A Mouse Model for Familial Dysautonomia

Familial Dysautonomia (FD) is a genetic disorder of the autonomic nervous system caused by a mutation in the gene IKBKAP on chromosome 9. This mutation leads to a decrease in expression of the IkB kinase complex associated protein (IKAP) and has detrimental effects on the development and function of autonomic and sensory neurons. FD is an autosomal recessive trait found mostly in Ashkenazi Jews, with approximately 1 in 27 being carriers of the trait. There is currently no cure for FD and half of all affected individuals die before they reach 40 years of age. To explore the disorder we have engineered a line of mice that express a conditional knockout of the IKBKAP gene in cells containing alpha-tubulin, a component of neurons. We are interested in analyzing the effect of this mutation on the nervous tissues as well as organ systems. Our methodology involves cryosectioning mutant and control mice tissues and using immunohistochemistry to stain for cells of interest. By examining the microanatomy displayed in this disease, we are able to further understand how this genetic mutation leads to the symptoms of FD and gain insight on which preventative measures and medications will have the best results in increasing the quality and length of life of FD patients.