

INVESTMENT IN RESEARCH SAVES LIVES AND MONEY

Newborn Screening (NBS)

Newborn screening (NBS) is a system that begins when a newborn baby is tested for certain disorders that may not be noticeable at birth.¹ Many of these disorders can cause long-term health problems or early death, so detection of disorders through newborn screening allows providers to diagnose the disease and start treatment before any serious or permanent problems occur. Much of this testing is done before the baby leaves the hospital or birthing center using only a few drops of blood.^{1,2} Each state in the U.S. has different requirements around which diseases are screened in NBS programs depending on disease prevalence, detectability, treatment availability, outcome, state legislation, and overall cost effectiveness.³ The Advisory Committee on Heritable Disorders in Newborns and Children nominates conditions to the Department of Health and Human Services (HHS) to create a recommended list of disorders for which every baby should be screened.⁴ Some core conditions on the list include cystic fibrosis, sickle cell disease, and hearing loss.

TODAY

More than
98%

of babies born in the United States are screened.⁵

Almost

1 out of 300

newborns screened is diagnosed with a condition.⁵

According to the CDC, NBS is one of the **top 10 public health achievements**

of the first 10 years of the 21st century.⁶

COST

Most screenings are roughly **\$100 per newborn** and screening is performed regardless of the parents' health insurance status or ability to pay.^{1,8}

\$500,000-\$1M:

Lifetime costs of developmental diseases that can be avoided with NBS.⁷

\$1-\$2 billion:

Yearly national savings for screening for phenylketonuria (PKU).¹⁰

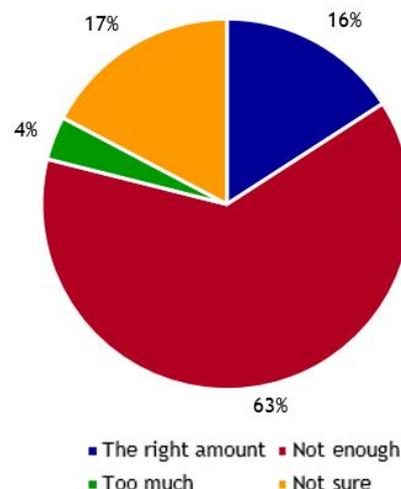
The U.S. spends about 5 cents of each health dollar on research to prevent, cure and treat disease and disability. Do you think that this is too much, the right amount, or not enough?

Research Delivers Solutions

Before the early 1990s, adding more tests to a NBS panel required more blood collected from the child and continuous assessment of data. Researchers tested how well **tandem mass spectrometry (MS/MS)**, an instrumental analysis, would be in detecting conditions. In one study of over 250,000 newborns, screening using MS/MS approximately doubled the disease detection rate compared to conventional methods. Now, testing in NBS using MS/MS is cheaper, faster, and more efficient in diagnosing more disorders at once.¹¹

Researchers wanting to determine the effectiveness of NBS for **sickle cell disease (SCD)** analyzed the screenings of more than 80,000 newborns born between 1975 and 1985. In a group of patients who were diagnosed with SCD in the newborn period, the overall mortality rate was 1.8%. In the second group of patients who were diagnosed after three months of age, the overall mortality rate was 8%. Both groups had extensive follow-up for 7.2 years and 9.4 years, respectively. The research concludes that NBS coupled with extensive follow-up can significantly decrease patient morbidity and mortality.¹²

One observational study between 2007 and 2013 found that there was a 33.4% decline in infant deaths from **critical congenital heart disease** in eight states with mandated screening policies compared to states without mandated screening policies.¹³ Their results show a significant decline, suggesting a likely correlation between implementing statewide policies and reduced infant cardiac deaths.



Source: A Research!America poll of U.S. adults conducted in partnership with Zogby Analytics in January 2019

Newborn Screening (NBS)

Then. Now. Imagine.

THEN

Between the 1960s and the 2000s, there were no national recommendations centered on what diseases should be included in NBS programs, and most NBS programs checked between three and 10 disorders.¹⁴

NOW

HHS issues a recommended newborn panel of screenings that is updated as new disorders are nominated and accepted. In 2018, HHS recommended screening for 35 conditions.¹⁴

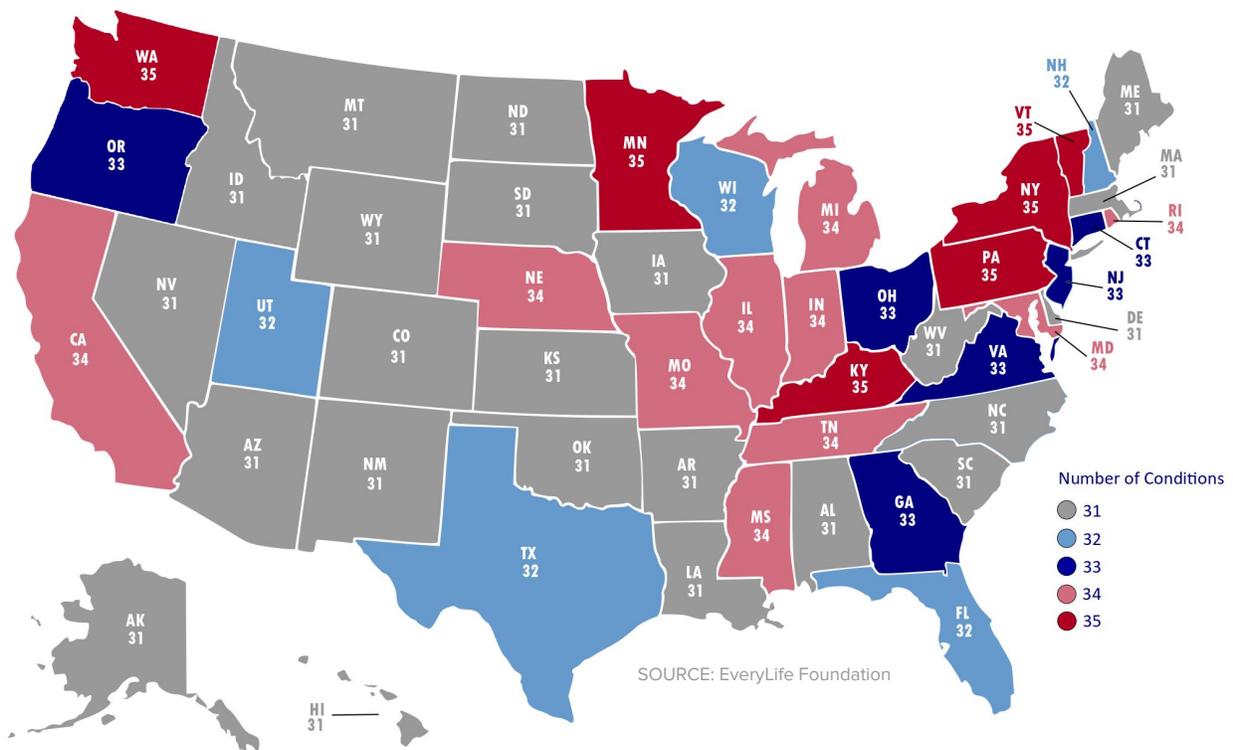
IMAGINE

A nation with more comprehensive NBS.

Quest for Diagnosis

HHS and the Advisory Committee on Heritable Disorders in Newborns and Children maintain the **Recommended Uniform Screening Panel (RUSP)**, a standardized list recommending which disorders states may want to screen for in their NBS programs. In order to add a condition to the RUSP, the disorder has to meet three criteria: the disorder must be detectable within 48 hours of birth, the screening must be sensitive and specific, and early detection must enable treatment to secure a proven benefit. States are not required to follow the RUSP, and it can take an average of three to five years before a state will adopt a new condition for their screening program.⁴

Number of Conditions Screened by State



1. "What is newborn screening?" NIH. 2020.

2. "Newborn screening." CDC. 2019.

3. "How many newborns are screened in the US?" NIH. 2017.

4. Xu et al. "Delay in state adoption of newborn screening tests." *Pediatrics*. 2018; 141(1): e20170300.

5. "Newborn screening and improved outcomes." CDC. 2012.

6. "Ten Great Public Health Achievements." CDC. 2011.

7. Grosse, Scott. "Does newborn screening save money? The difference between cost-effective and cost-saving interventions." *J of Ped*. 2005 Feb; 146:168-170.

8. Botkin et al. "Whole genome sequencing and newborn screening." *Curr Genet Med Rep*. 2016; 4(1): 1-6

9. Chan, Kee & Petros, Michael. "Simple test, complex system: Multifaceted views of newborn screening science, technology, and policy." *Global Ped Health*. 2019; 6(1-10).

10. "Newborn screening saves lives and money." March of Dimes. N.d.

11. Schulze et al. "Expanded newborn screening for inborn errors of metabolism by electrospray ionization-tandem mass spectrometry: results, outcome, and implications." *Pediatrics*. Jun 2003; 111(6): 1399-1406.

12. Vichinsky et al. "Newborn screening for sickle cell disease: Effect on mortality." *Pediatrics*. 1989; 83(5): 834.

13. Abouk, Rahi et al. "Association of US state implementation of newborn screening policies for critical congenital heart disease with early infant cardiac deaths." *JAMA*. 2017;318(21):2111-2118.

14. "Recommended Uniform Screening Panel." HHS. 2019.

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The Albert and Mary Lasker Foundation is a founding partner in this series of fact sheets. www.laskerfoundation.org

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