Prevalence, Treatment Patterns, and Healthcare Resource Utilization Among Patients With CDKL5 Deficiency Disorder: Retrospective Analysis of US Claims Data

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OVERVIEW

QUESTION

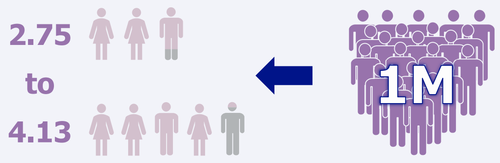
What is the prevalence of cyclin-dependent kinase-like 5 (CDKL5) deficiency disorder (CDD) in the United States? What are the clinical characteristics, treatment patterns, and healthcare resource utilization (HCRU) burden of patients with CDD?

INVESTIGATION

Patients with CDD (and comparator groups of patients with epilepsy and Dravet syndrome [DS]) were identified from the US Komodo Healthcare database. Patients had ≥2 diagnosis claims (CDD: G40.42; epilepsy: G40 excluding G40.42; DS: G40.83 excluding G40.42) during the study period (1/1/2020–6/30/2025) occurring ≥1 month apart, with the first claim falling within the patient selection window (1/1/2020–12/31/2024).

RESULTS

Estimated prevalence of CDD in the United States[a]

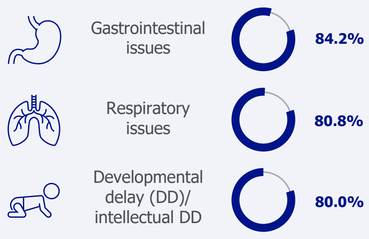


Cases of CDD/million population in 2024

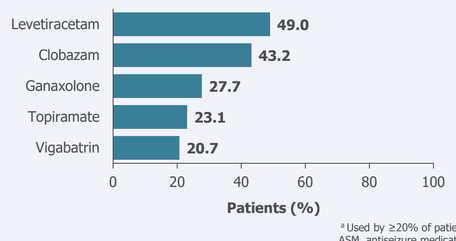
[a]Restricted to cases with ≥2 CDD diagnosis claims ≥1 month apart (prevalence is higher with 1 CDD diagnosis claim). Range reflects 40-60% claims capture rate.

Clinical characteristics and ASM treatment of CDD cohort (N=584)

Most common non-seizure comorbidities across the study period

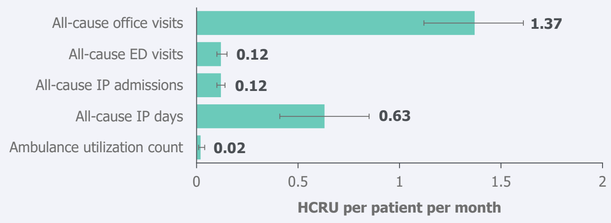


Use of medications to treat seizures over the study period[a]



[a] Used by ≥20% of patients.
ASM, antiseizure medication.

HCRU burden in the matched CDD cohort post-CDD diagnosis (N=368[a])



[a]Data from comparison with matched CDD and DS cohorts over the 6-month post-index period; CDD cohort shown.
CDD, cyclin-dependent kinase-like 5 deficiency disorder; DS, Dravet syndrome; ED, emergency department; HCRU; healthcare resource utilization; IP, inpatient.

CONCLUSIONS

- Prevalence of CDD in the United States was 2.75-4.13/million population and highest for females. Comorbidities among the CDD population were prevalent, particularly gastrointestinal issues, respiratory issues, and developmental delay (DD)/intellectual DD. Levetiracetam and clobazam were the most common medications used to treat seizures over the study period. Per patient per month HCRU among patients with CDD was high, suggesting substantial HCRU burden for this patient population.

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Background

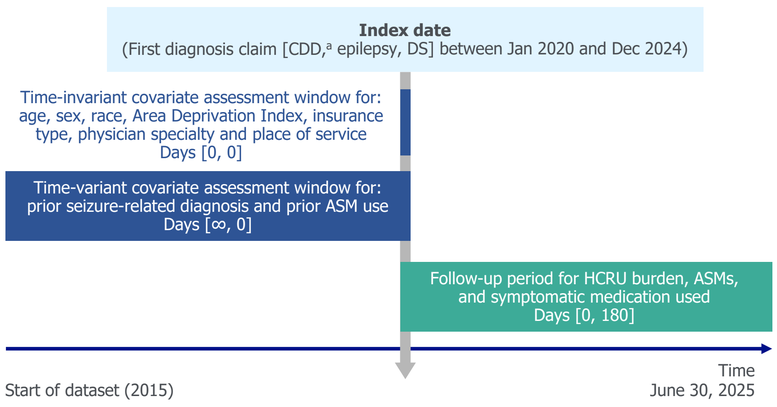
- Cyclin-dependent kinase-like 5 (CDKL5) deficiency disorder (CDD) is an ultra-rare developmental and epileptic encephalopathy (DEE) caused by pathogenic variants in the *CDKL5* gene.
- CDD is characterized by early-onset, antiseizure medication (ASM)-resistant seizures and significant developmental delays.[1]
- The incidence of CDD, based on limited information, is estimated at 1:40,000-1:60,000 live births.[2]
- Published evidence on CDD is currently limited, with notable gaps in evidence related to epidemiology, treatment patterns, and healthcare resource utilization (HCRU).

Objective

- To determine the prevalence of CDD and the clinical characteristics, treatment patterns, and HCRU burden in patients with CDD in the United States.

Methods

- In this retrospective study, patients with CDD, epilepsy, and Dravet syndrome (DS) were identified from the US Komodo Healthcare database, which captures 75-80% of US medical claims and 65-75% of pharmacy claims across regions.
- Eligible patients had ≥2 diagnosis claims (*International Classification of Diseases, Tenth Revision [ICD-10]* codes of G40.42 for the CDD cohort, G40 [excluding G40.42] for the epilepsy cohort, and G40.83 [excluding G40.42] for the DS cohort) occurring ≥1 month apart, with the first claim falling within the patient selection window (1/1/2020–12/31/2024).
- The study period was 1/1/2020–6/30/2025.
- Patients were followed until end of the study period, death, or disenrollment.



[a]The *ICD-10* code for CDD was approved in January 2020. Therefore, the selection period for patients with CDD was defined as January 1, 2020, through December 31, 2024. ASM, antiseizure medication; CDD, cyclin-dependent kinase-like 5 deficiency disorder; DS, Dravet syndrome; HCRU, healthcare resource utilization; *ICD-10, International Classification of Diseases, Tenth Revision.*

- Outcomes were annual CDD prevalence in the United States over the study period, clinical characteristics of patients with CDD, treatment patterns among patients with CDD, and HCRU burden for patients with CDD vs matched epilepsy and DS cohorts.
  - Prevalence required ≥2 CDD diagnosis claims ≥1 month apart (a conservative approach vs 1 claim).
- Descriptive analyses were used to summarize data.
- HCRU burden was compared between the CDD cohort and the matched (1) DS cohort and (2) non-CDD epilepsy cohort in the 6 months post-diagnosis.
  - The epilepsy cohort is larger and more heterogeneous than the other cohorts. The CDD cohort was matched with epilepsy and DS cohorts based on age, sex, race, insurance, state of residence, Area Deprivation Index, and quarter/year of diagnosis using propensity score matching.
  - T-tests and nominal p-values are reported for matched group comparisons.
- For HCRU outcome, patients ≥1 year of age were required to have 6 months of continuous enrollment post-index date, allowing up to 60-day gaps.
  - For patients <1 year of age, continuous enrollment was not required (Komodo assigns January 1 of the birth year as the diagnosis date for all diagnoses in that year, including the index date, so requiring enrollment would exclude infants born later in the year and introduce bias).

Results

PREVALENCE

- In 2024, CDD prevalence ranged from 2.75 to 4.13 per million US population (Komodo capture rate of 40-60%; 50% rate was 3.33/million).
  - CDD was highest in females <18 years of age (17.3/million).
  - CDD accounted for 0.03% of all epilepsy and DEE diagnoses.

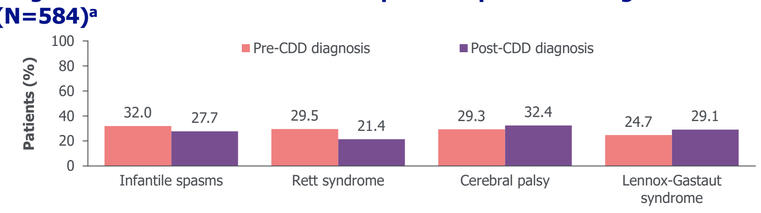
CLINICAL CHARACTERISTICS OF THE CDD COHORT

Patient demographics and characteristics

	CDD cohort (N=584[a])
Age, mean (95% CI), median, years	19.1 (17.2, 21.1); 10.0
Age category, n (%)	N=583
<1 year	29 (5.0)
1-2 years	94 (16.1)
3-11 years	198 (34.0)
12-17 years	92 (15.8)
≥18 years	170 (29.2)
Sex, n (%)	N=584
Female	415 (71.1)
Male	164 (28.1)
Undefined	5 (0.9)
Race/ethnicity, n (%)	N=425
White	229 (53.9)
Hispanic/Latino	79 (18.6)
Black/African American	54 (12.7)
Unknown/Other	44 (10.4)
Asian/Pacific Islander	19 (4.5)
Insurance group, n (%)	N=545
Medicaid	274 (50.3)
Commercial	182 (33.4)
Medicare	89 (16.3)

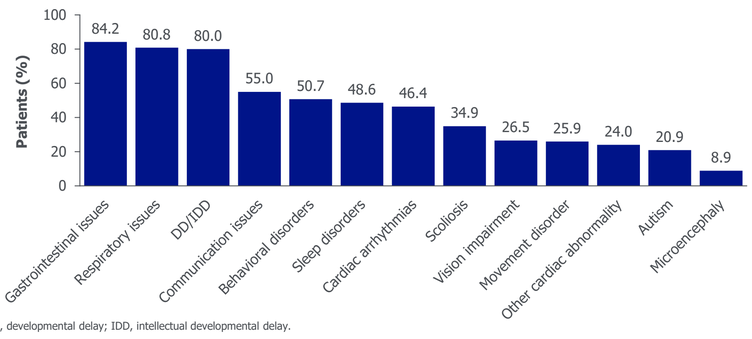
[a]Specific demographic data were missing for some patients and are reflected by lower N values for age category, race/ethnicity, and insurance group.
CDD, cyclin-dependent kinase-like 5 deficiency disorder; CI, confidence interval.

Diagnoses associated with seizures pre- and post-CDD diagnosis (N=584)[a]



[a]Occurring in ≥15% of patients. CDD, cyclin-dependent kinase-like 5 deficiency disorder.

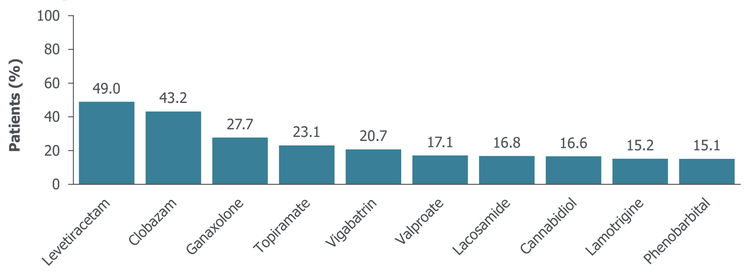
Non-seizure comorbidities across the study period (N=584)



DD, developmental delay; IDD, intellectual developmental delay.

TREATMENT PATTERNS IN THE CDD COHORT

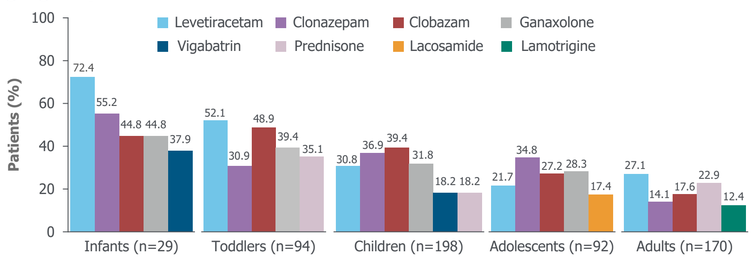
Use of medications to treat seizures over the entire study period (N=584)[a,b]



[a]Used by ≥15% of patients; [b]Clonazepam was excluded due to its use as a rescue medication.

- Levetiracetam (33.9%), clobazam (33.0%), and clonazepam (29.8%) were the most common seizure-related drugs utilized after CDD diagnosis.
- Increases in uptake from pre- to post-diagnosis were seen for ganaxolone (23.5%), fenfluramine (5.1%), and cenobamate (3.3%).

Five most common ASMs/rescue medications after CDD diagnosis, by age group[a]



[a]Infants, <1 year of age; toddlers, 1-2 years of age; children, 3-11 years of age; adolescents, 12-17 years of age; adults, ≥18 years of age.
ASM, antiseizure medication; CDD, cyclin-dependent kinase-like 5 deficiency disorder.

HCRU BURDEN IN CDD, EPILEPSY, AND DS COHORTS (6-MONTH POST-INDEX PERIOD)

- For the CDD (n=569) and epilepsy (n=1136) matched cohorts, there was significantly more burden among patients with CDD as measured by seizure-related HCRU claims, status epilepticus claims, concomitant ASMs, and rescue medication claims ($p<0.05$ for each).
- HCRU was generally similar between the matched CDD and DS cohorts. However, seizure-related claims and office visits were higher for the CDD cohort, while emergency department visits, ambulance use, and number of rescue medications were higher for the DS cohort.

HCRU analysis for matched CDD and DS cohorts[a]

HCRU variable[a]	CDD (n=368) Mean (95% CI)	DS (n=638) Mean (95% CI)	p-value
Seizure-related claims[b]	**11.77 (10.11, 13.43)**	7.69 (6.66, 8.72)	0.000
Status epilepticus claims[b]	1.53 (1.01, 2.05)	1.85 (1.43, 2.27)	0.342
All-cause ED visits[c]	0.12 (0.10, 0.15)	**0.18 (0.16, 0.20)**	0.001
Seizure-related ED visits[c]	0.08 (0.06, 0.09)	**0.14 (0.12, 0.16)**	0.000
All-cause IP admissions[c]	0.12 (0.10, 0.14)	0.11 (0.09, 0.13)	0.262
Seizure-related IP admissions[c]	0.10 (0.09, 0.12)	0.10 (0.09, 0.12)	0.884
All-cause IP days[c]	0.63 (0.41, 0.85)	0.54 (0.35,0.73)	0.560
Seizure-related IP days[c]	0.56 (0.34, 0.77)	0.45 (0.30, 0.61)	0.438
All-cause office visits[c]	**1.37 (1.12, 1.61)**	0.85 (0.74, 0.96)	0.000
Seizure-related office visits[c]	**0.58 (0.45, 0.71)**	0.40 (0.35, 0.45)	0.010
Ambulance utilization count[c]	0.02 (0.01, 0.04)	**0.07 (0.06, 0.09)**	0.000
Avg concomitant ASMs[d]	1.91 (1.74, 2.08)	1.85 (1.74, 1.97)	0.562
No. of rescue medication claims[b]	0.09 (0.07, 0.12)	**0.21 (0.18, 0.24)**	0.000

[a]Variables are reported per patient per month except for average number of concomitant ASMs, which reflects per patient over 6-month follow up; [b]Number of claims; [c]Number of claim days; [d]Number of unique molecules. ASM, antiseizure medication; CI, confidence interval; CDD, cyclin-dependent kinase-like 5 deficiency disorder; DS, Dravet syndrome; ED, emergency department; HCRU, healthcare resource utilization; IP, inpatient.

LIMITATIONS

- Database studies are prone to miscoding and other errors and omissions.
- An *ICD* code for CDD was only approved in January 2020; thus, low use of the CDD code and misclassification due to coding errors are likely.
- Comorbidities (eg, developmental delay [DD]/ intellectual DD) may be underrepresented; providers may have attributed them as part of CDD and are not inclined to code them individually.
- The Komodo database primarily includes individuals with commercial insurance, which may limit the generalizability of findings.
- HCRU burden was an exploratory outcome, as the entire patient journey may not be reflected due to a lag between seizure onset and first G40.42 code use.

CONCLUSIONS

- This real-world, retrospective analysis provides the first report of the prevalence of CDD in a large US claims-based population.
- Estimated CDD prevalence confirms the rarity of this disease; it will be of interest to reassess as the CDD diagnosis code is increasingly used.
- HCRU burden was similarly high for matched CDD and DS cohorts, and higher for CDD vs a matched general epilepsy cohort. Overall, seizure-related claims, status epilepticus claims, and all-cause office visits were the most common HCRU and high with the CDD population, suggesting significant HCRU burden for this population.
- High comorbidity, HCRU, and polypharmacy burden may be associated with lower quality of life and high financial burden among these patients and their caregivers. Studies are needed to assess potential associations.
- Future analyses of datasets are needed to confirm these findings as CDD becomes better diagnosed and properly coded in claims data.

References

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UCB-sponsored. UCB was involved in the design of the study, the collection, analysis, and interpretation of data, and review of the poster. The authors acknowledge Bobby Jacob, PharmD, MPH (UCB, Smyrna, GA, USA) for managing the development of the poster, and Carlee M Cunningham, PhD, and Lynne Isbell, PhD, CMPP (Envision Medical Communications, Fairfield, CT, USA) for writing assistance, which was funded by UCB. Author contributions: X Dong conducted the analysis. J Khushalani designed the study. J Khushalani and M Martin interpreted the results. All authors critically reviewed the poster and approved the final version for presentation. Author disclosures: S Amin received funding from BioMarin, Boston Scientific, CDKL5 UK, Desitin, Ipsen, Jazz, LivaNova, Medtronic, Novartis, Nutricia, PTC, TSA, and UCB. O Devinsky has received grant support from the Centers for Disease Control and Prevention, Multidisciplinary University Research Initiatives, National Institute of Mental Health, National institute of Neurological Disorders and Stroke (NINDS), and National Science Foundation; has equity and/or compensation from Actio, AJNA, Blackrock Neurotech, California Cannabis Enterprises, Empatica, Regel, Script, Tevard, and Tilray; received consulting fees from Emotiv, Praxis, Precision, Stoke, Tennex, UCB, and Ultragenyx; holds patents for use of cannabidiol in treating neurological disorders owned by GW and Jazz; he has waived all financial interests. He holds other patents in molecular biology and is the managing partner of the PhiFund Ventures. X Dong, J Khushalani, and M Martin are employees of UCB with stock ownership. P Öhagen is an external contractor for UCB. RR Rajaraman received speaker fees from Acadia and Jazz; served on scientific advisory boards for Stoke and UCB; received consulting fees from Marinus; received research support from the International CDKL5 Research Foundation and Marinus; received grant support from the Department of Defense and NINDS; was an investigator for GRIN, Jazz, Lundbeck, Marinus, Roche, UCB, and Ultragenyx. S Demarest consulted for BioMarin, Capsida, Encoded, Longboard, Mahzi, Marinus, Neurogene, Ovid, Tysha, UCB, and Ultragenyx; received funding from Mila's Miracle Foundation, the National Institutes of Health, and Project 8P; was an advisory board member for non-profit foundations (FamilieSCN2A, N of 1 Collaborative, Project 8P, Rare X, Ring14 USA, and SLC6A1 Connect).

55th Child Neurology Society Annual Meeting
Montréal, QB, Canada | October 14-17, 2026
Originally presented at American Academy of Neurology 78th Annual Meeting
Chicago, IL, USA, April 18–22, 2026