

# CDKL5 Deficiency Disorder



Registered Charity No. 1149099. CDKL5 UK

# What is CDKL5 Deficiency Disorder (CDD)

CDD is a rare X-linked genetic disorder that results in early onset, difficult to control, seizures, and severe neuro-developmental impairment. CDD is classed as a rare disease with an incidence rate believed to be 1:42,000.

CDD mostly effects females due to the location of the gene on the x-chromosome, but many males have also been diagnosed.

The CDD gene provides instructions for making a protein that is essential for normal brain development. Although little is known about the protein's function, it may play a role in regulating the activity of other genes, including the MECP2 gene (or Rett Syndrome). The CDKL5 protein acts as a kinase, which is an enzyme that changes the activity of other proteins by adding oxygen and phosphate atoms (a phosphate group) at specific positions. Researchers have not yet determined which proteins are targeted by the CDKL5 protein.

Most children affected by CDD suffer from seizures that begin in the first few months of life. Most cannot walk, talk or feed themselves, and many are confined to wheelchairs, dependent on others for everything. Many also suffer with scoliosis, visual impairment, sensory issues and various gastrointestinal difficulties. As time has gone on it appears that there might be other symptoms that play a role in the condition.

This is the current list of the most common symptoms however it is important to note that not every child will have what is listed, and therefore there appears to be a wide spectrum in terms of the severity of some children, from moderately affected to profoundly affected.

- Epileptic seizures starting in the first 8 months of life.
- Infantile spasms (in about 50%)
- Many different types of epilepsy usually including myoclonic jerks
- Hand stereotypies in the form of hand clapping, tapping, movements or mouthing
- Marked developmental delay
- Limited or absent speech
- Hypersensitivity to touch, for example dislike of hair brushing
- Lack of eye contact or poor eye contact, but may have intensive eye interaction
- Gastro-esophageal reflux, and other severe gastrointestinal problems such as low gut motility
- Constipation
- Small, cold feet
- Breathing irregularities such as hyperventilation
- Grinding of the teeth
- Episodes of laughing or crying for unknown reasons
- Low/Poor muscle tone
- Very limited hand skills
- Some autistic-like tendencies
- Scoliosis
- Cortical Visual Impairment (CVI), aka "cortical blindness"
- Apraxia
- Eating/drinking challenges, such as refusal to eat and drink, as well as giving up altogether
- Interrupted sleep
- Characteristics such as a sideways glance, and habit of crossing legs

# How is CDKL5 UK helping the CDD Community?

CDKL5 UK is a small charity supporting the research and medical community working in the field of CDD as well as families affected by CDD.

CDKL5 UK fundraises for research and support and is involved in many activities in the UK, Europe and the rest of the world. CDKL5 UK has supported the Rare Disease Collaborative Network (RCDN) - CDKL5 which is located in Bristol and has funded a post of Research Coordinator for a period of 5 years.

This post will assist Bristol in their goal of establishing the RCDN as the Centre of Excellence in the UK for CDD but also meeting the UK Rare Disease Strategy outcome of being a research focussed clinic.

The RCDN - CDKL5 can be accessed by contacting Dr Sam Amin, Consultant Paediatric Neurologist, the Clinical Lead for the RCDN at [samamin@nhs.net](mailto:samamin@nhs.net).

CDKL5 UK represents CDD within the EpiCare – a European Reference Network aimed at furthering knowledge, research and clinical trials into Rare and Complex Epilepsies as the Epilepsy Patient Advocacy Group (Epag).

## What is the CDKL5 Alliance

CDKL5 UK is a founding member of the CDKL5 Alliance. The CDKL5 Alliance is a community of over 18 patients groups worldwide who collaborate to bring the medical and scientific community together by working together in meaningful and respectful way. They organise an international family and research conference yearly which is well sponsored and attended and rotates yearly between North America and the rest of world.



# Where can I find more information about CDD

**CDKL5 UK**  
[www.curecdkl5.org](http://www.curecdkl5.org)  
[info@curecdkl5.org](mailto:info@curecdkl5.org)

**Twitter**  
[@CDKL5UK](https://twitter.com/CDKL5UK)

**Instagram**  
[cdkl5uk](https://www.instagram.com/cdkl5uk)

**Youtube**  
[cdkl5uk](https://www.youtube.com/cdkl5uk)

**Facebook**  
[@CDKL5UK](https://www.facebook.com/CDKL5UK)

**CDKL5 Alliance**  
[www.cdkl5alliance.org](http://www.cdkl5alliance.org)

**Youtube**  
[CDKL5Alliance](https://www.youtube.com/CDKL5Alliance)

**Twitter**  
[@CDKL5Alliance](https://twitter.com/CDKL5Alliance)

**EpiCare website**  
[epi-care.eu](http://epi-care.eu)

**Loulou foundation website**  
[www.louloufoundation.org](http://www.louloufoundation.org)

