Dear ICD-10-CM Coordination and Maintenance Committee,

I am writing to you on behalf of CureGRIN Foundation, an organization that is dedicated to finding treatments and cures for GRIN, GRIA, GRIK, and GRID disorders.

I'm writing to urge the CDC to support the proposed new ICD-10 category for Genetic Neurodevelopmental Disorders presented by Dr. Berglund at the March 2024 ICD-10-CM Coordination and Maintenance Committee Meeting.

These diseases are not adequately described by existing ICD-10-CM codes. Lack of a unique code for these disorders results in numerous challenges, including inability to accurately track incidence and prevalence, difficulty in researching genotype-phenotype correlations, and complicating the development of protocols for standard of care.

Rare Epilepsy

The inclusion of gene-based ICD codes for disorders representing rare epilepsy has the potential to improve patient care, improve communication amongst care teams, and allow for tracking of individual diseases. We want to specifically highlight a study by Dianalee McKnight et al. published in JAMA Neurology (doi:10.1001/jamaneurol.2022.365) that shows that a genetic diagnosis for patients with epilepsy changes patient care in half of all cases. Thus, an ICD-10-CM code that identifies the genetic underpinnings for epilepsies has the potential to improve patient care.

Treating Genomes

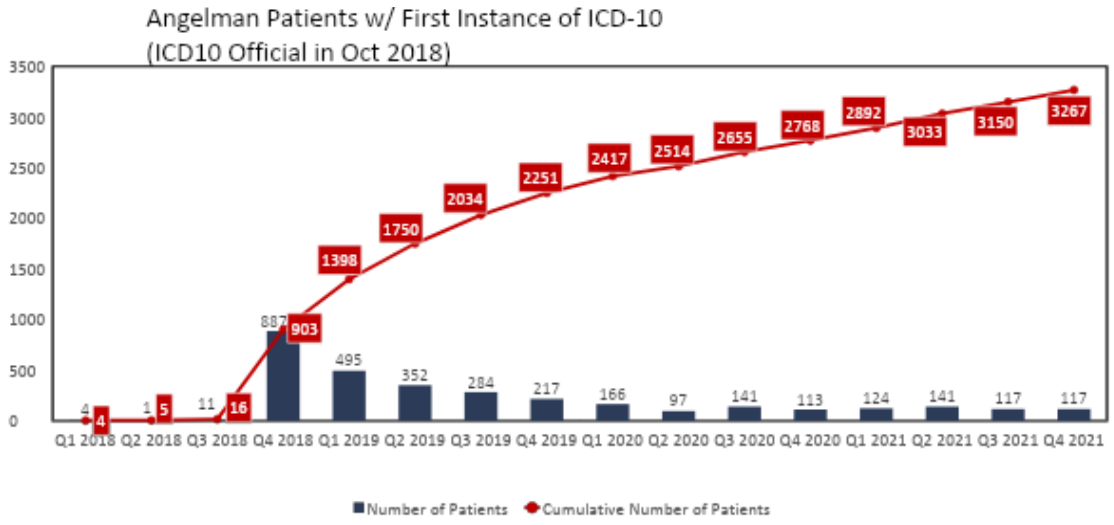
Tracking diseases by symptom or syndrome makes sense in a world where physicians are treating symptoms. But most experts predict that the future of healthcare for genetic conditions will focus as much on either i) targeting the genome through genetic medicine, or ii) targeting the mechanism, which is correlated with the affected gene. Gene-based ICD-10 codes are especially important in 2024, given the high number of precision therapies in development targeting rare epilepsies and neurological diseases. (American Society for Cell & Gene Therapy counts 1,038 pipeline gene therapies being developed for rare disease).

The genetic basis of rare diseases can impact treatment of symptoms in addition to the overall disease. Epilepsy caused by a sodium channel mutation, such as SCN2A mutation, will need to be treated with a sodium channel medication. Ketamine should never be administered to patients with NMDAR mutations, such as GRIN1. Developmental delay stemming from the protein misfolding caused by mutations in the STXBP1 gene will likely benefit from a chemical chaperone. And movement issues caused by a citrate transport deficiency, such as SLC13A5 mutations, will need to be treated with precision medications aimed at citrate transport. Without these specific genetic diagnosis codes in the medical record, it will be difficult to locate, treat, and track the health of patients with rare neurological diseases.

We Know Our Diseases

At the March 2023 ICD-10 Coordination and Maintenance Committee meeting, a clinician speaking on behalf of one of the major physician academies expressed that gene-based codes are irrelevant because parents wouldn't know which gene is affected. Nothing is further from the truth. Parents of children with rare neurodevelopmental diseases know the gene better than their child's birthday. In addition, these genes are represented by strong advocacy organizations primed to partner with all stakeholders to encourage adoption of ICD codes for rare, genetic diseases. This includes communication campaigns to educate our families so they know to mention the new codes to their clinicians at every visit.

The following table, compiled by Ambit Health Data, shows the rapid uptake of ICD-10 codes for Angelman's Disease after their ICD-10 code became official in 2018.



Physicians Want Codes

Physicians speaking on behalf of other organizations have advised the CDC against adding gene-based codes for rare neurological diseases. From our experience, however, the vast majority of physicians who work with our patients are very much in favor of adding these codes for the diseases outlined below, as demonstrated by their presentations to incorporate these codes into the ICD-10-CM at each of the ICD code meetings. These treating physicians tell patients they cannot understand why representatives of the societies that represent them would stand in the way of improving patient care by adding codes. Many have written letters to the CDC to express their views. In addition, other physician associations are fully in support of gene-based codes including the American College of Medical Genetics and Genomics.

We respectfully ask the CDC to support the proposal for the following neurodevelopmental disorders that was discussed at the March 2024 C&M meeting. Alternatively, at the very least, we ask that you do not oppose our application.

The following genes were presented to be considered for codes at the March 2024 C&M meeting.

DLG4	GRIN1	SLC6A1
GRIA1	GRIN2A	STXBP1
GRIA2	GRIN2B	FOXG1
GRIA3	GRIN2D	
GRIA4	SCN2A	
GRIK2	SCN8A	

My son Oliver was diagnosed with GRIN1 in September 2017. Some of the symptoms that he experiences include epilepsy, global developmental delay, intellectual disability, expressive language disorder, hypotonia, ulcerative colitis and cortical vision impairment. At 11 Oliver is non-ambulatory, non verbal, and receives most of his nutrition and medication through a G-tube. He loves the outdoors, music, and conversing in his own way with friends and family. His smile and infectious laughter can light up a room and he makes friends wherever he goes. The joy he brings our family is immeasurable and we will continue to be his advocate and champion, cheering loudly for every inchstone achieved.

Currently, there are no codes to fully describe Oliver's condition. There is an urgent need for ICD-10 to implement codes for GRIA1, GRIA2, GRIA3, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, and GRIN2D.

Thank you for your consideration.

Sincerely,

Rebecca Forsyth