

AUTOMATED ANALYSIS TO DETERMINE THE ASSOCIATION OF HUMAN MITOCHONDRIAL HAPLOGROUPS WITH PRESBYCUSIS Rhea Sanchez, Robert Parody, Dina L. Newman*, Michael Osier*, *rheasanchez47@yahoo.com*, *rjpeqa@rit.edu*, *dina.newman@rit.edu*, *michael@bioinformatics.rit.edu*

Presbycusis is the progressive deterioration of hearing that occurs over a lifetime. Based on recent research, there is reason to believe that susceptibility to presbycusis may be at least partially caused by the inherited variations in mitochondrial DNA. To investigate this hypothesis, a series of audiologic and genetic tests were conducted on 250 human subjects over the age of 58 with hearing abilities ranging from severe presbycusis to perfect hearing, also known as “golden ears”. The data collected from these tests were too large to analyze manually. This problem of efficiency revealed the need for conducting the analyses computationally. Perl (Practical Extraction and Report Language) is the programming language that was used to develop an analysis program: the Presbycusis Data Analysis Tool (PDAT). The program takes both the genetic and phenotypic data for each subject. First, each individual is categorized into one of nine major European mitochondrial haplogroups based on the known genotypes at the ten Single Nucleotide Polymorphisms (SNPs) which determine each haplogroup. After categorization, two statistical tests were performed on the phenotypic data to determine whether or not any one particular haplogroup is more susceptible to presbycusis relative to the remaining haplogroups. A two-sample t-test for identical means is used to test the significance of individual genotype-phenotype combinations. A test for differences in slope, as determined by linear regression, between individuals with or without a particular haplogroup is used to compare hearing ability across a range of frequencies.