Association of Phospholipase D3 Gene with Alzheimer's Disease

Alzheimer's Disease (AD) is the most common type of dementia worldwide. Currently, researchers have associated many different agents to the development of AD; however, this paper will be, specifically, investigating the major contributor to the development of Late-Onset Alzheimer's Disease (LOAD): genetics. In the study, the proposed research question is whether there is any association of Phospholipase D3 (PLD3) gene Val(Valine)232Met(Methionine) variant with LOAD. The role of mutated PLD3 gene needs to be studied because the normal product of the gene, PLD3 protein, plays a crucial role in the normal functioning of lysosomes (essentially the breakdown of cellular products) in the endosomal-lysosomal system in cells. It is hypothesized that the PLD3 gene variant will have an association with the cadaver specimens diagnosed with LOAD. Here, the study will utilize 10 cadaver specimens available at MSSU cadaver lab to observe the association of the gene variant with histologically diagnosed LOAD cadaver specimens through DNA sequencing. In the study, the presence of the gene variant in the cadaver specimens is most likely to be associated with LOAD, and the level of significance will be assessed using a Fisher's Exact Test. The association will further corroborate the current research on PLD3 gene's role in the development of LOAD and facilitate additional research involving this gene through different approaches to fully confirm the association and develop therapies that will target the abnormal protein or a novel gene therapy that will fix the mutation in the gene to decrease the likelihood of developing LOAD.