

May 28, 2021

VIA ELECTRONIC DELIVERY

Tiina K. Urv, Ph.D.
National Center for Advancing Translational Sciences
National Institutes of Health
9000 Rockville Pike
Bethesda, Maryland 20892

RE: Request for Information: Facilitating the Early Diagnosis and Equitable Delivery of Gene-Targeted Therapies to Individuals with Rare Diseases

Dear Dr. Urv,

The Institute for Gene Therapies (IGT) is writing to submit considerations for the National Institute of Health's (NIH) National Center for Advancing Translational Sciences (NCATS) Request for Information (RFI) titled, "Facilitating the Early Diagnosis and Equitable Delivery of Gene-Targeted Therapies to Individuals with Rare Diseases." Ensuring early identification of gene therapy patients and equitable access to gene therapy products is crucial to improving the lives of those living with these conditions. Our comments focus on considerations for identifying gene therapy patients, such as newborn screening programs and genetic testing, as well as considerations for facilitating patient access to gene therapies.

IGT was launched in February of 2020, with a focus on advocating for a modernized regulatory and reimbursement framework that encourages the development of transformative gene therapies and promotes patient access. Through a Corporate Advisory Council, Patient Advocacy Advisory Council, and Scientific, Academic & Medical Council, the Institute represents a wide array of patient advocacy groups, gene therapy manufacturers, and scientific, medical, and academic stakeholders seeking to advance the promise of gene therapies. IGT aims to inform the conversation regarding the value of transformative therapies and advocate for policies and practices to ensure patient access to these treatments. A full list of our members is available at <https://www.gene-therapies.org/advisory-councils>.

Identifying Gene Therapy Patients

Rare disease patients, as well as their families and caretakers, face significant challenges in receiving conclusive diagnoses. With over 6,000 rare diseases, 72% are genetic and 70% of those genetic diseases 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. Furthermore, delayed treatment and misdiagnoses causes severe decreases in quality of life due to the debilitating nature of such diseases, leading to chronic, degenerative,

progressive, and life-threatening issues. Without accessible screening for such conditions, an unnecessary increase in morbidity and mortality may occur for patients with rare diseases.

Bolstering Newborn Screening Programs

Due to the debilitating nature of these diseases, patients may face an unnecessary increase in morbidity and mortality without timely access to an accurate diagnosis through newborn screening (NBS). To mitigate unnecessary misdiagnoses, optimize outcomes, and accelerate the 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.

At the federal level, the Advisory Committee on Heritable Disorders in Newborns 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.

IGT supports reauthorization of the Newborn Screening Saves Lives Act, which expired in September 2019, and provides vital funding for continuing the program. Furthermore, IGT is urging Congress to consider opportunities to ensure the entire US 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.

Providing Genetic Screening to All Patients

Enhanced access is needed for screening tests beyond NBS that can facilitate diagnosis, monitoring, and treatment, which are all critical for patients with rare and serious diseases. Availability and affordability of genetic testing is key to ensuring that patients are aware of clinical studies and can obtain the benefits of approved gene therapies. Robust genetic screening should be available to newborns and all people at elevated risk or suspected of having a genetic disorder. All payers in US, including public and private insurers, should establish policies for automatic coverage of the appropriate genetic tests at the time of approving coverage for a new gene therapy for a genetic disease/disorder to prevent undue treatment delays.

Ensuring Patient Access to Gene therapies

After more than 20 years of careful study, gene therapies are now making their way through the FDA approval process and to patients who need them. In many cases, gene therapies halt but cannot reverse the effects of a disease by addressing the underlying genetic cause. Delays in access to approved gene therapies can result in patients suffering irreversible damage caused by their disease. Increased access to gene therapies benefits patients' short- and long-term health, their caregivers, the healthcare system, and society. IGT is working with policymakers and stakeholders to develop a sustainable, flexible, and permanent payment pathway reflective of the scientific advancements resulting in gene therapy breakthroughs.

Encouraging Greater Adoption of VBP Arrangements

IGT supports developing a reimbursement system equipped to adequately reimburse providers for administering groundbreaking gene therapies. A novel reimbursement pathway will drive sustainability for gene therapies while enabling patient access and provider payment. Value-based payment arrangements (VBPs), in which payment is tied to a patient's actual outcomes after receiving a therapy, are a critical component of a modern reimbursement system. VBPs are a vital tool that can support the flexibility that manufacturers, payers, and providers need to define payment terms and outcomes measures that will work best in the context of a given disease and patient.

Facilitating Pay-Over-Time Arrangements

Not all disease states that may be treated with a gene therapy are amenable to VBPs, such as those for rapidly progressive, degenerative diseases. IGT supports the development of mechanisms to facilitate payment-over-time arrangements for gene therapies, which may support expanded access to certain gene therapies.

Coordinating Out-of-State Care in Medicaid

Care coordination for out-of-state care in the Medicaid program is pivotal to actualizing the potential of gene therapies for children with medically complex conditions. Without appropriate care coordination, undue delays in access to treatment can result in detrimental impacts on patients and their families, including increased morbidity and mortality. Both the Medicaid and CHIP Payment and Access Commission (MACPAC) and American Academy of Pediatrics have recognized the need for updating and streamlining out-of-state care for children with medically complex conditions.^[1] Furthermore, care coordination in Medicaid is critical for children in rural areas, where there are significantly lower rates of specialists.^[2]

To facilitate better care coordination for out-of-state care for children with medically complex conditions, CMS should take steps beyond its 2020 Request for Information (RFI) on Coordinating Care from Out-of-State Providers for Medicaid-Eligible Children with Medically Complex Conditions to reduce administrative, fiscal, regulatory, and access barriers for families; make it easier for providers to enroll, obtain adequate reimbursement, and provide care for out-of-state children; and improve transparency and education for

^[1] Medicaid & CHIP Payment and Access Commission, *Issue Brief: Medicaid Payment and Policy for Out-of-State Hospital Services* (Jan. 2020), <https://www.law.georgetown.edu/wp-content/uploads/2018/07/Rule-18-Handout-1.Secara-1.pdf>; Council on Children with Disabilities and Medical Home Implementation Project Advisory Committee, *Patient- and Family-Centered Care Coordination: A Framework for Integrating Care for Children and Youth across Multiple Systems*, 133 PEDIATRICS e1451, e1451-60 (2014).

^[2] *Improving Healthcare Value in Rural America*, Altarum Healthcare Value Hub Res. Brief 19 (Oct. 2017), <https://www.healthcarevaluehub.org/advocate-resources/publications/improving-healthcare-value-rural-america>.

beneficiaries, CMS, states, and manufacturers alike. IGT urges CMS to integrate best practices for care coordination into its State Medicaid Program and Medicaid Managed Care Organizations (MCOs) guidance.

Conclusion

IGT appreciates the opportunity to submit these comments for NIH to consider when addressing early diagnosis and equitable delivery of gene-targeted therapies to individuals with rare diseases. IGT would be pleased to serve as a resource on gene therapy issues and answer any questions regarding these recommendations.

Sincerely,

A handwritten signature in black ink that reads "Erik Paulsen". The signature is written in a cursive style with a large, prominent initial "E".

The Honorable Erik Paulsen
Chairman
Institute for Gene Therapies