



# NEWBORN SCREENING & GENETICS

## UNMET NEEDS

- 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
- Coordinated efforts nationwide in leading novel advances (e.g., next generation sequencing, electronic data exchange, etc.) in public health laboratories for newborn screening



## BACKGROUND

Newborn screening (NBS) saves lives. Each year, over 12,000 newborn lives are changed because of the early detection and intervention NBS makes possible. NBS is one of the largest and most effective public health interventions in the US, saving and improving the lives of children, families and communities.

NBS is not a diagnostic test, but rather it determines a baby's risk for certain genetic, metabolic, congenital and/or functional disorders. Abnormal screening results cue healthcare providers to pursue additional diagnostic testing to determine if the baby has the disorder in question. If diagnosed early, these heritable conditions can be cured or successfully treated.

Almost all infants born in the US (about 98%) undergo NBS, however, the number and types of disorders for which newborns are screened varies from state to state. Most states follow the HHS Secretary's Recommended Uniform Screening Panel (RUSP), and 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 35 treatable disorders and reporting on an additional 26 conditions.

## NEWBORN SCREENING SAVES LIVES ACT

Recognizing the need for federal guidance and resources to assist states in improving their NBS programs, Congress enacted the Newborn Screening Saves Lives Act (P.L. 110-204) in 2008 and its reauthorization in 2014 (P.L. 113-240), ensuring:

- Enhanced state programs to provide screening, counseling and healthcare services to newborns and children.
- Assistance in educating healthcare professionals about screening and training in relevant new technologies.
- Development and delivery of educational programs about NBS counseling, testing, follow-up, treatment and specialty services to parents, families and patient advocacy and support groups.
- Establishment, maintenance and operation of a system to assess and coordinate treatment for disorders.

## CDC SUPPORTS QUALITY ASSURANCE

The US Centers for Disease Control and Prevention (CDC) Newborn Screening Quality Assurance Program (NSQAP) is the only comprehensive program in the world devoted



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to ensuring the accuracy of newborn tests. The Newborn Screening Coalition recommends Congress fund the NSQAP program at \$29.4 million in FY 2020 to:

- Strengthen and enhance lab quality assurance programs.
- Enable public health labs 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.
- Increase the use of modern testing platforms and create a “Data Hub” to facilitate data sharing and analysis.

The NSQAP is an essential component of the NBS system. This program has:

- Ensured that newborn screening provides accurate detection of disorders, timely diagnoses, minimal false positive reports, and sustains high quality performance in more than 500 labs nationwide.
- Conducted laboratory studies and testing improvements that supported the addition of spinal muscular atrophy (SMA) to the RUSP.
- Provided technical assistance, education and financial support to move six state NBS programs towards full implementation of severe combined immunodeficiency (SCID) screening, and supported the implementation of mucopolysaccharidosis-I (MPS-1), Pompe, SMA and x-ALD newborn screening in nine states.
- Supported the Molecular Assessment Program, which provides guidance and technical expertise on molecular testing techniques, improving disease detection.
- Initiated a program to improve testing for sickle cell disease and cystic fibrosis by expanding the diversity of available quality assurance materials, in partnership with California and Indiana.
- Lead the development of national guidance documents for cystic fibrosis, SCID, and critical congenital heart defect screening.

## CDC FUNDING

### NBS QUALITY ASSURANCE PROGRAM

FY 2019: \$16  
FY 2020: \$29.4 (necessary)

## HRSA FUNDING

### HERITABLE DISORDERS— NBS SAVES LIVES REAUTHORIZATION ACT

FY 2019: \$16.3  
FY 2020: \$32 (necessary)

(Dollars in millions)

## HRSA SUPPORTS NBS SERVICE DELIVERY AND QUALITY IMPROVEMENT

The US Health Resources and Services Administration (HRSA) 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 a number of NBS programs to facilitate the implementation of robust and sustainable screening and follow-up for SCID, MPS-1, x-ALD, Pompe and SMA.

HRSA is also engaged in activities to improve the quality practices within NBS programs and in enacting data-driven outcome assessments by supporting a national data repository and technical assistance center. The Newborn Screening Data Repository and Technical Assistance Center continues 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

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