

Locally-recurrent cutaneous squamous cell carcinoma lesions of head and neck: a case-control study based on genetic analysis.

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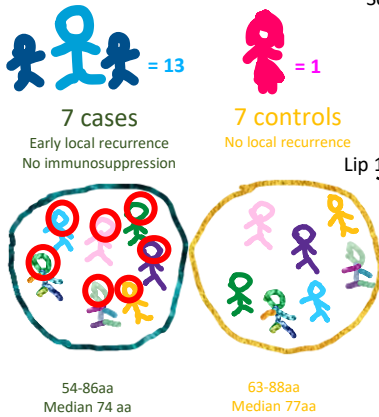
Background:

Cutaneous squamous cell carcinoma (cSCC) of the head and neck is a form of skin cancer with the proportion of local recurrence >5%, ranging up to 10% in the scalp. The prediction of recurrent cases in HnSCC lesions of such an anatomic district with intermediate dimensions and absence of any risk factor remains challenging. We here tried to define the main genetic features underlying the clinical behavior for local recurrency of HnSCC lesions

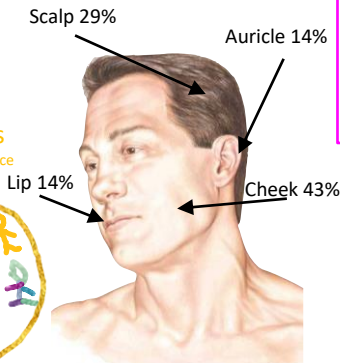
Patients and Methods:

We conducted a retrospective cohort study on patients with T2 HnSCC (as per AJCC 8 guidelines, tumors that are 2 cm or larger but less than 4 cm in size without any risk factors), treated with standard excision. Cases and controls were selected from a database of surgically treated cSCC at the University Hospital of Sassari, collected from 2017 to 2020. The study included seven cases with early local recurrence and seven controls with no recurrence during at least 3 years of follow-up. No patient had a history of immunosuppression.

Design studio:

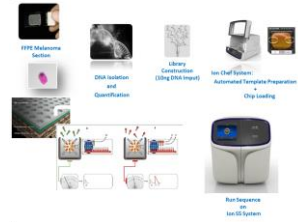


HnSCC localization:



Molecular analysis:

NGS analysis was used to assess the mutational status of 25 key genes involved in skin tumor pathogenesis



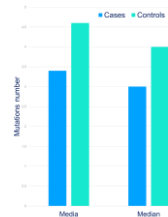
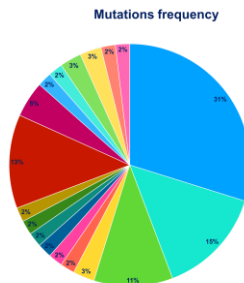
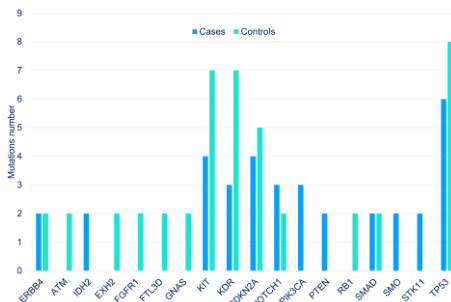
IMI SOMATIC PANEL



Amplification: 343
Amplification range: 125-175 bp
Puls Input DNA: 3
Coverage (%): 100
Panel Size: 35,113 bp

Results:

A total of 63 mutations were identified, with 37 (59%) in the control group and 26 (41%) in cases. The most frequently mutated gene was TP53, accounting for 31% of all mutations, followed by KIT (15%), KDR (13%), and CDKN2A (11%). The average mutation rate was 3.7 for cases and 5.3 for controls. Excluding mostly-prevalent TP53 mutations, distributed uniformly in both cases and controls, KDR mutations were more common in controls (6/7; 86%), while cases had a lower occurrence of KDR mutations (2/7; 29%). Additionally, mutated KDR was associated with mutations in CDKN2A or KIT or both in 2/7 (29%) of cases and in 6/7 (86%) of controls. Three cases had a history of non-melanoma skin cancer (NMSC) and developed NMSC at other locations during follow-up.



Conclusion:

Although the present collection of patients with T2 HnSCC is very limited, our findings seem to suggest that the occurrence of an increased rate of mutations might be associated with a lower tendency to develop local recurrency in such a cSCC subtype. Moreover, there is a hint about the existence of a pattern of mutated genes associated with reduced local cSCC recurrency.

Increased number of mutations
= Increased antigenic exposure

More effective
immune response

