

**INVESTIGATION OF CONNEXIN GENES *GJB2* AND *GJB6* FOR POTENTIAL INVOLVEMENT IN THE DEVELOPMENT OF PRESBYCUSIS.** Aziana Ismail, Rebecca Lebowitz, D. Robert Frisina, David A. Eddins, Dina L. Newman\*, Department of Biological Sciences and the International Center for Hearing and Speech Research, [axi4073@rit.edu](mailto:axi4073@rit.edu), [dina.newman@rit.edu](mailto:dina.newman@rit.edu)

Presbycusis, or age-related hearing loss, is a hearing disability due to deterioration within the ear commonly found among old people. Although no genes are yet known to cause presbycusis in humans, many genes are implicated in various forms of congenital deafness. Mutations in gap junction genes *GJB2* (connexin 26) and *GJB6* (connexin 30) are the most common cause of nonsyndromic sensorineural hearing impairment; therefore, these genes were chosen as candidate genes for possible role in presbycusis. We have sequenced sections of *GJB2* and *GJB6* from 20 human subjects with presbycusis. Both exons of *GJB2* have been completely sequenced, and 1500 bp upstream of the gene has been partially sequenced in the screening set. For *GJB6*, all three exons have been entirely sequenced, and 1500 bp upstream of the gene has been partly sequenced in the screening set. To date, 12 single nucleotide polymorphisms (SNPs, i.e. individuals differ in their DNA sequence by a single base) have been found in *GJB2* and seven SNPs in *GJB6*. Three SNPs (two in *GJB2* and one in *GJB6*) were analyzed for association with presbycusis in the entire sample set; however, no correlation between phenotype and genotypes was found. Future work will involve completing the sequencing of the screening set for these two genes as well as genotyping additional SNPs for the entire population in order to determine whether or not variation in these genes is associated with presbycusis.