Breaking the Mitochondrial–Synaptic Pathogenic Loop to Restore Neural Network in CDKL5 Deficiency

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CDKL5 Deficiency Disorder (CDD) is a severe condition that causes early seizures and lifelong neurological challenges. The brain's communication network, the "information highways" of the brain, is responsible for cognitive functions. In CDD children, a disrupted brain's communication network leads to both seizures and neurological difficulties, debilitating CDD symptoms due to a harmful cycle in the brain where damaged "energy factories" (mitochondria) and faulty connections between brain cells (synapses) worsen each other. Although FDA-approved Ztalmy helps to control seizures, it does not break the disease cycle. Therefore, to interrupt this cycle, we will use novel patented brain imaging to track these root causes in real time and test two already FDA-approved drugs, to see if supporting both energy function and brain cell connections will restore brain communication networks, consequently, better cognitive functions. By targeting the root problem - the disrupted brain's communication network – this work aims to go beyond just controlling seizures, but instead improve brain development and everyday functioning in children with CDD.