

Study of *KCNE1* gene in patients with Ménière's Disease

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Introduction

The Ménière's Disease (MD) is characterized by vertigo, tinnitus, hearing loss and aural fullness, symptoms caused by the increased volume of endolymph, present in the inner ear labyrinth. The MD has unknown etiology, but changes in some genes involved in the endolymph homeostasis may be associated with the disease.

Observing that the scientific literature does not present much regarding the subject, the objective of this research is to verify if genetic variations in *KCNE1* gene may be associated with MD unleashing. For the study, 30 patients with MD clinical diagnosis were selected and submitted to clinical, otorhinolaryngologic and complementary exams at the Federal University of São Paulo otoneurology ambulatory and the *KCNE1* molecular analysis of these patients were performed at Molecular Biology and Genetic Engineering Center (CBMEG/UNICAMP).

Results and Discussion

In exon 3 of the *KCNE1* gene, it was found the rs1805127 (c.112A>G) and rs17173510 (c.84G>A) Simple Nucleotide Polymorphisms (SNPs) and a *missense* mutation, N75H (c.223A>C).

Genetic alteration	Regular patients	Heterozygous patients	Homozygous patients
c.112A>G	3	12	15
c.84G>A	29	1	0
c.223A>C	29	1	0

Table 1. Number of patients according to the alterations found in exon 3 of *KCNE1* gene.

The c.112A>G variation, found in heterozygosis and homozygosis, causes the replacement of the amino acid serine for glycine at codon 38 of the protein while the c.84G>A alteration does not cause any change.

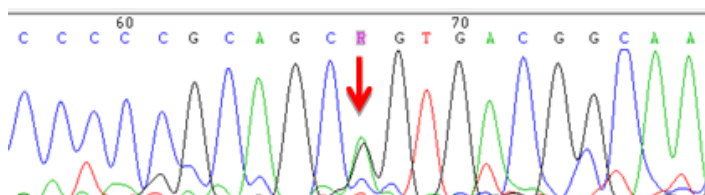


Figure 2. Electropherogram indicating the c.112A>G alteration in heterozygosis.

A patient also presented the c.223A>C alteration, in a heterozygosis. This mutation is characterized by the replacement of Asparagine for Histidine at codon 75 of the protein.

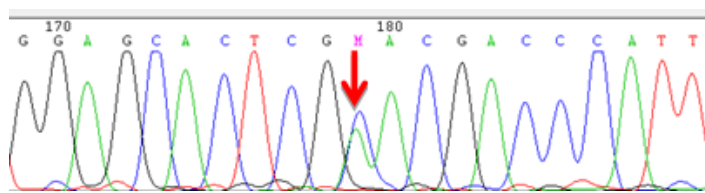


Figure 3. Electropherogram indicating the c.223A>C alteration in heterozygosis.

Furthermore, the N75H mutation has not yet been described in the literature and was not detected in control sample. Therefore, this variation requires studies to prove its possible pathogenic value and involvement with MD.

Although the rs1805127 SNP has been associated with increased susceptibility to MD, our results did not confirm this relation and were similar to those obtained by Campbell et al. (2010) and Hietikko et al. (2012). However, the analysis of the clinical parameters indicates association with the presence of comorbidity nephropathy in the sample studied.

For rs17173510 SNP, it was not observed any statistical evidence of association among MD and this variation.

Conclusions

It was detected the rs1805127 and rs17173510 SNPs, but statically it was not found any association among MD and *KCNE1* gene alterations. However, the rs1805127 was related to comorbidity nephropathy. In addition, it was found the a *missense* mutation, N75H, still not reported in previous studies.

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