USP6 GENE REARRANGEMENT TESTING OF GNATHIC ANEURYSMAL BONE CYSTS: A MULTICENTER ANALYSIS OF TEN CASES.

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Background: The jaws are an uncommon location for primary aneurysmal bone cysts (ABCs), and few gnathic cases have been tested for USP6 rearrangement. Rearrangements of CDH11 and/or USP6 are identified in approximately 70% of primary extragnathic ABCs.

MATERIALS/METHODS: Herein, this multi-institutional, IRB-approved study investigates the USP6 status and clinicopathologic characteristics of cases histopathologically diagnosed as primary gnathic ABC. This study retrospectively identified 11 cases from four Oral & Maxillofacial Pathology Services and submitted them for USP6 analysis. Evaluation of one case failed, but the results of FISH testing, histomorphology, patient age and sex, lesion location, lesion duration, and clinical impression were abstracted for the remaining 10 cases.

Results: The patients ranged in age from 10 to 43 years (mean: 25.4 years), with a male-to-female ratio of 1:1 (5:5). Nine cases occurred in the mandible and one case in the maxilla. The majority of lesions were present for an unknown duration or more than one month. Morphologically, five cases exhibited classic ABC features while five exhibited few cystic spaces with "solid" morphology. None of the tested cases demonstrated rearrangement of the USP6 locus by FISH.

Conclusion: In the jaws, lesions that morphologically mimic primary extragnathic ABC rarely show USP6 abnormalities, and may be genetically distinct lesions. Lesions with USP6 rearrangement are neoplastic; however, the etiology of lesions lacking rearrangement is less certain and may represent primary ABC without USP6 rearrangement, secondary ABC that have effaced the lesion of origin, or ABC-like degenerative lesions.

GENETIC POLYMORPHISM OF TUMOR NECROSIS FACTOR ALPHA (TNF-A) AND TUMOR NECROSIS FACTOR BETA (TNF-B) GENES AND RISK OF ORAL PRE CANCER AND CANCER..

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Background: Inflammatory cytokines such as TNF-α andTNF-β may play a pathogenic role in the development of oral precancerous lesions and oral cancer. Genetic polymorphisms of in these genes are known to predispose to malignant disease. We hypothesized that the risk of oral precancerous lesions and oral cancer might be associated with polymorphisms in these two inflammatory genes.

Methodology: A total 500 patients with oral pre cancer &cancer and 500 healthy volunteers were genotypes for the TNF-α (-238) G/A and TNF-β (252) A/G gene polymorphism. Genotypes were identified by polymerase chain reaction (PCR) restriction fragment length polymorphism(RFLP). Genotype frequencies were evaluated by Chi-square test.

Results: Compared to the GG genotype the GA genotype of TNF-α (G238A) polymorphism has been found to significantly increase the risk of oral disease (OR= 1.99) and especially the risk of Lichenplanus and oral malignancy (OR= 2.805 and 5.790 respectively). The risk of overall oral disease, Lichenplanus and oral malignancy were also high with allele A compared to allele G of TNF-α (G238A) polymorphism (OR =1.88, 2.34, 4.42) and respectively). Similarly, the risk of oral disease was also more in the heterzygote (AG) than the common allele homozygote (AA) of TNF-β (A252G) polymorphism (OR= 1.483).

Conclusions: We conclude that the TNF-α (-238) G/A, TNF-β(252) A/G polymorphism were significantly associated with Oral pre cancer & cancer.

THE "OLD SAILORS " ILLNESS MAKES A RETURN.

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Vitamin C, also known as ascorbic acid, is a co-factor in multiple enzymatic reactions including that of collagen synthesis. Due to the absence of the enzyme L-gulonolactone oxidase, humans are unable to synthesise ascorbic acid; hence it is recognised as an essential nutrient. Scurvy refers to the clinical presentation of a deficiency of ascorbic acid, which occurs as a result of inadequate dietary intake. It has long been considered an illness of historical rather than contemporary significance. However, a tendency towards Western diets rich in processed foods and lacking in fresh produce has given rise to the re-emergence of the condition in the developed world. Current evidence suggests that there is a resurgence of scurvy in Sydney. A case is highlighted of an otherwise healthy 35year old male who presented to a hospital emergency dental clinic with generalised red, boggy gingivae. Clinical examination revealed bilateral involvement of the buccal and lingual/palatal gingivae in both the mandibular and maxillary arches, predominantly affecting the interdental papillae. Radiographic examination confirmed there was no associated bone loss or bony pathology. The clinical differential diagnoses included leukaemia, Kaposi sarcoma or possible scurvy. A battery of serological investigations yielded the eventual diagnosis of severe ascorbic acid deficiency (vitamin C level $<5\mu$mol/L, HIV negative and blood films and blood counts that did not show features of leukaemia). Dietary intake was immediately instituted in addition to 250mg daily vitamin C supplementation. The condition improved within weeks and completely resolved within 3 months (progress vitamin C level 55 μmol/L) and the patient was educated to henceforth maintain an adequate nutritional intake of ascorbic acid. Though these cases will be infrequent, prudent clinicians need to re-familiarise themselves with the signs and symptoms of scurvy and maintain a wide diagnostic radar in order to ensure a speedy and accurate diagnosis.