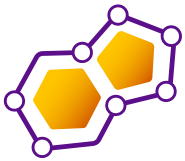
CDKL5 Deficiency Disorder (CDD) Clinical Documentation Reference

This reference is provided as an example of the type of clinical documentation typically requested by payers to support a Medical Necessity request. This reference may also serve to support the creation of a Letter of Medical Necessity, which is a clinical summary of your patient's medical record and should state your rationale for prescribing ZTALMY® (ganaxolone) CV and the medical necessity request for patient coverage.

CDD DISEASE OVERVIEW

CDD is a serious and rare genetic disorder caused by a **mutation of the cyclin-dependent kinase-like 5 (*CDKL5*) gene**, which is located on the X chromosome and encodes proteins essential for normal brain function[1,2]



Diagnosed by **genetic testing** to determine if there is a mutation in the *CDKL5* gene[2]

Incidence is approximately **1:40,000 live births** and predominantly affects females[3]

Characterized by early-onset, **difficult-to-control seizures** and severe neurodevelopmental impairment[1,2,4]

SIGNS AND SYMPTOMS OF CDD[1]

Clinical notes typically present the patient history, including comorbidities, medications, neurodevelopment, and milestones. This is not a complete list of all signs and symptoms associated with CDD.

- Epilepsy, early onset and refractory
- Cortical visual impairment/lack of eye contact
- Motor and cognitive developmental delay/ intellectual disability
- Limited hand skills
- Gastrointestinal disturbances/constipation
- Poor muscle tone
- Limited ability to walk
- Sleep disturbances

CONFIRMATION OF DIAGNOSIS

ICD-10 G40.42 Cyclin-dependent kinase-like 5 deficiency disorder. If applicable, add secondary diagnosis codes.

Provide documentation from patient records including:

- Age at seizure onset
- Age at CDD diagnosis
- Confirmed genetic test indicating variant in *CDKL5* gene and date completed
- Current patient weight
- Other patient labs or diagnostic tests completed (MRI, EEG, CT)

CLINICAL COURSE AND DISEASE MANAGEMENT

Provide clinical notes about:

- Seizure types and frequency
- All previous and current therapies, duration of treatment, and rationale for discontinuation or change in therapy
- Other interventions such as diet, occupational therapy, surgical interventions, etc.
- Developmental delays
- Other clinical features such as gastrointestinal disturbances, visual impairment, muscle weakness, or sleep disturbances
- Impact on quality of life for patient and family
- Any other relevant documentation about how CDD is treated or managed in this patient

References: 1 Olson HE, Demarest ST, Pestana-Knight EM, et al. *Pediatr Neurol*. 2019;97:18-25. **2** Leonard H, et al. *Lancet Neurol*. 2022;21(6):563-576 **3** Symonds JD, Zuberi SM, Stewart K, et al. *Brain*. 2019;142(8):2303-2318. **4** Demarest ST, Olson HE, Moss A, et al. *Epilepsia*. 2019;60(8):1733-1742.

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