

***ALDH18A1*- A POSSIBLE ROLE IN AGE RELATED HEARING LOSS. Danielle Cunningham, Dina L. Newman\***, Department of Biological Sciences, Rochester Institute of Technology, [dx4534@rit.edu](mailto:dx4534@rit.edu), [dina.newman@rit.edu](mailto:dina.newman@rit.edu)

*ALDH18A1* (Aldehyde Dehydrogenase 18 family, member A1), also known as *PYCS* (Pyrroline 5-Carboxylate Synthetase), catalyzes the reduction of glutamate to delta1-pyrroline-5-carboxylate. Mutations in this gene have been known to cause neurodegeneration, cataracts, connective tissue diseases, and a multitude of other disorders. Glutamate has been shown to be the main excitatory neurotransmitter involved in hearing. Previous studies have show that *Aldh18A1* is downregulated in the auditory nervous system of elderly mice. Thus we are examining variants within the gene to determine if any could be correlated with age-related hearing loss in humans (presbycusis). Several variants have been published that cause amino acid changes or frame shift mutations: T297I, T297P, Q363STOP, S370Y, V592I, frameshift at amino acid 3, R38K, R84Q, and S264P. We have begun screening each of these sites to see if both alleles are present in our population of >600 European Americans over age 58. For those SNPs that are present, we will genotype the entire population and then look for correlations between genotype and hearing phenotypes. So far several SNP sites have been examined, but only T297I has a significant frequency in the population.