# Consanguinity & Periodontal Disease: A Case Report

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## Abstract

The relationships among the different forms of aggressive periodontitis are not clear, and the way in which they progress with time remains unknown. Periodontal diseases in children occur in localized and generalized forms. In the current case report, four children belonging to a single family, suffering from an incisor molar pattern of periodontal disease, were examined. Preliminary investigations revealed no significant dental or medical history and systemic findings, other than a clinical presentation similar to Localized Aggressive Periodontitis (LAP). This maybe suggestive of familial aggregation of a recessive trait, in the consanguineous family, leading to periodontal disease.

**Keywords:** Periodontitis, Consanguinity, Familial Aggregation.

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## Introduction

Certain forms of periodontitis clinically and microbiologically differ from chronic periodontitis.[1] The American Academy of Periodontology (AAP)workshop in 1999 replaced the term early-onset periodontitis with Aggressive Periodontitis (AP).[1] Diagnosis of AP is made on clinical and radiographic findings which show rapid attachment loss and bone destruction, and possible familial aggregation of disease. Disease susceptibility to periodontitis in apparently healthy pre-pubescent children has been associated withleukocyte dysfunction or other defects in the host defense system.[1]Literature has reported a high correlation between the presence of neutrophil chemotaxis defects in vitro and the occurrence of AP and many of these cases appear to have a familial pattern of occurrence.[2,3,4]

'Consanguinity,' implies the inheritance of genes which are identical by descent, i.e., inherited from the common ancestor(s).[5] The highest consanguineous marriages (20% to over 50%) are reported in North of Africa, Asia etc, usually associated with low socioeconomic status, illiteracy, and rural residence.[6] The off spring of consanguineous couples are at higher risk for autosomal recessive disorders due to risk of homozygosis by descent.[5] There are many reports in the literature describing families with multiple AP affected individuals, suggesting familial aggregation.[7] The patterns of disease in these families have led investigators to postulate both dominant and recessive modes of Mendelian inheritance of this disease.[7]

## Description of Case

A total of four children (probands) with their respective parents, reported to the Department of Periodontics, in October 2018. All the fourprobands belong to one family, with history of consanguineous marriage within the family. The consanguinity is seen among two couples, couple 1 and couple 2. Couple 1 has two male offsprings, ages ten and seventeen years old. Couple 2 reported with one male and one female offspring, ages sixteen and fourteen years old respectively. Since both couple 1 and couple 2 are first cousins, their inbreeding coefficient was 0.0625(F). Inbreeding coefficient represents a measure of the proportion of loci at which the offspring of a consanguineous union is expected to inheritidentical gene copies from both parents.[8]In clinical genetics, a consanguineous marriage is defined as a union between two individuals who are related assecond cousins or closer, with the inbreeding coefficient (F)equal or higher than 0.0156.[8] Figure 1

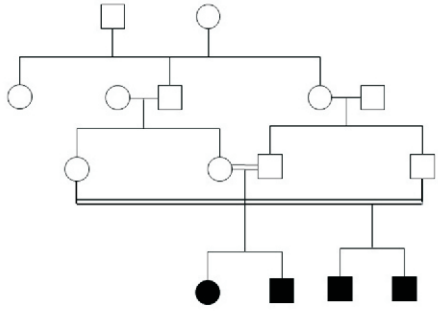

Figure 1:The family pedigree with the shaded boxes representing the probands. Circle: Female; Square: Male, Double line: Consanguineous Marriage among first cousins.

## Clinical Findings

All the patients with a suspected diagnosis of AP were examined by a single clinician. Full-mouth measurements were obtained using UNC-15 probe (University of North Carolina) for probing pocket depth, recession (measured as distance from the cement–enamel junction, CEJ, to the gingival margin) and lifetime cumulative attachment levels (LCAL, measured either as a direct measurement of CEJ to the base of the pocket, or as a calculation of probing pocket depth and recession). Six sites were measured for each natural tooth. Appropriate radiographs were recordedfor each patient. The suspected genetic background of their condition was explained to them. In this context, the importance of examining their first-degree blood relatives was highlighted. All available first-degree relatives were invited for a dental examination.A thorough medical and dental history was recorded for each patient. There was no history of deleterious habits such as smoking. All the patients presented with mild to moderate plaque accumulation. Simplified Oral Hygiene Index (OHI-S) score for all patients was fair.

The first patient, a ten year old boy, on examination, revealed the upper and lower anterior teeth and first molars of all quadrants completely erupted into the oral cavity with the rest of the dentition in various stages of eruption. The patient complained of bleeding from gums and pus discharge from the lower front teeth. He presented with deep pockets with exudation in relation the upper and lower anterior teeth and grade 3 mobility of 31 & 41, grade 2 mobility of 32 & 42 and grade 1 mobility of teeth 12, 11, 21, 22,16,26,36,46. The gingiva was inflamed with a tendency to bleed spontaneously with respect to 33, 32, and 31,41,42,43. The probing depth around upper and lower anterior teeth was 10-12mm from the CEJ at the mesiofacial, mid-facial and distofacial sites. The OPG revealed significant bone loss around all the erupted first molars and anterior teeth. The patient's plaque and calculus deposits were scanty and could not be correlated to the amount of periodontal destruction observed. Figure 2,3& 4

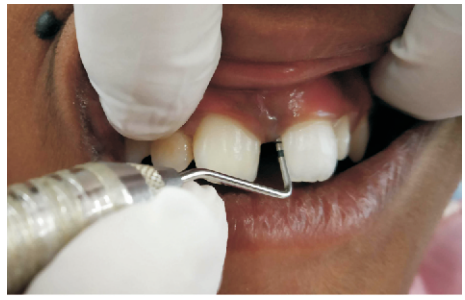

Figure 2: Probing depth = 12mm on mesial of tooth 21 in 10 year old patient, reveals a periodontal pocket.

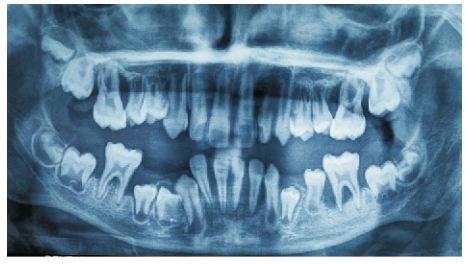

Figure 3: Orthopantomogram of 10 year old male patient revealing bone loss around erupted molars and incisors.

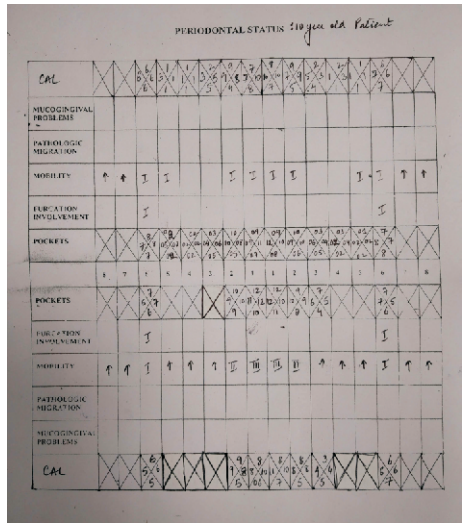


Figure 4 : Periodontal Charting of 10 year old male patient.

The second patient, a 17 year old boy, reported with history of fixed orthodontic treatment for malocclusion and maxillary dentition protrusion of over six months. He clinically presented with a uniform probing

depth of 2-3mm but significant attachment loss with respect to upper and lower anterior teeth and maxillary first molars due to recession of gingiva. The patient presented with plaque accumulated in areas of gingival recession. Orthopantomogram (OPG) revealed horizontal bone loss around all the teeth with greater bone destruction visible around the upper and lower anterior teeth and the maxillary first molars. Supra-eruption and premature occlusal contact was also present in relation to tooth 26. The marginal gingiva of the maxillary anterior teeth and maxillary first molars was inflamed and showed bleeding on probing. The rest of sites did not bleed on probing. Figure 5

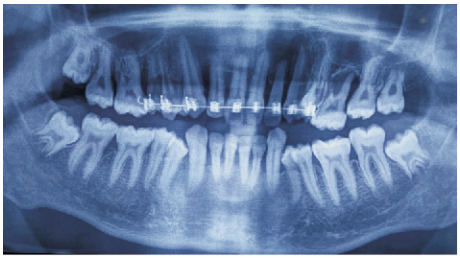

Figure 5: Orthopantomogram of 17 year old male patient revealing bone loss around first molars and incisors.

The third patient, a 16 year old boy, reported with an extruded, grade 3 mobile 11. IOPA revealed severe bone destruction till the apex of 11. His clinical picture also reveals more than 5mm of attachment loss (Grade 3 recession) around the upper and lower anterior teeth and first molars in all quadrants. Probing depth was an average of 2-3mm uniform on all the teeth. The OPG revealed bone destruction on the mesial of upper first molars and thin, tapering root morphology of 15,25 apart from horizontal bone destruction seen with respect to the upper and lower anterior teeth. There was generalized bleeding on probing from all sites with concomitant inflammation.Figure 6&7

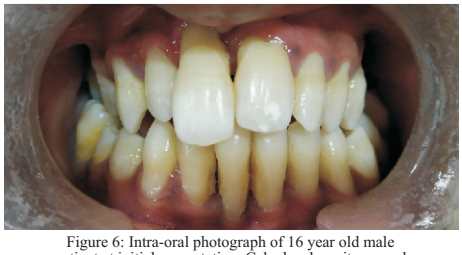

Figure 6: Intra-oral photograph of 16 year old male patient at initial presentation. Calculus deposits around teeth can be appreciated. Gingival recession and malocclusion in the anterior teeth can be seen. Extrusion of tooth 11 is seen.

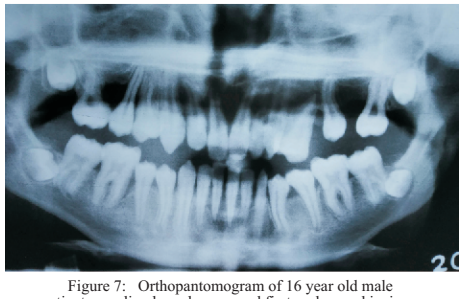

Figure 7: Orthopantomogram of 16 year old male patient revealing bone loss around first molars and incisors.

The fourth patient, a 14 year old girl, did not report with any significant chief complaint. On examination, she had crowding of lower anterior teeth with cross bite between teeth 43 and 12 and negligible overjet between the anterior teeth. The posterior teeth did not occlude revealing an open bite with premature cusp contacts. Plaque and calculus was present, covering the cervical and middle thirds of her posterior teeth. She presented with chronic gingivitis and normal probing depth. Loss of attachment on all sites (Mid-palatal, mesifacial, facial & distofacial) was present on her 16, 12, 11 and 26. Her OPG showed onset of bone destruction involving the furcation of 46 and severe horizontal bone loss with respect to 16 &26. Horizontal bone loss was also seen with respect to upper and lower anterior teeth along with a mesiodens present between 11 and 21.Figure 8

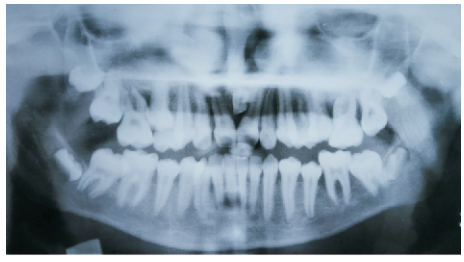

Figure 8:Orthopantomogram of 14 year old female patient revealing bone loss around first maxillary molars and incisors.

## Investigations and Treatment

Complete Blood Count was performed for all the patients. The reports did not show any significant finding except low hemoglobin value and reduced MCV, MCH and MCHC in patients 1, 2 and 4. A provisional diagnosis of Iron Deficiency Anemia was made. The neutrophil and lymphocyte count was normal for all the patients except patient 3, whose reports showed a neutrophil count of 35 % (40-75%), which is mild neutropenia.

Since patient 1, the 10 year old boy reported with the most severe periodontal disease, we analyzed his complete blood count at a frequency of twice a week for 3 consecutive weeks, for cyclic neutropenia. The neutrophil count remained normal throughout 3 weeks. Leukocyte Adhesion Deficiency 1 and Leukocyte Adhesion Deficiency 2 were also investigated for in the same patient using flow cytometry. Four markers including CD18, CD11a, CD11b and CD15s were tested for. Normal expression of all the four markers was reported.

Plaque samples from each patient were collected and cultured. The microflora successfully cultured comprised of Streptococci spp., Actinomyces spp. and other commensals.

The role of trauma from occlusion in exacerbating the periodontal pathogenesis and bone destruction around affected teeth was also suspected in the children with malocclusion or history of orthodontic treatment. The patients were treated by scaling and root planing, debridement of pockets and systemic host modulation therapy with doxycycline (20 mg OD x 3 months) to arrest what seemeda rapid rate of bone loss in these young patients. The patients were scheduled on regular recall for monitoring and treatment.

## Discussion

In the reported family, no significant history of periodontal disease was obtained in any of their first degree relatives. However, among the four children presenting with periodontitis we do observe a few similarities. Firstly, all the four children present with significantly greater attachment loss around the upper and lower anterior teeth and the first molars. The other teeth either show no attachment loss or cannot be evaluated since they are erupting (10 year old patient). Secondly, OPG of all the children consistently shows horizontal bone loss around the upper and lower anterior teeth correlating with the clinical attachment loss around these teeth. Thirdly, all the three adolescents' show comparatively greater bone loss around the maxillary first molars, while the mandibular first molars show incipient bone loss or no bone loss at all. While deep pockets are not present in three out of four patients, advanced bone loss is seen in all the patients, which is not characteristically arc shaped or angular as described in literature.[9] However, the disease predilection towards the first molars and incisors does exist, as seen in Localized Aggressive Periodontitis (LAP). One explanation for this pattern might be that these first permanent teeth that erupt (first molars and incisors) get colonized first, and there is initiation of destruction of periodontal tissues by various mechanisms.[10]

Since in these patients, trauma from occlusion may be one of the contributing/disease modifying factors one may diagnose the patients to have incidental attachment loss in some teeth.[11] However, since isolatedareas of attachment loss may also represent initial clinical presentations of LAP, patients with this clinical diagnosis should be considered as a high-risk group for AP.[11]

No isolation of the more pathogenic anaerobes from the plaque samples could be attributed to dependence of bacteria on chemical signals in mixed communities such as a biofilm, making in-vitro culture difficult.[12]

Family linkage studies and segregation analyses of families with AP support a major locus hypothesis and potential inheritance models include autosomal dominant, autosomal recessive & X-linked dominant.[7] Despite the familial aggregation of AP, the mode of inheritance is still unclear.[7] Since, the disease has been reported in young patients, with teeth yet to erupt in the 10 year old patient, it is a possibility that the probing depths might decline after eruption and periodontal stability may increase with age. However, the loss of bone and clinical attachment will need to be treated and followed-up.

## Conclusion

The case report indicates that there was no evidence for defects in the immunological functions of the four LAP patients, and that the underlying cause of LAP is not always related to leukocyte dysfunction. The basic disorder of LAP remains meagerly understood.

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**More References are available on request at editor@healtalkht.com.**