Washington University in St. Louis
SCHOOL OF MEDICINE

Progress Report

Infantile Neuroaxonal Dystrophy (INAD) Summary of the Research Plan

June 2016

Report by
Paul T. Kotzbauer, M.D., Ph.D.
Associate Professor of Neurology

During the first year of our INAD research project, the Kotzbauer lab has pursued two strategies to develop INAD treatments.

The Kotzbauer Lab

For the first strategy, we developed laboratory methods to screen for drugs that improve the function of PLA2G6 proteins with INAD-causing mutations. We used these methods to demonstrate that INAD mutations cause the PLA2G6 protein to be unstable and have also demonstrated that under the right conditions it is feasible to improve the stability and enzyme function of a PLA2G6 protein with an INAD mutation.

For the second strategy, we made progress in developing laboratory methods to screen for drugs that stimulate other enzymes to compensate for the loss of PLA2G6 function. Further studies of this strategy have directed us to test one particular group of compounds that might achieve this goal, which we plan to begin testing soon.

We are working to further develop the screening methods in both strategies in order to begin screening compounds over the next year. These initial screening efforts could identify molecules that are suitable for further optimization and testing as therapeutic agents.