

# International CDKL5 Disorder Database Newsletter



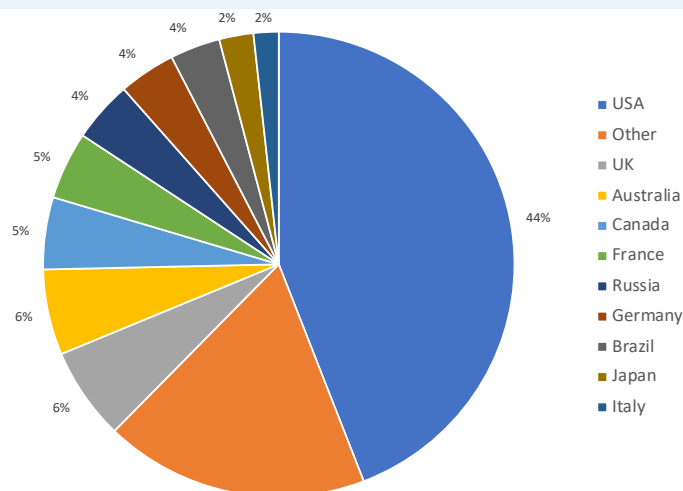
Issue 9, May 2023

## THANK YOU!

Our team would like to thank you very much for your contributions and commitment to this valuable research! The success of our research would not be possible without your support and efforts.

- **416** families have now completed a baseline questionnaire with **52** completing since early 2022. If you have not yet had a chance to complete please contact us on [CDKL5@telethonkids.org.au](mailto:CDKL5@telethonkids.org.au) or Helen at [Helen.Leonard@telethonkids.org.au](mailto:Helen.Leonard@telethonkids.org.au) and we can resend your login.
- **190** families have completed a follow-up questionnaire either in 2018/2019 or 2022/2023.
- Families have now provided us with **148** sets of hand function videos and **144** sets of gross motor videos.
- Since the last newsletter we have talked to **250** families registered with the International CDKL5 Disorder Database (ICDD).
- Your data informed **8** research articles in peer-reviewed medical journals in 2022.

## CDKL5 Families Around the World in our database



## Initial and Follow-up Questionnaires

We thank all the families who have completed the initial and follow-up questionnaires! If you have misplaced the email with your login details or would like any assistance with the questionnaires, please get in touch by emailing us on [CDKL5@telethonkids.org.au](mailto:CDKL5@telethonkids.org.au). The more information that we can collect from our families around the world helps us to learn more about CDKL5.

## NIH "1 in 200" Study Progress - PREPARING FOR CLINICAL TRIALS IN CDKL5

We are part of the International CDKL5 Clinical Research Network, developing strong outcome measures for CDD to enable good quality clinical trials. We are modifying and validating measures of severity and functional abilities specifically for children and adults with CDD. We aim for accurate measurement of the new treatments in upcoming clinical trials.

Stage 1 of this study involved initial validation of the following measures: Video Gross Motor, Video Hand Function, Clinical Severity Assessment (parent and clinician reported), Sleep, Communication and Quality of Life. Recently we described our results in reports for the FDA and NIH which received great reviews and the tick of approval to continue to the next stage of the project. All measures look to be working well for individuals with CDD, except for communication which we are working to rectify (information on this on the next page).

We have now begun the 2<sup>nd</sup> stage of the project – the longitudinal phase. This phase includes families in the US who attend CDKL5 Centre of Excellence Clinics who complete our CDKL5 measures periodically over 2 years. If you do not attend US Center of Excellence clinics, we still have video and questionnaire studies that you can join.

If you would like some more information or copies of our measurement papers please contact us through [CDKL5@telethonkids.org.au](mailto:CDKL5@telethonkids.org.au)

## Communication in children and adults with CDD

Our new team member Jess has enjoyed completing interviews with 23 parent caregivers of children with CDD to talk about the different ways their children are communicating. This is to help modify available communication measures to suit CDD, to make sure we have a measure that can measure the subtle and nuanced ways your children are communicating that are not currently recognised in available measures. Three main categories were found: (1) Mode, the different communication methods (e.g., words, gestures, AAC device, eye movements); (2) Purpose and meaning, the reasons for communication (e.g., choice making, expressing emotions, showing affection); (3) Reciprocal or sharing exchanges, two-way interactions (e.g., initiating, engaging). Communication was also influenced by the child's health including seizures, cortical visual impairment and medications, and their environment including familiar place and people. We are designing a new measure and will be testing it shortly.

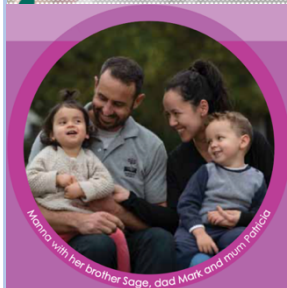
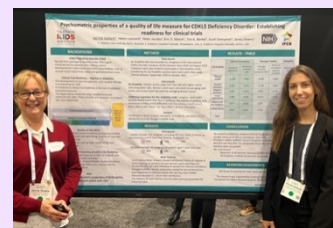
## PRESENTATIONS

Helen presented posters at the 2022 November CDKL5 Forum held in Boston. Her presentations were on growth patterns in CDD, combinations of medications used and CDD milestones data. She also gave an oral presentation relating to adults with CDD which was a collaboration with Dr Ángel Aledo-Serrano from Spain and other international centres.

In December 2022, Jenny, Helen and Jacinta travelled to Nashville to present an update at a CDKL5 meeting of CDD outcome measure results, hosted by IFCR.

Jenny and Jacinta presented at the American Epilepsy Society conference about measurement of quality of life in CDD and what better looks like following new treatments in rare disorders. They also attended the Rare Epilepsy Network Meeting which included discussion on solutions for developing new and supporting existing multidisciplinary clinics.

The trip was a fantastic opportunity for the NIH Study team to connect and discuss future research steps for the longitudinal stage of our grant. It was also lovely for some team members to meet for the first time in person!



## Impact Report

CDKL5 was a focus in the 2022 Telethon Kids Institute Impact Report. A beautiful photo of Manna and her father, Mark, featured on the cover. Within, there is a section on our clinical trial readiness research and the International CDKL5 Disorder Database. Also, a beautifully written article details the experience of Manna's parents, Patricia and Mark, from the first seizure their daughter experienced, through to diagnosis, Manna's milestones and hopes for the future. If you would like to read these articles, the TKI 2022 Impact Report is available [here](#).



**Congratulations** Jacinta for receiving the 2022 Loulou Foundation Junior Fellowship Award for her contributions to the current program of research for CDKL5.

## Recent Publications

### **CDKL5 deficiency disorder: clinical features, diagnosis, and management**

*Leonard H, Downs J, Benke TA, Swanson L, Olson H, Demarest S, Lancet Neurology 2022*

We wanted to make sure that clinicians have access to the best available information about CDD. This paper:

- Provides updated data about epilepsy patterns, development, functioning and medical problems associated with CDD;
- Discusses variation in the presentation of symptoms and genetic background;
- Looks at how CDD impacts the quality of life of your children as well as how it affects you, their caregivers;
- Describes how genetic testing had changed over the last two decades since CDD was first identified;
- Summarises what is known about the clinical management and describes the clinical trials in progress or planned.

### **Factors influencing the attainment of major motor milestones in CDKL5 deficiency disorder**

*Wong K, Junaid M, Demarest S, Saldaris J, Benke TA, Marsh ED, Downs J, Leonard H, Eur J Hum Genet 2023*

This study aimed to tease out the factors which affected the age at which a child with CDD would attain the milestones of sitting and walking. Two thirds of girls but fewer boys could sit independently by the age of seven years and a quarter of boys and a quarter of girls each learned to walk independently. Influencing factors were mutation group, mosaicism (when only a proportion of cells have the CDKL5 mutation), age at seizure onset and experience of a honeymoon period. Gender was an influencing factor for sitting but not walking.

## GET IN TOUCH!

If you would like to see more of our CDKL5 publications, they can be found [here](#) OR contact us at at [CDKL5@telethonkids.org.au](mailto:CDKL5@telethonkids.org.au) for a full copy.

### **If you are interested in:**

- 1) Participating in our research or would like more information
- 2) Completing a follow-up questionnaire and haven't done so recently
- 3) Being a part of our Consumer Reference Group meetings to give us your thoughts and feedback on our research projects prior to data collection (a very important part of our research process and your input is very valuable)  
or
- 4) Would like to send us an email about what you would like to know about CDD. Your thoughts and opinions help us to investigate important research questions.

Please contact us through [CDKL5@telethonkids.org.au](mailto:CDKL5@telethonkids.org.au)

### **How to Register the CDKL5 Database:**

If you are interested, or know someone that is interested, in participating in the database, you can register using one of two ways:

- 1) Visit our registration page at: <https://rettregister.telethonkids.org.au/CDKL5/Register>
- 2) Email your contact details (including telephone number) to [CDKL5@telethonkids.org.au](mailto:CDKL5@telethonkids.org.au)

Until next time,

The CDKL5 team 😊