New to CDKL5 Deficiency Disorder? What you need to know

What is CDKL5 Deficiency disorder (CDD)

CDD is a neurological disorder with seizure onset during typically in the first 3 months of life, but can be later and for some children they never experience seizures.

Is CDD under recognised?

World-wide it is estimated the incident of CDD is around 1:40000, but there are still some challenges to diagnosis and reporting as some older patients may have been mis-diagnosed with other conditions such as cerebral palsy and others who may

What causes CDD?

CDD is caused by mutations in the CDKL5 (Cyclin-Dependent-Kinase-Like 5) gene. The CDKL5 gene is supposed to provide instructions for making a protein that is essential for brain and neuron development. Mutations interfere with this process, causing seizures and a range of neuro-developmental delays.

CDD was first identified in 2004 with CDD becoming recognized as a distinct condition in 2012. The gene's location on the X chromosome means CDD affects girls more often than boys, although there are many boys who are affected. The X chromosome is one of the sex chromosomes; females have two X chromosomes, and males have one X and one Y.

How is CDD diagnosed?

A genetic diagnosis test is now available in the UK from the NHS. Early diagnosis and achieving some early seizure control can made a huge difference – this does represent a significant challenge, and can improve long-term outcomes. conditions such as cerebral palsy and others who may have gone un-diagnosed.

What is the course of the CDD?

Generally, the first symptoms which infants experience is seizures, and along with this they will experience either development arrest or significantly slower development. These development delays increase with age and many children will have difficulty in meeting milestones. They may also have a range of other symptoms such as respiratory, gastrointestinal, and mobility problems. But it is important to note there is a spectrum like many disorders, and these range from to profound to mildly effected and it really is an unknown at the start of someone's journey which path they will follow.

What treatments are available?

At this current time there is no cure for CDD. Treatments which focus on controlling the seizures or at least minimising them are often used, including medically prescribed dietary therapies. There is some hope on the horizon with at least three gene therapy programmes currently in pre-clinical phase of development. There are also numerous pharmaceutical companies who are in clinical trials with patients with CDD with some showing good effect in controlling seizures and improving the quality of life for those living with CDD.

Is there a specialist centre in the UK for CDD?

A new Centre of Excellence, as recognised by NHS England, has been set up at Bristol Royal Hospital for Children, specifically for children and adults with CDKL5 CDD in the UK, led by Paediatric Neurologist Dr Sam Amin. The initiative hopes to help support the holistic care of your child, in partnership with your local medical team, by providing specialist clinical input as well as focused pastoral care and support. You can contact the centre via CDKL5 UK funded research coordinator Emma Claydon via email Emma.Claydon@uhbw.nhs.uk or by calling 0759 6274 442.

How CDD may impact you are a family

You are likely to have many questions as to how this happened to your child, and generally the answer is we don't know, mutations which cause CDD are generally referred to as "de-novo" mutations which roughly translates to "new" and means it happens randomly and not passed down through families. Therefore, there is nothing you could have done to prevent it.

Many parents and families of children who are diagnosed with CDD will experience a sense of loss and will grieve for the child they thought they would see grow up and this is entirely normal. As with any grief, it will get easier to deal with, but it will always be there, and it is OK to grieve even if you have been living with CDD in your life for a number of years. It is not unusual for life milestones to re-trigger grief such: educational – such as proms, or the marriage of a sibling so take time to recognise it and try and be kind to yourself.

There are many support groups out there on Social Media, and you will see hundreds of children and adults with CDD and they will all have their own story. It is so important not to try and compare your child to others or your life living with CDD to others as although we share the journey of CDD, they path we follow will be very different. Learning to understand your child and how they communicate even in the smallest way, will help you to see your child first and not CDD – don't let CDD define you or your child.

Where can I find more information about CDD?

www.curecdkl5..org.uk
www.cdkl5alliance.org
www.foulioufoundation.org

Youtube:

CDKL5 UK
CDKL5 Alliance

Support Groups:

Facebook
CDKL5 World – United in Hope,
CDKL5 UK & Ireland Working Group