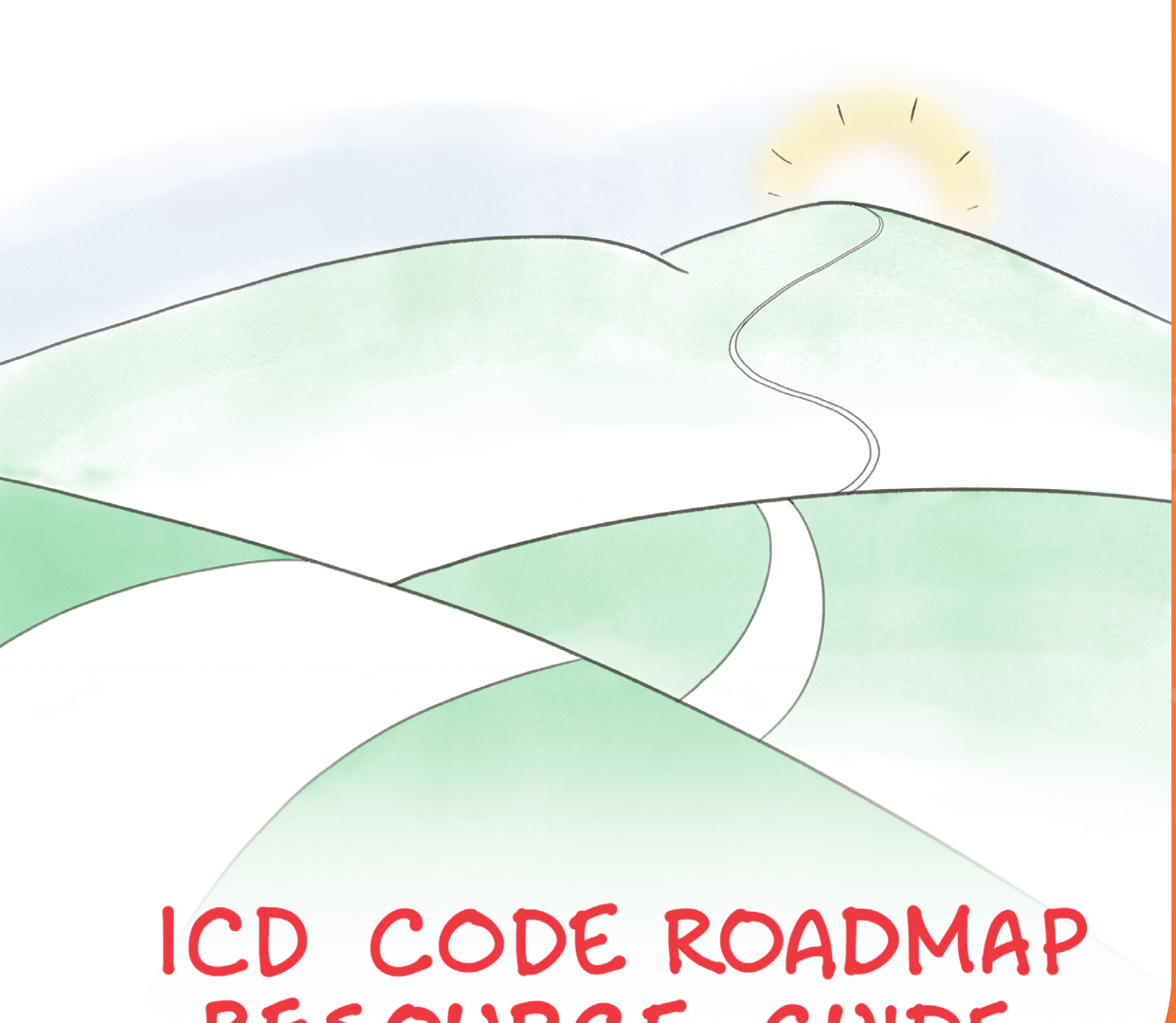

ICD CODE ROADMAP RESOURCE GUIDE

EVERYLIFE FOUNDATION FOR RARE DISEASES

INTRODUCTION

A first-of-its-kind resource for the rare disease community – and beyond

As I reflect on all the advocacy experiences I've had, navigating the nomination of new ICD codes was among the most complicated journeys I've traversed. If not for the friends and colleagues who had traveled the path before me and who so generously shared their insights and expertise, I'm not sure how we would have managed.

It's not that developing the evidence to support an ICD code nomination was necessarily more difficult than other systems patient advocacy groups are working within, it's that it is an ecosystem that currently has few points of access or navigational resources. There are no formal guidances, nor toolkits for nominating groups to follow. Communities often only learn of the importance and utility of ICD codes through hindsight, and the process of nominating a code can be lengthy and resource-intense.

It seems that every week, someone reaches out seeking information and support for their community's navigation of this process. And every conversation includes the question, *"Is there a roadmap or guide that explains ICD codes, the process and where we should begin?"* The answer was always, *"Not that I'm aware of."* Until now…

Over the last 18 months, we worked with dozens of rare disease patient advocacy community partners who've blazed this trail and nominated codes for their communities. We worked with partners from federal agencies, representative biopharmaceutical industry organizations and payer groups to include ICD code considerations for rare disease through the lenses of the many stakeholders who utilize these codes. And importantly, many of our partners graciously shared their experiences in the form of case studies and templates – so that we could all learn from one another and be strengthened by the experiences of those who have gone before us.

Our hope is that this **ICD Code Roadmap** serves as a navigational resource for you and your community. This guide is not intended to replace the rich exchange of direct mentorship and collaboration among partners, but rather to provide you with a firm foundation upon which to base these engagements.

Thank you to all those who helped to make the ICD Code Roadmap possible. Demonstrating – once again – that our rare disease community is stronger together.

Annie Kennedy

Annie Kennedy
Chief of Policy and Advocacy
EveryLife Foundation for Rare Diseases

EVERYLIFE FOUNDATION FOR RARE DISEASES

About the EveryLife Foundation

The EveryLife Foundation for Rare Diseases is a 501(c)(3) nonprofit, nonpartisan organization dedicated to empowering the rare disease patient community to advocate for impactful, science-driven legislation and policy that advances the equitable development of and access to lifesaving diagnoses, treatments and cures. We do not speak for patients. We provide the training, education, resources and opportunities to make their voices heard. By activating the patient advocate, we can change public policy and save lives.

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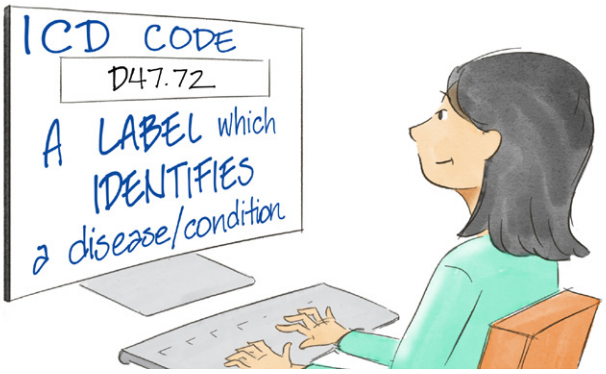
For all who seek to better understand how diagnosis codes are assigned, updated and revised in the United States' health information system, this guide is for you.

It starts with basic information about the coding system and then describes the process by which patient advocacy organizations and others can propose changes to existing codes or introduce new codes, including the necessary components of a proposal.

Case examples and links to additional resources provide the information you will need to determine whether and how to get involved in the continuous process of refining the diagnostic coding system used in the United States.

SECTION 1: THE BASICS

This introductory section will familiarize you with the global system for coding diagnoses, how the U.S. system fits within it and how it is governed. It also provides the building blocks of a specific coding structure and the general process by which the coding system is refined in the U.S. in between major overhauls of the international system.



Overview of the ICD Coding System

The "International Statistical Classification of Diseases and Related Health Problems" (ICD) is a standard classification system used around the world throughout the health care system and by government agencies and researchers to track and report diseases, disorders, injuries and other health conditions. It is maintained by the World Health Organization (WHO) and has roots dating back to 1893.

According to the WHO, "the ICD is important because it provides a common language for reporting and monitoring diseases." ICD codes are used to compile public health statistics, including data about the prevalence and incidence of diseases and medical conditions.

ICD codes are used to compile public health statistics, including data about the prevalence and incidence of diseases and medical conditions.

ICD codes facilitate:

- Easy storage, retrieval and analysis of health information for evidenced-based decision-making
- Sharing and comparing health information and disease surveillance between medical specialties, hospitals, health care systems, regions, settings and countries
- Data comparisons in the same location across different time periods
- Measurement of health care quality, safety and efficiency, including costs and outcomes

Consider the power of understanding which types of medical specialties are diagnosing a particular rare disease, or having the ability to track changes in patients' health outcomes after a new therapy is approved for a specific disease. A payer's medical policy decisions for a new therapy might be based on an assessment of how many individuals covered by their plans have a particular diagnostic code in their record. These are the types of queries made possible through ICD codes. They are also the types of assessments that can be distorted by imprecise or out-of-date codes.

While the intention is to have a standard set of codes in use around the world, in actuality each country governs use of the coding system and can modify the WHO's codes to suit its purposes. The ICD coding system has gone through 11 major revisions to reflect advances in health, medical science and data capture and tracking over time. WHO will make the newest version, ICD-11, available on January 1, 2022. Migration to a new ICD revision occurs at different rates across the world, and it can take several years to fully implement. There is no mandatory deadline for individual countries to adopt a new version; the previous version (ICD-10) will remain in effect in each country until the new one is adopted. The exact timeline for ICD-11 adoption in the U.S. in currently unknown but is likely to be several years after WHO implementation.

Maintenance of ICD Codes in the United States

In the U.S., codes are presently based on the ICD-10 set of codes issued by WHO. ICD-10 went into effect in the U.S. on October 1, 2015, 23 years after it was approved by WHO in 1992. At the time of the transition, ICD-9-CM included 13,000 codes and ICD-10-CM started with 69,000 codes. This demonstrates the laborious process required to adapt information systems and coding practices to implement such a major code revision. There has not yet been an estimated date for U.S. adoption of ICD-11, but it is not anticipated to go into effect in the U.S. for several more years.

While the change from ICD-9 to ICD-10 introduced more codes and generally more specific codes, some diseases lost specific ICD codes in the transition. See page 25 for the case study on Friedreich's ataxia and how this was rectified through action led by the **Friedreich's Ataxia Research Alliance**.

Today there are two distinct types of ICD codes in use in the U.S.:

ICD-10-CM The CM stands for "Clinical Modification" and it includes codes used to classify **diagnoses and reasons for clinical visits** in all health care settings, including hospital stays. These codes are maintained by the National Center for Health Statistics (NCHS), a division of the U.S. Centers for Disease Control and Prevention (CDC). NCHS serves as the WHO Collaborating Center for the Family of International Classifications for North America. It is responsible for coordinating all official disease classification activities in the U.S. relating to the ICD and its use, interpretation and periodic revision.

ICD-10-PCS The PCS stands for "Procedural Classification System." It includes codes classifying **procedures** performed in hospital inpatient health care settings. ICD-10-PCS codes are maintained by the Centers for Medicare and Medicaid (CMS).

This resource guide focuses on the process for assigning ICD-10-CM codes used in the U.S. to classify diagnoses and reasons for visits in all health care settings. It does not address ICD-10-PCS codes or codes for countries outside of the U.S.

ICD-10-CM and ICD-10-PCS codes are updated once each year, on the first day of the federal government's fiscal year, October 1. Updates to both sets of codes are managed by the ICD-10 Coordination and Maintenance (ICD C&M) Committee, a federal interdepartmental committee. NCHS is responsible for managing the diagnosis codes and CMS is responsible for procedure code management. The process for engaging in updates to diagnosis codes is described later in this guide (see page 9). For information about the process for proposing updates to procedure codes, please refer to the CMS website.

With each annual update of ICD-10-CM diagnosis codes, the government issues "Guidelines for Coding and Reporting Using the ICD-10-CM." These guidelines are considered to be a companion to the official version of the listing of ICD-10-CM codes as published on the NCHS website. Guidelines are approved by four organizations that make up the Cooperating Parties for the ICD-10-CM: the American Hospital Association (AHA), the American Health Information Management Association (AHIMA), CMS and NCHS. These are considered to be **the** authoritative entities with respect to ICD-10-CM codes.

The **National Center for Health Statistics** (NCHS) is responsible for coordinating all official disease classification activities in the U.S., including maintaining the ICD-10-CM codes for diagnoses. NCHS is part of the U.S. Centers for Disease Control and Prevention (CDC).

The Guidelines describe the purpose and importance of use of ICD-10-CM codes by health care professionals and professionals who assist with coding medical records (coders) as follows:

> "The diagnosis codes have been adopted under HIPAA for all healthcare settings. A joint effort between the healthcare provider and the coder is essential to achieve complete and accurate documentation, code assignment and reporting of diagnoses and procedures. These guidelines have been developed to assist both the healthcare provider and the coder in identifying those diagnoses that are to be reported. The importance of consistent, complete documentation in the medical record cannot be overemphasized. Without such documentation accurate coding cannot be achieved. The entire record should be reviewed to determine the specific reason for the encounter and the conditions treated."

Anatomy of an ICD-10-CM Code

The ICD-10-CM is highly structured and specific codes must conform to tightly managed classification conventions. The diagnosis coding system itself is divided into two parts:

- **Alphabetic Index**: An alphabetical list of terms and their corresponding code
- **Tabular List**: A structured list of codes divided into chapters based on a body system or condition that contain categories, subcategories and codes that use a combination of letters and numerals to create unique identifiers. The ICD-10-CM uses an indented format for ease of reference.
    - **Categories**: Three characters – a letter followed by two numerals. If there is no further subdivision of a category, it is equivalent to a code.
    - **Subcategories**: The category (a letter plus two numerals), followed by a decimal point and then a subcategory (letter(s) or numeral(s)) for a total of either 4 or 5 characters.
    - **Codes**: May be a total of 3, 4, 5, 6 or 7 characters. The 6th and 7th characters are generally used to provide additional context, for instance with regard to timing or treatment status (controlled or uncontrolled). For example, a five-digit code for a condition that occurs during pregnancy might include a 6th character to indicate whether it occurs during the first, second or third trimester.

This is an example of the coding structure for Castleman disease within ICD-10-CM:

CHAPTER	Chapter 2: Neoplasms (COO-D49)
CATEGORY	D47 Other neoplasms of uncertain behavior of lymphoid, hematopoietic and related tissue
SUBCATEGORY	D47.Z Other specified neoplasms of under uncertain behavior of lymphoid, hematopoietic and related tissue
CODE	D47.Z1 Post-transplant lymphoproliferative disorder (PTLD)
CODE	**D47.Z2 Castleman disease**
CODE	D47.Z9 Other specified neoplasms of under uncertain behavior lymphoid, hematopoietic and related tissue

You can use the CDC's ICD-10-CM browser tool to look up the coding structure for any disease of interest. If the search yields a response, "No results found," or a result where the disease is misclassifed based on current consensus of scientific and/or clinical understanding, then keep reading this guide. **Because our understanding and classification of human health and disease is constantly evolving, the ICD coding system requires constant tending, especially when it comes to rare diseases.**

Dr. David Fajgenbaum almost lost his life to a mystery illness before doctors diagnosed it as Castleman disease. Once it had a name, he worked to get it an ICD-10-CM code. **Learn more on page 22.**

Overview of the Process for Proposing Changes to ICD-10-CM Codes

This section provides an overview of the mechanics and the usual timeline that proposed changes to ICD-10-CM codes follow. Section 2 (see page 9) provides guidance for developing the content of a coding proposal.

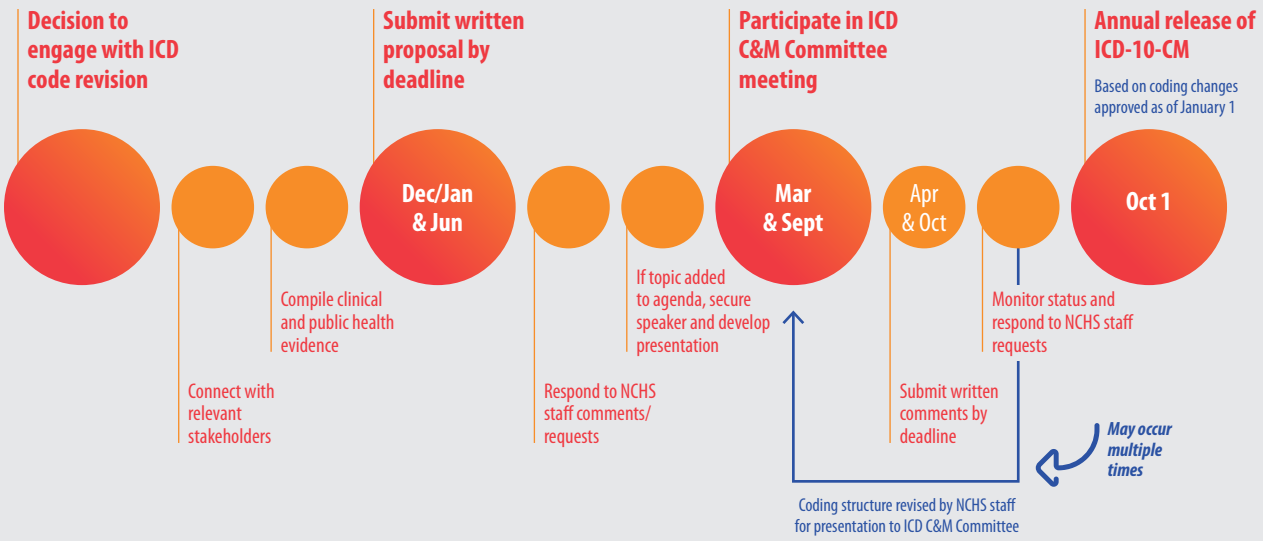
First, it is important to acknowledge that given the importance of ICD codes, the process for updating codes – by design – is evidence-based, deliberative and thorough. Those who oversee the coding system must balance the needs of individuals and organizations seeking to revise codes with the integrity of the coding system as a whole. The rationale for presenting a particular proposal (or not presenting it) and adopting a revised code (or not) may not always be clearly communicated. It's wise to approach the process with this understanding, as well as an expectation that engaging in the process will require both patience and persistence. Under optimal circumstances, a code can be approved within six months, but this is unusual. Plan for a 13-18-month process at least, from date of the initial submission to the effective date of a new code and prepare for the possibility that many proposals take even longer. The ICD Code Roadmap Readiness Assessment Tool may be useful in helping gauge whether the right conditions are in place to dive into the process.

> Engaging in the process of refining the ICD coding system requires patience and persistence. The usual timeline from initial submission to effective date of a new code is 13-18 months but can be longer.

Updates to the ICD-10-CM coding system may originate with NCHS staff or within other federal agencies or from external organizations. All updates, whether they involve amending an existing code or creating a new code, generally follow these steps, which are described in more detail in Section 2:

- **Compile evidence** to support creation of a new or revised ICD-10-CM code. (See page 11 for more information about the type of evidence that may support a nomination.)
- **Develop a formal proposal**, including a proposed coding structure, in collaboration with other interested stakeholders. (See page 9 for details about types of stakeholders you may wish to engage.)
- **Submit the proposal** to the ICD Coordination and Maintenance Committee (ICD C&M Committee) via email in time to meet one of the two deadlines set each year (December/January for the March meeting and June for the September meeting); the deadline by which the proposal is submitted determines – but does not guarantee – the first possible meeting at which the proposal could be presented.

TYPICAL TIMELINE: 13–18 MONTHS

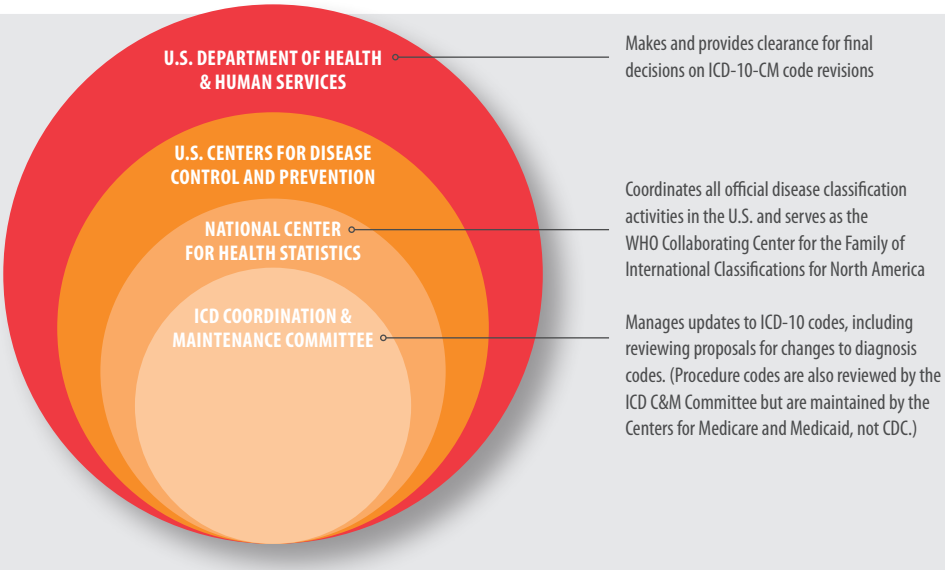


- **Await contact from NCHS staff**. They will notify the contact person for a submission if the topic will be included on an upcoming ICD C&M Committee agenda. Some interim clarification or revision may be requested by NCHS staff prior to the meeting. In the Committee's initial consideration of a particular disease, it is commonplace for the submitter to identify an authoritative speaker (generally someone with clinical expertise) to provide a brief oral overview of that disease (including any supporting data used to construct the coding proposal) and to participate in the meeting. ICD C&M Committee meetings are open to the public but do require pre-registration.
    - If the proposal is determined to be incomplete or inadequate for presentation to the Committee, the submitter will be notified. Depending on the nature of the deficits, it may be worthwhile to revise the proposal and resubmit in advance of a future deadline.
- At the ICD C&M Committee meeting, **NCHS staff will present a proposed coding structure** which may differ from the coding structure outlined in the initial proposal from the external organization(s). Discussion periods following each presentation are brief but may allow for questions and comments from both the ICD C&M Committee and those in attendance. Coding professionals and representatives of medical specialty societies regularly attend and comment on proposals. It is very important to understand that no decisions will be made during ICD C&M Committee meetings, nor are you likely to receive any feedback that indicates whether the proposal will be approved.
- Following each ICD C&M Committee meeting is a comment period. **Consider responding in writing** to the NCHS staff's proposed coding structure and questions or comments that arise during the meeting.
- Between meetings, it is wise to proactively **attempt to stay in contact with NCHS staff** about the status of the proposal, including future ICD C&M Committee meetings at which revised coding structures for the disease may be discussed. There may be opportunities to submit additional evidence or letters of support. In some cases, NCHS staff may consult authoritative experts about the coding structure. The volume of inquiries fielded by the small NCHS staff may result in there being less interim communication than you might prefer.
- **Watch for final decisions on code revisions**. These are made through a clearance process within the Department of Health and Human Services.

Coding changes approved through this process become effective as of the next ICD-10-CM update issued each year on October 1. NCHS staff may or may not notify the submitter in advance of publication that a code has been approved.

Iterations of a proposal initially submitted by the Dravet Syndrome Foundation were reviewed at three separate meetings of the ICD C&M Committee. The third iteration was vetted by NCHS staff with the American Academy of Neurology before it was presented. See page 23 for the Dravet syndrome case study.

Federal maintenance of ICD-10-CM codes



SECTION 2: THE APPROACH

This section provides guidance for assembling a formal written proposal to the NCHS for consideration by the ICD C&M Committee to create a new code or revise an existing code. The process of developing a compelling and well-supported proposal can take several months; be mindful of the twice-annual deadlines for new submissions (December/January and June) as motivation to keep a proposal initiative moving forward.

Organizations across the limb girdle muscular dystrophy community came together to develop a proposal for a set of new ICD codes, with the Muscular Dystrophy Association playing a central organizing role. Read more about their "big tent" approach to this initiative on page 27.

Collaboration

Like most worthwhile initiatives conducted on behalf of individuals with rare diseases, approaching a change to the ICD-10-CM system is best accomplished through a coalition (formal or informal) of relevant patient advocacy organizations, clinical experts, researchers (including those at academic institutions and government agencies) and life science companies developing and/or marketing medical products to diagnose and/or treat the disease. The different resources, skills and perspectives that each entity brings to this process will enhance likelihood of success, certainly in comparison to one entity going it alone or fragmenting into groups with different (competing) proposals for the same disease. It is vital to gather as much support as possible in the process of developing the proposal and to keep the alliance together so any follow-up responses at meetings or through written comments are as cohesive as possible.

Here are some of the ways that different stakeholders can contribute to such an initiative:

- **Patient advocacy organizations**: Foremost, advocacy leaders have the pulse of the community and understand the pain points patients and caregivers experience, including in their care journeys. They can survey community members and/or may have registries or other data platforms that could be used to understand which ICD codes are being used and any associated challenges with access or reimbursement that may exist. Organizations often have connections to other experts through their scientific and medical advisory boards. They may also know of federal research efforts and databases that could be leveraged. Finally, these organizations often

serve an important role as the natural connector between other stakeholders, helping to forge consensus on the optimal approach to a topic like ICD coding and can help manage the timeline and workflow among various parties. If a new/revised code is approved, they will be crucial to helping get out the news to community members and health care professionals.

- **Clinical experts**: The NCHS staff and ICD C&M Committee's assessment of coding proposals relies on solid clinical evidence, so it's wise to involve health care professionals who have a deep understanding of the subject condition, especially in terms of what differentiates it from other conditions that it may be linked to within the ICD coding system. They may have first-hand knowledge of challenges they experience (or their staff members experience) getting patients access to care and service with the current codes used for the disease of interest. Clinical team members can also help vet and socialize a draft proposal with others within their discipline, especially professional societies that may be actively involved in ICD coding activities (such as the American Academy of Pediatrics and the American Academy of Neurology). They, too, will be helpful in affirming the need for a new or revised code, promoting use of it, monitoring usage and identifying any new or unforeseen challenges that it might create in delivering patient care. Consult the Council of Medical Specialty Societies if you're unsure about which medical specialties might be appropriate to approach.
- **Researchers**: In additional to bolstering clinical evidence to support a proposal, epidemiologists and health care services researchers familiar with the disease can be very helpful in terms of providing documentation for its prevalence and incidence and information about health care utilization. Health economics researchers might be able to provide information about how an ICD code would sharpen the ability to understand patient outcomes and costs, both to the individual and the healthcare system. Don't forget to include investigators and administrators working with large health surveillance networks (like the CDC's surveillance program, MD STARnet, in the case study of limb girdle muscular dystrophy, see page 27) and in federal agencies.
- **Industry**: Life science companies may use ICD-code-derived data to inform a variety of decisions, including determining clinical trial feasibility and site location by understanding referral patterns and within which medical specialties and centers patients are being cared for. For this reason, they may be able to provide data that highlights the need for a new coding structure or supports the request for one. Their data may be useful in monitoring changes after a new code is introduced. Additionally, companies may have more familiarity with the process of getting an ICD code revised or added, based on prior experience working in other diseases. They may also be able to provide financial resources through unrestricted grants to a patient advocacy organization to help offset the staff time dedicated to such an initiative, as well as to provide support for direct costs, such as engaging a coding expert (see below and "Proposed Coding Structure" on page 13).
- **Coding Experts**: Consider engaging a professional who can advise on the technical development of a proposed coding structure (as described on page 13), liaise with NCHS staff and inform responses at meetings and through written comments. You may have such an individual within your community; otherwise seek professionals who are credentialed through or affiliated with the American Health Information Management Association (AHIMA) or the American Academy of Professional Coders (AAPC).

Building a coalition of willing collaborators may require education about ICD codes and the potential value of revising coding for the disease of interest. Feel free to broadly share the ICD Code Roadmap video, this guide, and the FAQs with those who want or need to know more about the process before getting involved.

Here's how the AAPC describes the work that coders do: "Medical coding professionals help ensure the codes are applied correctly during the medical billing process, which includes abstracting the information from documentation, assigning the appropriate codes, and creating a claim to be paid by insurance carriers. . . That documentation is not only the patient's ongoing record, it's how the healthcare provider gets paid."

Proposal Format and Evidence

According to information on the NCHS website, proposals for a new code(s) should include:

- A description of the code(s)/change(s) being requested
- Rationale for why the new code/change is needed (including clinical relevancy)
- Supporting clinical references and literature

Proposals should be submitted by a patient advocacy organization or a recognized medical/clinical entity such as a professional society. A cover letter to the proposal should specify all the collaborating organizations and individuals who have contributed to and support the proposal. It should also designate a single point of contact. Ensure that it is someone who monitors email and phone messages regularly and is able to be highly responsive to NCHS staff, sometimes on short notice.

The proposal itself must be concise – no more than 2-3 pages in length. In light of the need for brevity, it is important to focus on aspects of the disease (and the state of understanding about it) that are directly connected to the need for a specific code. Requests based solely on the need to validate the existence of a disease by assigning it a code or, generally, "to aid research or patient care" may carry less weight; specifics are important. Draw on facts, statistics and figures published in reputable peer-reviewed journals and provide citations.

The Friedreich's Ataxia Research Alliance utilized chart review data shared by an industry partner to support a coding proposal. The data documented how the loss of specificity in coding for Friedreich's ataxia (FA) from ICD-9-CM to ICD-10-CM led to 56% of FA patients being coded incorrectly. See page 25 for the case study.

As you develop the proposal, it may be prudent to keep this statement from the ICD-10-CM Guidelines in mind: "The diagnosis codes have been adopted…to achieve complete and accurate documentation, code assignment and reporting of diagnoses and procedures." Coding professional Linda Holzman of Clarity Coding, speaking at a 2018 webinar hosted by the National Organization for Rare Disorders (NORD), summarized the benefits of a specific ICD-10-CM code for a rare disease as:

- Higher quality information for measuring healthcare service quality, safety and efficiency
- Improved ability to measure outcomes, efficacy and costs of new medical technology
- Enhanced review of medical necessity and fewer claims denials
- Reduced need for manual review of health records to perform research and data mining and to adjudicate reimbursement claims
- Reduced need for corroborating documentation to support information reported on claims
- More detailed information for clinical research

"The diagnosis codes have been adopted…to achieve complete and accurate documentation, code assignment and reporting of diagnoses and procedures."

–ICD-10-CM Guidelines

Here are some specific rationales put forward by other rare disease advocates to consider as you think through the approach that might be most persuasive:

- **Differentiate the subject disease from other conditions in the same ICD grouping**: In the ICD-10-CM, Friedreich's ataxia (FA) was grouped under a code (G11.1) for "Early-onset cerebellar ataxia" along with conditions for which the diagnosis, disease course, management and treatment approaches vary significantly from those for FA. While symptom onset for FA may occur early in life (between ages 5 and 18), this is not the case for all the conditions listed under G11.1. Also, several comorbidities, including cardiomyopathy and diabetes, are unique to FA as compared to these other cerebellar ataxias. The proposal for a distinct code for FA focused on these differences and their impact on patient care. Read the FA case study on page 25.

- **Reduce the occurrence of specific care delivery challenges**: The coding proposal submitted for Dravet syndrome (DS) drew attention to specific pharmacological sensitivities that individuals with SCN1A mutations experience. Antiepileptic drugs whose mechanism of action involves the sodium channel exacerbate seizures in DS patients with this mutation and is contraindicated. They proposed that having an ICD-10 code specific to DS would inform medication choices and other aspects of patient care. Learn more about the DS proposal on page 23.
- **Comparatively prevalent condition within the same ICD grouping**: A proposal to designate specific ICD codes for three types of muscular dystrophy (MD) emphasized that these three subtypes were among the four most common types of MD and that, "Assigning individual ICD-10-CM codes…will facilitate the surveillance of these diseases; will allow more accurate estimates of their incidence, prevalence, survivorship, mortality and its causes, injuries, symptoms and health visits; will help to identify factors that influence health status and will assist in the recognition and management of secondary conditions, impacting both mortality and morbidity." The proposal included the following data to support a request for a combined ICD-10-CM code for Duchenne/Becker MD and a separate code for facioscapulohumeral MD. (Myotonic MD had already been identified with a distinct code.) The case study on page 31 provides more information about this proposal and what was learned after the codes were approved.

A proposal to designate specific ICD codes for three types of muscular dystrophy (MD) emphasized that these three subtypes were among the four most common types of MD.

There are several major types and dozens of subtypes of muscular dystrophy. Most types and subtypes, however, are extremely rare. The 4 most common types of muscular dystrophy are:

MUSCULAR DYSTROPHY	PREVALENCE	MODE OF INHERITANCE	AGE AT ONSET/SURVIVAL
Becker	1.5 per 100,000 males	X-linked	Adolescence / Up to middle age
Duchenne	4.8 per 100,000 males	X-linked	2-6 years/ Mid 20s
Facioscapulohumeral	4.0 per 100,000 population	Autosomal dominant	Childhood / Decades after onset
Myotonic	8.3 per 100,000 population	Autosomal dominant	20-40 years / Decades after onset

- **Targeted drugs in the pipeline**: The 2017 proposal for Duchenne and Becker codes also referenced the approval of therapies for specific subtypes of muscular dystrophy as another justification. A 2020 proposal for distinct subtypes of limb girdle muscular dystrophy went a step further, identifying the drug/program name and type of intervention to illuminate that therapies under development are targeted to specific genotypes. This proposal used a combination of the relative prevalence of the subtype and the potential for a targeted therapy within five years as a pragmatic strategy for narrowing the number of distinct ICD codes requested from 34 specific subtypes to 12 codes. Read more about this proposal on page 27.

LGMD SUBTYPE	ESTIMATED PREVALENCE* (PER MILLION)	NOMINATED FOR CODE? (BASIS)	COMPANIES DEVELOPING THERAPIES	DRUG/PROGRAM NAME AND APPROACH	STAGE OF DEVELOPMENT	SOURCE
Autosomal Recessive LGMD due to Anoctamin-5 dysfunction (2L or R12)	17.63 per million	Yes (Prevalence and Rx)	Sarepta	SRP-9006 - Gene therapy	Pre-clinical	Link
Autosomal Recessive LGMD due to calpain-3 dysfunction (2A or R1)	8.38 per million	Yes (Prevalence and Rx)	Sarepta and Genethon	"Calpain 3" - Gene therapy and an unnamed gene therapy	Pre-clinical	Link, and Link
Autosomal Recessive LGMD due to collagen-VI dysfunction (bethlem myopathy recessive or R22)	Unknown but likely over 4 per million**	Yes (Prevalence)				
Autosomal Recessive LGMD due to dysferlin dysfunction (2B or R2)	7.457 per million	Yes (Prevalence and Rx)	Sarepta	SRP-6004 - Gene therapy	Clinical	Link
Autosomal Recessive LGMD due to fukutin-related protein dysfunction (2I or R9)	4.516 per million	Yes (Prevalence and Rx)	ML Bio, Genethon, and AskBio	BBP-418 (Ribitol) – Small molecule, and two unnamed gene therapies	Clinical and Pre-clinical	Link, Link, and Link
Autosomal Recessive LGMD due to alpha-sarcoglycan dysfunction (2D or R3)	3.361 per million	Yes (Prevalence and Rx)	Sarepta	SRP-9004 - Gene therapy	Clinical	Link
Autosomal Recessive LGMD due to beta-sarcoglycan dysfunction (2E or R4)	.7967 per million	Yes (Rx)	Sarepta	SRP-9003 - Gene therapy	Clinical	Link
Autosomal Recessive LGMD due to gamma-sarcoglycan dysfunction (2C or R5)	.1169 per million	Yes (Rx)	Sarepta and Genethon	SRP-9005 - Gene therapy and an unnamed gene therapy	Pre-clinical	Link, and Link
Autosomal Dominant LGMD due to calpain-3 dysfunction (1I or D4)		Yes (analogous dominant form)				
Autosomal Dominant LGMD due to collagen-VI dysfunction (bethlem myopathy dominant or D5)		Yes (analogous dominant form)				
Autosomal Recessive LGMD due to delta-sarcoglycan dysfunction (2F or R6) [Included for Reference]	.0659 per million	No (Does not meet prevalence or Rx threshold)				
Autosomal Recessive LGMD due to telelthonin dysfunction (2G or R7) [Included for Reference]	.0396 per million	No (Does not meet prevalence or Rx threshold)				

Proposed Coding Structure

One of the trickiest components of a proposal is the specific recommendation for how to revise the coding structure itself to reflect a new or amended code. This is where guidance from a coding professional can be extremely helpful.

The proposal must be consistent with the "established structure and conventions of the classification," as stated on the ICD C&M Committee website (see general description on page 6). It is important to understand that topics presented at ICD C&M Committee meetings will include a coding structure modification proposed by NCHS staff. This proposed structure may differ from the original proposed structure submitted by an external organization (or group of organizations). The coding community will pay close attention to the proposed modification. Their questions, comments and concerns will focus on the topic at hand, as well as the precedent it sets for other similar or related conditions and potential consequences for other condition-code combinations. Again, this is why a coding professional should ideally be part of the team that helps to construct the proposal.

As you work through this part of the proposal, it may be prudent to view the specific coding modification through a pragmatic lens, both in terms of the initial proposal to be submitted and the process that follows. In particular, there might be a solid case to support a request for multiple codes to distinguish between important subtypes of a specific condition (or group of conditions). Yet, as

> The proposal submitted by a coalition led by Parent Project Muscular Dystrophy requested a single code for Duchenne and Becker and a separate code for facioscapulohumeral muscular dystrophy. The decision to combine Duchenne and Becker was based on a concern that separating them might put access to services and treatments at risk, given the fast-evolving scientific and clinical understanding of the two subtypes. Learn more through the case study on page 31.

*Liu, W., Pajusalu, S., Lake, N.J. et al. Estimating prevalence for limb-girdle muscular dystrophy based on public sequencing databases. Genet Med 21, 2512–2520 (2019). https://doi.org/10.1038/s41436-019-0544-8

**Nallamilli, B., Chakravorty, et al. (2018). Genetic landscape and novel disease mechanisms from a large LGMD cohort of 4656 patients. Annals of clinical and translational neurology, 5(12), 1574–1587. https://doi.org/10.1002/acn3.649

several of the case studies presented in this guide illustrate, multi-code requests often get pared down in the process, sometimes in unexpected ways. For this reason, it's also wise to identify at the outset what would be an acceptable outcome of the process, recognizing that you will not be in control of that final outcome. If there are some potential negative consequences of a coding change, weigh the benefits and risks early.

For those developing a request for a code to recognize one or more rare diseases (or subtypes), be aware that among the coding community there is a sense of protection to prevent the overall set of ICD-10-CM codes from becoming too large and unwieldy. There is no established minimum for the requisite "commonness" of a disease, a subtype or a reason for seeking healthcare services, but the phrase "one in a million"[1] has been used during ICD C&M Committee meetings as an informal threshold for whether a specific code is merited. (See the case studies for limb girdle muscular dystrophy and SYNGAP1, on pages 27 and 29, respectively.) Factor this trend into the structure for the coding modification and provide supporting evidence whenever possible to justify the classification(s).

In some cases, the decision about where to insert new codes will be fairly straightforward, as it was with Duchenne, Becker and facioscapulohumeral subtypes of muscular dystrophy. The proposal suggested adding four new codes to the subcategory for G71.0 Muscular dystrophy as shown below:

In some cases, the decision about where to insert new codes will be fairly straightforward.

ICD 10 (current)

VI DISEASES OF THE NERVOUS SYSTEM

- **G00-G09** Inflammatory diseases of the central nervous system
- **G10-G14** Systemic atrophies primarily affecting the central nervous system
- **G20-G26** Extrapyramidal and movement disorders
- **G30-G32** Other degenerative diseases of the nervous system
- **G35-G37** Demyelinating diseases of the central nervous system
- **G40-G47** Episodic and paroxysmal disorders
- **G50-G59** Nerve, nerve root and plexus disorders
- **G60-G64** Polyneuropathies and other disorders of the peripheral nervous system
- **G70-G73** Diseases of myoneural junction and muscle
  - **G70** Myasthenia gravis and other myoneural disorders
  - **G71** Primary disorders of muscles
    - **G71.0** Muscular dystrophy
    - **G71.1** Myotonic disorders
      - + **G71.11** Myotonic muscular dystrophy
      - + **G71.12** Myotonia congenita
      - + **G71.13** Myotonic chondrodystrophy
      - + **G71.14** Drug induced myotonia
      - + **G71.19** Other specified myotonic disorders
    - **G71.2** Congenital myopathies
    - **G71.3** Mitochondrial myopathy, not elsewhere classified
    - **G71.8** Other primary disorders of muscles
    - **G71.9** Primary disorder of muscle, unspecified
  - **G72** Other myopathies
  - **G73** Disorders of myoneural junction and muscle in diseases classified elsewhere
- **G80-G83** Cerebral palsy and other paralytic syndromes
- **G90-G99** Other disorders of the nervous system

ICD 10 (proposed change)

VI DISEASES OF THE NERVOUS SYSTEM

- **G00-G09** Inflammatory diseases of the central nervous system
- **G10-G14** Systemic atrophies primarily affecting the central nervous system
- **G20-G26** Extrapyramidal and movement disorders
- **G30-G32** Other degenerative diseases of the nervous system
- **G35-G37** Demyelinating diseases of the central nervous system
- **G40-G47** Episodic and paroxysmal disorders
- **G50-G59** Nerve, nerve root and plexus disorders
- **G60-G64** Polyneuropathies and other disorders of the peripheral nervous system
- **G70-G73** Diseases of myoneural junction and muscle
  - **G70** Myasthenia gravis and other myoneural disorders
  - **G71** Primary disorders of muscles
    - **G71.0** Muscular dystrophy
      - + G71.01 Duchenne or Becker muscular dystrophy
      - + G71.02 Facioscapulohumeral muscular dystrophy
      - + G71.08 Other specified muscular dystrophies
      - + G71.09 Muscular dystrophy, unspecified
    - **G71.1** Myotonic disorders
      - + **G71.11** Myotonic muscular dystrophy
      - + **G71.12** Myotonia congenita
      - + **G71.13** Myotonic chondrodystrophy
      - + **G71.14** Drug induced myotonia
      - + **G71.19** Other specified myotonic disorders
    - **G71.2** Congenital myopathies
    - **G71.3** Mitochondrial myopathy, not elsewhere classified
    - **G71.8** Other primary disorders of muscles
    - **G71.9** Primary disorder of muscle, unspecified
  - **G72** Other myopathies
  - **G73** Disorders of myoneural junction and muscle in diseases classified elsewhere
- **G80-G83** Cerebral palsy and other paralytic syndromes
- **G90-G99** Other disorders of the nervous system

Figure from the proposal submitted by Parent Project Muscular Dystrophy, June 2017

[1] Discussion at the March 10, 2021 meeting of the ICD C&M Committee referenced "one in a million" beginning at the 5:33 mark. A link to the webcast is available on the Committee's webpage.

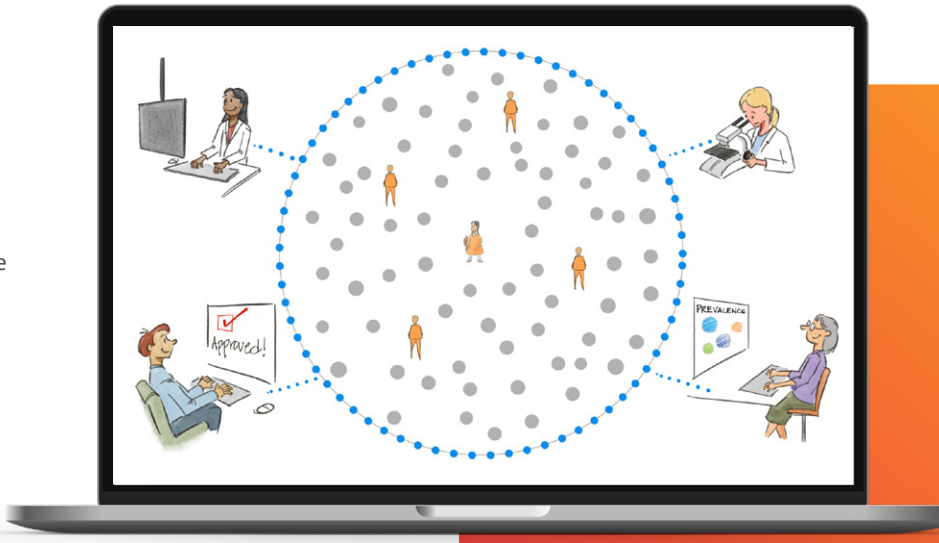
For other diseases, the decision about which category or subcategory in which to place new codes might be more challenging, due to the state of current scientific/clinical understanding of the disease or there being different, but equally plausible, ways of classifying the disease within the existing structure. This was the case for SYNGAP1. A proposal submitted on behalf of the SynGAP Research Fund and Bridge the Gap, two patient advocacy organizations, provided NCHS staff with two possibilities. One would place a new code within the section for "G93.49 - Other encephalopathy" as "G93.4902 SYNGAP1 Encephalopathy." The second option would place the new code within "F78 – Other intellectual disabilities" as "F78.02 – Intellectual disability secondary to pathogenic SYNGAP1 variant." Learn more about the SYNGAP1 proposal on page 29.

A final factor to consider in developing the proposed modification is whether there is a code for the disease in the WHO's ICD-11 that is available for countries ready to implement it as early as January 1, 2022. Use the WHO browser tool to check; if there is a code in ICD-11 (but not ICD-10-CM), it is worth considering whether the same (relative) placement within the ICD-10-CM would serve the intended purposes. The WHO designation could then be cited as a supporting factor in favor of the change. (Note: There has not yet been a target date announced for implementation of ICD-11 in the U.S., but it is likely to be several years away.) If the placement within ICD-11 doesn't serve your purposes, rectifying it will require a separate effort, working through a process managed by the WHO.

Letters of Support

Beyond the signatories to the proposal itself, other organizations and groups of individuals can be encouraged to express written support of a proposed modification of the ICD-10-CM with the initial submission through a brief letter that reiterates the core rationale for the change. Cast a wide net and consider requesting letters of support from the following:

- Patient advocacy organizations, including those that might represent a broader array of related conditions and coalition partners
- Individual and groups of clinical experts, including relevant medical societies
- Individual and groups of researchers, including professional associations for relevant research disciplines
- Life science companies with marketed medical products or therapies and/or diagnostics in development for the disease



If the proposed topic is selected for presentation at a meeting of the ICD C&M Committee, an open comment period will follow the meeting date that closes about one month later. Organizations, groups and individuals can be encouraged to restate their support of the proposal in writing. This would also be the time to rally organizations and groups to express reservations or concerns about any potentially problematic change in the coding structure presented by NCHS staff at the meeting. Keep in mind that it might be necessary to gather the organizations and individuals collaborating on a proposal to submit a coherent, aligned response to the Committee. Divergent viewpoints may work against any change to the ICD-10-CM.

As you reach out to gather letters of support, it may be helpful to share the ICD Code Roadmap video and FAQs in case those receiving the request want or need to know more about the process before getting involved.

Ana Mingorance, Ph.D., of the LouLou Foundation took the lead on developing a proposal seeking a distinct ICD-10-CM code for CDKL-5 Deficiency Disorder (CDD). In a blog post about the process they followed, Ana writes, "We got letters from some of the major medical societies (others don't produce letters but call and talk to the [NCHS] review team, so we still wanted to make sure they would be supportive), from relevant umbrella patient organizations like the Epilepsy Foundation and NORD, and from all of the pharmaceutical companies working on CDD. The message was always the same: 'CDD is a unique disorder with unique medical needs that therefore needs a unique code, and we support the proposed code.'" Their proposal was submitted in June 2019; a new code for CDD went into effect on October 1, 2020.

Submitting the Proposal and Monitoring Progress

There are two deadlines each year for submitting proposals for ICD-10-CM codes:

- **Winter: December or January for the meeting in March**
- **Summer: June for the meeting in September**

Be sure to check the schedule posted to the ICD C&M Committee Meeting web page for the exact deadlines. It can be beneficial to reach out to NCHS staff in the weeks or months before the deadline to make them aware of the intention to submit a proposal and, possibly, to introduce the topic and the approach you're taking. It is better to do this after you have landed on an approach, but before it is finalized in the event you gain new information through your interactions that inform proposal development. Given the small size of the NCHS team, it's possible you won't successfully connect before the deadline; there is no requirement for pre-submission contact, so move forward to submit it as soon as the proposal is deemed ready by those collaborating to develop it.

Many organizations submit proposals with a transmittal cover letter that identifies the collaborating entities (organizations, institutions, individuals, companies, etc.). Be sure to allow enough time to get appropriate permission from each party. In that letter and in the body of the email message to which the letter/proposal are attached, be sure to designate a reliable, responsive contact person and provide email and telephone information for reaching that individual. Protect the upcoming meeting date and ask your preferred clinical expert to do the same.

Once the proposal is submitted, there are about three months between the submission deadline and the next committee meeting date. NCHS staff can contact the submitter at any point during that period to pose questions or seek clarification about the proposal. Staff may also request that a clinical expert prepare to give a short clinical presentation (generally five minutes or so) that supports the proposal. Note that a member of the NCHS staff will present the proposed coding structure, which may differ from the way it was presented in the original proposal. Revisions are based on their assessment of the evidence presented and research they conduct independently. You may receive confirmation that the topic will be on the meeting agenda as late as a day or two before the meeting itself. This can be nerve-wracking, so prepare for the possibility in advance! Proposals that are determined to be incomplete will be returned to the submitter; you will also be notified if the proposal will not be reviewed at the next meeting.

The best way to prepare for the ICD C&M Committee meeting is to attend or watch (live or recorded) webcasts of prior meetings. Each topic is covered in a short amount of time (10-15 minutes), which includes time for questions and comments from participants. The most important thing to understand is that **no decisions will be made at the meeting itself**. (This bears repeating!) One veteran of the process described it as, "attending the Oscars without learning who the winners in each category are."

Proposals for ICD code additions/revisions must be submitted to NCHS by email to nchsicd10CM@cdc.gov. Once proposals are reviewed, all requestors will be contacted as to whether the proposal has been approved for presentation at the ICD-10 C&M Committee meeting or not. Incomplete proposals will be returned to the submitter.

The most important thing to understand is that no decisions will be made at the meeting itself.

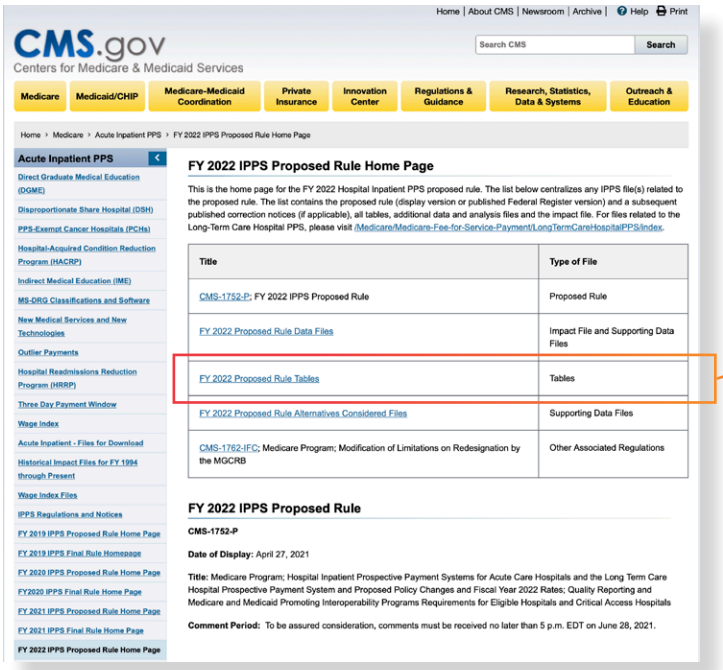
An open comment period of roughly one month follows each meeting. This is a good time to submit written comments or clarifications to the proposal itself, based on discussion at the meeting, or in response to the NCHS staff's revision of the coding structure. It also provides an(other) opportunity to encourage others to send letters of support for a code.

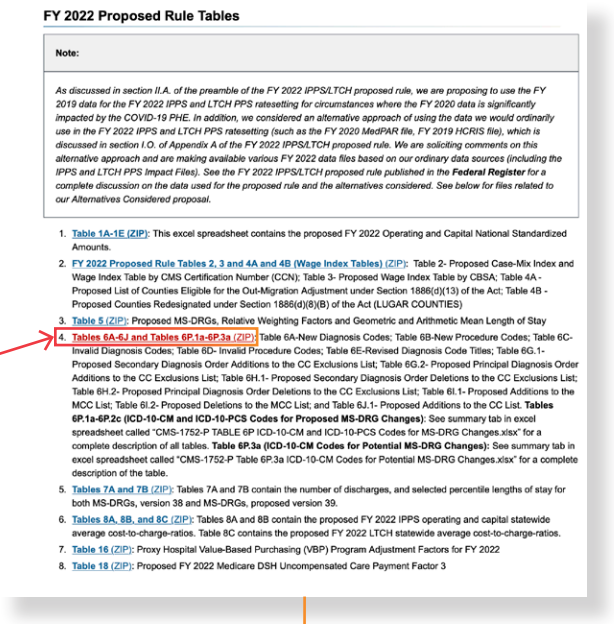
Following the close of the comment period, communication from NCHS staff will depend on additional clarification needed or other questions that may arise. They may let the submitter know that the topic will be on the agenda for a future meeting, if another version of the coding structure is to be presented and reviewed. For this reason, it's wise to block the ICD C&M Meeting dates on the calendar until a final outcome is determined.

Each April, a notice of proposed rulemaking for the "Health Insurance Prospective Payment System Codes" (HIPPS Codes) is published in the Federal Register and posted to the CMS website. On CMS.gov, click to the "Proposed Rule Tables" and then look for the table with "New Diagnostic Codes." You will have to download this table to view it. In the FY2022 notice posted on April 27, 2021, table 6A displayed new codes that will become effective on October 1, 2022, as shown in the figure below.

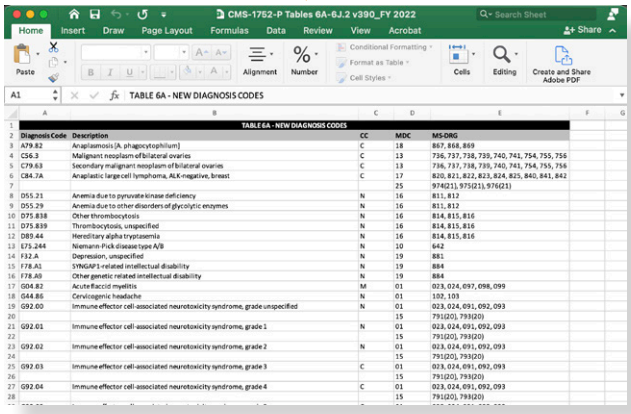
"Proposal packets" from past ICD C&M Committee meetings provide a great orientation to the format of these meetings and topic presentations. Committee meetings held in 2020 and 2021 were conducted remotely due to restrictions imposed during the Coronavirus pandemic; those meeting recordings provide a good opportunity to hear the discussion of other topics. The list of presenters in the "Diagnosis Topics" at the front of each packet may help guide selection of an appropriate expert to provide the clinical background, if requested. These materials can be found on the **ICD C&M Committee webpage**.

Locating new ICD-10-CM codes in the IPPS Proposed Rules at CMS.gov





TIP: When you click, you will have to download this table to view it



By early May, you could also contact NCHS staff to request an update (if you haven't already received one from them), since the information will be in the public domain by then. It is possible that a decision may be delayed until the next cycle, or further revision is needed. It may be challenging to obtain information about the exact status of a proposal that is pending, as well as details about specific issues that remain in question.

Upon Approval & Implementation

As soon as you learn that a new code has been approved, develop a plan to help spread the word through as many channels as possible so the news reaches healthcare professionals and their staff members who support the coding process, along with all the other parties who will benefit from advance notice. Engage all the coalition partners who helped develop the coding proposal – each has a network of potentially helpful contacts who can spread the word and make sure the code is implemented as soon as it goes into effect. Coding and decision-support software packages generally update automatically with the coding changes that go into effect on October 1 each year but getting the code into appropriate use begins with community and provider awareness and frequent reminders so that it has maximum utility.

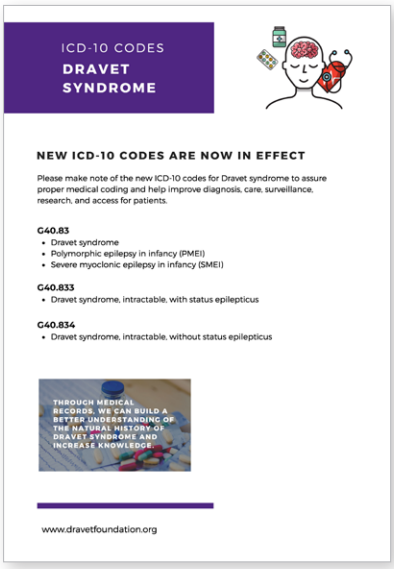
The Friedreich's Ataxia Research Alliance posted a short, informational video to YouTube to explain the new ICD code and its importance to the community.

Patient advocacy organizations can issue press releases to highlight the effort's success and the partners who contributed to achieving it. A press release may also spur coverage by relevant publications and will make the information accessible in web search results. Medical and scientific advisory board members should be alerted so that they can help spread the news to colleagues, healthcare systems and professional societies. Similarly, hosting a webinar for staff of specialty clinics connected through and by the organization can prepare them for the upcoming change. If there are existing relationships with public, commercial and private payers, make sure they know a change is coming; policy documents may need to be updated to reflect a new code. It's also important to let researchers know about the change.

Patients and families can help bring this information to their healthcare professionals' attention, so include the information in blog posts, electronic newsletters and websites to involve them in spreading the word. Consider making fact sheets or cards to distribute electronically and in print

The above ideas are worth repeating (or highlighting) when the code goes into effect on October 1. Here are links to examples of awareness-raising activities that rare disease communities have led to spread the word about a new code approval:

- Angelman syndrome:
  - The Atlantic (magazine)
  - Spectrum News
- Castleman disease
- CDKL5 deficiency disorder
- Dravet syndrome:
  - Dravet Syndrome Foundation
  - Neurology Live
- Duchenne and Becker muscular dystrophies
- Friedreich's ataxia
- Prader-Willi syndrome



The Dravet Syndrome Foundation provided fact sheets and cards to families to help spread the word.

Life science company partners can help to spread the word through their networks of clinical experts, field staff and medical science liaisons (MSLs) who are in frequent contact with hospital and clinic staff and administrators. Companies also have ongoing communication with payers, employer groups and pharmacy benefit managers and can use these channels to announce the new code so that coverage and reimbursement policies can be updated.

The work doesn't end when the code goes into effect. The understanding of disease is ever-evolving, as are the ways a particular disease is described, diagnosed, treated and reported, so it's important to stay attuned to changes that might warrant another update of the ICD-10-CM code(s). Touch base periodically with collaborators who might detect or be able to document changes that signal reasons to take another look at the codes.

It's possible that a new U.S. ICD-10-CM code could prompt inquiries from colleagues in other countries about what they can do to update the coding system used in their country. Each country has its own process, but the proposal materials and precedent in the U.S. may be useful to advocates interested in working on coding changes in other countries, too.

Two years after a new code was implemented for Duchenne and Becker muscular dystrophies, a review of data showed that the code is widely used and is cited in coverage policies for drug therapies, genetic testing and nerve conduction testing. Learn more about how Parent Project Muscular Dystrophy rallied community stakeholders to propose the coding change and other observations at the two-year mark following implementation. **See page 31 for the case study.**

SECTION 3: TIPS & PITFALLS

Much of what has been learned by those familiar with the process of proposing a new or revised ICD-10-CM code has been passed through informal channels, in the form of lessons learned. Here are some of the most important learnings shared by veterans of the process and references to more detailed information:

Tips to improve chances of success:

- Rely on data and evidence, not storytelling or heartstrings, for proposals. (See "Proposal Format e and Evidence" on page 11.)
- Prepare to live with a potentially uncomfortable level of uncertainty around this process once you enter into it. There are weeks and months where you will receive little new information and other times when you might have to respond on short notice. The outcome is unpredictable – both in terms of how long it might take to reach a conclusive answer and the final decision itself.
- Ensure that the disease (or disease subtype) you are focused on is listed in the National Institute of Health Genetic and Rare Disease (GARD) Information Center. Sites like this are consulted to verify the legitimacy of a disease, so work to get it added before submitting a proposal for a new or revised ICD-10-CM code.
- Build a coalition that speaks to coding issues with a consistent voice. (See "Collaboration" on page 9.)
- If possible, work with an experienced professional coder or someone who has gone through the process before. (See "Coding Experts" on page 10.)
- Attend one or more meetings of the ICD C&M Committee to establish relationships (if/when meetings are held in person), learn the process and become familiar with types of questions asked. (See the Committee's webpage for dates and other information.)
- Review the "Diagnosis Topics" listing at the front of proposal packets from past ICD C&M Committee meetings to get an idea of the types of clinical experts who present disease-area background during these sessions.
- Know the history (ICD-9-CM) and the future (WHO ICD-11) of how the disease of interest has been reflected in diagnostic codes. (See "Proposed Coding Structure" on page 13 for how you might make use of this information.)
- Try to keep tabs on the process, even during periods between ICD C&M Committee meetings when there may be little interim communication and information from NCHS staff. If the person who submits the proposal changes their contact information or becomes unavailable for more than a week (especially in the lead-up to a meeting date), ensure the NCHS staff contact gets updated email/phone information and/or knows who they can rely on to respond to requests. (See the visual timeline on page 7 for an overview of the process.)
- Plan ahead to foster adoption of the new code(s) (if granted) with healthcare professionals, healthcare systems, coders and payers. Amplify awareness and education about the code(s) on the effective date and repeat the message often; it can take time to change practices and achieve full implementation. (See "Upon Approval" on page 18.)

Attend one or more meetings of the ICD C&M Committee to establish relationships (if/when meetings are held in person), learn the process and become familiar with types of questions asked.

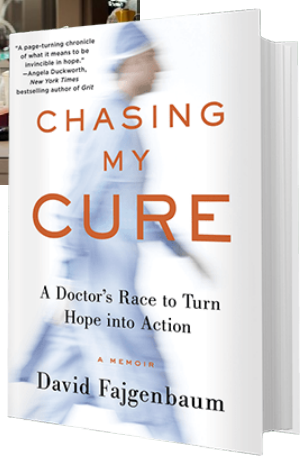
Pitfalls that can delay progress or deter approval:

- Don't waste time looking for a way to short-cut the formal process. It is designed to be deliberative and proceeds according to a highly structured calendar of events. (See "Overview of the Process for Proposing Changes to ICD-10-CM Codes" on page 7.)
- Seeking a code solely as a means to validate a disease will likely not be sufficiently compelling on its own. See "Proposal Format and Evidence" (page 11 and the case studies (beginning on page 22) for ideas about how to develop a public-health based rationale that can stand up to the kind of scrutiny it will receive.
- If there is a lack of consensus about the name of or criteria for diagnosing the disease you're focused on, it is probably wise to address those issues first. The coding community seeks clarity and precision, so signs of open discord and disagreement among clinical experts or community members may be reason enough to defer the decision or decline it outright. (See "Collaboration" on page 9.)
- Similarly, if there are competing proposals in development for the same condition that suggest different placements for a new code or different coding structures, work to find consensus before submitting. (See "Collaboration" on page 9 and "Proposed Coding Structure" on page 13.)
- Don't rush the initial proposal. Getting the appropriate stakeholders on board to develop an evidence-based, consensus proposal is a must, as stated above. Even though there are only two submission deadlines each year, the process could string out through several cycles if you have to recover from early missteps. It's also worth stating that it can extend for several cycles even if you do everything you can on the front end, so make sure you have sufficient time and resources for planning, preparation and responding to (potentially) short notice requests over an extended period of time (at least 13 months, and possibly much longer from the date following submission of the proposal) before you commit to the process.
- Avoid demanding updates or action from members of the NCHS staff. It's a small team responsible for coordinating and maintaining updates to the entire coding structure. This is a collaborative and iterative process.

The ICD Code Readiness Assessment Survey can help you explore whether pursuing a specific ICD code is right for your community at this time.

SECTION 4: CASE STUDIES

CASTLEMAN DISEASE



Eleven weeks passed between the date **David Fajgenbaum, M.D.**, was first hospitalized and when he was diagnosed with HHV-8 negative, idiopathic multicentric **Castleman disease** and given his first chemotherapy treatment in 2010. Even with the vast healthcare resources David and his physician father could harness, David came close to death numerous times over those 11 weeks and the next 3 years, a harrowing story he chronicles in his memoir, **"Chasing My Cure."**

Once he had a name for the disease he was fighting, David set out to tackle it. One discovery of many in this quest was that Castleman disease wasn't part of the formal health information nomenclature – it lacked a specific ICD code. David understood from his medical training that accurate codes are crucial to tracking, reimbursement, gaining access to therapies and much more. Yet Castleman disease was swept into a "miscellaneous code, one that covered a number of hard-to-categorize diseases," as he described it. In 2014, he and Dr. Frits van Rhee, along with colleagues from the Castleman Disease Collaborative Network he co-founded, went to work to secure a new code specific to Castleman disease.

"I emailed the folks at CDC multiple times for guidance, and they were very helpful when I finally received a response," he recalled. "I drafted a proposal that was improved upon by Dr. van Rhee and other members of our Scientific Advisory Board. In it, we requested the addition of three codes for specific subtypes of Castleman disease. Letters of support were sent by 15-20 patients, physicians and researchers, and I kept the heat on. I presented our proposal at the ICD C&M meeting held in September 2014; after that, long periods went by without any information or response from the CDC staff. The status of our request wasn't clear. We had not engaged any coding experts to help us, so I had no idea what to expect or when we might hear something."

More than two years later, in 2017, David was looking through his medical reports following a recent doctor visit. He noticed an unfamiliar ICD code, D47.Z2, on a billing statement. He recalls that moment in his book, "Confused, I googled it. It was Castleman disease. Our very own code." Upon further exploration, David learned that a single code for Castleman disease had been approved and went into effect on October 1, 2016. The subtype codes were not accepted as proposed, based on the Committee's determination that Castleman disease was too rare to warrant three separate codes. Although chagrined at the idea that a "burn due to water skis on fire" warranted three codes, he decided one code for Castleman disease was better than no code. "It wouldn't exist if we hadn't lobbied for it. Someone had to turn hope into action to make this a reality. If you don't do it, oftentimes no one else will. We've come a long way."

> "The code specific to Castleman disease wouldn't exist if we hadn't lobbied for it. Someone had to turn hope into action to make this a reality. If you don't do it, oftentimes no one else will."
>
> – David Fajgenbaum, M.D.

Visit the **CDCN ICD code page** for more information.

DRAVET SYNDROME

Like many rare disease advocates, **Mary Anne Meskis's** determination to problem-solve is rooted in personal experience. The youngest of her three children, Elliot, has **Dravet syndrome**, a rare catastrophic form of epilepsy that begins in the first year of life. She was among a group of parents who founded the Dravet Syndrome Foundation in 2009, and she became the organization's first executive director in 2012, shepherding its mission to expedite research and find better treatments and a cure.

One of the challenges Mary Anne recognized was the manner in which Dravet syndrome was coded in health information systems – it was included in a general code for "Other epilepsy and recurrent seizures," G40.8. "This broad category didn't encompass the many symptoms of Dravet syndrome, which include behavioral and developmental delays, movement and balance issues, orthopedic conditions, chronic infections, sensory integration disorders and dysautonomia. It made it more difficult to secure coverage for indicated medicines and medical testing to address all healthcare needs of a person with Dravet syndrome," she stated. By 2017, there were several promising therapies for Dravet syndrome in various life science companies' pipelines, including some for a subtype associated with a mutation of the SCN1A gene, which accounts for over 80 percent of those diagnosed. She knew from other advocacy leaders that coding issues can make it more difficult for patients to access therapies (if and) when they are approved.

She initiated discussions about seeking a specific ICD code for Dravet syndrome with the Foundation's Medical Advisory Board. In January 2018, they submitted a proposal to establish a new subcategory and four subtype-specific codes for Dravet syndrome. It reflected the importance of differentiating between Dravet syndrome with and without evidence of a SCN1A mutation, as well as with and without status epilepticus.

Over the next two years, Mary Anne and the group monitoring and responding to developments in the ICD C&M Committee's deliberations worked to address comments. Dr. Ian Miller, a pediatric epileptologist, presented to the Committee on behalf of the Foundation twice in person. Mary Anne accompanied him too. "We went to Baltimore not knowing what time our proposal would come up. We listened intently to the other presentations and the discussion that followed ours and others, but we received little information about the status of the proposal between meetings," she recounted.

The Committee staff's presentation at the meeting held in March 2019 indicated that the third iteration of the Dravet syndrome proposal released on that date had been "reviewed and supported" by the American Academy of Neurology at the Committee staff's request. Dravet syndrome was not discussed at the meeting held in September 2019; it wasn't clear what that meant. However, in January 2020, two years after submitting the initial proposal, Mary Anne received word that the Committee had approved a new subcategory for Dravet syndrome under the G40 Epilepsy code, along with two codes for specific subtypes. These would become effective as of October 1, 2020.

As Mary Anne notes, the process didn't end with that news, as welcome as it was. "We immediately went to work to spur awareness and adoption of the new codes. They won't benefit anyone if healthcare professionals don't know they exist. We issued an announcement in February 2020 to set the stage and a press release after they went into effect, in November 2020, using the heightened visibility of Epilepsy Awareness Month as leverage to spread the word. We have one-page fact sheets and business-card-size reminders that our families can use to make sure their healthcare providers know about and use the codes. We keep a steady drumbeat going."

Elliot Meskis, pictured with his sister Sophie, is among those who inspired Mary Anne to help establish the Dravet Syndrome Foundation and to take on projects like proposing a unique ICD-10-CM code for Dravet syndrome.

> "Upon learning that new codes for Dravet syndrome were going to become effective, we immediately went to work to spur awareness and adoption of the new codes. They won't benefit anyone if healthcare professionals don't know they exist."
>
> – Mary Anne Meskis

The timeline and iterations of the Dravet syndrome proposal are mapped below to illustrate how the process unfolds when comments offered at the meeting, received in writing afterwards or actively sought out from authoritative sources prompt revisions.

DATE	ACTION	PROPOSED REVISION OF ICD CODE AS PRESENTED (RED INDICATES NEW ADDITIONS TO THE CODING STRUCTURE)
1/5/18	DSF proposal submitted	G40 Epilepsy G40.8 Other epilepsy and recurrent seizures G40.83 Dravet syndrome G40.831 ..... intractable, without SCN1A mutation, without status epilepticus G40.832 ..... intractable, without SCN1A mutation, with status epilepticus G40.833 ..... intractable, with SCN1A mutation, without status epilepticus G40.834 ..... intractable, with SCN1A mutation, with status epilepticus
1/8/18	Deadline for new proposals to be reviewed at March 2018 meeting	
3/6/18	Proposal presented by ICD C&M Committee Staff	G40 Epilepsy G40.8 Other epilepsy and recurrent seizures G40.83 Dravet syndrome Polymorphic epilepsy in infancy (PMEI) Severe myoclonic epilepsy in infancy (SMEI) G40.831 Dravet syndrome, intractable, with SCN1A mutation, with status epilepticus G40.832 Dravet syndrome, intractable, without SCN1A mutation, without status epilepticus G40.833 Dravet syndrome, intractable, without SCN1A mutation, with status epilepticus G40.839 Dravet syndrome, intractable, with SCN1A mutation, without status epilepticus and Dravet syndrome not otherwise specified
5/11/18	Deadline for comments on proposals discussed at March 2018 meeting	
9/11/18	Proposal presented by ICD C&M Committee Staff	G40 Epilepsy G40.8 Other epilepsy and recurrent seizures G40.83 Dravet syndrome G40.831 Dravet syndrome, intractable, with status epilepticus G40.832 Dravet syndrome, intractable, without status epilepticus G40.839 Dravet syndrome unspecified, polymorphic epilepsy in infancy (PMEI), severe myoclonic epilepsy in infancy (SMEI), Dravet syndrome not otherwise specified
11/13/18	Deadline for comments on proposals discussed at September 2018 meeting	
3/5/19	Proposal presented by ICD C&M Committee Staff	G40 Epilepsy G40.8 Other epilepsy and recurrent seizures G40.83 Dravet syndrome Polymorphic epilepsy in infancy (PMEI) Severe myoclonic epilepsy in infancy (SMEI) G40.831 Dravet syndrome, intractable, with status epilepticus G40.832 Dravet syndrome, intractable, without status epilepticus and Dravet syndrome not otherwise specified
10/1/20	**New codes go into effect**	G40 Epilepsy G40.8 Other epilepsy and recurrent seizures G40.83 Dravet syndrome Polymorphic epilepsy in infancy (PMEI) Severe myoclonic epilepsy in infancy (SMEI) G40.833 Dravet syndrome, intractable, with status epilepticus G40.834 Dravet syndrome, intractable, without status epilepticus

FRIEDREICH'S ATAXIA

When the U.S. converted from ICD-9 to ICD-10 in 2015, the intent was for the diagnostic codes to become more specific and more accurate. In spite of this intent, the ICD code for Friedreich's ataxia (FA) became less specific in the transition. FA went from having a specific code (334.0) in ICD-9-CM to being grouped under a code for "Early-onset cerebellar ataxia" (G11.1) along with conditions for which the diagnosis, disease course, management and treatment approaches vary significantly from those for FA. Several comorbidities, including cardiomyopathy and diabetes, are unique to FA as compared to these other cerebellar ataxias.

Recognizing the need to recover that greater degree of specificity for research purposes, the benefit of patient care and for facilitating payer approval of approved treatments, **Susan Walther, M.S., L.C.G.C.**, director of patient engagement for the Friedreich's Ataxia Research Alliance (FARA), worked with physicians in the Collaborative Clinical Research Network for FA to initiate a request for a specific ICD-10-CM code.

> "We had information that illustrated just how messy the coding had become under ICD-10-CM. Of 1,276 FA patients who had been correctly identified under the ICD-9-CM code for FA (334.0), only 558 (44%) were subsequently coded with the new ICD-10-CM code, G11.1." – Susan Walther, M.S., L.C.G.C.

Among the evidence they compiled to support the request was a review of diagnostic codes cited in the health records of 1,276 FA patients. Susan recalls, "This data had been shared by an industry partner months before we determined the need to submit a proposal for a new code. As I was drafting our request, I remembered we had information which illustrated just how messy the coding had become under ICD-10-CM. These 1,276 patients had been correctly identified under the ICD-9-CM code for FA (334.0), but only 558 (44%) were subsequently coded with the new ICD-10-CM code, G11.1." There were eight other ICD-10-CM codes used in charts of 404 patients and there was no additional data in the records for the remaining 25 percent, as shown below:

ICD-9 CODE	ICD-10 CODE	DIAGNOSIS CODED AS:	NUMBER (%) OF FA PATIENTS' RECORDS REVIEWED
334.0		Friedreich's ataxia	1,276 (100%)
	G11.1	Early-onset cerebellar ataxia	558 (44%)
	G11.9	Hereditary ataxia, unspecified	148 (12%)
	G11.8	Other hereditary ataxias	82 (6%)
	G71.0	Muscular dystrophy	79 (6%)
	G11.2	Late-onset cerebellar ataxia	28 (2%)
	G11.3	Cerebellar ataxia with defective DNA repair	18 (1%)
	G11.0	Congenital nonprogressive ataxia	16 (1%)
	G60.0	Hereditary motor and sensory neuropathy	14 (1%)
	No data		314 (25%)

Susan used this data and other evidence sources to prepare the proposal in collaboration with advisors from the NCHS. They recommended a subcategory and a code that would move FA out of the group of early-onset cerebellar ataxias, given it is not an accurate placement. After approximately six months of discussion and proposal revisions with NCHS staff, FA was placed among the topics to be discussed at the ICD C&M Committee's September 2019 meeting. Susan presented the clinical overview; in it she included the data from the chart review. David Berglund, M.D., presented the agency's proposal for a revised coding structure. He proposed maintaining the G11.1 category for "early-onset cerebellar ataxia" and creating new codes for early-onset cerebellar ataxia, unspecified (G11.10), Friedreich's ataxia (with two subtypes listed at G11.11) and other early-onset cerebellar ataxia (with four subtypes listed at G11.19).

"The time I had to present was very short, so I drew on my experience as a genetic counselor and focused on details about FA that would support ways in which a new code would bring greater clarity to the coding structure as a whole. The lunch break followed our segment, and I was approached by one of the coding professionals in attendance who said my presentation was convincing. She also told me we probably wouldn't hear anything else about the status of our request until the new approved codes came out the following year. It was an eye-opening experience as to how difficult it would be to find information on approval or denial of our request," Susan said.

A year later, on October 1, 2020, a new ICD-10-CM code went into effect for FA, G11.11. The new code was too similar to the previous code, but it felt to Susan like a victory, nonetheless. To help spur adoption, FARA issued a press release, posted a short video describing the importance of using G11.11 for FA and created fact sheets for use by families and health care professionals to migrate to the new code. "Working to remind providers to simply add another '1' to the code is worth it, if by doing so patients will experience improved clinical management. Over time, we hope it helps us better track the overall prevalence of FA, the onset of various related symptoms and hospitalizations, and that it will expedite coverage and reimbursement of the care FA patients rely on, especially when disease-modifying treatments for FA come to market."

Visit FARA's ICD code resources.

Ambassadors for the Friedreich's Ataxia Research Alliance actively raise awareness about the patient perspective of what it's like to live with FA. With approval of a new ICD-10-CM code for FA in 2020 came the need to spread the news to families, health care professionals, researchers and others.

LIMB GIRDLE MUSCULAR DYSTROPHIES

With 34 known distinct genetic subtypes of limb girdle muscular dystrophy (LGMD), **Paul Melmeyer**, vice president for public policy and advocacy at the Muscular Dystrophy Association (MDA), knew he had his work cut out for him as he set out to develop and advance a consensus proposal for an evidence-based ICD-10 coding structure. His prior work on ICD coding issues while on staff at the National Organization for Rare Disorders (NORD) prepared him for the task. The burgeoning investment in developing gene-specific therapies for the LGMDs made it an urgent one.

Other forms of muscular dystrophy, namely myotonic, Duchenne, Becker and facioscapulohumeral, had specific codes, but LGMD was listed under a code for "Other specified muscular dystrophies." To develop a strawman structure, Paul relied on proposals developed by Parent Project Muscular Dystrophy (for Duchenne and Becker) and other advocacy organizations, as well as coding experts who understood both the science of medical coding and the ICD C&M Committee's expectations and evidence requirements. He reached out to various patient advocacy organizations serving the LGMD community, physicians, researchers and federal agency partners. Paul reported, "I found varied levels of interest in and understanding of ICD codes and their importance, so I followed with education about the process, why it mattered and why they should participate. Ultimately, it yielded a 'big tent' coalition of stakeholders willing to work with MDA, weigh in on proposal drafts and lend their ongoing support to the effort."

> "Among the community stakeholders I approached about developing an ICD-10-CM coding structure, I found varied levels of interest in and understanding of ICD codes and their importance, so I followed with education about the process, why it mattered and why they should participate. Ultimately, it yielded a 'big tent' coalition of stakeholders willing to work with MDA, weigh in on proposal drafts and lend their ongoing support to the effort."
>
> – Paul Melmeyer

The proposal submitted to the ICD C&M Committee in December 2020 recommended a total of 12 subcodes: this includes six for the most prevalent LGMD subtypes and four for other subtypes for which there are advanced clinical therapeutic programs that could result in approved therapies within the next five years. These 10 new subtype specific codes would capture more than half of the LGMD community. According to the proposal, "The remaining LGMD population members will be captured in the two remaining codes we are nominating for LGMD: 'Other genetically confirmed limb girdle muscular dystrophy subtype' and 'Limb girdle muscular dystrophy, unspecified.' The remaining 24 subtypes can be considered for unique ICD-10-CM codes at a later date as therapeutic development advances."

In further support of the nomination, the proposal included a table with prevalence estimates and citations for 8 of the 34 identified LGMD subtypes (see page 13). The proposal states, "It is estimated that each of the other 24-identified LGMD subtypes affect fewer than three per one million individuals, with most if not all affecting fewer than one individual per million. This natural division in prevalence of LGMDs makes for a fairly apparent division between the more prevalent LGMDs that we are nominating for ICD-10-CM codes and the LGMD subtypes that do not meet our prevalence threshold." Two other subtypes, both autosomal dominant forms linked to specific genetic mutations for which the recessive forms met their inclusion criteria. This was done to avoid potential confusion if only the recessive forms had been included.

In keeping with the big-tent strategy set from the beginning, the proposal was submitted under the auspices of nine patient advocacy organizations: Coalition to Cure Calpain 3; CureLGMD2i; Jain Foundation; Kurt+Peter Foundation; LGMD Awareness Foundation; LGMD2i Research Fund; LGMD2L Foundation; Muscular Dystrophy Association; and The Speak Foundation. It was also signed by the 10 clinical experts, most of whom lead the Limb-Girdle Muscular Dystrophy Clinical Research Network (LGMDCRN). It acknowledged contributions made to the process by the team at the Centers for Disease Control and Prevention responsible for MD STARnet, staff of biopharmaceutical companies developing therapies for LGMD subtypes, physical therapists who routinely treat individuals with an LGMD, experts in ICD-10-CM coding practices, medical societies representing medical professionals who treat individuals with an LGMD and other stakeholders.

Creation of codes for LGMD subtypes was among the topics discussed at the March 9, 2021 meeting of the ICD C&M Committee. One of the LGMDCRN signatories, Nick Johnson, M.D., M.S.C.I., F.A.A.N., provided the 10-minute clinical overview of LGMD. David Berglund, M.D., from the ICD C&M Committee staff presented the agency's proposed coding structure, which reflected a different grouping strategy for the 34 subtypes under a total of 11 added codes. At times Dr. Berglund questioned whether the combined subtypes within a grouping would result in combined prevalence of more or less than one case per million. He also invited comments in writing on the overall approach to creation of ICD-10-CM codes for rare conditions.

As this publication went to press, the organizations and individuals involved in developing the initial proposal were coalescing support for the proposal across all LGMD stakeholders by circulating a petition within the patient community, collecting endorsements from medical professionals familiar with LGMD and seeking support from all relevant stakeholder groups.

Click here to read the proposal for ICD codes for LGMDs

Click here to read the community letter of support

The three-minute ICD Code Roadmap video, "How ICD Codes Impact the Rare Disease Community Journey," portrays the story of Sara and her family. After a long odyssey to find a specialist who can diagnose Sara's disease, they learn it doesn't have a diagnostic code, which may complicate the process of getting appropriate care and treatment. The video goes on to describe what an ICD code is and why it matters. Feel free to share it, and all the ICD Roadmap resources, broadly.

SYNGAP1

Hans Schlecht, M.D., M.M.Sc., added "patient advocate" to his list of titles after his son, Erich, was diagnosed via whole exome sequencing with a pathogenic SYNGAP1 variant. This deficiency causes a spectrum of intellectual and developmental delays, seizures, low muscle tone and disordered sleep and behavior. Putting his medical training and professional network to use for the community, Hans joined fellow SYNGAP1 advocates in efforts to advance research and care.

It was at a 2017 meeting hosted by Global Genes where Hans first recognized the opportunity to leverage big healthcare systems' databanks to achieve a better understanding of the prevalence of rare conditions and help families affected by them to connect with one another. "I talked to dozens of people, each of whom attended in hopes of finding another person who might be able to help their loved one with a rare disease. I knew that health systems like Kaiser have huge data warehouses that could be used to identify cases, both for the benefit of research and the individual. Then came the news that Angelman's syndrome had been assigned a unique ICD code. It made me think that ICD codes could be a key to more efficiently identifying SYNGAP1 families and families with other rare forms of intellectual disabilities."

When Erich was diagnosed with SYNGAP1 deficiency, Hans Schlecht put his medical training and professional network to use for the community to advance research and care. Inspired by the example of the Angelman's disease community, Hans set out to secure an ICD-10-CM code for SYNGAP1. Three years later, a code was approved and will go into effect on October 1, 2021.

Upon learning about the painstaking process by which ICD-10 codes in the U.S. are assigned, Hans initially conceived of a means to link different types of coding systems together. For example, the Human Gene Organization (HUGO) Gene Nomenclature Committee (HGNC) is the global authority for approving unique symbols and names for all the building blocks of the human genome to allow for "unambiguous scientific communication." Hans' vision was to tie the symbols used by HGNC together with codes issued under the ICD. Some experts with whom he shared this idea found it intriguing, but ultimately advised him that the ICD-10 code was too "brittle" for such a sweeping change, especially with an international update of the entire coding system coming into effect in 2022.

With that, in 2019 Hans set out to develop a proposal for SYNGAP1 to be assigned a unique ICD code and he helped about a dozen other advocates prepare separate proposals for related causes of intellectual disability. Initial feedback from the ICD Coordination & Maintnenace Committee staff prompted revisions and some of the advocates dropped from the process. Hans submitted a proposal on behalf of the SynGAP Research Fund and coordinated with another proposal from Bridge the Gap, another SYNGAP patient advocacy organization, offering two distinct options for the tabular modifications. One would place it within the section for "G93.49 - Other encephalopathy" as "G93.4902 SYNGAP1 Encephalopathy." The second option would place the new code within "F78 – Other intellectual disabilities" as "F78.02 – Intellectual disability secondary to pathogenic SYNGAP1 variant." This time SYNGAP1 was added to the agenda for the March 18, 2020 meeting of the ICD C&M Committee. Codes adopted as a result of presentations at this meeting would go into effect on October 1, 2021.

At the meeting, physician-researcher Constance L. Smith-Hicks, M.D., Ph.D., presented a five-minute review of the scientific and medical understanding of SYNGAP1. David Berglund, M.D., of the ICD C&M Committee staff followed with a coding proposal based on the second option in Hans' proposal to create a new subcategory, "F78.A – Other genetic-related intellectual disabilities." It also proposed two new codes: "F78.A1 – SYNGAP1-related intellectual disability" and "F78.A9 – Other genetic-related intellectual disability," which would index several genetic variants, including: CHAMP1, HNRNPH2, SATB2, SETBP1 and STXBP1. These were some of the variants for which other advocates had submitted initial proposals. [Note: See page 55-56 of the proposal packet for the specific proposal presented by the agency at the meeting.]

While presenting the SYNGAP1 proposal, Dr. Berglund questioned the known prevalence of the genetic variants that would be indexed under the F78.A9 code. He stated, "Although we do not have absolute cut-offs as to when a rare disease should have a specific separate code, we will invite comment from the public about the issue of how common – as opposed to rare – something should be when we are considering assigning a separate, specific code." He also raised a question about the implications for SYNGAP1 and related conditions if a code was created within the section for intellectual disabilities, as opposed to the one for encephalopathies. "There are over 100 genetic mutations that can cause intellectual disabilities; which ones should we index? How do we choose which to index if we place the SYNGAP1 code here?," he asked. A member of the audience posed a question about such implications, which brought the discussion to an end due to time constraints.

"The news that Angelman's syndrome had been assigned a unique ICD code made me think that ICD codes could be a key to more efficiently identifying SYNGAP1 families and families with other rare forms of intellectual disabilities. I learned first-hand that it's a painstaking process." -- Hans Schlecht, M.D., M.M.Sc.

The COVID-19 pandemic placed new demands on the ICD C&M Committee to keep pace with the rapidly evolving understanding of that disease's effect on human health and how to capture it in the health information system. Hans waited a few months before following up on the March 18, 2020 meeting and subsequent comment period to check in with Committee staff. "I was told no decisions had been made," he said. "Based on their deadlines and once-a-year update process, now the earliest effective date for a SYNGAP1 code could be October 1, 2022. I understand that it's a struggle to determine the best way to implement rare diseases in the ICD code, but we're losing time. The prevalence of SYNGAP1 and health outcomes related to it are being obscured because patients are classified in any number of general or 'not-otherwise-specified' codes. Yet, SYNGAP1 and other genetically determined types of intellectual disabilities are disadvantaged from being assigned a specific code because the prevalence is unknown and possibly below some minimum threshold. Without a code, it is difficult to document their prevalence. That seems like a solvable problem to me."

TABLE 6A - NEW DIAGNOSIS CODES

Diagnosis Code	Description	CC	MDC	MS-DRG
A79.82	Anaplasmosis [A. phagocytophilum]	C	18	867, 868, 869
C56.3	Malignant neoplasm of bilateral ovaries	C	13	736, 737, 738, 739, 740, 741, 754, 755, 756
C79.63	Secondary malignant neoplasm of bilateral ovaries	C	13	736, 737, 738, 739, 740, 741, 754, 755, 756
C84.7A	Anaplastic large cell lymphoma, ALK-negative, breast	C	17	820, 821, 822, 823, 824, 825, 840, 841, 842
			25	974(21), 975(21), 976(21)
D55.21	Anemia due to pyruvate kinase deficiency	N	16	811, 812
D55.29	Anemia due to other disorders of glycolytic enzymes	N	16	811, 812
D75.838	Other thrombocytosis	N	16	814, 815, 816
D75.839	Thrombocytosis, unspecified	N	16	814, 815, 816
D89.44	Hereditary alpha tryptasemia	N	16	814, 815, 816
E75.244	Niemann-Pick disease type A/B	N	10	642
F32.A	Depression, unspecified	N	19	881
F78.A1	SYNGAP1-related intellectual disability	N	19	884
F78.A9	Other genetic related intellectual disability	N	19	884
G04.82	Acute flaccid myelitis	M	01	023, 024, 097, 098, 099
G44.86	Cervicogenic headache	N	01	102, 103
G92.00	Immune effector cell-associated neurotoxicity syndrome, grade unspecified	N	01	023, 024, 091, 092, 093
			15	791(20), 793(20)
G92.01	Immune effector cell-associated neurotoxicity syndrome, grade 1	N	01	023, 024, 091, 092, 093
			15	791(20), 793(20)
G92.02	Immune effector cell-associated neurotoxicity syndrome, grade 2	N	01	023, 024, 091, 092, 093
			15	791(20), 793(20)
G92.03	Immune effector cell-associated neurotoxicity syndrome, grade 3	C	01	023, 024, 091, 092, 093

New diagnosis codes published on the CMS website on April 27, 2021 included codes for SYNGAP1 and "Other genetic related intellectual disability."

Neither the September 2020 nor March 2021 ICD C&M Committee meetings addressed comments or revised proposals related to SYNGAP1 or the other genetic variants causing intellectual disabilities listed in the agency's March 2020 proposal. Yet, on April 27, 2021, the CMS Inpatient Prospective Payment Systems (IPPS) Proposed Rule notice (see page 17 for more about this annual announcement) included a new ICD-10-CM code for SYNGAP1, F78.A1. It also included a code, F78.A9, for "Other genetic related intellectual disability." There are no indexed conditions listed under F78.A9 in this publication, although other updates and indexes may be announced in May or June, consistent with the usual schedule for full information on new codes.

Looking ahead to implementation of these codes and the potential benefit to SYNGAP1 patients, Hans reflected on the collaboration to get to this point. "So much of scientific research is beyond the control of the patient advocate but getting an ICD code is within their direct ability and, as such, should be a high priority for time and effort," he said.

DUCHENNE AND BECKER MUSCULAR DYSTROPHIES

Annie Kennedy became familiar with ICD codes early in her career when she was working in a neuromuscular clinic. She saw how codes were applied, singularly and in combination, to document patients' health statuses and to help them gain access to needed treatment and services. Years later, in 2010, while working on a study of the economic burden of ALS, spinal muscular atrophy (SMA), Duchenne muscular dystrophy and myotonic muscular dystrophy, Annie first came to appreciate how vital these codes are to research. "Our analysis of direct and indirect costs attributable to the four diseases relied on ICD codes. There wasn't a specific code for Duchenne, so those costs were 'hiding' within the general muscular dystrophy code and impacted our ability to develop distinct evidence around each disease community," she said.

A couple years after that, she was working with the CDC to attempt to identify 100 percent of the cases of Duchenne in South Carolina as a pilot project under the MD STARnet surveillance system. Annie saw firsthand how slow and expensive the process was. De-identified information was being manually extracted from physician's offices and then a sophisticated algorithm had been developed to confirm cases of Duchenne for data entry. "The next phase of that project was going to involve additional states. This surveillance data was critical and powerful but to expand beyond the few states we currently were working in, we would need to get a specific ICD code for Duchenne. That lit a fire." Annie joined Parent Project Muscular Dystrophy (PPMD) at about this same time, and PPMD founder and CEO, Pat Furlong, immediately gave Annie the green light to work on getting Duchenne a code.

While she was assembling the coalition of stakeholders, attending ICD C&M Committee meetings as background and talking with other advocacy leaders (including Dr. David Fagjenbaum, see page 22) about their experiences developing coding proposals, the first genetically targeted therapy for Duchenne was approved in September of 2016 and launched into a complex access environment. "While we'd understood that patient access wasn't going to be automatic, we had not anticipated just how difficult it would be. We found ourselves engaging closely with public and private payers around the available data that could help inform decision-making around patient community eligibility and clinical outcomes. In one instance, a state's Medicaid program was reviewing an FDA-approved therapy for a small subpopulation of Duchenne patients and had estimated that 500 patients in that state would be eligible for the therapy. Given the potential budgetary impact, they told us it would be unlikely the product would be covered because their state just couldn't absorb the costs for the 500 patients they estimated would be eligible for treatment. Shocked by that number, we learned they had used the ICD code for "general" muscular dystrophy to make their determination. That included a lot more people than would have the specific genetic subtype of Duchenne that the therapy was targeted to treat," Annie recounted. Working with MD STARnet and PPMD's provider network, they determined that just **three** individuals in the state would be eligible for the new therapy. That crucial information led to a commitment to cover the treatment; it also underscored the urgent need for a unique ICD code to avoid this happening in 49 other states.

PPMD worked swiftly with other advocacy organizations, clinicians, researchers and a coding professional to develop a proposal. "We had to determine whether to request two separate codes for Duchenne and Becker or to combine them under one code." Annie explained further, "The clinical understanding of the two subtypes of muscular dystrophy was evolving fast, and we didn't want to risk the possibility that there might be unintended consequences around treatment and benefit eligibility for individuals living with Becker if they had a separate code. Ultimately, we decided to request a single code for Duchenne or Becker (combined), along with a separate code for facioscapulohumeral muscular dystrophy." The proposal was submitted, reviewed and approved in 2017, and the codes became effective on October 1, 2018.

> "It is gratifying to have data as confirmation of the importance of the work we – and so many other leaders – have done on behalf of their communities to improve access to care for individual patients, better understand care outcomes and improve research efforts. While few may know what a powerful tool an ICD code can be, the time we dedicated to refining the ICD code for Duchenne served as a force-multiplier for our community."
>
> – Annie Kennedy, Chief of Policy and Advocacy, the EveryLife Foundation

Annie Kennedy (second from left, among Duchenne community advocates) led Parent Project Muscular Dystrophy's effort to compile the evidence and engage a wide array of community stakeholders to support the proposal for ICD-10-CM codes for Duchenne, Becker and facioscapulohumeral muscular dystrophies. The proposal was submitted, reviewed and approved in 2017, and the codes became effective on October 1, 2018.

To help understand the impact of the new code over the first two years of implementation, Sarepta, the company who has developed and secured approval for three targeted muscular dystrophy therapies to-date conducted an analysis, drawing on consultations with clinical experts, publications, claims data and coverage policies. They determined that the code for Duchenne and Becker is widely used and is cited in coverage policies for drug therapies, genetic testing and nerve conduction testing. Clinicians report 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 also reduces administrative burdens by making billing easier. Comparative studies of Duchenne and Becker are hampered by the use of a single code for both, but the combined data set is growing and the overall benefits, so far, seem to outweigh drawbacks. Additionally, the code has helped to develop a more complete understanding of the patient journey, identify physicians and care centers with newly diagnosed patients and refine patient censuses by state and age.

"As a patient organization, it is sometimes hard to know which efforts to direct the precious commodities of time and resources towards. It is gratifying to now have this data as confirmation of the importance of the work we – and so many other leaders – have done on behalf of their communities to improve access to care for individual patients, better understand care outcomes and improve research efforts. While few may know what a powerful tool an ICD code can be, the time we dedicated to refining the ICD code for Duchenne served as a force-multiplier for our community," Annie stated with conviction.

SECTION 5: RESOURCES

The following is a list of resources referenced in this guide and other components of the ICD Code Roadmap.

- **ICD Code Roadmap video**
- **ICD Code Roadmap Frequently Asked Questions**
- **ICD Code Roadmap Readiness Assessment Tool**

Resource	Purpose
American Academy of Professional Coders (AAPC)	Learn more about coding and coding experts
American Health Information Management Association (AHIMA)	Learn more about coding and coding experts
BoardSource	Learn more about best practices for non-profit board governance
Council of Medical Specialty Societies	Ensure you are coordinating with all the medical specialty societies related to a condition
Federal Register	Find notices of new codes when announced annually in April
Guidelines for Coding and Reporting Using the ICD-10-CM (2021)	Updates to the ICD-10-CM official guidelines for 2021 (including COVID-19 updates)
ICD-10-CM Browser	Locate existing codes in the US ICD-10-CM system
ICD-10-CM Official Guidelines for Coding and Reporting	Obtain details on ICD-10-CM coding structure, conventions and implementation
ICD-9-CM	Identify whether the disease has a code history from the prior version of ICD codes
ICD-11 WHO Browser	Locate codes in the WHO ICD-11 system that will be available to countries ready to implement it as of January 1, 2022
Instructions for revising ICD-10-PCS (procedure codes)	Learn more about the process for revising procedure codes (ICD-10-PCS) managed by the Centers for Medicare and Medicaid (CMS)
NCHS Information on Proposals for New Codes	Understand the review team's expectations and process for handling ICD-10-CM proposals
NCHS Prior Proposals	View examples of how the ICD C&M Committee meetings are structured and proposed coding structures for other diseases
NIH Genetic and Rare Diseases Information Center (GARD)	Determine if a clear description of the condition is available through this important NIH resource that is used by the coding community
PubMed.Gov	Search the published medical literature for diagnostic criteria and clinical descriptions
WHO ICD Homepage	Learn more about the international context for ICD codes

Rare Disease Patient Advocacy Organization Experiences with the ICD-10-CM Process	Source
Angelman syndrome	Article from The Atlantic magazine
Castleman Disease	News announcement from Castleman Disease Collaborative Network
CDKL5	Blog from Dracaena Consulting (written by Ana Mingorance of the LouLou Foundation)
Dravet Syndrome	Blog from the Dravet Syndrome Foundation
Duchenne Muscular Dystrophy/Becker Muscular Dystrophy	News announcement from Parent Project Muscular Dystrophy
Friedreich's ataxia	Friedreich's Ataxia Research Alliance new code resources
Limb-Girdle Muscular Dystrophy ICD code proposal and community letter of support	Proposal materials from the Muscular Dystrophy Association
Prader-Willi Syndrome	Blog from the Foundation for Prader-Willi Research
SYNGAP1	Blog from SYNGAP Research Fund

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ALEXION

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