Costs, Healthcare Resource Utilization, and Patient Characteristics Associated With CDKL5 Deficiency Disorder: A Retrospective Analysis of the US Closed Claims Database MarketScan

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Poster P5-008

Background

- Cyclin-dependent kinase-like 5 (CDKL5) deficiency disorder (CDD) is an ultra-rare X-linked developmental and epileptic encephalopathy caused by pathogenic variants in the *CDKL5* gene.
- Literature on CDD is currently very limited, with specific evidence gaps around patient and clinical characteristics, healthcare resource utilization (HCRU), and costs.
- Syndrome-specific International Classification of Diseases (ICD) codes for CDD and Dravet syndrome (DS) were introduced in 2020, permitting research from large national US claims databases.

Objective

- To describe patient characteristics, HCRU, and costs associated with a cohort of patients with CDD captured in the US MarketScan database.
- To help contextualize results for CDD, outcomes were compared with:
  - A cohort of patients with non-CDD epilepsy, and
  - A cohort of patients with DS.

Methods

DATA SOURCE AND STUDY DESIGN

- This was a retrospective cohort study using de-identified administrative claims data from the Merative MarketScan Commercial, Medicare, and Medicaid databases.
- Patients were selected for inclusion if they had:
  - ≥2 claims with 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,
  - Claims occurring ≥1 month apart during the patient selection period (7/1/2020-3/31/2024 for Commercial and Medicare, 7/1/2020-6/30/2023 for Medicaid).
  - ≥6 months of continuous enrollment (medical and pharmacy coverage) prior to first recorded claim for CDD/epilepsy/DS, allowing for ≤60-day gaps.
- The index date was the first date of CDD (epilepsy/DS for other cohorts) diagnosis captured during the patient selection period.
- The follow-up period was from index to 6 months post index; patients were followed until end of continuous enrollment or database end (9/30/2024 for Commercial/Medicare and 12/31/2023 for Medicaid), whichever was earliest.

ANALYSES

- The CDD cohort was matched to the comparator cohorts by payer type and US geographic region using the exact matching method.
  - Up to 5 patients with non-CDD epilepsy/DS for each patient with CDD.
- Patient characteristics, HCRU, and costs of patients with CDD were compared with patients with non-CDD epilepsy, as well as patients with DS.
- Cost analyses were restricted to patients enrolled in non-capitated insurance plans (ie, where healthcare providers are reimbursed based on services delivered; therefore, costs reflect true healthcare utilization).
- An analysis of covariance (ANCOVA) model was used to test for group differences after adjusting for sex, age, and insurance plan.
- Costs were converted to 2024 US dollars using the medical care component of the Consumer Price Index.[1]

Results

PATIENTS

- Overall, 56 patients with CDD met the eligibility criteria, of whom 52 completed 6 months of follow-up.
- Among 45 patients enrolled in non-capitated insurance plans, 42 had 6 months of follow-up.

Overview

QUESTION

What are the patient characteristics, healthcare resource utilization (HCRU), and costs associated with cyclin-dependent kinase-like 5 deficiency disorder (CDD), and how do they compare with non-CDD epilepsy and Dravet syndrome (DS)?

INVESTIGATION

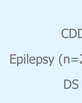
Retrospective cohort study used de-identified administrative claims data from the Merative MarketScan Commercial, Medicare, and Medicaid databases. Cohorts for CDD, non-CDD epilepsy, and DS were identified (7/1/2020-3/31/2024 for Commercial and Medicare and 7/1/2020-6/30/2023 for Medicaid) and compared.
Patient characteristics, HCRU, and costs are reported.

RESULTS

Cohorts (pre matching)

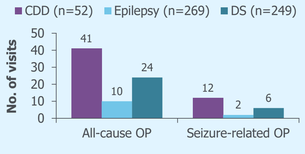
	CDD	Epilepsy	DS
Median age (years):	11.5	34	11
Female:	76.8%	52.3%	49.2%

Payer type

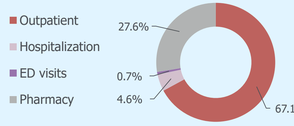


HCRU over 6 months of follow-up

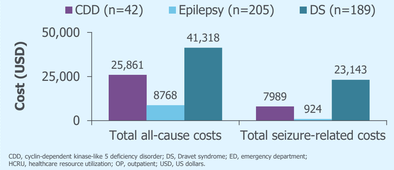
Number of outpatient visits



HCRU contributions to total costs for patients with CDD (n=42)



Median total HCRU costs per patient over 6 months of follow-up



CDD, cyclin-dependent kinase-like 5 deficiency disorder; DS, Dravet syndrome; ED, emergency department; HCRU, healthcare resource utilization; OP, outpatient; USD, US dollars.

CONCLUSIONS

- Total HCRU and costs were high for patients with CDD and DS compared with the general epilepsy population, reflecting a high burden of these developmental and epileptic encephalopathy populations for patients, families, payers, and health systems.
- Patients with CDD had more outpatient visits than the other cohorts, with home health contributing substantially to outpatient HCRU.
- 30.9% of total costs were seizure related for patients with CDD, demonstrating the need for better treatment for both seizure and non-seizure symptoms to reduce the burden for patients, families, and health systems.

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Patient characteristics

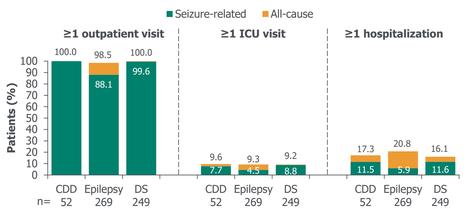
		PRE MATCHING		POST MATCHING[a]	
	CDD	EPILEPSY	DS	EPILEPSY	DS
N	**56**	**236,156**	**453**	**280**	**264**
Age, years, median	11.5	34	11	30.5	11
Age, years, mean (SD)	13.8 (13.1)	35.5 (21.3)	12.1 (8.1)	32.5 (19.6)	12.3 (8.2)
Age group, years, n (%)					
<2	6 (10.7)	2537 (1.1)	23 (5.1)	5 (1.8)	17 (6.4)
2 to <6	9 (16.1)	10,969 (4.6)	86 (19.0)	17 (6.1)	45 (17.0)
6 to <12	13 (23.2)	21,424 (9.1)	135 (29.8)	19 (6.8)	79 (29.9)
12 to <18	14 (25.0)	24,144 (10.2)	95 (21.0)	33 (11.8)	50 (18.9)
18 to <25	6 (10.7)	28,181 (11.9)	85 (18.8)	36 (12.9)	58 (22.0)
≥25	8 (14.3)	148,901 (63.1)	29 (6.4)	170 (60.7)	15 (5.7)
Female, n (%)	43 (76.8)	123,579 (52.3)	223 (49.2)	133 (47.5)	127 (48.1)
Race, n (%) (available for Medicaid only)					
White	20 (35.7)	67,191 (28.5)	168 (37.1)	91 (32.5)	92 (34.8)
Black	2 (3.6)	29,770 (12.6)	36 (7.9)	37 (13.2)	17 (6.4)
Hispanic	4 (7.1)	8121 (3.4)	33 (7.3)	13 (4.6)	17 (6.4)
Other	1 (1.8)	5703 (2.4)	25 (5.5)	3 (1.1)	16 (6.1)
Missing	7 (12.5)	17,788 (7.5)	61 (13.5)	26 (9.3)	28 (10.6)
Missing, not Medicaid	22 (39.3)	107,583 (45.6)	130 (28.7)	110 (39.3)	94 (35.6)
Geographic region, n (%)					
Northeast	2 (3.6)	15,077 (6.4)	21 (4.6)	10 (3.6)	10 (3.8)
North Central	6 (10.7)	30,068 (12.7)	30 (6.6)	30 (10.7)	25 (9.5)
South	8 (14.3)	46,898 (19.9)	59 (13.0)	40 (14.3)	40 (15.2)
West	6 (10.7)	15,211 (6.4)	19 (4.2)	30 (10.7)	19 (7.2)
Unknown	0	329 (0.1)	1 (0.2)	0	0
Missing (Medicaid)	34 (60.7)	128,573 (54.4)	323 (71.3)	170 (60.7)	170 (64.4)
Payer type, n (%)					
Commercial	21 (37.5)	92,136 (39.0)	130 (28.7)	105 (37.5)	94 (35.6)
Medicaid	34 (60.7)	128,573 (54.4)	323 (71.3)	170 (60.7)	170 (64.4)
Medicare	1 (1.8)	15,447 (6.5)	0	5 (1.8)	0

[a]Matched by payer type and US geographic region. CDD, cyclin-dependent kinase-like 5 deficiency disorder; DS, Dravet syndrome.

- In the CDD cohort, 29 (51.8%) patients had a prior seizure-related claim:
  - 15 (26.8%) patients had a prior claim for Lennox-Gastaut syndrome and 14 (25.0%) patients had a prior claim for infantile spasms.
- Additionally, 27 (48.2%) patients had a prior claim for Rett syndrome and 3 (5.4%) patients had a prior claim for Angelman syndrome.
- Median follow-up time was shorter in the CDD cohort (17.1 months) than in the matched epilepsy (25.6 months) and DS (26.4 months) cohorts.

HCRU AT 6 MONTHS POST INDEX

HCRU among patients with 6 months of follow-up



CDD, cyclin-dependent kinase-like 5 deficiency disorder; DS, Dravet syndrome; HCRU, healthcare resource utilization; ICU, intensive care unit.

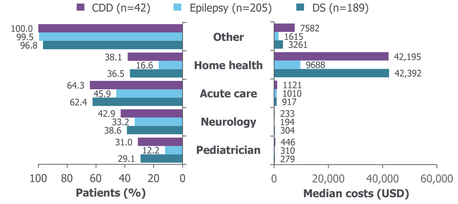
Hospital duration and number of outpatient visits among patients at 6 months of follow-up[a]

	CDD (n=52)	EPILEPSY (n=269)	DS (n=249)
Hospitalization, days, median			
All-cause	6	5	5
Seizure-related	5	4	4
Outpatient visits, number, median			
All-cause	41	10	24
Seizure-related	12	2	6

[a]Patients were matched by payer and geographic region. CDD, cyclin-dependent kinase-like 5 deficiency disorder; DS, Dravet syndrome.

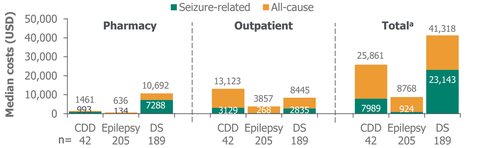
COSTS AT 6 MONTHS POST INDEX

Median outpatient HCRU and costs per patient over 6 months (patients on non-capitated plans with 6 months of follow-up)



CDD, cyclin-dependent kinase-like 5 deficiency disorder; DS, Dravet syndrome; HCRU, healthcare resource utilization; USD, US dollars.

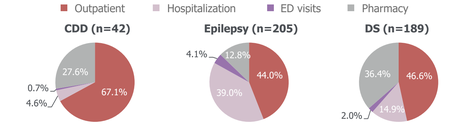
Median total HCRU costs per patient over 6 months (patients on non-capitated plans with 6 months of follow-up)



[a]Medical and pharmacy. CDD, cyclin-dependent kinase-like 5 deficiency disorder; DS, Dravet syndrome; HCRU, healthcare resource utilization; USD, US dollars.

- For patients with CDD, median total medical costs per patient over 6 months of follow-up were $25,861, driven largely by outpatient costs ($13,123).
- Based on the ANCOVA analysis, differences in log-transformed total costs between the CDD cohort and the non-CDD epilepsy cohort remained after adjustment for sex, age, and insurance plan (least squares mean difference [95% CI]: 1.03 [0.46, 1.61]; p=0.0005).

HCRU contributions to total costs (patients on non-capitated plans with 6 months of follow-up)



CDD, cyclin-dependent kinase-like 5 deficiency disorder; DS, Dravet syndrome; ED, emergency department; HCRU, healthcare resource utilization.

- Among patients with CDD, 93.0% of pharmacy costs were for an antiseizure medication (ASM) claim.
- In the epilepsy and DS cohorts, ASM claims comprised 51.2% and 58.7% of pharmacy costs, respectively.

Strengths

- To our knowledge, this is the first real-world study of patients with CDD utilizing a US closed-claims database. Closed payers' claims provide a robust timeline of all events.[2]

Limitations

- Unlike the long-established epilepsy code, the ICD-10 code (G40.42) for CDD is relatively new (approved in 2020), thus CDD outcomes are likely to be underestimated due to underuse of the ICD-10 code. Additionally, for patients presenting with CDD prior to the ICD-10 code availability, there will be a lag between true CDD onset and use of the CDD code.
- This limitation has been mitigated by using a second comparator group, the DS cohort, which also has a newly introduced ICD code (approved in 2020).
- Interpretation is also limited by the small size of the CDD cohort and thus lack of statistical power and the limited length of follow-up.

Conclusions

- Total medical costs were higher among the CDD cohort compared with the non-CDD epilepsy cohort.
- Costs in the outpatient setting were the main drivers of the higher costs associated with CDD, with home health having the largest contribution.
- Cost differences persisted after adjusting for sex, age, and insurance type.
- Pharmacy costs in CDD were predominantly driven by ASMs.
- These data suggest an unmet need for effective seizure control and disease management in patients with CDD, given the high cost burden associated with the disease compared with the non-CDD epilepsy comparator group.
- Future analysis from claims-based datasets are necessary to confirm these findings as CDD becomes better recognized, diagnosed, and properly coded in claims data.

References

1. Bureau of Labor Statistics. Consumer Price Index: How BLS measures price change for medical care services in the Consumer Price Index. https://www.bls.gov/cpi/factsheets/medical-care.htm Accessed March 16, 2026. 2. Baer O, et al. *J Health Econ Outcomes Res.* 2023;10(2):44-52.

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 thank the patients and their caregivers in addition to the investigators and their teams who contributed to this trial. The authors acknowledge Katerina Kumpan, PhD, CMPP (UCB, Slough, UK) for managing the development of the poster, and Lynne Isbell, PhD, CMPP (Envision Spark, an Envision Medical Communications agency, a part of Envision Pharma Group, Fairfield, CT, USA) for writing assistance, which was funded by UCB. Author contributions: J Khushalani was involved in study design, analysis, and interpretation. B Sile and J Roberts contributed to study design and data interpretation. A Kuba analyzed the data. M Brunnert contributed to study design and data interpretation. SE Marsh contributed to study design. M Martin contributed to study design and data interpretation. RR Rajaraman and S Demarest contributed to data interpretation. All authors critically reviewed the poster and approved the final version for presentation. Author disclosures: J Khushalani, B Sile, J Roberts, A Kuba, M Brunnert, SE Marsh, and M Martin are employees of UCB.
RR Rajaraman has served as a speaker for 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 the National Institute of Neurological Disorders and Stroke; and served as an investigator for GRIN, Jazz, Lundbeck, Marinus, Roche, UCB, and Ultragenyx.
S Demarest has 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 BP; and is an advisory board member for non-profit foundations FamillesSCN2A, N of 1 Collaborative, Project BP, Rare X, Ring14 USA, and SLC6A1 Connect.

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American Academy of Neurology 78th Annual Meeting
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