

P67: Age related hearing loss in Portugal: methods and mutations

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Introduction: Presbycusis is a multifactorial disease that represents the damage of intrinsic and extrinsic factors, acting in the inner ear through life. Presbycusis or age-related hearing loss (ARHL) is estimated to affect up to 1.5 billion of people aged 60 by 2025.

Objectives: The aim of this study was to genetically characterize individuals with presbycusis, for *GJB6*, *NAT2*, *GRM7* genes and mtDNA haplogroups in order to evaluate contributions to ARHL diagnosis

Materials and Methods: DNA was obtained from blood samples of Portuguese individuals >65. All subjects signed an informed consent, fill a survey and perform audiological evaluation. *HVS1* region was sequenced for mtDNA haplogroup (mtDNA Haplogroup Analysis) estimation. *GJB6* analysis was assessed by Multiplex PCR. *NAT2* and *GRM7* analysis was performed by qPCR. *NAT2* phenotype was determined using software NAT2PRED

Results and Discussion: We screen samples from 318 individuals (25.5% man and 74.5% women). The del(*GJB6*-D13S1830) was present in 0.87% of the individuals. Haplogroup H was the most common observed (54.3%) in our sample. This haplogroup was associated with an average severity of HL of 39.42 dB. Significant differences ($p < 0.001$, binomial exact test) were found between both sexes. T allele at *GRM7* was the most observed (94.3%) in our sample. Slow acetylation phenotype was the most common (55.7%) observed in *NAT2* gene, and corresponds to an average HL of 29.40 dB in our sample.

Conclusion: Haplogroup H is associated to average severity hearing loss. Results for *NAT2* gene were in accordance with expected for European populations. The T allele of *GRM7* gene associated to ARHL, was the most common in our sample. Consequently the A allele, observed only in 6 individuals with an average HL of < 30 dB, points to a possible protective role of A allele of *GRM7* gene.