



**July 1, 2019**

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

**Re:** Edward Schuchman Research Fellowship Progress

**Report Period:** January 1 – June 30, 2019

**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 evaluated three new subjects - two subjects with Type A and one with Type B, and re-evaluated one existing subject at the Children's Hospital at Montefiore (CHAM). We currently have one additional subject scheduled for an assessment this Summer. 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 ongoing. We continue to collect elastography data, and now have information on 12 patients including two patient who has had multiple measurements over time. We are analyzing the elastography data and writing up the findings. We continue to collaborate with the International Niemann-Pick Disease Alliance on streamlining data collection for ASMD. We are working on updating our research website to facilitate increased visibility of our program for ASMD. Overall, we have had a very productive period. We look forward to continuing to learn from the ASMD community in order to improve understanding of this rare, complex disease.

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**To:** National Niemann-Pick Disease Foundation  
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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 (December 31, 2018), we have assessed two Type B and two with Type A subjects. This past two quarters, we continued to dedicate time and personnel to improving the data points of the longitudinal RedCap database. In collaboration with International Niemann-Pick Disease Alliance, we are re-evaluating, cleaning, and revising the electronic case report forms to incorporate the new assessments and harmonize data points being collected from CHAM. 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 ongoing.

Relating to these new activities, we are reviewing the data collected from the 12 liver elastographies collected to date, which was funded by NNPDF money. If there is evidence of abnormality, we will apply for a separate funding mechanism to support additional work in this area. In addition, we proposed studying if there are cerebral electrophysiologic changes in patients with type B disease, focusing on those with suspected intermediate, or type A/B, disease. This will be done at the Human Phenotyping Core of Einstein's Rose F. Kennedy Center in conjunction with Dr. Sophie Molholm under a separate protocol and informed consent. To date, we have not had an intermediate type assessed. Similar to the elastography studies, we will use this NNPDF award to fund about four studies; if there is evidence of abnormality, we will apply for a separate funding mechanism to support additional work. Overall, we have had a very productive funding period. We have begun updating our website to increase visibility of this important study. We look forward to continuing to learn from the ASMD community in order to improve understanding of this rare, complex disease.



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