

PARTICIPATE IN THE ExCeeD STUDY for CDKL5



If your child is 18 months or older and has a genetic diagnosis of CDKL5 deficiency disorder (CDD), he or she may be eligible to participate in our research study. The goal of the study is to understand the clinical features of individuals with CDD and independent functional abilities or seizure freedom, and to establish related genetic and molecular patterns



Eligibility



1

Children who can walk, communicate (word, sign or picture-based), and pick up small objects with thumb and forefinger

OR

2

Children who have never had seizures, or who have been seizure free for several years (can be on 1 anti-seizure treatment)



The study involves filling out caregiver questionnaires remotely (approximately 2 hours total) and one 60-minute virtual interview with the study team. No on-site visits are required for study completion. Participants who complete the study will be offered two gift cards valued at \$100 total as a token of appreciation for their participation.



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Expanding CDKL5 Deficiency Disorder (ExCeeD) Study FAQs

ExCeeD Study	
<i>Official Study Title</i>	Expanding CDKL5 Deficiency Disorder (ExCeeD) Study
<i>Sponsor</i>	LouLou Foundation
<i>Principle Investigator</i>	Dr. Heather Olson
<i>Patient Population</i>	Patients 18 months or older who have a genetic diagnosis of CDKL5 deficiency disorder (CDD).
<i>Is my child eligible?*</i> <i>*additional eligibility criteria will be reviewed with the study team</i>	<ol style="list-style-type: none"> 1. Children who can walk, communicate (word, sign or picture based), and pick up small objects with thumb and forefinger <p style="text-align: center;">OR</p> <ol style="list-style-type: none"> 2. Children who have never had seizures, or who have been seizure free for several years (can be on 1 anti-seizure treatment)
<i>What will I be asked to do?</i>	<ul style="list-style-type: none"> • Complete caregiver questionnaires remotely (approximately 2 hours total) • Complete one 60- to 90-minute virtual interview with the study team • No on-site visits are required for study completion • Additional optional opportunities will include: • For female, U.S. based participants saliva and blood sample collections to be analyzed for a specific genetic pattern For U.S. based participants, blood collections to store for future biomarker analysis
<i>Will participants be compensated?</i>	Yes, participants who complete they study will be offered two gift cards valued at \$100.00 total as a token of appreciation for their time.
<i>What are the risks of the study?</i>	<ul style="list-style-type: none"> • Breach of confidentiality • Suicidality or self-harm behaviors may be identified in some of the standardized assessments • Frustration, fatigue or upset as result of certain questionnaires and/or interview questions • Minor discomfort, bruising, fainting and/or infections have been associated with blood draws
<i>Topics discussed in questionnaires</i>	<ul style="list-style-type: none"> • Communication skills • Daily living • Social communication skills and relationships • Physical activity • Problem solving • Your child/dependent's: <ul style="list-style-type: none"> ○ Activities ○ Behaviors ○ Emotions ○ Epilepsy history ○ Cognition ○ Therapies ○ Other medical issues

Date

Dear parents of ____,

We would like to tell you about an opportunity to participate in the Expanding CDKL5 Deficiency Disorder (ExCeeD) Study, a research study for patients with disease-associated variants in the *CDKL5* gene who have independent functional abilities or seizure freedom. The ExCeeD study is being conducted at Boston Children's Hospital, but you do not have to be a patient at Boston Children's Hospital to participate. Dr. Heather Olson, the director of the CDKL5 Center of Excellence at Boston Children's Hospital, is the principal investigator for this study.

We are currently recruiting individuals 18 months of age or older who meet the following criteria:

- They have a *CDKL5* gene variant thought to be the cause of their neurologic presentation by their clinician (the gene variant may be classified as pathogenic, likely pathogenic or variant of uncertain significance).
- Participants must **either** have independent functional abilities (such as walking, communicating, and picking up small objects) **or** be seizure-free.

The study involves filling out caregiver questionnaires remotely (approximately 2 hours total) and one 60-minute virtual interview with the study team. No on-site visits are required for study completion. Participants who complete the study will be offered two gift cards valued at \$100.00 total as a token of appreciation for their participation.

Additional, optional opportunities throughout the study will include the choice to record virtual research visits, for the purpose of data analysis only. For female participants who are located in the United States, participants will have the option to provide blood and saliva samples that will be analyzed for a genetic pattern called X-inactivation skewing, conducted by Greenwood Genetic Center. U.S. participants will have the opportunity to provide blood samples that will be saved in a blood bank at Coriell Institute for Medical Research, to be used in future studies, after all samples are de-identified. You can still participate in this study even if you do not consent to these additional, optional opportunities.

The goal of the study is to understand the clinical features of individuals with this presentation and to establish related genetic and molecular patterns. We expect to enroll 30-50 individuals in the study and are recruiting internationally.

The first step to participating in this study is to speak with our study team to understand the details of the study and decide whether you would like to participate. If you decide to enroll your child in the study, you will be required to sign a consent form prior to initiation of any research activities.

If you are interested in participating in the ExCeeD Study, have any questions, or would like more information, please feel free to contact our study team at ExCeeD@childrens.harvard.edu. You can also telephone our study coordinator, Kathryn Mansour, directly at 617-355-1494.

We would like to contact you about this study, but do not want to bother you if you are not interested. If you do NOT want to be contacted, please call or e-mail us indicating that you do NOT want us to contact you any further about this study.

If we do not receive an email or call from you within the next two weeks, a study coordinator will telephone you to discuss the study. Allowing us to contact you does not mean that you have agreed to participate in the study, but only that you are willing to discuss it with us further.

We look forward to the opportunity to speak with you.

**Translational Neuroscience Center
2 Brookline Pl, Brookline, MA 02445
Tel: 617-355-0601/617-355-1494**

Sincerely,

Heather Olson, MD MS
Director, CDKL5 Center of Excellence
Boston Children's Hospital