



**December 31, 2018**

**To:** National Niemann-Pick Disease Foundation

**Re:** Edward Schuchman Research Fellowship Progress

**Report Period:** Final Report

**Lay Summary**

Our National Niemann-Pick Disease Foundation's Edward Schuchman Research project provides comprehensive evaluations for families affected with acid sphingomyelinase deficiency (ASMD), also known as types A and B Niemann Pick disease. Since transferring this study to the Children's Hospital at Montefiore (CHAM) in 2017, we have evaluated 15 subjects with ASMD – 13 with Type B and two with Type A. In addition, because of travel restrictions, we have had phone consultations with international patients (Pakistan, China). Importantly, all studies included in the original protocol, including radiologic studies, laboratory evaluations, biomarker studies, pulmonary tests, and consultations with experts have been performed at CHAM. New study activities focusing on enhancing our understanding of lung, brain, and liver disease have been ongoing. We continue to collect elastography data, and now have information on 12 patients including one patient who has had multiple measurements over time. We are now analyzing the elastography data and writing an abstract for submission to the 2019 ACMG meeting. We are also collaborating with the International Niemann-Pick Disease Alliance on streamlining data collection for ASMD. Overall, we have been very productive and look forward to continuing to learn from the ASMD community in order to improve understanding of this rare, complex disease.