Papillon-Lefèvre Syndrome – A Case Report.

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INTRODUCTION:

Papillon-Lefèvre syndrome (PLS) belongs to a heterogeneous group of skin diseases that are characterized by hyperkeratosis of palms and soles and presence of severe and early onset periodontitis. Genetic studies have shown that mutation in the major gene locus of chromosome 11q14 with the loss of function of cathepsin C (CTSC) gene is responsible for PLS. Loss of CTSC function is responsible for the severe periodontal destruction seen clinically. This report represents classical signs and symptoms of PLS in a six year old girl.

CASE REPORT:

A six year old girl presented to Department of Periodontics, Govt. Dental College and Hospital, Srinagar. Her father noticed mobility in relation with some of her permanent teeth. The patient was referred to the Department of Periodontics for general dental care by the Department of Dermatology, where she had been diagnosed with PLS. She was the first child born to apparently healthy non consanguineous parents. Typical clinical signs of the disease were seen during the child’s first year of life. However, he had not sought any treatment until now.

General and extra-oral examination

The family history revealed consanguineous marriage of the parents. The parents and other family members were not affected. Patients had overall normal physical and mental development. Extra-oral examination revealed yellowish, keratotic, confluent plaques affecting the skin of her palms and soles. Well circumscribed, psoriasiform, erythematous, scaly plaques were also present on the elbows and knees bilaterally along with dystrophy and transverse grooving of the nails (Figure 1).

ABSTRACT:

Papillon-Lefèvre syndrome (PLS) belongs to a heterogeneous group of skin diseases that are characterized by hyperkeratosis of palms and soles and presence of severe and early onset periodontitis. Genetic studies have shown that mutation in the major gene locus of chromosome 11q14 with the loss of function of cathepsin C (CTSC) gene is responsible for PLS. Loss of CTSC function is responsible for the severe periodontal destruction seen clinically. This report represents classical signs and symptoms of PLS in a six year old girl.
Intraoral examination
Intraoral examination revealed severe gingival inflammation, abscess formation and deep periodontal pockets. Severe mobility affecting all the permanent teeth, with heavy deposits of plaque and calculus and halitosis were also present (Figure 2). All primary teeth were exfoliated.

Radiographic findings
Orthopantomogram showed extensive alveolar bone loss in all remaining teeth. The alveolar bone around the mobile teeth was devoid of definable lamina dura. An extensive alveolar bone loss was noted, a “floating in air appearance,” which were extracted afterwards (Figure 3).

Laboratory investigation
Laboratory investigation was carried out, which included hematological and biochemical assessment. The results were within normal limits.

Figure 1 (A,B,C,D): Patient presenting yellowish, keratotic, confluent plaques affecting the skin of palmar surfaces of hands, knees and keratotic plaques on soles and dorsal surfaces of feet.

Figure 2: Intraoral view.

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Figure 2: Intraoral view.

Treatment
A multidisciplinary approach involving Dermatologist, Periodontist, Pedodontist and a Pediatrician is important for the overall care of patient with PLS.

Periodontal treatment
The aim of periodontal treatment is to eliminate the reservoir of causative organisms. It is generally agreed that the response to local debridement or to systemic antibiotic alone or in combination provide at best a transitory response.22-24

Treatment given:
• Conventional periodontal treatment in the form of scaling and root planning.
• 0.2% chlorhexidine gluconate mouthwash and oral hygiene instruction was employed to control disease activity.
• Systemic antibiotic treatment was given for 4 weeks amoxicillin (20-50 mg/kg/d) + metronidazole (15-35 mg/kg/d) in divided doses every eight hours as an adjunctive with conventional treatment.
• Teeth with hopeless prognosis were extracted.
• In teeth having deep periodontal pockets, periodontal flap surgery was done.
• To restore masticatory function, partial dentures were inserted.
• Maintenance visits of 2- 3 months were planned.

In recent years, dental implant offers not only considerable better stability and retention of prosthesis, but also improved comfort, masticatory efficiency and esthetics. There is data available that dental implants are successful mode of rehabilitation in patients with PLS.25 Implant supported prosthesis are also planned in patients after their growth period is over.

Dermatological treatment
The skin manifestations of PLS are usually treated with emollients. Salicylic acid and urea may be added to enhance their effect. Oral retinoids including acitretin, etretinate and isotretinoin are the mainstay of the treatment of both keratoderma and periodontitis associated with PLS.26 After 8 weeks of oral acitretin, there was a dramatic improvement with marked reduction of keratodermas. Treatment may be more beneficial if it is started during eruption and maintained during the development of the permanent teeth.

DISCUSSION:
PLS can adversely affect growing children psychologically, socially and aesthetically. Typically the parents are not affected and there is no family history of the disease. Higher prevalence has been reported when parental consanguinity is involved, but no predilection for gender or race has been documented.27,28 Phenotypically, the parents were healthy and there was no family history of the disease, suggesting an autosomal recessive pattern of inheritance. In case of PLS, the inflammatory infiltrate at the sites of periodontal infection is not under regulatory control.
Increase neutrophil influx and retention of inflammatory infiltrate and their proteases play a significant role in continued periodontal destruction. It makes difficult to control and limit periodontitis once lesions are established and disease becomes unresponsive to traditional periodontal treatment.

The clinical manifestations observed in our patient were hyperkeratosis of the palms, soles, elbows, and knees and generalized aggressive periodontitis, which resulted in loss of the primary and permanent teeth. Because the etiology and pathogenesis of PLS periodontitis is directly related to high levels of Actinobacillus actinomycetemcomitans, the use of an antibiotic that acts specifically on this pathogen has been claimed to be important for a successful treatment.28

Prosthetic replacement in such patients is an age specific specialty treatment involving initial replacement with complete or partial dentures and future consideration for an implant supported prosthesis. It would provide immediate satisfaction to the patient in terms of esthetics and function. In the present case, prosthetic rehabilitation was considered in order to provide immediate satisfaction to the patient in terms of esthetics and function

CONCLUSION
PLS is a rare autosomal recessive disorder. The conflicting findings of PLS management could be related to the severity of the condition, the age at which treatment was instituted, timing and duration of antibiotic therapy, professional supervision, supportive treatment plan and home care.

The complex etiopathogenesis of PLS means that successful treatment of the periodontal component of this syndrome remains challenging. After identification of the gene defect, better treatment modalities can be developed. In cases where patient reports late or is not responding to periodontal treatment, dental implants are successfully advised. PLS threatens children and their parents with the prospect of edentulism if left untreated. Hence, early diagnosis and intervention is essential. Osseointegrated implants are an option for the future and can have a great impact psychosocially by restoring esthetics as well as function.

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CONFLICTS OF INTEREST:
There are no conflicts of interest.

REFERENCES:


