



Category: Cancer Genomics

# Integrated analysis of whole exome and RNA sequencing for Neo-epitope peptide prediction in buccal cancer

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## Abstract

Head and neck squamous cell carcinoma (HNSCC) is the sixth most common malignancy globally, but ranks first in India because of the extensive use of tobacco, betel-quid and alcohol. Despite, the availability of aggressive treatments, the survival for HNSCC patients remains relatively poor. High-throughput sequencing technologies have identified several mutant genes and pathways in cancer genomes but the transcription patterns of these genetic alterations remain unclear. We performed an integrated analysis of whole exome and RNA sequencing to gain a better understanding of the transcriptional consequences of these genetic mutations, thus paving the way to identifying biomarkers and neo-epitopes.

To this end, we conducted a prospective study on patients diagnosed with HNSCC from Gujarat, India. Detailed clinical data was collected from a total of 541 consented patients. Additionally, DNA and RNA was extracted from a subset of nine buccal cancer patients from both tumor and blood (Age  $44 \pm 9$ ; M: 7, F: 2). Whole exome and RNA sequencing was performed on an Illumina platform followed by data analysis using bioinformatics tools. Normalized expression of all coding variants were compiled and variants with Alt allele depth in RNA-seq  $\geq 1$  was prioritized for HLA binding and neo-epitope prediction using our proprietary pipeline OncoPeptVAC.

As previously reported, DNA sequencing results revealed recurrent frameshift/nonsense and missense mutations in the *TP53*, *PIK3CA*, *CASP8*, *FAT1*, *TTN*, *FSIP2* and *CDKN2A* genes. Additionally, unique mutations were observed in the *CA6*, *FGD3*, *FLG2*, *COX4I1*, *CD58*, *MAPK1*, *HLA-A* and *HLA-DQB1* genes, previously not associated with head and neck cancers. A positive correlation between the mutant allele frequency in Exome and RNA-seq data confirmed the expression of both missense and frameshift mutant genes. Our results show that the majority of the mutation derived peptides were not predicted to be immunogenic for the HLA types considered. Peptides derived from variants in *TP53*, *ANKRD12*, *EPHB2*, *ZNF200*, *TRPM4*, *CHD4*, *PLEC*, *KMT2C*, *GLTSCR1* were predicted to have neo-epitopes properties in buccal cancer. These findings provide a platform to predict immunogenic neo-epitopes which could be used in the development of personalized vaccines for patients with buccal cancer.

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