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gene-therapies.org

September 15, 2025

VIA ELECTRONIC DELIVERY

CDR Leticia Manning, MPH
Designated Federal Officer
Maternal and Child Health Bureau
Health Resources and Services Administration
Department of Health and Human Services
5600 Fishers Lane
Rockville, Maryland 20852

RE: Notice With Request for Comment: Consideration of Adding Duchenne Muscular Dystrophy to the Recommended Uniform Screening Panel

Dear Ms. Manning:

The Institute for Gene Therapies (IGT or "the Institute") is pleased to submit these comments to the Health Resources and Services Administration (HRSA or "the Agency") regarding the Notice With Request for Comment: Consideration of Adding Duchenne Muscular Dystrophy to the Recommended Uniform Screening Panel ("Notice").[1] IGT supports HRSA's efforts to consider and evaluate the addition of Duchenne Muscular Dystrophy (DMD) to the Recommended Uniform Screening Panel (RUSP). IGT is dedicated to supporting efforts that ensure the Panel's recommendations keep pace with the rapidly evolving therapeutic landscape, and we submit feedback in response to the Notice.

IGT supports efforts to realize the value of transformative gene therapies for patients, caregivers, the healthcare system, and society at large. More specifically, IGT advocates for efforts that mitigate unnecessary misdiagnoses, optimize outcomes, accelerate new cures development, and modernize newborn screening (NBS). Enhanced access to screening tests facilitates diagnosis, monitoring, and treatment, which are all critical for patients with rare and serious diseases. It is imperative that HRSA recognize that diagnosis must keep pace with innovation in genomic medicine, as failure to do so will limit the ability to deliver these transformative treatments to patients in a timely manner to ensure maximum benefit.

IGT was launched in February 2020 to advocate for a modernized regulatory and reimbursement framework that encourages the development of transformative gene therapies and promotes patient access. Through our Patient Advocacy Advisory Council, Corporate 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. Our response to this Notice represents our perspective as a membership group and focuses on areas where our members have firsthand experience or knowledge.[2]

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[1] U.S. Department of Health and Human Services, Health Resources and Services Administration. Notice With Request for Comment: Consideration of Adding Duchenne Muscular Dystrophy to the Recommended Uniform Screening Panel. 2025. Accessed from: https://www.federalregister.gov/documents/2025/08/14/2025-15433/notice-with-request-for-comment-consideration-of-adding-duchenne-muscular-dystrophy-to-the.

[2] A complete list of our members is available at https://www.gene-therapies.org/about-igt.

I. Recommendations Regarding the Addition of DMD to the RUSP

Of the 10,000+ rare diseases, a majority are linked to genetic causes, and approximately 70% affect children.[3,4] DMD is one example of a rare, genetic disease that has a variety of signs and symptoms that manifest differently in patients, often resulting in misdiagnoses and delayed treatment. DMD is a rare, progressive genetic neuromuscular disease, which limits life expectancy to a median of 28.1 years.[5] DMD typically presents in early childhood with progressive muscle weakness, along with difficulty walking, frequent falls, enlarged calf muscles, and delayed motor milestones. As the disease progresses, respiratory and cardiac complications may develop due to muscle degeneration, substantially impacting the patient's quality of life.[6]

Availability of NBS for DMD is key to ensuring that patients and their families are aware of available clinical studies, which allow patients to obtain benefits of approved gene therapies. It is critical for patients to be screened and diagnosed early, as to allow the full benefits of gene therapies. When implemented consistently, NBS offers actionable knowledge for DMD, ensuring that a family can pursue early diagnostic confirmation and potential intervention before symptom onset or progression.

As our healthcare system prepares for a wave of life-changing therapies for pediatric genetic conditions over the next decade, early diagnosis will be crucial to achieving optimal patient outcomes. IGT urges HRSA through its RUSP policies to keep pace with innovation in genomic medicine. Robust genetic screening should be available to all newborns, and all people at elevated risk or suspected of having a genetic disorder like DMD. To this aim, IGT supports the addition of DMD to the RUSP.

II. Conclusion

IGT greatly appreciates HRSA's interest in soliciting public feedback regarding the addition of DMD to the RUSP. There are now over 10,000 rare genetic diseases that have been described – a number that will only continue to grow as we learn more about how genetic alterations drive disease processes. Achieving early diagnosis for rare diseases is critical to giving patients the chance to benefit from early treatment. NBS is a vital tool that can help eliminate a burdensome diagnostic odyssey and ensure equitable access to early diagnosis and treatment. Thank you again for the opportunity to provide this information, and we look forward to continued engagement on the issue.

Sincerely,

John R. Feore, III
Director, Health Policy and Advocacy
Institute for Gene Therapies

[3] National Organization for Rare Diseases. NORD Rare Disease Database. 2025. Accessed from: http://rarediseases.org/rare-diseases/.
[4] Health TL. The landscape for rare diseases in 2024. The Lancet. Global health. 2024 Mar;12(3):e341. Accessed from: https://www.thelancet.com/journals/langlo/article/PIIS2214-109X(24)00056-1/fulltext.
[5] Broomfield J, Hill M, Guglieri M, Crowther M, Abrams K. Life Expectancy in Duchenne Muscular Dystrophy: Reproduced Individual Patient Data Meta-analysis. Neurology. 2021 Dec 7;97(23):e2304-e2314. doi: 10.1212/WNL.0000000000012910. Epub 2021 Oct 13. PMID: 34645707; PMCID: PMC8665435.
[6] Muscular Dystrophy Association. Duchenne Muscular Dystrophy. 2025. Accessed from: https://www.mda.org/disease/duchenne-muscular-dystrophy.