



January 9, 2018

To: National Niemann-Pick Disease Foundation

Re: Edward Schuchman Research Fellowship Progress

Report Period: August 19, 2016 - December 19, 2017

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 our last progress report, we have dedicated time to fully transition this study to the Children's Hospital at Montefiore (CHAM). We gained institutional approval to begin the study in April 2017 and have evaluated 12 subjects (11 Type B, 1 Type A) at CHAM between May and November 2017, with 6 subjects waiting to be scheduled for assessments. In addition, because of travel restrictions, we had three phone consultations with international patients from Russia, China, and Pakistan. Importantly, all studies included in the original protocol, including radiologic studies, laboratory evaluations, biomarker studies, pulmonary tests, and consultations with experts have continued at CHAM. New study activities focusing on enhancing our understanding of lung, brain, and liver disease have been added on to the protocol. Relating to these new activities, nine of the 12 subjects had liver elastography to assess the hepatic fibrosis in patients with ASMD, which was funded by NNPDPF money. Overall, we have had a very productive interval period, despite a substantial move. We look forward to continuing to learn from the ASMD community in order to improve understanding of this rare, complex disease.