2016 Million Dollar Bike Ride
Pilot Grant Program

**Application Title:** Role of mitochondria and ROS in etiology of SRS

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Synder-Robinson Syndrome (SRS) is an X-linked intellectual disability caused by mutations in SMS, spermine synthase; which results in a lack of spermine. Neurological manifestations present in SRS patients require an understanding of the neuropathy of SMS-deficiency. We have preliminary data that indicate cells from SRS patients may have a mitochondrial dysfunction. In order to explore this possibility and perhaps develop a potential treatment, we plan to conduct some studies.

First, since spermine, absent in patients with SRS, facilitates calcium’s uptake by mitochondria, we will determine the levels of calcium within the cell and the mitochondria of patients and controls. Second, mitochondrial function will be determined by how well certain substrates are utilized by the mitochondria of patients and controls. We will also measure the level of reactive oxidative species (ROS) in the same cells. Finally, based on the results, we will examine if compounds affecting calcium balance, ROS accumulation and mitochondrial transport can rescue the metabolic phenotype we have observed in patient cells.