CDKL5 Program of Excellence
2020 Pilot Grant Program

Project Title: "Characterization and correction of Cdkl5 nuclear function in human CDD neurons during maturation"

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CDKL5 deficiency disorder is a severe neurological condition without a cure characterized by seizures, developmental delay and intellectual disability that affects children from early on after birth. Understanding the early molecular alterations that *CDKL5* mutations cause in human neurons is essential to develop new molecular therapies to restore normal function in the nervous system before damage that cannot be repaired has occurred. This research proposal aims to systematically investigate and correct via modern genome editing approaches the initial changes induced in the nucleus of human neurons from CDKL5 syndrome patients. These experiments will allow to clarify the early modifications in gene expression that affect neuronal function of CDKL5 patients that can be targeted for pharmacological or gene therapy strategies