



SCREENING OF SNPS IN AMELX GENE IN CLEFT LIP AND PALATE PATIENTS WITH ENAMEL MALFORMATION

OLIVEIRA F^{***1}, Garbieri T^{*2}, Dionísio T^{***1}, Neves LT^{1,3}, Santos C¹, Machado MAAM¹, Oliveira TM^{1,3}

 Bauru School of Dentistry, University of São Paulo, Bauru, Brazil.
Sagrado Coração University, Bauru, Brazil.
Hospital for Rehabilitation of Craniofacial Anomalies, University of São Paulo, Bauru, Brazil.

PURPOSE: The enamel development involves the expression of multiple genes necessary to control the complex process of mineralization. Mutations in enamel protein and protease gene have been associated to dental malformation. The AMELX gene encodes a member of the amelogenine family of extracellular matrix protein and has an important role in biomineralization during the tooth enamel development. A single-nucleotide polymorphism is a DNA sequence variation occurring when a single nucleotide in the genome between members of a biological species or paired chromosomes in a human. These genetic variations underlie differences in the susceptibility to disease. It is known that patients with cleft lip and palate have high frequency of enamel malformation. The purpose of this research was to evaluate the presence of SNPs in AMELX gene in cleft lip and palate patients with and without enamel malformation. **METHODS:** The saliva of 80 patients with cleft lip and palate was collected and divided into 2 groups: Group 1 (with enamel malformation, n = 46) and Group 2 (wihtout enamel malformation, n = 34). The genomic DNA was extracted from this saliva, followed by PCR and DNA direct sequencing. **RESULTS:** The SNP rs2106416 was detected in 19 samples (23%) of total of 80 samples of saliva, being 13 in Group 1 (28% of 46 samples) and 6 in Group 2 (17% of 34 samples). This SNP rs2106416 is a silent mutation (c.261C>T p.87H>H) distributed throughout exons 2 and 6 in the AMELX gene. **CONCLUSION:** According to the results obtained from the DNA direct sequencing of AMELX gene, the SNP rs2106416 were found in the sequence of nucleotides of this gene, in subjects with cleft lip and palate and with and without dental malformation.

Support: FAPESP 2011/21190-7