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The EveryLife Foundation for Rare Diseases is a 501(c)3 organization
Tax ID # 26-4614274
1012 14th Street, NW
Washington, DC 20005
T (202) 697-RARE (7273)
www.everylifefoundation.org

May 15, 2026

National Center for Health Statistics
Centers for Disease Control and Prevention
3311 Toledo Road
Room 7122
Hyattsville, MD 20782

To Whom It May Concern,

Thank you for the opportunity to provide input to guide the development of principles that clarify the ICD-10-CM scope and process for genetic conditions. As genomic medicine advances, the ICD-10-CM system must clearly and consistently represent diseases defined by molecular etiology.

The EveryLife Foundation for Rare Diseases is an evidence-based policy organization comprised of a robust coalition of nearly 400 patient advocacy organizations, biopharmaceutical partners, coalition groups, and other relevant stakeholders with a shared interest in rare disease, many of whom have already or soon plan to navigate the ICD-10-CM nomination process. At the urging of our coalition, the EveryLife Foundation developed the ICD Code Roadmap to help patient organizations assess their nomination readiness level and navigate the evidence collection and submission process. Through this activity and our experience with the limitations in developing evidence to guide rare disease policy due to the limited number of ICD-10 codes corresponding to rare conditions, we have a unique appreciation for the need to identify a streamlined process that can keep pace with advances in genomic medicine.

In FY2022, the EveryLife Foundation worked with Members of Congress to include language in the Labor, Health and Human Services Appropriations Report requesting that the National Center for Health Statistics (NCHS) and the Centers for Medicare and Medicaid Services (CMS) identify a streamlined process for rare disease code evaluation.

FY 2022 Labor HHS Report Language (p. 236)[1]:

Improving Accuracy of Diagnosis of Rare Diseases.—The Committee is aware that the lack of a precise diagnostic code makes disease surveillance, including understanding the true prevalence of a condition, more difficult and hinders diagnosis, treatment, and access to approved therapies. The Committee encourages HHS to work with federal agency partners to establish a pathway for rare disease stakeholders to pursue a diagnosis code, including resources to advise stakeholders as to data and

[1] .7/crpt/hrpt96/CRPT-117hrpt96.pdf

other needs and to help facilitate the application process for rare disease codes.

The lack of specific codes creates challenges for clinicians, researchers, and health systems seeking to accurately document these conditions. When cases are captured under broad or nonspecific categories, insurance coverage and reimbursement are challenged, the true prevalence of disorders is obscured, and our ability to conduct epidemiologic research and monitor outcomes is reduced. In addition, the lack of ICD-10 codes hinders efforts to generate real-world evidence, which is increasingly important for medical product development for rare conditions.

Our Process

To inform EveryLife's response to the CDC's request for information, we convened an ad hoc meeting of our Community Congress coalition member organizations. Over 150 leaders from coalition member organizations joined the discussion, the largest ad hoc convening since COVID-19-related meetings, indicating significant interest and involvement by patient organizations in coding discussions.

Additionally, the EveryLife Foundation team evaluated multiple proposals from external organizations and partnered with COMBINEDBrain and other coding experts to evaluate the needs and opportunities related to streamlined ICD-10-CM organizing principles.

Following these discussions, EveryLife Foundation determined that the proposal submitted by MONDO, and endorsed by dozens of organizations and individuals, reflected the process improvements for submitting and evaluating new code requests that the community desired. ***After thoughtful review, EveryLife Foundation has endorsed the MONDO proposal as a solid starting point for further conversations.***

Additionally, EveryLife Foundation signed the letter submitted to the CDC by COMBINEDBrain, highlighting the benefits of the MONDO proposal and a proposal submitted by the Novagen Research Fund (NGRF). While EveryLife did not formally evaluate and endorse the NGRF proposal due to time constraints, we support incorporating elements of both approaches into the next stages of CDC's process to create new organizing principles.

Based on input from discussions with Community Congress coalition members and the experiences shared with us by member organizations, we highlight several considerations for advancing the final organizing principles in three domains:

- **The Value of Specific ICD Codes and the Impact When None Exist**
- **The Role of Rare Disease Patient Advocacy Organizations & Rare Disease Experts**
- **Process Considerations and Stakeholder Engagement to Focus on Next Steps**

1. The Value of Specific ICD Codes and the Impact When None Exist:

The CDC's RFI asks, *"What factors should be used to determine whether unique codes should be created for rare and ultra-rare diseases? What factors, in addition to prevalence or incidence, are relevant?"* As these factors are determined, they should account for the real-world implications of a specific code to patient care, research, and public health surveillance.

While there are disagreements about how many rare diseases are identified, depending on the source and approach used, there is agreement that advances in genomic medicine are leading to more precise identification of gene-disease relationships, resulting in a coding system that can't keep up with the need in its current form.

The significant gap in ICD-10-CM coverage of rare diseases has implications at every level of the system, including public health research, disease-level biomedical and care delivery research, the length of the diagnostic odyssey, coverage and reimbursement for care and treatments, and an emerging impact on Medicaid eligibility.

Fewer than 10% of the identified rare diseases have a corresponding ICD-10 code.[2] About 200 new diseases are identified per year by just one classification system.[3] As a result, physicians are left to choose from existing codes for other conditions. In cases where a code exists, it is often not specific enough to capture the significant variability in disease manifestations across the affected population or to account for specific within-gene mutations.

For rare disease patients and families, there are real consequences of inaccurate or nonspecific coding, including:

- Diagnostic Process: Physicians may be less likely to consider conditions without an ICD code, in part due to the limited inclusion of most rare diseases in medical education curricula and limitations in decision-support and medical records systems. Additionally, as the use of AI and natural language processing systems to support diagnostic decision-making becomes more widely adopted, without greater code specificity, there is less information available to train the systems on gene-disease relationships.
- Care Delivery Challenges: More specific codes help to recognize and manage mutation-specific impacts and secondary conditions, enabling better tracking of health outcomes and more precise identification of changes in health status that require timely intervention. Over time, as care approaches evolve and new treatments advance, the lack of specific codes makes it more challenging to ensure the right patients are notified of innovations and to track the results of interventions they receive.
- Coverage and Access to Care: At all levels of coverage decision-making, more specific codes enable more seamless access for patients. Payers assess how many individuals covered by their plans have specific diagnostic codes and use general prevalence estimates to determine overall coverage policies for new procedures and treatments. Individual coverage decisions that rest on a medical-necessity determination are guided by diagnostic codes and their associated evidence or care standards.
- Public Health Surveillance: Assigning more specific ICD codes facilitates disease surveillance, allows for more accurate estimates of their incidence, prevalence, disease course, utilization patterns, and enables rare diseases to be accurately accounted for in public health prioritization. Nearly every available analysis attempting to document the public health impacts, economic implications, and care patterns associated with rare diseases in aggregate or by disease groups

[2] McMurry, J., Matentzoglu, N., Bailey, K., Berg, J., Jason, C., Chute, C., Fish, P., Hamosh, A., Lee, Y., Madlock-Brown, C., Moffitt, R., Pandya, A., Pfaff, E., Rajkovic, A., Schaper, K., Sharathkumar, A., Sikora, T., Smedley, D., Son Rigby, C., … Powell, B. (2026). Which Rare Diseases Are Best Suited to Diagnostic AI? A Systematic Selection Framework. Zenodo. https://doi.org/10.5281/zenodo.19489582

[3] Haendel M, Vasilevsky N, Unni D, Bologa C, Harris N, Rehm H, Hamosh A, Baynam G, Groza T, McMurry J, Dawkins H, Rath A, Thaxton C, Bocci G, Joachimiak MP, Köhler S, Robinson PN, Mungall C, Oprea TI. How many rare diseases are there? Nat Rev Drug Discov. 2020 Feb;19(2):77-78. doi: 10.1038/d41573-019-00180-y. PMID: 32020066; PMCID: PMC7771654.

contains significant limitations, noting the results are likely vastly underestimated due to the lack of codes.

- Clinical Research and Therapy Development: Codes play an important role in the therapy development process, from early-stage decisions about what to move forward with, to the ability to assess how the product works in the post-approval phase. Advances in precision medicine and technology platforms mean that more decisions about which products to advance into clinical-phase research are made at the specific mutation level. These decisions are guided in part by what is known about the prevalence and incidence of a disease. When that information is unclear or unavailable, promising science may never reach patients. More specific codes also enable the appropriate patients to be matched to clinical trial opportunities and more retrospective analysis to be conducted. Lastly, the limited number of codes prevents the full benefits of real-world data advancements from reaching patients with rare diseases, even as products for ultra-rare diseases are the very ones that most need to rely on post-market confirmatory sources of evidence for safety and efficacy.
- Emerging Implication – Medicaid Community Engagement Requirement Compliance: Starting in 2027, most states are required to implement community engagement requirements for the adult Medicaid expansion population. While the law includes broadly defined exemptions for medically frail individuals or complex medical situations, states are expected to have leeway in determining which conditions qualify. The first state to implement these requirements, Nebraska, has elected to index the list of eligible conditions to diagnosis codes[4], and more states are likely to adopt this approach. Limited coverage under ICD-10-CM could leave rare disease patients who are unable to comply with the community engagement requirement without healthcare coverage or, at best, require them to navigate burdensome, ill-defined bureaucratic processes.

There is a growing list of examples documenting the benefits of obtaining specific ICD-10 codes, including:

- The COMBINEDBrain submission notes just 3 of the examples documenting how new codes have enabled epidemiologic tracking immediately after they were issued, all 3 of which now have active clinical development pipelines.
- Dravet Syndrome, a severe form of epilepsy that also causes a range of developmental disabilities, obtained a specific code and associated sub-categories in 2020. The existence of the codes has led to the inclusion of Dravet Syndrome in an effort to map real-world knowledge of the disease to inform a large language model and methods to identify and connect patients to care[5]. That is especially relevant for Dravet syndrome because of the rapid expansion of targeted therapies, including antisense oligonucleotides and gene therapy programs.

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[4] https://dhhs.ne.gov/Documents/Nebraska%20Medicaid%20Work%20Requirements%20-%20Medically%20Frail%20and%20SUD%20Conditions.pdf

[5] E. Kim, M. Shrestha, R. Foty, T. DeLay and V. Seyfert-Margolis, "Structured Extraction of Real World Medical Knowledge using LLMs for Summarization and Search," *2024 IEEE International Conference on Big Data (BigData)*, Washington, DC, USA, 2024, pp. 3421-3430, doi: 10.1109/BigData62323.2024.10825160.

- An analysis conducted two years after a new code was issued for Duchenne and Becker muscular dystrophy in 2018 found that the code is widely used and cited in coverage policies for drug therapies, genetic testing, and nerve conduction testing. Clinicians reported that the unique code is helpful in identifying and tracking patients for purposes of delivering clinical care, recruiting patients into studies, and measuring health outcomes that enable quality improvement. The code reduced administrative burdens by making billing easier and helped develop a more complete understanding of the patient journey, identified physicians and care centers with newly diagnosed patients, and refined patient censuses by state and age.

2. The Role of Rare Disease Patient Advocacy Organizations & Rare Disease Experts:

While developing the ICD-10 Code Roadmap and in the engagement we conducted to inform this response, numerous patient advocacy organizations at various stages in the request process provided insights. Whether these groups were in the early stages of understanding what evidence and information is required, in the process of assembling a submission, or had previously engaged with the ICD-CM committee, the two common threads were (1) overall confusion about the committee's adjudication process, and (2) the immense time and resource constraints placed on organizations leading evidence collection and nomination processes.

The current system for requesting and evaluating new codes requires significant time and resources. Often, patient advocacy organizations lead efforts to navigate the evidence development and collection process, assemble experts, identify resources, evaluate evidence, and make recommendations in their submissions. These organizations are often small, sometimes volunteer-run, and, in many cases, formed to support patients and parents and/or to ensure that scientific and therapeutic discovery advances.

Patient organizations would welcome a new process that provides greater clarity and predictability and streamlines the overall processes.

However, a new process must include a meaningful role for patient advocacy organizations and condition-specific experts. In most cases, patient organizations should be consulted in the evaluation of new codes, drawing on their expertise in the condition, clinical and research experience, and relationships with scientific and clinical experts as needed. We were pleased to see the MONDO proposal clearly commit to retaining a role for patient organizations. We encourage the CDC to continue engaging directly with rare disease organizations as you evaluate and advance new organizing principles.

3. Process Considerations and Stakeholder Engagement to Focus on Next Steps

The decision to pause consideration of new code requests during CDC's evaluation, while understandable, creates a substantial gap at a time of rapid growth in genomic diagnostics and precision medicine. For communities affected by rare genetic diseases, an additional year without specific coding may further limit the visibility of these conditions within healthcare systems and national data sources, reduce access to diagnostics and clinical interventions, and negatively impact the development of new medical products.

We urge CDC to move swiftly to advance the consideration of the MONDO and NGRF proposals while committing to active and sustained participation of patient organizations, expert clinicians, researchers in rare disease natural history, surveillance, and epidemiology, and experts from other federal agencies, in addition to the planned public feedback meetings. This engagement will be essential to ensure that

the organizing principles and process resulting from the current evaluation process are patient-centered and aligned with stakeholder priorities.

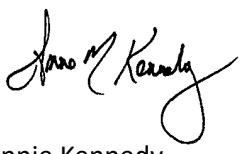
The EveryLife Foundation urges prioritizing further stakeholder engagement to drive consensus around:

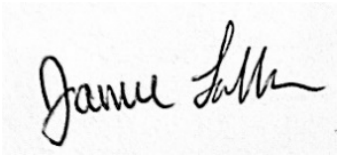
- **The benefits and feasibility of pre-assigning codes to identified rare diseases, submitting code request evaluations in batches, and the ultimate role for the ICD-10-CM Coordinating Committee in either approach.**
- **An approach to align the existing codes for ultra-rare and genetic diseases to be consistent with the new process or procedures that will make the transition smoother for the coding and research communities.**
- **The qualifying criteria for a rare disease to be considered for its own code, including consideration of the framework-based approach described in the NGRF proposal.**
- **The best approaches to multi-system and syndromic disease classification.**
- **How to account for increasing knowledge about genetic susceptibility and disease manifestation in a way that enhances timely diagnosis, enables scientific discovery, and protects patients from possible genetic discrimination.**
- **How to ensure the sustainability and stability of the new processes, including internal agency operations, knowledge management systems to buffer institutional knowledge losses that occur in staff transitions, and procurement of stable funding for any non-governmental resource included in the evaluation process.**

Separately, we urge the resumption of discussions to advance the adoption of ICD-11, which already includes additional rare disease codes. We recognize the current challenges inherent in this transition and suggest that, at a minimum, efforts be made to ensure that ICD-10 batch submissions for already identified rare diseases proceed in a manner that enables parallel adoption into ICD-11.

Thank you for your continued work maintaining and improving the ICD-10-CM classification system. We appreciate the opportunity to provide input and would welcome the chance to contribute further to discussions on how best to represent genetic diseases to support patient care, research, and public health. We would also welcome the opportunity to discuss this issue directly.

Sincerely,





Annie Kennedy
Chief Mission Officer
EveryLife Foundation for Rare Diseases

Jamie Sullivan, MPH
Senior Vice President of Policy and Advocacy
EveryLife Foundation for Rare Diseases

CC: Michael Pearlmutter, Chief Executive Officer
Amy Gaviglio, Chairman, Board of Directors