Executive Function in Cystinosis

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Seminal behavioral studies by Trauner and others have suggested the presence of cognitive dysfunction in Cystinosis. Our own work suggests some behavioral and neural differences and difficulties with sensory memory in this population. However, the neurocognitive phenotype associated with CTNS mutations and its developmental path are still poorly understood, and the prime areas of neurocognitive vulnerability in this population are in need of much more thorough characterization. This is critical to developing effective therapies to compensate for or improve on areas of cognitive vulnerability. To this end, we have been characterizing different components of executive functioning in Cystinosis using high-density electrophysiology (EEG)—a non-invasive method that allows one to directly measure functional brain activity at the millisecond scale and thus reliably assess the integrity of information processing at the neural level—and standardized cognitive functional assessments.

Executive function refers to the set of cognitive processes that: (1) guide action and behaviors essential to aspects of learning and everyday human performance; (2) contribute to the monitoring or regulation of performance; and (3) relate not only to the cognitive domain, but also to socioemotional and behavioral domains. Executive function abilities—such as memory updating, set shifting, conflict monitoring, and inhibition—are critical for academic, professional, and social achievements. To define, plan, and execute daily goals, for example, one must engage memory to maintain objectives, inhibit responses to task-irrelevant distracting information, and set shift to adapt to the changing demands of one's environment.

In this talk, we will present preliminary findings on different components of executive functioning in Cystinosis and discuss how those compare to what one would expect to see in the general population. Relatedly, we will review strategies that may be used to address executive functioning differences. Lastly, given that a significant number of participants is visiting us for the second time, we will briefly discuss longitudinal changes in cognitive and neural function in Cystinosis.