

CORRELATION BETWEEN MITOCHONDRIAL HAPLOGROUPS AND PRESBYCUSIS Andrea C. Braganza, Sesquilé Ramón, Robert D. Frisina, David A. Eddins, Dina L. Newman*, Department of Biological Sciences and the International Center for Hearing and Speech Research, acb8467@rit.edu, dina.newman@rit.edu

Presbycusis, or age related hearing loss, is a disease that affects the elderly. It starts with loss of hearing in the higher frequencies and then the lower frequencies. People with presbycusis have problems hearing and telling apart high pitched sounds such as ‘s’ and ‘th’. Certain sounds seem annoying or overly loud. Since it is a progressive disease, people with presbycusis sometimes do not realize that their hearing is diminishing. Previous experiments have shown that people who are maternally related have similar risks of developing presbycusis, and we know that mitochondrial (mt) DNA is maternally inherited. Mt lineages cluster into haplogroups, with each haplogroup representing the maternal-line descendants of a single person who first showed a particular mutation. Therefore we are searching for a correlation between a certain haplogroup and the risk of developing presbycusis. The subjects were phenotyped with classical and advanced hearing tests, and blood was drawn at ICHSR for DNA extraction. Using Custom TaqMan Assays (Real-Time PCR) for nine SNP sites and a combination of RFLP and direct sequencing for a tenth, we classified the subjects into the nine major European mt haplogroups (and “other”). Controls were confirmed by direct sequencing. Preliminary results show a significant difference between subjects belonging to different haplogroups; subjects from haplogroup H seem to have better hearing as compared to subjects from the other haplogroups. We are currently working on increasing the sample set to improve our statistical analyses. Future work will help to pinpoint the important difference(s) in the mt DNA between the different haplogroups that cause differences in hearing. This knowledge will be essential in counseling patients, implementing prevention strategies and developing new treatments for a disease that affects the day-to-day life of millions of Americans.