What is CDKL5 Deficiency Disorder?

CDKL5 Deficiency Disorder (CDD) is a rare developmental & epileptic encephalopathy caused by changes to a gene called CDKL5.

The CDKL5 gene provides instructions for making proteins that are essential for normal brain and neuron development.

Changes to the gene cause both epileptic activity as well as severe impairment of development. Hallmarks of CDD are the onset of seizures at a very early age and severe neurodevelopmental disability impacting cognitive, motor, speech, and visual function.

There is a spectrum of severity and not all symptoms listed here are experienced by all who are diagnosed.

Learn more about CDD and the work being done to treat and cure this rare disease at CDKL5.COM.

Symptoms

- Seizures starting early in life
  - Epileptic spasms often occurring without hypsarrhythmia
  - Multiple different types of seizures
- Poor muscle tone (hypotonia)
- Cortical Visual Impairment (CVI)
- Gastrointestinal Difficulties
  - Constipation
  - Reflux
- Sleep difficulties
- Limited ability to walk
- Inability to speak but may use complex gestures/vocalization
- Limited hand skills
- Purposeless hand movements (stereotypies)
- Teeth-grinding (bruxism)
- Intellectual disability
- Breathing irregularities (such as hyperventilation)
- Respiratory infections
- Scoliosis
- Behavioral symptoms such as anxiety and social avoidance

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