GJB3 AND ITS RELATIONSHIP TO PRESBYCUSIS

Mia Johnson, Stephanie Techney, D. Robert Frisina, David A. Eddins and Dina L. Newman*, Department of Biology at Monroe Community College, Department of Biological Sciences and the International Center for Hearing and Speech Research at RIT, mxjsbi@rit.edu, sjtsbi@rit.edu, dina.newman@rit.edu

Presbycusis, or age-related hearing loss, is a common disorder among the elderly community. Previous studies have shown a significant genetic component to presbycusis. The specific gene that causes susceptibility to presbycusis has yet to be discovered, so we are conducting research on the gene *GJB3* for an association between this gene and presbycusis. *GJB3* codes for connexin 31, a gap junction protein found in the cochlea of the inner ear. Mutations in *GJB3* can cause congenital deafness by disrupting the flow of potassium in the ear. We hypothesize that single nucleotide polymorphisms (SNPs) in *GJB3* maybe associated with the related phenotype, presbycusis. Previously the coding region and surrounding sequence were examined for variation in a screening set of 20 human subjects. We sequenced the promoter region and the 5' untranslated exon in 24 subjects to search for additional variation that might have regulatory consequences on the gene's expression. Three additional SNPs and one insertion/deletion were identified. Future work will investigate whether any of these genetic variants is associated with hearing loss. Finding the causative gene for presbycusis could possibly lead to early detection and new treatments.