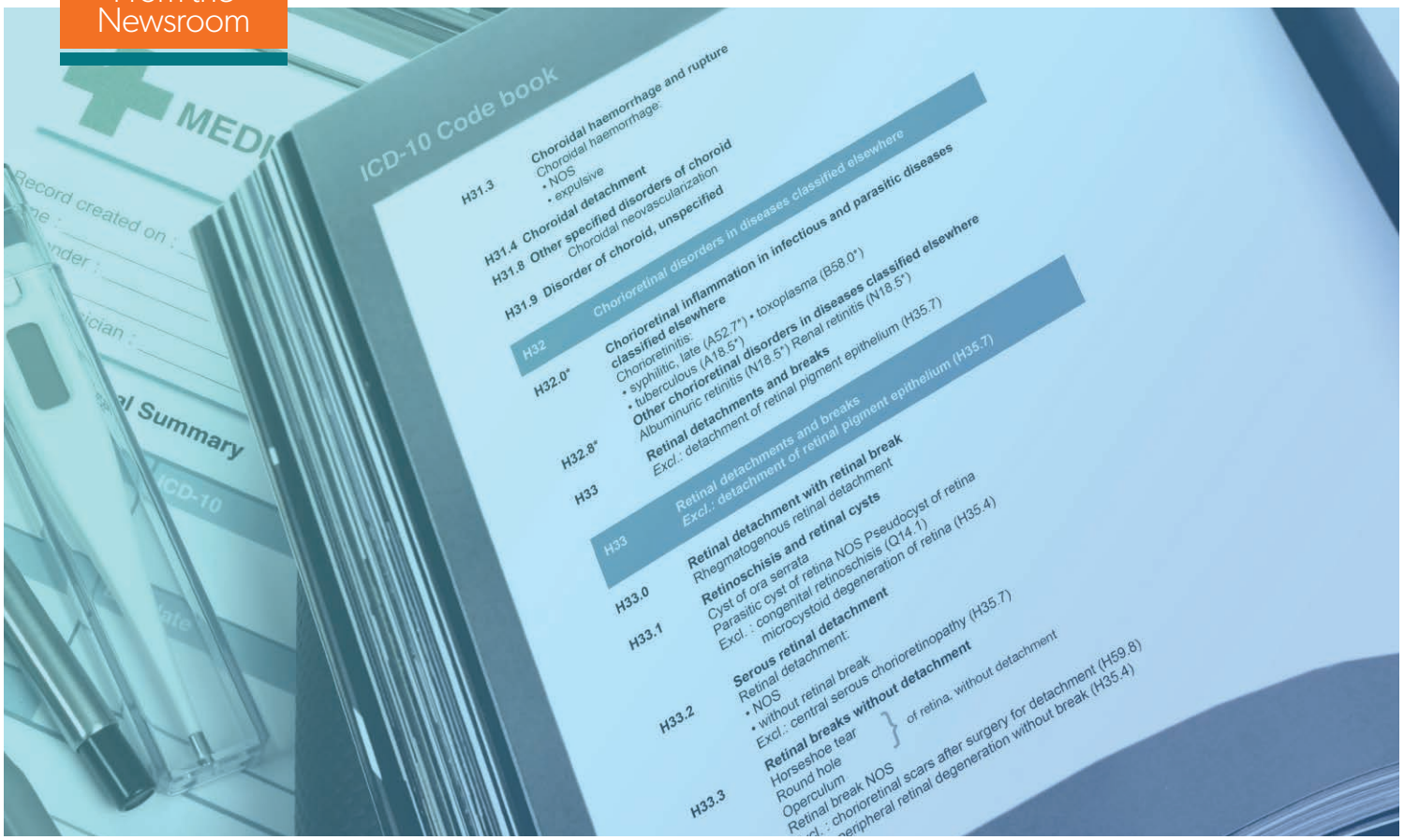

Genomic Medicine Adoption May Be Hindered by Shortcomings in ICD-10 Coding

Medical geneticists and bioinformaticians seek coding systems that can support accurate genotyping, disease causation, and reimbursement alike.

By Neil Versel

THE INTERNATIONAL Classification of Diseases 10th Revision (ICD-10) and its predecessors have been essential code sets in medical records for decades, but geneticists, medical informaticians, and coders alike are finding the current system to be wholly inadequate for the age of precision medicine.

Many genetic and rare diseases do not have corresponding ICD-10 codes, creating a barrier to wider adoption of precision medicine because a lot of healthcare providers are not able to bill for genetic testing or for treatment of certain rare genetic diseases.

"A lot of times even when there is an ICD code, the clinicians don't use them because they won't get billed. It's for insurance purposes," said Melissa Haendel, chief research informatics officer at the University of Colorado Anschutz Medical Campus. "We need … to have a standard that allows you to code patients, but not at the expense of insurance."

The system used in the US, called ICD-10-CM – for Clinical Modification – contains more than 90,000 codes.

In a 2021 paper in *NPJ Genomic Medicine*, researchers affiliated with Illumina discussed difficulties in diagnosing rare hereditary diseases in children after reviewing the entire ICD-10-CM code set. Only about 500 codes could be confidently associated with a genetic disorder, and those tended to be more common ailments like sickle-cell disease and cystic fibrosis.

"In the rare disease area, we can't even count how many patients there are with most genetic disorders because they're not coded in the administrative databases," said one of the authors, John Belmont, a former senior principal medical scientist at Illumina who now consults on genomic and precision medicine. "We can only use indirect measures of doing the epidemiology."

Haendel said that she often gets approached by patient foundations and other rare-disease advocacy groups about getting their conditions added to ICD-10-CM. While she called this "not necessarily a bad thing," it is inefficient and unsustainable "if every single individual group has to go and beg for their code."

Even when ICD codes do exist, they are not always useful for diagnostics or for ontology developers like Haendel. "It does me no good to have [an ontological] term for Fanconi anemia," she said. Although there is an ICD-10-CM code for Fanconi anemia, none of the nearly two dozen subtypes of that disease are there, making it difficult to use ICD-10 for differential diagnoses.

Plus, because ICD-10-CM is largely used for billing, coders often will pick the code that will produce the highest allowable reimbursement for a given patient's insurance plan. "If your insurance will not be billed for Fanconi anemia, then the person doing the annotation might put a different code," said Haendel.

Sometimes, the coding is done simply to support a higher level of reimbursement, regardless of whether it is medically accurate.

This shortcoming in ICD-10-CM reflects the environment in which it was developed.

"ICD-10 is really a classification, and it's not intended to specifically identify every clinical concept," explained Sue Bowman, senior director of coding policy and compliance at the American Health Information Management Association (AHIMA).

"It's not going to have a unique code for every medical concept because classifications, by definition, group things into buckets," Bowman said. Many rare conditions do end up in what ICD-10 considers "residual" categories.

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That said, the current ICD-10-CM contains thousands more codes than it did when it became mandatory in the US in 2015. Interest groups can petition for the creation of new codes.

"There have been a fair number of rare genetic-based diseases added to the classification over the years," Bowman said, however. "There is quite an extensive section on chromosomal abnormalities."

The Centers for Disease Control and Prevention's (CDC) National Center for Health Statistics is in charge of maintenance for diagnostic codes in ICD-10-CM. The Centers for Medicare and Medicaid Services (CMS) manages procedure codes in ICD-10-CM. The process is rather *ad hoc*, as not every request is accepted due to insufficient public support for a change.

Conditions that do not have a unique ICD-10-CM code are indexed. "At least coders are consistently assigning the same code for the condition and have some direction on doing that," Bowman noted. "But, of course, there's other things that are also grouped into that same code." That creates ambiguity.

Meanwhile, vendors of electronic health records and leaders of precision medicine programs alike have been lamenting a lack of adequate standards to support efficient use of genomic data in clinical care. There is no standardization of genetic phenotypes, making it difficult to express genotypes in EHRs.

Some of this problem is being addressed by the Global Alliance for Genomics and Health (GA4GH) and by Fast Healthcare Interoperability Resources (FHIR), an interoperability standard from Health Level Seven International (HL7). While FHIR has had a genomics add-on since 2019 and major vendors have incorporated it, adoption is spotty among the end users – hospitals and healthcare networks.

"The question is, how do you represent genomic data?" according to Christopher Chute, chief research information officer at Johns Hopkins University. "I don't think it should be how do you represent genetic data in the codes. I think it should be how do you represent genomic data in patient records."

The goal is to "represent unambiguously rare diseases in patient records," with exact variants associated with genetic diseases, according to Chute, who chaired the World Health Organization (WHO) committee in charge of revising ICD-10 into version 11.

Chute said that ICD-11 is architected differently than earlier revisions, underpinned by an acyclic graph semantic network, making it a "strict monohierarchy" for statistical calculation. He called ICD-10 "really a warmed-over version of the architecture of 9," an issue addressed in Revision 11.

ICD-11 can also postcoordinate, which means that it can group together disparate concepts to create meaningful answers to medical questions. As an example, the system can take findings of lung cancer in the upper-right lobe and match it with characteristics of adenosquamous cell carcinoma to "compose a clinical sentence," according to Chute.

"There's no reason why you couldn't postcoordinate it with genomic characteristics as well," added Chute, who chairs the Medical Scientific Advisory Committee for the WHO.

"ICD-11 has the capacity to squeeze that all into its coding system, but it's not entirely clear to me that that's the best way to capture this information," Chute said.

It may take years for it to become the standard in the US, though. At the international level, ICD-11 went into effect at the beginning of 2022, but it is up to each country to adopt the new revision.

The transition from ICD-9 to 10 in America was slow, painful, and expensive. ICD-9-CM had been the American coding standard

since 1979, and it took until 2015 for CMS to require the switch to 10 after years of lobbying-induced delays.

"It was an unfunded mandate," Chute said. He estimated that it cost the US healthcare industry $150 billion to upgrade its technology and processes to handle ICD-10, which initially had about five times the number of codes as Revision 9 and now has more.

AHIMA's Bowman is more optimistic about the chances of ICD-11 coming to the US in the next few years. "ICD-11 does solve a lot of the problems [regarding genomics] because the structure of ICD-11 is quite different from ICD-10," she said.

Notably, it assigns a unique identifier to every distinct clinical concept. While there may not be an ICD-11 code for every genetic disease, the greater specificity regarding clinical concepts makes it easier to link the coding database to a nomenclature such as Orphanet, which covers rare diseases and orphan drugs. ICD-10 lacks this so-called "foundation layer."

Bowman believes that much of the resistance that existed during the transition from ICD-9 to ICD-10 last decade has dissipated. "I think a lot of the medical community has seen the value of greater detail and specificity in a coding system," she said.

Bowman suggested that the COVID-19 pandemic may have slowed the transition to ICD-11 because the WHO and governments around the world have been preoccupied for nearly three years now.

"I don't think it's going to be 20 years away," Bowman said. But major issues that need to be addressed include whether the US needs its own clinical modification for ICD-11, as it did for revisions 9 and 10.

Money still talks

Regardless of progress with interoperability standards like GA4GH and HL7 FHIR, healthcare providers in the US cannot be reimbursed by third-party payors without ICD-10 codes.

Since they expect it to be a generation or more before ICD-11 takes hold in America, Haendel said that she and Chute have been proposing a hybrid approach, where ICD-10 is a "launching spot" for insurance authorization, while another system provides richer clinical descriptions in EHRs.

Haendel is principal investigator of the Monarch Initiative, an open-source bioinformatics platform for matching phenotypes to genotypes. That initiative has produced Monarch Disease Ontology, or MonDO, a coding system that, among other things, characterizes rare diseases.

MonDO is trying to coordinate with rare-disease communities to develop a consensus on how to name new diseases and then disseminate that information in a computational tool that is compatible with ICD-11 when that becomes the standard in the US, according to Haendel. "In that way, we overcome that challenge of each foundation having to go to ICD one at a time to ask for their disease to be added," she said.

In the meantime, Chute said that the informatics community should not have to look to express everything in a single code. "We're really talking about a matrix of information," he said.

That matrix might include MonDO, Orphanet, Online Mendelian Inheritance in Man (OMIM), the Human Phenotype Ontology (HPO), or Phenopackets.

> But unless the EHR and billing systems are integrated and automated so that coding is simple, physicians are not going to want to take on an extra administrative burden. Clinicians have pushed back for years about additional steps that EHRs require of them.

Phenopackets, a standard for sharing disease and phenotype information for diagnosing and treating rare and hereditary diseases including cancer, was approved by the International Organization for Standardization (ISO) last year. Both Haendel and Chute were involved in its development.

"You really want to be able to have a Phenopacket-esque structure in a standardized framework inside the electronic health record," Chute said, adding that FHIR is a close approximation.

That might not happen as long as ICD-10-CM is the standard in coding, according to Chute. "Can 10-CM be fixed in its current form? I wouldn't know how to do it," Chute said.

Belmont, vice chair of the Health Economics Committee for the American College of Medical Genetics and Genomics (ACMG), said that ICD-10 classifies conditions based on "type of" relationships, such as which bone in the leg is broken. Genetics is more about "something is caused by," he added.

"You have a gene variant and it causes cardiomyopathy, for instance, or it causes intellectual disability," Belmont explained.

This causal focus on disease mechanisms is where medicine is headed, according to Belmont. "My belief is that we need to reorganize the ontology so that it's built more about our way of thinking about diseases," he said.

While he spoke to GenomeWeb as an independent consultant rather than a representative of ACMG, Belmont said that the Health Economics Committee has an interest in genomics-related ICD-10 questions, including how medical geneticists can be properly reimbursed for their services.

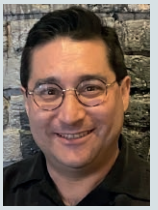
Belmont, who maintains an academic appointment in pediatrics and molecular and human genetics at Baylor College of Medicine in Houston, said that there needs to be more "long-term conversation and integration" between the medical genetics and the bioinformatics communities. "In some ways, we've been developing in parallel, but not really interacting enough," he said.

Belmont noted that in his clinical practice at Texas Children's Hospital, physicians do not handle coding. "There are other practices where the doctor is actually picking the codes," he said. "That's the opportunity, I think, for the doctor to pick a more specific code for a genetic disorder if they have found one."

But unless the EHR and billing systems are integrated and automated so that coding is simple, physicians are not going to want to take on an extra administrative burden. Clinicians have pushed back for years about additional steps that EHRs require of them.

Belmont said that there is no consensus yet within the medical genetics community about how to express genetic subtypes of diseases in EHRs.

He called MonDO, HPO, OMIM, and Orphanet "really excellent," but not used in EHRs often enough. "When I could, I would write down the OMIM number that was associated with the diagnosis I was making, but people typically don't do that," Belmont said. PMQ



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Neil Versel is an editor at GenomeWeb, specializing in bioinformatics and data management in life sciences and healthcare. He has covered the healthcare technology industry for more than 20 years. Prior to joining GenomeWeb, his work appeared in various publications, including *US News & World Report*, Forbes.com, and the *Chicago Sun-Times*. He was an invited delegate to the Rockefeller Foundation's 2008 Making the eHealth Connection conference series in Bellagio, Italy, and is a former board member of Health eVillages and of the Multiple System Atrophy Coalition. A graduate of Washington University in St. Louis, Neil is based in Chicago.