CDKL5 UK
Empowering Families
Embracing Hope

NHS
CDKL5
Rare Disease
Collaborative Network

Caregivers' Voice in Leveraging CDD Burden

Carol-Anne Partridge
Mum to Amber aged 20 with CDD

On Behalf of the Impact of CDKL5 Deficiency Disorder Group

Affiliations

Co-Founder and Chair of CDKL5 UK
CDKL5 Community Advisor at the Bristol CoE
ePAG - ERN Epicare
Director of Complex and Rare Epilepsies Alliance (CREA - A newly formed European Alliance)

Disclosures

Consultancy Fee received from Orion Pharma - Donated to CDKL5 UK
CDKL5 UK Funding Grant from Orion Pharma
Honorarium Received from Ultragenx - Donated to CDKL5 UK

Content Warning

This presentation includes data that may be *upsetting*. It contains terms like “worse than death”, which is used in health research.

This does not *reflect* how we value our children.

Please take care and step away if you need to

The Background

Orion Pharma brought together experts in the field of CDD to form a steering committee, which included clinicians, researchers and patient groups. Over three years, with the support of Carentity and Axon, the steering group collaborated on expanding the knowledge of CDD, by creating:

Review > Epilepsia Open. 2024 Jun;9(3):832-849. doi: 10.1002/epi4.12914. Epub 2024 Mar 7.

Providing quality care for people with CDKL5 deficiency disorder: A European expert panel opinion on the patient journey

Sam Amin [1], Rikke S Møller [2] [3], Angel Aledo-Serrano [4], Alexis Arzimanoglou [5], Patrick Bager [6], Sergiusz Jóźwiak [7], Gerhard Josef Kluger [8] [9], Sandra López-Cabeza [10], Rima Nabbout [11], Carol-Anne Partridge [12], Susanne Schubert-Bast [13] [14] [15], Nicola Specchio [16], Reetta Kälviäinen [17]

Affiliations + expand

PMID: 38450883 PMCID: PMC11145618 DOI: 10.1002/epi4.12914

Abstract

Cyclin-dependent kinase-like 5 (CDKL5) deficiency disorder (CDD) is a developmental and epileptic encephalopathy caused by variants in the CDKL5 gene. The disorder is characterized by intractable early-onset seizures, severe neurodevelopmental delay, hypotonia, motor disabilities, cerebral (cortical) visual impairment and microcephaly. With no disease-modifying therapies available for CDD.

Published on 10.06.2025 in Vol 9 (2025)
This is a member publication of University of Bristol (Jisc)

Preprints (earlier versions) of this paper are available at https://preprints.jmir.org/preprint/72489, first published February 12, 2025.

Caregivers' Perceptions of Clinical Symptoms, Disease Management, and Quality of Life Impact in Cases of Cyclin-Dependent Kinase-Like 5 Deficiency Disorder: Cross-Sectional Online Survey

Sam Amin[1]; Carol-Anne Partridge[2]; Helen Leonard[3]; Jenny Downs[3]; Helen Allvin[4]; Valentine Ficara[5]; Emilie Pain[5]; Minna A Korolainen[4]

Abstract

Introduction - Why this study matters

When you live with CDKL5 Deficiency Disorder (CDD), you don't just live with seizures or developmental challenges.

Families live with uncertainty, exhaustion, and relentless caregiving.

This study was designed to do something rare: let caregivers tell us the true story, not just through words, but through data that regulators, pharma, and policy makers will listen to.

What is CDKL5 Deficiency Disorder (CDD)

A rare, devastating neurological condition.

- Seizures often begin in the first months of life.
- Many children live with profound disabilities: cannot walk, talk, or care for themselves.
- It doesn't get easier with age, it simply changes.

Evie and Mum

> *“You adjust to the seizures. You don’t adjust to watching your child struggle every single day.”* - CDKL5 parent

Why Caregiver Data Matters

Most studies focus on clinical symptoms.

- But who carries the invisible burden? The caregivers.

We need hard data to quantify the daily reality, so that:

- Drug developers
- Health technology assessors (HTAs)
- Regulators
- Policymakers

…can understand the real impact — not just seizure frequency, but life.

How we did this study

132 caregivers of people with CDD took part.

They completed a detailed online survey focusing on the last 12 months:

- Symptoms
- Medical care
- Family and financial impacts
- Their child’s quality of life (using EQ-5D-5L).

Most families came from Europe, but also from many other regions — and their stories showed some big differences.

Introducing EQ-5D-5L

EQ-5D is a standard tool used worldwide to measure quality of life. Caregivers rated:

- Mobility
- Self-care
- Usual daily activities
- Pain/discomfort
- Anxiety/depression

This gives us a score that regulators and payers understand — from 1 (perfect health) to below 0 (worse than death).

What we found

- Median EQ-5D score: 0.18 (incredibly low).
- 13% of caregivers reported scores below zero, describing their child's health state as "worse than death."
- 9 out of 10 children need full support for self-care.
- 8 out of 10 have severely limited daily activities.
- Mobility is highly impaired in two-thirds.

“She can’t move on her own. Every bath, every meal, every position change is on me.” – CDKL5 parent

Younger vs. Older Parents

Parents of younger children face:

- More intense medical appointments.
- More multidisciplinary support.
- The rollercoaster of trying treatments, diets, therapies.
- The raw grief of lost milestones.

Older parents (teenagers & adults with CDD):

- Fewer medical options.
- Shrinking professional teams.
- Chronic exhaustion.
- Growing fear about what happens when they can no longer care.

“We’re not getting any younger. Who will look after her when we’re gone?” - CDKL5 parent

The Financial Burden

- Professional life impact: median 9 out of 10.
- Financial strain: median 7 out of 10.
- Some regions (outside Europe) faced even higher financial burden - scoring 10 out of 10.

Why?

- Lost employment or reduced hours.
- Out-of-pocket medical costs.
- Adapted housing, transport, equipment.
- Private therapy or care services.

“It can feel like having a second mortgage. But this one never gets paid off.” - CDKL5 parent

Why this data is so important

Real-world evidence like this can be used:

- In pricing and reimbursement decisions.
- To justify new therapies.
- To help regulators understand true disease burden.

For a rare disease like CDKL5, we need data like this to make our children's invisible struggles visible.

Aurelia aged 6 months

Call to Action

Seizures continue to play a big role in influences disease burden, but also the resulting lifelong disability, family sacrifice, and financial strain.

EQ-5D helps us speak a language that funders, insurers, regulators understand.

But we must keep listening to caregivers' voices, in trials, in care planning, in policy.

Families have carried this data inside their hearts for years. Now we've put it into numbers.

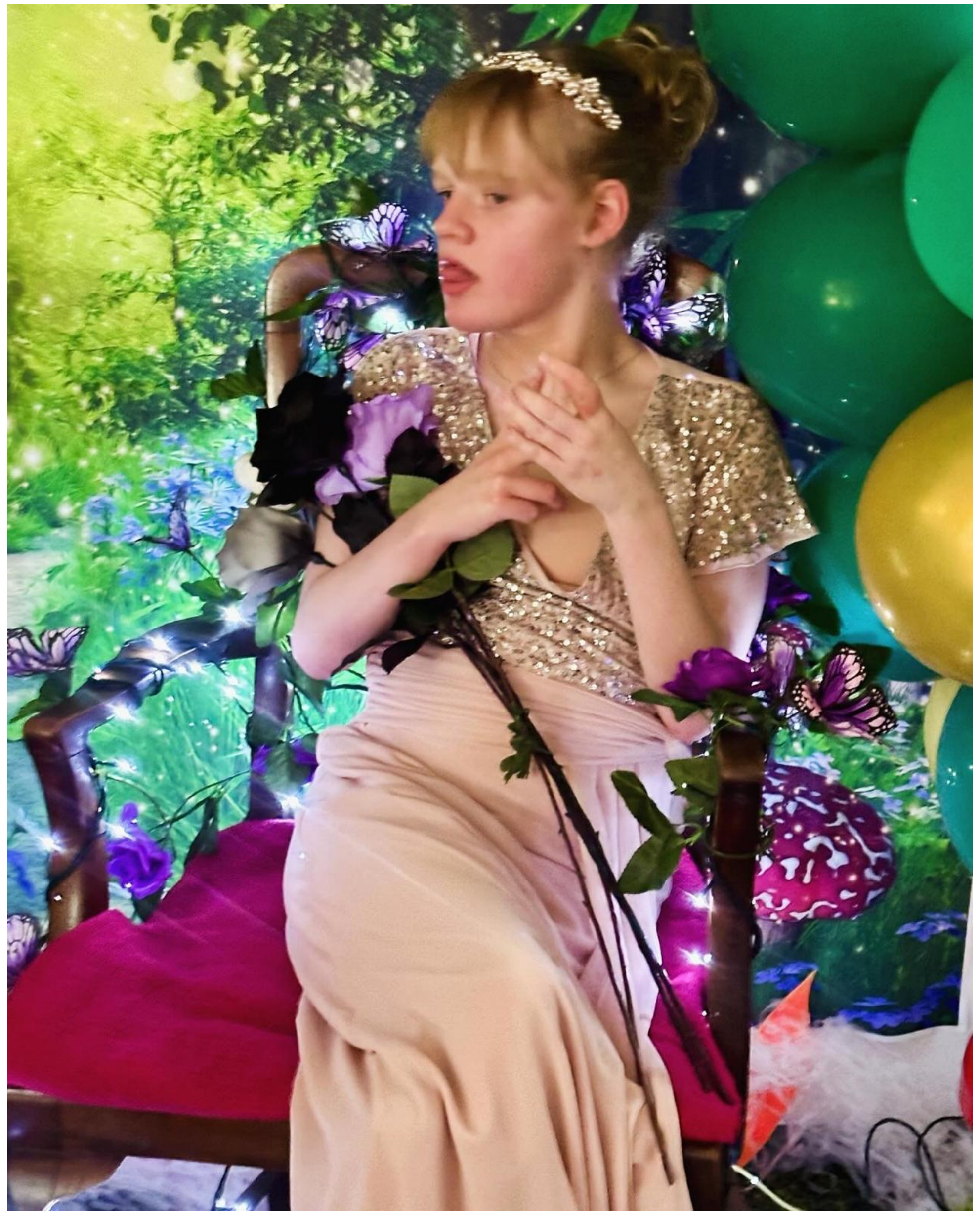
Thank You & Acknowledgements

Huge thanks to the families who shared their lives so openly.

Thanks to the collaboration of:

- CDKL5 Patient Groups
- Clinical and research Partners

And a big thank you to Orion Pharma, Carenity, and Immedica



My Amber aged 20