Centre of Excellence for CDKL5 Deficiency Disorder

NEWSLETTER

CDKL5 Collaborative Network Newsletter - Autumn 2020

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New Centre of Excellence for CDKL5 Deficiency Disorder at Bristol Royal Hospital for Children

Despite the restrictions we have all faced due to the Coronavirus Pandemic (Covid-19), 2020 has seen the formation of a new Centre of Excellence, as recognised by NHS England, specifically for children with CDKL5 deficiency disorder in the UK.

This service is being led by Paediatric Neurologist Dr Sam Amin at Bristol Royal Hospital for Children.

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 at a single point of contact.

We need your input to help us shape our service to meet the needs of your family.

We want to know what kind of support will be helpful and how best to deliver this. Please get in contact!

Meet the team

Dr Sam Amin

Sam is the clinical lead for the CDKL5 Centre of Excellence and Rare Disease Collaborative Network and has worked tirelessly to get the project off the ground.

Sam is a consultant paediatric neurologist at Bristol Royal Hospital for Children.

He has a special interest in CDKL5.

He has done a lot of clinical and academic work in the field of CDKL5.

He is in the process of writing the international guidelines for the management of CDKL5 with colleagues from the Centre of Excellence in Colorado and is a scientific advisor for CDKL5 UK.

CDKL5 ALLIANCE

CDKL5 UK
www.curecdkl5.org

Meet the team

Helen Bowden

Helen started working for the CDKL5 Centre of Excellence in March 2020, at the very start of the pandemic.

This post is funded by CDKL5 UK. Helen has been using the last 6 months to build an understanding of CDKL5 Deficiency

Disorder by speaking to parents, attending conferences and reading as much as she can.

Helen supports Dr Amin by providing a single point of contact for all CDKL5 related queries, setting up and administering research projects and providing practical support and advice to parents.

You can contact Helen by emailing helen.bowden2@nhs.net or calling 07592 083 001.

CDKL5 Forum 12-14 October 2020

The Loulou Foundation's 6th International Scientific Forum for CDKL5 Research took place in October virtually.

The forum was attended by over 200 researchers, doctors, parents, patient advocacy groups and members of the Biotech industries from across the world.

Many aspects of CDKL5 treatment and cure were discussed with further work developed and working groups set up to ensure the path to treatments are the most direct and can happen quickly.

Promising Results from Early Phase 2 treatment trials Zogenix—Fenfluramine Small trial (max. 10 participants) at NYU for patients with CDKL5 following positive results using the drug to treat seizures in Dravet Syndrome.

Current results are positive and a Randomised Controlled Trial with a larger number of participants is suggested.

Ovid Pharmaceuticals—Soticlestat Following positive outcomes for seizure control in Dravet Syndrome, a small cohort of patients with CDKL5 took part in a Phase 2 study of this treatment.

Talks are under way to ascertain whether a Randomised Controlled Study can take place.

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Treatment News

The CDKL5 Centre of Excellence has successfully applied for and treated, one patient with the drug Ataluren.

Two further applications for compassionate usage are in progress.

The team hopes to be able to support further applications for compassionate drug use in the future.

Get your voice heard!

The British Paediatric Neurology Association (BPNA) and the James Lind Alliance (JLA) have come together to organise a Priority Setting Partnership on Childhood Neurological Conditions.

This partnership aims to shape future research on these conditions by bringing together patients, their parents and carers and health care professionals to identify and prioritise unanswered questions around treatments, therapies or procedures to present to researchers and funders.

The CDKL5 Rare Disease Network is really keen to ensure the voices of CDKL5 families are heard within this consultation and their personal priorities addressed.

We would encourage anyone who is interested and able to complete the survey by following the link: https://www.surveymonkey.co.uk/r/BPNA_JLA_RESEARCH_PSP

Research News

Marinus Pharmacuticals has completed its Phase 3 trial of the drug Ganaxolone (The Marigold Study), a trial specifically looking to reduce seizure activity in children with CDKL5.
Results:

- Trial met primary endpoint, with a median 28-day major motor seizure frequency reduction of 32.2 percent compared to 4.0 percent for placebo (p=0.002)
- Ganaxolone was generally well tolerated by participants and the discontinuation rate in the active treatment arm was less than 5 percent
- New Drug Application (NDA) submission planned for mid-2021; commercial launch targeted for 2022.

For further information please use our link

The CDKL5 Centre of Excellence in Bristol are working with Marinus to include UK participants on the extended access programme. This will enable patients who meet the trial inclusion criteria to access the treatment.If you would like to be considered for the use of this drug, please get in contact with us by emailing helen.bowden2@nhs.net or calling 07592 083 001.