January 7, 2020

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 23 subjects with ASMD – 16 with Type B, one with Type A/B and six with Type A. In 2019, we performed 11 evaluations – four new Type A, one new Type A/B, and three new Type B, as well as three re-assessments on existing Type B subjects. 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 13 patients including one patient who has had multiple measurements over time. We are also finishing the ASMD Natural History Database rebuild, and expect to begin chart review and data entry in early 2020. 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.

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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 transferring this study to the Children's Hospital at Montefiore (CHAM) in 2017, we have evaluated 23 subjects with ASMD – 16 with Type B, one with Type A/B and six with Type A. In 2019, we performed 11 evaluations – four new Type A, one new Type A/B, and three new Type B, as well as three re-assessments on existing Type B subjects. Additionally, we had five consultations. One in person with a family who will come back next year for assessments, one who decided not to consent to the Natural History Study at this time, and due to travel restrictions, three phone consultations with international patients from Pakistan and China. 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 have begun re-evaluating, cleaning, and revising the electronic case report forms to incorporate the new assessments and data points being collected from CHAM. We expect the database to be completed in January 2020. 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 13 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. Additionally, due to the transfer of Institutions, negotiation of research rates, and timing of the distribution of funds we have approximately $45,930 of leftover funds to continue this important work. We look forward to continuing to learn from the ASMD community in order to improve understanding of this rare, complex disease.

The Children's Hospital at Montefiore

Department of Pediatrics
Division of Genetic Medicine

Division Chief
Melissa Wasserstein, M.D.

Clinical Genetics
Paul A. Levy, M.D.
Robert W. Marion, M.D.
John Greally, Ph.D., MB.B.Ch

Biochemical Genetics
Paul A. Levy, M.D.
Melissa Wasserstein, M.D.

Cancer Genetics
Robert Burk, M.D.

Reproductive Genetics
Susan Klugman, M.D.
Kumal Bajaj, M.D.
Siobhan Dolan, M.D.
Tamar Goldwaser, M.D.

Genetic Counselors
Kaitlyn Brown, M.S.
Ellen Hantman, M.S.
Nicolette Ioele, M.S.
Katie Gallagher, M.S., C.G.C.
Monisha Sebastin, M.S.

Metabolic Dietician
Susan Brodlie R.D., M.S.

Research Manager
Nicole Kelly, M.P.H.

Research Coordinators
Jessica Fischetti, M.A.
Jessenia Lopez, M.S.
Estefany Maria, B.A.
Shanice Hamilton, B.S.

Contact us
Pediatric Genetic Medicine
Children's Hospital at Montefiore
3415 Bainbridge Avenue
Bronx, New York 10467

(718) 741-2323 Office
(718) 920-6506 Fax

EINSTEIN
Albert Einstein College of Medicine