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Identification of genes regulated by IKBKAP: Investigating why neurons die in the disease Familial Dysautonomia

Familial Dysautonomia is a disease of the peripheral nervous system caused by a mutation of the IKBKAP gene on human chromosome nine. The goal of this research project is to determine what effect the knock-out of this gene has on transgenic model mice. In particular, it examines why proteins previously found to have altered concentrations in the mutant mice are present in different amounts compared to the control. To look at the expression of these genes, which include neuropeptide Y, parvalbumin, and substance P, the polymerase chain reaction was used to amplify these genes from reverse transcribed RNA isolated from both mutant and control mouse tissue. The PCR products were then run in agarose gel electrophoresis to determine expression levels. Due mostly to a lack of time, the project has yet to produce any conclusive results but work continues in order to obtain evidence concerning gene expression levels in the model mice.