May 15, 2026

Carolyn M. Greene, MD
Acting Director
National Center for Health Statistics
Centers for Disease Control and Prevention
3311 Toledo Road
Room 7122
Hyattsville, MD 20782

Re: Organizing Principles for Classification of Ultra-rare and Genetic Conditions

Dear Dr. Greene,

In service of the neuromuscular disease (NMD) patient community, the Muscular Dystrophy Association (MDA) thanks the ICD-10 Coordination and Maintenance Committee (the Committee) for the opportunity to comment on the "Organizing Principles for Classification of Ultra-rare and Genetic Conditions". While some rare neuromuscular diseases have ICD-10 codes, including several limb-girdle muscular dystrophies (LGMD) in which MDA led a coalition of stakeholders to obtain these codes in 2022, many other neuromuscular diseases do not yet have their diagnostic code.[1] Consequently, the approach the Committee chooses to take is of great importance and relevance to our community.

MDA is the #1 voluntary health organization in the United States for people living with muscular dystrophy, ALS, and related neuromuscular diseases. For over 75 years, MDA has led the way in accelerating research, advancing care, and advocating for the support of our community. MDA's mission is to empower the people we serve to live longer, more independent lives.

Most rare neuromuscular diseases under MDA's umbrella are genetic diseases, and most of these are ultra-rare conditions affecting fewer than 1,000 individuals in the United States. Due to the advent of genetic medicine, more and more of these communities are witnessing promising therapeutic development efforts, often genetically-targeted therapies that address the root cause of the disease. ICD-10 codes specific to these diseases are paramount to ensuring equitable access and coverage of not only these promising therapies, but also the multidisciplinary care tailored specifically to the neuromuscular disease obtained at MDA's over 150 care centers across the country. Furthermore, ICD-10 codes facilitate surveillance and epidemiological research, critical endeavors to understanding these under-researched diseases. Our community must also see many clinicians and medical professionals, making care coordination particularly difficult.

[1] https://www.mda.org/press-releases/the-lgmd-community-celebrates-adoption-of-icd-10-diagnostic-codes-for-lgmd

For these reasons and more, we strongly urge the Committee to preserve our community's ability to obtain an ICD-10 code specific to their rare disease. Any effort that compromises the possibility of rare disease communities obtaining ICD-10 codes while preserving the same functionality for common disease communities is unacceptable and discriminatory against those with rare diseases.

Below are MDA's recommendations to the Committee as it deliberates on the best way forward.

Please lift the pause on new rare genetic diseases ICD-10 code nominations: As the Committee deliberates, we strongly urge you to lift the pause on consideration of new rare genetic disease ICD-10 code nominations. We do not see any reason why the Committee cannot concurrently deliberate on the future approach while still accepting nominations. There are communities within the neuromuscular disease population interested in nominating their condition for an ICD-10 code, but now are in limbo as they await the Committee's lifting of the pause. We urge the Committee to lift the pause and accept new nominations.

Preserve the ability to nominate a condition using the underlying genetic cause: As MDA was collaborating with the LGMD stakeholder community, we faced the question of whether we should nominate the LGMD subtypes based upon the genetic underpinnings of the disease or upon the clinical nomenclature. We decided upon using the genetic underpinning (ex. LGMD due to dysferlin dysfunction) for several reasons that may prove instructive to the Committee as it deliberates:

1. <u>The genetic underpinning of disease does not change while the clinical nomenclature often does:</u> We collectively chose to identify each disease based upon the protein dysfunction resulting from the genetic mutation because this underlying dysfunction does not change as the clinical nomenclature may. There are now over 30 recognized LGMDs that have been called different names at different times. In fact, we expect continued evolution on how the LGMDs are clinically classified. Thus, if the LGMD ICD-10 codes were based upon clinical nomenclature, they could soon be out of date.

2. <u>The genetic underpinning may result in varying phenotypes:</u> The same genetic mutation underlying an LGMD may also result in varying clinical diagnoses depending on the disease severity, manifestations, or progression. For example, certain genetic mutations resulting in LGMDs may also result in a clinical diagnosis of a myopathy (for example, Limb girdle muscular dystrophy due to anoctamin-5 dysfunction can also be diagnosed as Miyoshi myopathy type 3). Furthermore, mutations in the gene that codes for fukatin-related proteins can result in LGMDs, congenital muscular dystrophies, and myopathies. The ways in which the field classifies these diseases may evolve, but the underlying genetics likely will not.

3. <u>Treatments are evolving to target the underlying genetic cause of the disease:</u> Genetic medicine continues to evolve to facilitate the development of new treatments that treat the underlying genetic cause of the disease. Consequently, ICD-10 codes that also reflect the underlying genetic cause align with the direction of scientific discovery and medicine.

ICD-10 codes classified by genetic cause may not work for all genetic diseases - while neuromuscular diseases are often monogenic (originating from a single gene mutation), many other diseases are polygenic in nature. Consequently we do not have an overarching framework to recommend, but simply urge the Committee to preserve the ability to create ICD-10 codes based upon the underlying genetic cause of the disease.

Thank you for the opportunity to provide our viewpoints on the Committee's ongoing deliberations. For questions regarding MDA or the above comments, please contact Paul Melmeyer, Executive Vice President, Public Policy and Advocacy, at pmelmeyer@mdausa.org,

Sincerely,

Paul Melmeyer, MPP
Executive Vice President, Public Policy and Advocacy
Muscular Dystrophy Association