompe.

HRSA is also engaged in activities to improve the timeliness of newborn screening programs and in the development of a data repository and technical assistance center. In 2017, the initiative on Improving Timeliness of Newborn Screening Diagnosis 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. APHL's Newborn Screening Technical assistance and Evaluation Program (NewSTEPs) 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. ■

CONTACT

Peter Kyriacopoulos, senior director of public policy
240.485.2766 | peter.kyriacopoulos@aphl.org