

August 19, 2016

To: National Niemann Pick Disease Foundation

Re: Edward Schuchman Research Fellowship Progress Report

Period: October 1, 2015-August 18, 2016

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 evaluated another 11 subjects at Mount Sinai between October 2015 and February 2016. In addition, we created a RedCap database for ASMD and retrospectively entered more than 10 years' worth of patient data. In April, 2016, Dr. Wasserstein moved from Mount Sinai to become the Chief of Pediatric Genetic Medicine at the Children's Hospital at Montefiore (CHAM). Since that time, significant progress has been made towards transitioning this project to CHAM. All studies included in the original protocol, including radiologic studies, laboratory evaluations, biomarker studies, pulmonary tests, and consultations with experts will continue at CHAM. New study activities focusing on enhancing our understanding of lung, brain, and liver disease are being added on to the protocol. 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.