2016 Million Dollar Bike Ride
Pilot Grant Program

Application Title: Carboxyl lysosphingomyelin as a Biomarker for Diagnosis and Assessment of Treatment of NPC Disease

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A major challenge for developing treatments for Niemann–Pick C (NPC) disease is difficulty in evaluation of efficacy. This is complicated by limited patient numbers, and heterogeneity in age, severity, and stage of disease progression. Biomarkers that reflect NPC disease status could provide a valuable surrogate endpoint for assessment of treatment effect and reasonably predict clinical benefit. We have identified a new lipid biomarker that is elevated in the cerebrospinal fluid (CSF) and plasma of NPC patients and correlates with disease severity. In this proposal, we will examine a new biomarker to improve diagnosis of NPC and to facilitate drug development. This will be accomplished by developing an assay for the biomarker and testing whether the marker is predictive of clinical outcomes in 2-hydroxypropyl-β-cyclodextrin (HPβCD)-treated NPC1 cats and human subjects enrolled in NIH NPC Natural History Study and the Phase 1/2a trial of intrathecal HPβCD. Our goal is to develop a robust biomarker that can serve as an outcome measure to evaluate efficacy of promising therapeutics.