CDKL5 Program of Excellence
2020 Pilot Grant Program

Project Title: "Ascertaining the cell-autonomous effects of CDKL5 mutations in every brain cell type"

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Institution: Harvard Medical School

We will systematically identify the effects of *CDKL5* mutations on each of the cell types in the human brain to inform the discovery of therapies and biomarkers for CDKL5 deficiency disorder (CDD). To do this, we will analyze RNA expression in more than 150,000 individual cells sampled from *post mortem* brain tissue from female CDD patients. Using an analytical approach we recently developed, we will compare, for each cell type, cells that express normal *CDKL5* and cells that (due to X-inactivation) express mutated *CDKL5*. We will make the resulting data widely available and use it to identify affected cell types, key biological pathways for therapeutic intervention, and candidate biomarkers.