CDKL5 Program of Excellence
2019 Pilot Grant Program

Project Title: "CDKL-5Deficiency Disorder (CDD) Biomarkers Discovery Through System Biology"

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CDKL5 deficiency disorder (CDD) is a devastating disease where early onset neurological symptoms dominate the affliction. The common view of this disease is 'neurocentric'. However, we argue that the biology of CDKL5 as well as some of the disease characteristics suggest that the disease affects the whole patient. This is a novel idea that we have termed the 'systemic hypothesis'. We argue that the systemic hypothesis is important to define experimental strategies for the identification of molecules, also now as biomarkers, useful to measure disease severity and response to treatments. In this project, we will quantify simultaneously the expression of thousands of proteins in brain and peripheral tissues of a mouse model of CDKL-5DD. We will use these data to identify biomarkers of disease focusing on molecules that can be easily accessed in patient samples such as plasma, urine, and cerebrospinal fluid.