

6/8/18

Dear CDKL5 Families,

We would love your help with a study evaluating anatomic connectivity in the brain of patients with CDKL5 deficiency disorder. This study will use a technique called diffusion tensor imaging (DTI). The data is often collected but not processed during routine clinical MRI scans of the brain.

At Boston Children's Hospital we have preliminary data from patients with CDKL5 deficiency disorder enrolled in the CDKL5 Clinic Study at Boston Children's Hospital. We are now teaming up with basic scientist Dr. Michela Fagiolini and clinician-scientist Jurriaan Peters, also at Boston Children's Hospital/Harvard Medical School, for a parallel study in patients and mice to evaluate functional and anatomic connectivity. This is funded through the University of Pennsylvania Orphan Disease Center/Lou Lou Foundation. To make the human study feasible we need to recruit patients who have had MRIs. It is important to first evaluate this finding using data that has already been collected in order to avoid sedating patients for research MRI scans.

If you would like to share your child's MRI(s), we will ask you to enroll in our CDKL5 Clinic Study so that we can collect identifiable information. This will include MRI CDs and other clinical records that allow us to identify your child's CDKL5 genotype and phenotype. If you are interested, you can contact our CDKL5 research team at cdkl5@childrens.harvard.edu or 617-355-5230.

We appreciate your time and consideration concerning this research study.

Sincerely,



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