

Dr. Stephanie M. Cologna –

Differential Protein Expression Profiling in Cerebrospinal Fluid from Niemann-Pick Disease, type C1 Patients

Lay Summary

The goal of this research is to identify and quantify potential protein biomarkers in the cerebrospinal fluid from individuals with Niemann-Pick type C1. A comprehensive evaluation of those proteins that change in the brain may provide understanding of the cellular mechanisms causing the neurological problems in Niemann-Pick type C1. The protein biomarkers will provide a measurement to evaluate therapeutic interventions for Niemann-Pick type C or the protein biomarkers may display pathways that can be studied as potential therapeutic targets.

Mass Spectrometry will be used for protein identification. We propose to expand a technology known as “isobaric Tags for Relative and Absolute Quantitation” (iTRAQ) and develop analysis software to allow this technique to be applied across a large group of individuals. This experimental design will provide a means to evaluate CSF proteins with respect to age, disease severity, and changes with treatment (e.g., miglustat and 2-hydroxypropyl- β -cyclodextrin).