

Improving Access to Newborn Screening



Rare Disease Patients Face Significant Challenges In Receiving Diagnoses

Of the **6,000+** rare diseases, **nearly three-quarters are genetic** and of those **70%** begin in childhood.¹ These rare, genetic diseases have a variety of signs and symptoms that manifest differently in patients, resulting in misdiagnoses and delayed treatment.



Delayed Treatment & Misdiagnoses Result in Severe Decreases in Quality of Life

Due to the debilitating nature of these diseases, which often cause chronic, progressive, and life-threatening issues, patients may face an unnecessary increase in morbidity and mortality without timely access to an accurate diagnosis through newborn screening (NBS).

TO LEARN MORE, VISIT [GENE-THERAPIES.ORG](https://www.gene-therapies.org).



Access to Newborn Screening Will Save Lives

To mitigate unnecessary misdiagnoses, optimize outcomes, and accelerate availability of new cures, NBS must be modernized. Enhanced access to NBS can facilitate diagnosis, monitoring, and treatment, which are all critical for patients with rare and serious diseases. Access to NBS may also help address the inequities that play a role in delaying early diagnoses for people of color. The pace at which gene therapies are advancing through the pipeline will quickly outstrip existing federal and state NBS capacity. It currently can take more than a decade to achieve nationwide screening of a new condition, which is unacceptable for patients.



Federal & State Support is Critical

At the federal level, the Advisory Committee on Heritable Disorders in Newborn and Children (ACHDNC) is responsible for recommending disorders for newborn screening. The ACHDNC maintains a Recommended Uniform Screening Panel (RUSP), which consists of a standardized list of disorders that are recommended for states to implement in their NBS programs. The Committee engages in lengthy processes for evaluating the disorders to include on the list. While the majority of states screen for most disorders on the list, some states screen for additional disorders and others are at various stages of adopting more recent recommendations. State public health departments are tasked with making determinations on which tests to include within the state NBS program, resulting in a lack of consistency across states. For example, a newborn diagnosed with a rare disease in one state may be able to receive prompt care and treatment, but if a neighboring state does not screen for the same disease, a newborn would likely go undiagnosed and untreated.



Congressional Action is Necessary

IGT encourages Congress to reauthorize the Newborn Screening Saves Lives Act, which expired in September of 2019, and provides vital funding for continuing the program. Furthermore, we urge Congress to consider opportunities to ensure the entire U.S. newborn screening ecosystem, including the federal RUSP process and states, can keep pace with transformative new technologies, which could include:

- Modernization of the RUSP process to eliminate redundancies and accelerate the ability to recommend new conditions, including preliminary RUSP inclusion/or RUSP expansion for conditions with gene therapies in development or that received marketing approval;
- Additional funding and support to states to accelerate state compliance with RUSP recommendations; and,
- Public-private partnerships for financing newborn screening pilots and implementation of new conditions.

RARE DISEASE PATIENTS DESERVE A FIGHTING CHANCE - WE MUST IMPROVE ACCESS TO NEWBORN SCREENING TO HELP PATIENTS RECEIVE THE GENE THERAPIES THEY NEED.