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 includes a clinical summary of your patient's medical record and your rationale for this treatment. Include the medical necessity request for patient coverage.

CDD DISEASE OVERVIEW

CDD is a serious and rare genetic disorder caused by **pathogenic, or likely pathogenic variants in 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]

Genetic testing for variants of the *CDKL5* gene may help confirm the diagnosis of CDD[2]

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

Characterized by early-onset, **drug-resistant epilepsy** 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
- Motor and cognitive developmental delay/ intellectual disability
- Breathing disturbances
- Gastrointestinal disturbances/reflux, constipation
- Hypotonia
- Dyskinetic movements
- 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 pathogenic or likely pathogenic variant in *CDKL5* gene and date completed, if available
- 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, cortical visual impairment, hypotonia/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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