

NEWBORN SCREENING 101

Each year, approximately 4 million babies are born in the United Statesⁱ, and nearly every one of those infants is screened by state newborn screening (NBS) programs for certain serious or life-threatening heritable disorders and medical conditions that can be treated.ⁱⁱ **Screening saves or improves the lives of more than 12,000 babies each year.**ⁱⁱⁱ

NBS isn't just a test. It's an interconnected public health system that relies on the coordinated activities of health care providers, laboratories, public health professionals, and parents. The system includes six critical components:

- Screening for congenital conditions
- Rapid follow-up and referral for infants with positive test results
- Confirmatory testing and diagnosis
- Implementation of treatments and therapies for diagnosed infants
- Education for all stakeholders
- Evaluation of the success of the system

This system not only saves lives, but also saves money for both the health care system and the taxpayer by preventing severe and permanent disabilities because infants receive early treatment.

In the United States, NBS is a federal and state collaboration. Each state controls its own program by determining which conditions to include on its panel of tests, collecting specimens, performing screening tests, and developing follow-up systems to identify and treat infants who screen positive for a disorder on the panel. The federal government supports state NBS programs by providing technical assistance, facilitating collaboration across states, and allocating financial resources to implement screening for new disorders and improve the quality of programs, among other activities. For example, the Centers for Disease Control and Prevention (CDC) provides from \$300,000 to \$500,000 to support states when adding conditions to their NBS panels, with the award varying by the number of births in the state. The National Institutes of Health (NIH) and the Health Resources and Services Administration (HRSA) also have programs to help states improve and enhance NBS programs. **Federal support and funding are essential to the success of our nation's NBS programs, but current federal efforts fall short in ensuring this success.**

The federal government maintains the Recommended Uniform Screening Panel (RUSP), a list of conditions that it recommends every state include on its NBS panel. The Secretary of the Department of Health and Human Services (HHS) adds conditions to the RUSP after receiving evidence-based recommendations from the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC). The RUSP currently includes 35 core disorders. In order to be added to the RUSP, a condition must be (1) identifiable within one to two days after birth; (2) have a screening test available; (3) benefit from early detection and intervention; and (4) have an effective treatment.

SAVINGS FROM NBS

- Every dollar California spends on tandem mass spectrometry NBS yields a benefit of \$9.32 in medical costs avoided and immeasurable value in lives saved.^{iv}
- Treating an infant with Severe Combined Immunodeficiency (SCID) can cost Medicaid as much as \$2 million. However, if diagnosed at birth, infants can be cured with a bone marrow transplant costing about \$100,000.^v

CITATIONS

- ⁱ Hamilton B, Martin J, Osterman M, et al. Births: Preliminary Data for 2015. National vital statistics reports; vol 65 no 3. Hyattsville, MD: National Center for Health Statistics. 2016.
 - ⁱⁱ CDC. CDC Grand Rounds: Newborn Screening and Improved Outcomes. Morbidity and mortality weekly report 2012;61(21);390-393.
 - ⁱⁱⁱ CDC. CDC Grand Rounds: Newborn screening and improved Outcomes. Morbidity and mortality weekly report 2012;61(21);390-393.
 - ^{iv} Feuchtbaum L, Cunningham G. Economic evaluation of tandem mass spectrometry screening in California. Pediatrics 2006;117:280-286.
 - ^v Buckley R. The long quest for neonatal screening for severe combined immunodeficiency. Journal of Allergy and Clinical Immunology 2012;129:597-604.
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