



January 12, 2024

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**ASPE, Science and Data Policy**  
**Department of Health and Human Services**  
**Science and Data Policy**  
**Humphrey Building, Room 442E.2**  
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**Washington, DC 20024**

**Federal Registrar, Volume 88, Number 198: Response from the National Organization for Rare Disorders Regarding ICD-11 Request for Information**

Dear Dr. Arnold,

On behalf of the more than 30 million Americans living with a rare disease, the National Organization for Rare Disorders (NORD) thanks the National Committee on Vital and Health Statistics (NCVHS) for the issuing the Request for Information (RFI) regarding ICD-11 codes. Meaningful progress in research and care for our community depends on rigorous research, made possible by robust, granular and fit-for-purpose data. Timely and efficient implementation of ICD-11 codes will play a key role in unlocking the potential of real-world data (RWD) and real-world evidence (RWE) for rare diseases, many of which continue to be inadequately captured by current coding practice.

NORD is a unique federation of non-profits and health organizations dedicated to improving the health and well-being of people living with rare diseases by driving advances in care, research, and policy. NORD was founded 40 years ago, after the passage of the Orphan Drug Act (ODA), to formalize the coalition of patient advocacy groups that were instrumental in passing that landmark law. Since that time, NORD has been advancing rare disease research and funding to support the development of effective treatments and cures; raising awareness and addressing key knowledge gaps; and advocating for policies that support the availability of and access to safe and effective therapies.

For the rare disease community, more accurate representation of disease states through timely and efficient implementation of ICD-11 codes will be crucial. Appropriately specific ICD codes are pivotal in allowing researchers and physicians to track how many individuals are impacted by a specific disease, which is vital for rare diseases which often lack validated disease prevalence or incidence estimates; it also is also indispensable for gaining a better understanding of natural history and disease progression, including morbidity and mortality estimates, as well as

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streamlining coverage and reimbursement issues.<sup>1</sup> Given that over 95% of the more than 7,000 known rare diseases do not have an FDA approved treatment, leveraging real-world evidence effectively, including through appropriate coding practices, is vitally important.<sup>2,3</sup> Appropriate implementation of the new ICD-11 codes is critical to unlocking its full potential, in particular for rare diseases, given the large number of diseases and pervasive issues with rare disease diagnostic, research, and care. NORD thanks the NCVHS for drafting this RFI, including its emphasis on leveraging the codes to help collect better data on historically underrepresented populations to ensure real-world evidence can be fully leveraged to bring more effective rare disease therapies to market.

NORD is pleased to offer the specific recommendations below for how to maximize the impact of this draft guidance, informed by our 40 years of experience working constructively with all key stakeholders to help bring new and better rare disease therapies to more patients more quickly.

**Recommendation 1: Ensure the implementation of ICD-11 codes for rare diseases is timely, smooth, and allows for robust, high-fidelity datasets that are fit for purpose and can support meaningful improvements in rare disease research and care**

To date, only 7% of rare diseases have disease-specific ICD-10 codes, and without disease-specific codes, physicians are often left to use codes that only describe (some of) the symptoms a person is experiencing,<sup>4</sup> making it difficult to identify rare disease patients for clinical trials and leverage RWD and RWE to ultimately improve patient care.<sup>5</sup> Implementation of ICD-11 will substantially increase the number of disease specific codes to 24% of all rare diseases. Importantly, another 71% of rare diseases will fall under broader, not disease-specific codes in ICD-11; although these diseases will experience remaining uncertainty in coding, it will represent a meaningful improvement over current coding available in ICD-10.<sup>6</sup>

Considering that more than 5,500 rare diseases will be represented at some level in ICD-11, a drastic increase from 500 through ICD-10, education and technical support around the new rare

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<sup>1</sup> Feinstein JA, Gill PJ, Anderson BR. Preparing for the International Classification of Diseases, 11th Revision (ICD-11) in the US Health Care System. *JAMA Health Forum*. 2023 Jul 7;4(7):e232253. doi: 10.1001/jamahealthforum.2023.2253. PMID: 37505488; PMCID: PMC10495107.

<sup>2</sup> Office of the Commissioner. (n.d.). *Rare diseases at FDA*. U.S. Food and Drug Administration. <https://www.fda.gov/patients/rare-diseases-fda>

<sup>3</sup> World Health Organization. (n.d.). *International Classification of Diseases (ICD)*. World Health Organization. <https://www.who.int/standards/classifications/classification-of-diseases>

<sup>4</sup> Luxner, L. (2019, February 5). *ICD-10 codes, "really important" to rare disease patients, soon up...* Pompe Disease News. <https://pompediseasenews.com/news/icd-10-codes-really-important-to-rare-disease-patients-soon-up-for-fresh-consideration/>

<sup>5</sup> June 2023 - World Health Organization. World Health Organization. (2023, June). <https://cdn.who.int/media/docs/defaultsource/classification/icd/icd-10/icd-10-to-meddra-mapping-conventions.pdf?sfvrs>

<sup>6</sup> A. Rath, personal communication, January 10, 2024

disease ICD-11 codes for providers, hospitals, and the broader healthcare ecosystem will be vital to success. This should include raising awareness about the new ICD codes that are available for rare diseases patients and about how their use will help better meet the needs of our community.<sup>7</sup> NORD's Rare Disease Center of Excellence (COE) Program, as well as organizations that represent the rare disease community, including NORD and its member organizations, can play a key role in these efforts.<sup>8,9</sup>

As a part of its mission, NORD's Rare Disease COE Program is committed to sharing knowledge and best practices to improve rare disease care and advance rare disease research, while solving the greatest medical challenges and unmet needs of the rare disease patient community. These centers are a unique network of 40 academic medical centers, children's hospitals, clinics, and institutions with the shared goal of advancing care and expanding access for rare disease patients. Through collaboration and knowledge sharing, the network aims to create a scalable model of treatment and research for all rare diseases that would otherwise be unattainable, providing a much-needed national infrastructure to help accelerate advancements in rare disease diagnosis, treatment, and research. Given NORD's Rare Disease COEs have extensive ties to the rare disease community and to providers, researchers, and other experts in the fields, and are tracking the implementation of ICD-11 closely, NORD would like to work together with the Centers for Disease Control and Prevention (CDC) and the key stakeholders to ensure appropriate coding is available to all medical entities and researchers to uplift rare disease research and bring more effective therapies to market.

**Recommendation 2: Start engaging the rare disease community now as you look ahead to ICD-12 to further improve coding for rare diseases, and to address any challenges with the ICD-11 implementation that may need more fundamental corrections.**

Critical stakeholders in the rare disease community, including NORD and its 330+ member organizations, and medical experts in the rare disease field can also play a key role in the planning for ICD-11. Recognizing the importance of accurate coding for the rare disease community, NORD has already committed to working with Orphanet, our Rare Disease COEs, and other patient organizations to help bridge existing gaps in rare disease coding, and to collect evidence to help prioritize which rare diseases would benefit most from disease-specific codes in future iterations. The experiences from the NORD's Rare Disease COE program to date clearly emphasize the importance of community ties and multi-sector partnerships to achieve these goals. NORD is looking forward to working with CDC, the Office of the National Coordinator, and other key stakeholders to further improve coding for rare disease patients as part of ICD-12.

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<sup>7</sup> The EveryLife Foundation. (n.d.). *ICD Code Roadmap Resource Guide*. <https://everylifefoundation.org/wp-content/uploads/2021/06/ICD-Code-Roadmap-Guide-FINAL.pdf>

<sup>8</sup> Feinstein JA, Gill PJ, Anderson BR. Preparing for the International Classification of Diseases, 11th Revision (ICD-11) in the US Health Care System. *JAMA Health Forum*. 2023 Jul 7;4(7):e232253. doi: 10.1001/jamahealthforum.2023.2253. PMID: 37505488; PMCID: PMC10495107.

<sup>9</sup> Bearryman, E. (2015, January). Does your rare disease have a code?. EURORDIS. <https://www.eurordis.org/does-your-rare-disease-have-a-code/#:~:text=Nearly%20500%20rare%20diseases%20have,available%20in%20over%2050%20countries.>

Without implementing ICD-11 codes for rare diseases, providers will only be able to report codes that describe (some of the key) symptoms but not the rare disease, thus making it impossible for researchers to access symptoms and manifestations under the specific diagnosis.<sup>10</sup> By only reporting symptoms of a disease without more specific ICD code, researchers have difficulty tracking health care quality and outcomes for rare disease patients, costs of certain rare diseases, as well as struggle to find patients to access clinical trials.<sup>11</sup>

### **Recommendation 3: Learn from past experiences including the implementation of ICD-9 and ICD-10 codes as well as experience with ICD-11 implementation internationally**

To ensure the transition from ICD-10 to ICD-11 is timely, smooth and not unnecessarily burdensome for healthcare providers and the broader healthcare ecosystem, lessons learned from the ICD-9 to ICD-10 transition should be applied where possible. For example, in a study conducted on emergency departments in Illinois on the transition from ICD-9 to ICD-10, researchers found that 27% of a subset of ICD-9-CM codes billed to Medicaid were convoluted, while 8% of these codes were found to be incorrect.<sup>12</sup> Errors such as those observed during the ICD-9 to ICD-10 transitions can have significant impacts on the lives of rare disease patients, not only by creating negative clinical research implications, but also implications on the reimbursement and coverage of rare diseases, which have severe financial impacts on patients and families.

Similarly, experience with the implementation of Z-codes, which are designed to capture social determinants of health (SDOH) and can give vital insight into the social, environmental, and economic barriers patients experience, show the challenges associated with the implementation of novel codes, including the impact of limited incentives for use, and suggest the need for training physicians on the appropriate use of these codes.<sup>13</sup> The lack of incentives and coding for SDOH bares similarities with the lack of disease-specific coding rare disease patients, and provides useful lessons learned. Given the greater complexity of rare diseases, it is sensible to expect an even longer learning curve and larger disruptive impacts during the transition period, in particular for those providers that do not regularly care for rare disease patients.

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<sup>10</sup> Luxner, L. (2019, February 5). ICD-10 codes, “really important” to rare disease patients, soon up... Pompe Disease News. <https://pompediseasenews.com/news/icd-10-codes-really-important-to-rare-disease-patients-soon-up-for-freshconsideration/#:~:text=%E2%80%9CFor%20one%2C%20it%20help>

<sup>11</sup> The EveryLife Foundation. (n.d.). *ICD Code Roadmap Resource Guide*. <https://everylifefoundation.org/wp-content/uploads/2021/06/ICD-Code-Roadmap-Guide-FINAL.pdf>

<sup>12</sup> Krive J, Patel M, Gehm L, Mackey M, Kulstad E, Li JJ, Lussier YA, Boyd AD. The complexity and challenges of the International Classification of Diseases, Ninth Revision, Clinical Modification to International Classification of Diseases, 10th Revision, Clinical Modification transition in EDs. *Am J Emerg Med*. 2015 May;33(5):713-8. doi: 10.1016/j.ajem.2015.03.001. Epub 2015 Mar 7. PMID: 25863652; PMCID: PMC4430372.

<sup>13</sup> *Utilization of Z codes for Social Determinants of health among Medicare ...* Centers for Medicare and Medicaid Services: Office of Minority Health. (2021b, September). <https://www.cms.gov/files/document/z-codes-data-highlight.pdf>

To date, more than 60 countries have transitioned to using ICD-11 codes<sup>14</sup> and learnings from such international implementations can also be useful. For instance, a study from Canada on developing effective training materials for ICD-11 by measuring coder performance suggested the value of standardized training in this subject.<sup>15</sup> Effective training included a user guide, line codes, healthcare-related harms, as well as medical-surgical cases followed by a quiz for coders. This allowed researchers to see which areas needed more clarity and training, as well as create feedback on which codes are missing or need to be more thorough.<sup>16</sup> Since ICD-11 has major updates from ICD-10, proper guidance and training is needed to ensure all end-users of ICD-11 are engaged on how to use transition tools and other technologies associated with the update. The CDC should engage with users on how ICD-11 will impact their current systems and develop mechanisms for transition that meets the needs of the healthcare system.

Ensuring appropriate tracking and accountability, in particular for the implementation of rare disease codes, will be equally important to assess in near-real time how the transition from ICD-10 to ICD-11 is going, whether course-corrections will be needed, and to ensure appropriate data quality and fidelity during the transition. This will ensure that CDC vital statistics and surveillance systems will remain intact during the transition.

NORD again thanks NCVHS for the opportunity to provide comments on this important RFI, and we look forward to continuing the dialogue around ICD-11. For questions regarding NORD or the above comments, please contact Hayley Mason, Policy Analyst, at [hmason@rarediseases.org](mailto:hmason@rarediseases.org)



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<sup>14</sup> Feinstein JA, Gill PJ, Anderson BR. Preparing for the International Classification of Diseases, 11th Revision (ICD-11) in the US Health Care System. *JAMA Health Forum*. 2023 Jul 7;4(7):e232253. doi: 10.1001/jamahealthforum.2023.2253. PMID: 37505488; PMCID: PMC10495107.

<sup>15</sup> Eastwood CA, Southern DA, Doktorchik C, et al. Training and experience of coding with the World Health Organization's International Classification of Diseases, Eleventh Revision. *Health Information Management Journal*. 2023;52(2):92-100. doi:[10.1177/18333583211038633](https://doi.org/10.1177/18333583211038633)

<sup>16</sup> Eastwood CA, Southern DA, Doktorchik C, et al. Training and experience of coding with the World Health Organization's International Classification of Diseases, Eleventh Revision. *Health Information Management Journal*. 2023;52(2):92-100. doi:[10.1177/18333583211038633](https://doi.org/10.1177/18333583211038633)