

INVESTIGATION OF THE MITOCHONDRIAL GENOME FOR VARIANTS THAT PROTECT AGAINST HEARING LOSS. *Andrea Braganza, Amanda Souza, Laura Birdsall, Eric Stevens, Dina L. Newman**, Human Genetics Laboratory, Department of Biological Sciences, acb8467@rit.edu, als2389@rit.edu, lab5164@rit.edu, els9572@rit.edu, dina.newman@rit.edu

Presbycusis is a disease of the elderly, where as people age they start to lose their hearing, first in the higher frequencies and later in the lower frequencies too. Genetic as well as environmental factors cause presbycusis. However the genetic component is unclear. Previous studies have shown a correlation between mother and child but not father and child in the development of presbycusis. Since mitochondrial DNA (mtDNA) is a non-recombining molecule that is inherited maternally; we hypothesize that there are DNA variations in the mt genome which cause certain people to either be susceptible to or protected from developing presbycusis. Differences in mtDNA sequences are only due to mutations. Therefore as time passes, mutations accumulate sequentially along less and less related molecules that constitute independent lineages known as haplogroups. In previous work done in the lab on the nine European haplogroups and presbycusis, we have found that men who belong to haplogroup K seem to have better hearing than men from all other haplogroups. Then we sequenced the entire mitochondrial genomes of twelve subjects from the five most common haplogroups, and found 141 polymorphisms. We are currently genotyping SNPs that appear to be specific to haplogroup K in order to find the genetic differences that cause the phenotypic differences. From recent studies we found a new set of SNPs that can be used to more accurately classify people into the nine major European haplogroups. Therefore we have begun genotyping our samples for these new SNPs as well. With this new information, along with improved statistical methodology, we will redo the analysis.