Improving Access to Newborn Screening

IGT Institute for Gene Therapies

Rare Disease Patients Face Significant Challenges In Receiving Diagnoses

Of the **10,000+ rare diseases**, about 70% are genetic and begin in childhood. 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 newborn screening (NBS) diagnosis.

Access to Newborn Screening Will Save Lives

To mitigate unnecessary misdiagnoses, optimize outcomes, and accelerate new cures development, 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. The pace at which gene therapies are advancing through the pipeline will quickly outstrip existing federal and state NBS capacity. It currently can take up to 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 Committee includes representatives from across HHS, as well as a variety of healthcare experts. 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 the 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 is able to receive prompt care and treatment; if a neighboring state does not screen for the same disease, a newborn would likely go undiagnosed and untreated.

Congressional Action is Necessary

The Newborn Screening Saves Lives Act of 2021 provides vital funding for continuing this program, but Congress can do more to help 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:

- Public-private partnerships for financing newborn screening pilots and implementation of new conditions;
- 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; and,
- Additional funding and support to states to accelerate state compliance with RUSP recommendations.

Rare disease patients deserve a fighting chance – we must improve access to newborn screening to help patients receive the gene therapies they need.

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