

CDKL5 Program of Excellence 2018 Pilot Grant Program

Project Title: "Single-cell Transcriptional Profiling of Mosaic CDKL5 Deficiency Disorder Brain Tissue"

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The full effects of CDKL5 loss on neural function remain unclear, and this knowledge would greatly aid in the development of new therapeutics for CDKL5 Deficiency Disorder (CDD). To address this issue, we propose to systematically profile gene expression changes in the CDD-affected brain using a novel technique that allows us to distinguish mutant and unaffected cells within complex tissue samples. Similar studies will also be conducted in a mouse model of CDD to establish the degree to which this model recapitulates changes observed in the human disorder. Together, these studies should provide critical new insights into the downstream effects of CDKL5 loss and open new avenues for the development and evaluation of therapeutic strategies to combat this devastating disorder.