

PI3K MUTATIONAL STATUS IN ORAL SQUAMOUS CELL CARCINOMA.

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Background: Squamous cell carcinoma (SCC) of the oral cavity and oropharynx is the sixth most common cancer in the world, with approximately 350,000 deaths and 650,000 cases new diagnoses per year. Identification of targetable mutations can potentially benefit patients care. PI3K is a potential molecular target in head and neck squamous carcinoma.

Hypothesis: The objective of this study was to evaluate the prevalence of PI3K oncogene mutations and correlate molecular clinical and histological characteristics of individuals with local and advanced oral cavity and oropharynx squamous cell carcinoma.

Methods: Observational, retrospective study. Exon 9 and 20 of the PI3K oncogenes were studied with PCR and Sanger sequencing. Most common hotspots were evaluated: E542K, E545G, E545K, E545Q, Q546L, M1043V, H1047L, H1047R, R1049H, H1049S in 49 samples of patients with local and advanced oral cavity SCC. In a second phase we extended the analysis to the full sequence of both exons. Mutational status was correlated with clinical and histopathologic features in paraffin-embedded samples.

Results: 15 samples presented PI3K mutations (30.6%), 1 E542K, 6 E545K, 1 F550L and 8 M1043I. In the expanded study, F550L and M1043I mutations were encountered. One sampled presented concomitant mutations (E542K and M1043I). Patients mean age was 68 years, 51% females, old, all Caucasian, 56% were smokers. Stage I 42%, II 38%, III 5%, IV 5%, in situ 10%. There were no correlations between the clinical and histologic characteristics of the tumors of individuals with the mutational status of the codons 9 and 20 of the PIK3CA gene.

In conclusion, In our study the prevalence of mutations of the PI3K oncogene was 30%. We found a mutation (M1043I) not described previously in this disease The method chosen for the study of the codon 545 must discriminate the pseudogene.