GET TO KNOW

# CDKL5 DEFICIENCY DISORDER

CDD IS A RARE GENETIC CONDITION THAT AFFECTS BRAIN DEVELOPMENT. IT OFTEN BEGINS WITH SEIZURES EARLY IN LIFE AND CAN IMPACT MOVEMENT, COMMUNICATION, LEARNING, AND OVERALL DEVELOPMENT.

## SYMPTOMS

Symptoms and severity vary widely between individuals but typically begin in early infancy.

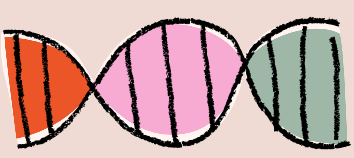
### NEUROLOGIC
Early-onset Seizures
Developmental Delay
Cognitive Differences

### SEIZURE TYPES
Infantile Spasms
Tonic Seizures
Myoclonic Seizures

### OTHER SYSTEMS
Low Muscle Tone
Vision Impairment
Feeding and GI Challenges

## CAUSE

CDD is caused by a change in the ***CDKL5* gene** located on the X chromosome, which plays an important role in early brain development.



## INHERITANCE

Most cases occur spontaneously, meaning they are **not** inherited from a parent. Rarely, CDD may be inherited from a parent who carries the gene change.

## DIAGNOSIS

- MEDICAL EVALUATION
- ELECTROEN-CEPHALOGRAM (EEG)
- MAGNETIC RESONANCE IMAGING (MRI)
- GENETIC TESTING

Diagnosis typically includes a medical evaluation and genetic testing to confirm a change in the *CDKL5* gene.

## TREATMENT

There is currently no cure for CDD. Treatment focuses on managing symptoms and providing supportive care to help improve quality of life.

- TREATMENT OPTIONS
  - ANTI-SEIZURE MEDICATIONS
  - FEEDING AND GI MANAGEMENT
  - CLINICAL TRIALS
  - PHYSICAL THERAPY
  - OCCUPATIONAL THERAPY
  - SPEECH THERAPY

## PREVALENCE

CDD is considered a rare disorder. It is estimated to affect about 1 in 40,000–60,000 children worldwide. Exact numbers may vary as research continues.

## RESOURCES

- CDKL5 ALLIANCE
- HOPE FOR CDKL5
- EPILEPSY FOUNDATION
- INTERNATIONAL FOUNDATION FOR CDKL5 RESEARCH
- NATIONAL ORGANIZATION FOR RARE DISORDERS (NORD)

Medical information adapted from GeneReviews® and the NIH Genetic and Rare Diseases Information Center.