October 8, 2018

To: National Niemann-Pick Disease Foundation
Re: Edward Schuchman Research Fellowship Progress
Report Period: February 15 - August 15, 2018

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 one subject withType Aand one with type B at the Children's Hospital at Montefiore (CHAM), with 3 subjects scheduled for upcoming assessments. In addition, because of travel restrictions, we have had phone consultations with international patients (Pakistan, China) and have another scheduled next month. 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. We continue to collect elastography data, and now have information on ten 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 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.

August 15, 2018

To: National Niemann-Pick Disease Foundation
Re: Edward Schuchman Research Fellowship Progress
Report Period: February 15 - August 15, 2018

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, this study has been fully implemented at the Children's Hospital at Montefiore (CHAM). Institutional review board approval was regained on March 18, 2018, 1 subject (Type A) has been assessed under this protocol between February and August 2018, with 3 subjects scheduled for assessments in the Fall. In addition, because of travel restrictions, we had two phone consultations with international patients from Pakistan. We also continue to dedicate time to improving the data points of the longitudinal RedCap database. In collaboration with International Niemann-Pick Disease Alliance, we have begun re-evaluating, cleaning, and revising the electronic case report forms to incorporate the new assessments and 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 have begun 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 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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