Rare Epilepsies:
CDKL5 Deficiency Disorder (CDD)

Inspired by **patients.** Driven by **science.**

Living with CDKL5 deficiency disorder
key facts

What is CDD?

An ultra-rare, severe developmental and epileptic encephalopathy that begins in early infancy and is characterized by multiple types of drug-resistant seizures, plus neurodevelopmental delays that impact cognitive, motor, speech, sleep, gastrointestinal, and visual function.

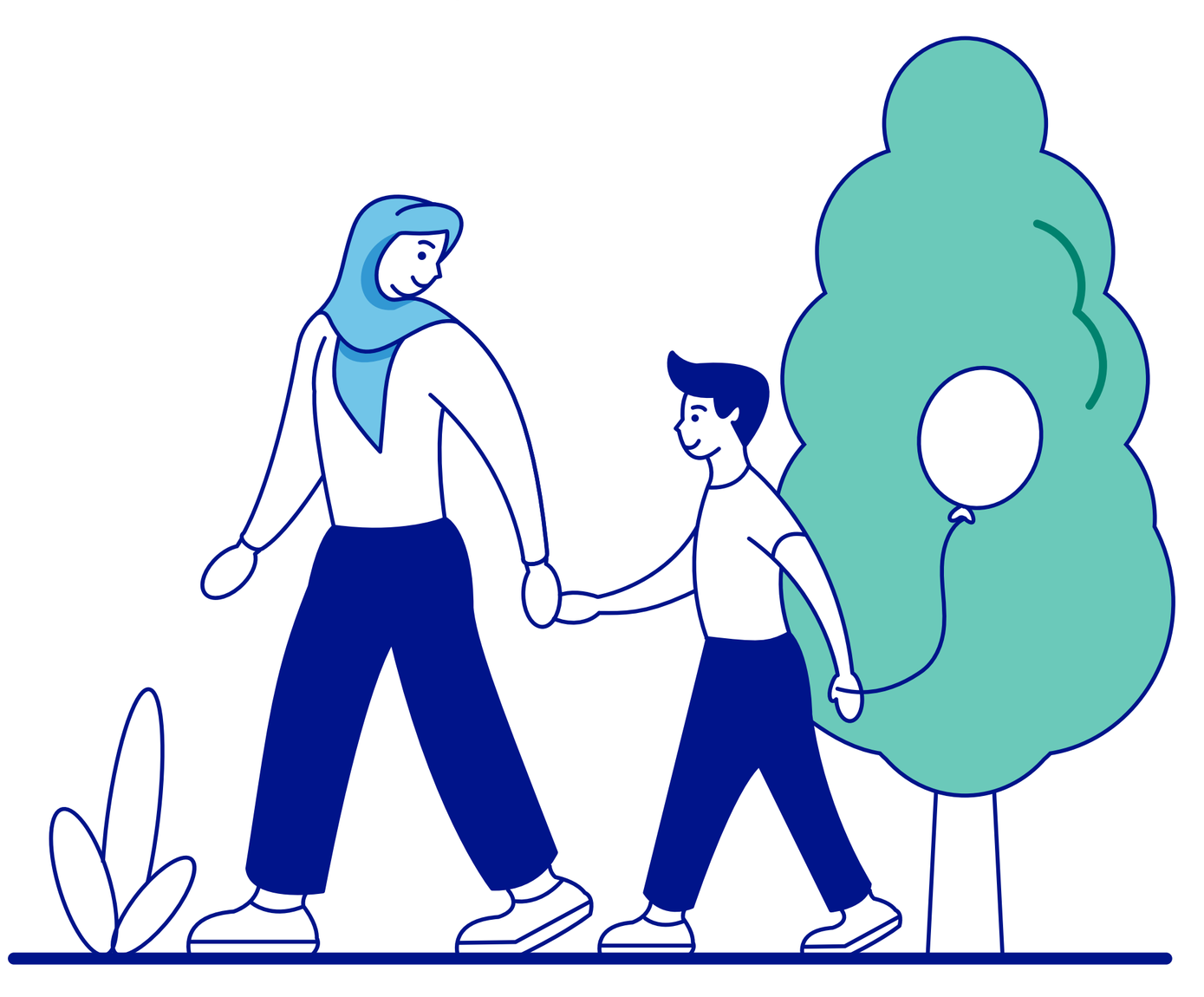
What causes CDD?

Cyclin-dependent kinase like-5 (CDKL5) deficiency disorder is a genetic condition that is caused by changes (pathogenic variants) in the *CDKL5* gene, which is located on the X chromosome.[1,2]

The *CDKL5* gene instructs the body how to make the CDKL5 protein which is required for normal brain development and function.[2] It is characterized by seizures that begin in infancy, followed by significant delays in many aspects of development.[1,2]

CDD may be underrecognized across multiple conditions

CDKL5 gene mutations have been found in children diagnosed with Infantile Spasms, West Syndrome, Lennox-Gastaut syndrome, Rett Syndrome, cerebral palsy, autism, and intractable epilepsy of unknown origin.[4]



The inheritance and epidemiology of CDD

Prevalence: Ultra-rare condition affecting approximately between 1 in 40,000 to 60,000 live births.[1-3]

Ethnicity: Impacts people of many different ethnicities.[2]

Gender: Affects four times as many females as males.[1,2] The course of the disease is often fatal in males in the first or second decade of life. Few female patients have been reported to live into the fourth decade of life.[1,3]

Family: Although CDD is generally not inherited from either parent (de novo mutations), cases of family history of CDKL5 mutations have been reported.[2]

What are the symptoms?

CDD leads to a broad, complex range of clinical symptoms that can differ in severity between patients.[4] Early diagnosis has an impact on the quality of life of patients and their families, as timely identification allows for earlier intervention and support.[5,6]

Seizures

In more than 90% of patients, seizures begin in the first year of life and often as early as at six weeks of age and persist into adulthood.[5,7] Despite available medication, CDD remains drug resistant and most patients continue to experience 1 to 5 seizures per day.[5,7]

The type of seizures experienced can vary throughout a CDD patient's lifetime.[5,7]

At disease onset: Most common seizure types include tonic seizures, infantile (or epileptic) spasms, generalized tonic-clonic seizures, and focal seizures.[5,7]

Over time: Epileptic spasms, tonic, myoclonic, and generalized tonic-clonic become the most common seizure types.[5,7]

Intellectual and Developmental problems

Developmental milestones are severely delayed in affected individuals including:[1,5]

- Little or no development of speech.[2]
- Musculoskeletal problems such as scoliosis.[2]
- Delays or failure to achieve gross motor skills and the use of larger muscles needed for whole-body movements such as sitting, standing, and walking.[1,2]
- Challenges with fine motor skills, the coordination of smaller muscles for everyday tasks such as the ability to pick up small objects.[2]

Other non-seizure symptoms

Other symptoms can include:

- Problems with behavior, vision, breathing, sleeping, feeding, and teeth grinding.[2]
- Gastrointestinal symptoms are also common and may include constipation, reflux, and air swallowing.[2,3]

Impact on caregivers:

Frequent and severe epileptic episodes and non-seizure symptoms such as increased sleep disturbance and behavioral issues can significantly impact family and caregiver's quality of life.[8] These individuals often have to give up their careers to provide their children with a wide range of treatment and multidisciplinary care to manage the symptoms of CDD.[8]

As the diagnosis of CDD and the subsequent access to syndrome-specific family support are often considerably delayed, this can further amplify the emotional burden of the condition on those supporting patients.[9]

How is CDD managed?

Current management of the condition is primarily symptom-based and requires a multidisciplinary approach to care, including:[6]

- Neurologist[6]
- Physiotherapy[6]
- Occupational therapy[6]
- Speech therapy[6]
- Nutritional guidance[6]

Seizures show high drug resistance and are often difficult to control.[10]

47% of individuals (N=122) are on three or more antiseizure medications (ASMs)[10]

6 The median number of ASMs taken throughout a patient's life.*[10]

Despite this, many patient's seizures remain uncontrolled.[11]

Other treatment options include dietary therapy, neurostimulation or callosotomy.[7]

* Data taken from a cohort of the international CDKL5 disorder database

References
1. Zuberi SM, et al. ILAE classification and definition of epilepsy syndromes with onset in neonates and infants: Position statement by the ILAE Task Force on Nosology and Definitions. Epilepsia. 2022;63(6):349-97. 2. Epilepsy Foundation. CDKL5 Deficiency Disorder. Available at: https://www.epilepsy.com/sites/default/files/2023-03/CDKL5_Deficiency_Disorder_March2023.pdf. Accessed: May 2026. 3. Rodak M, et al. CDKL5 Deficiency Disorder (CDD)—Rare Presentation in Male. Children (Basel). 2022;9(12):1806. 4. International Foundation for CDKL5 Research. About CDKL5. Available at: www.cdkl5.com/about-cdkl5. Last accessed: May 2026. 5. Jakimiec M, et al. CDKL5 Deficiency Disorder—A Complex Epileptic Encephalopathy. Brain Sci. 2020;10(2): 107. 6. Amin S, et al. Providing quality care for people with CDKL5 deficiency disorder: A European expert panel opinion on the patient journey. Epilepsia Open. 2024;9(3):832-849. 7. Hong W, et al. CDKL5 Deficiency Disorder-Related Epilepsy: A Review of Current and Emerging Treatment. CNS Drugs. 2022;36(6):591–604. 8. International Foundation for CDKL5 Research and Loulou Foundation. The Voice of the Patient Report: CDKL5 Deficiency Disorder (CDD). Available at: https://www.cdkl5.com/wp-content/uploads/2023/05/CDD-VoP-REPORT.pdf. Accessed: May 2026. 9. Mori Y, et al. Impacts of caring for a child with the CDKL5 disorder on parental wellbeing and family quality of life. Orphanet J Rare Dis. 2017;12(1):16. 10. Leonard H, et al. CDKL5 deficiency disorder: clinical features, diagnosis, and management. Lancet Neurol. 2022;21(6):563–76.

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