Papillon-Lefèvre Syndrome – A Case Report.

Abhima Kumar,1* Suhail Majid Jan,2* Rafiya N Khan,1* Roobal Behal.2*

1Post Graduate Student, 2Professor & Head of Department, 3Consultant, 4Department of Periodontics, Government Dental College and Hospital, Srinagar, Jammu and Kashmir, India.

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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.

INTRODUCTION:

Papillon-Lefèvre syndrome (PLS) is characterized by erythematous palmoplantar hyperkeratosis and severe periodontal disease. Dermatological as well as oral signs vary considerably between affected subjects.1 The condition is inherited as an autosomal recessive trait2 and linked to mutations of the cathepsin C gene.3,4 Cathepsin C is a lysosomal cysteine protease that activates several granule serine proteases expressed in bone marrow derived effector cells of myeloid and lymphoid series.5 These proteases are implicated in a variety of immune and inflammatory processes, including cell-mediated cytotoxicity, phagocytic destruction of bacteria, local activation and deactivation of cytokines and other inflammatory mediators, and extracellular matrix degeneration.6 Cathepsin C is normally expressed in palmar, plantar, and gingival epithelium,7 but its involvement in epithelial desquamation or its significance in gingival epithelium is unknown.8 While several cathepsin C gene mutations have been identified,9 the correlation to the disease’s phenotypic expression is still obscure. The aggressive periodontal inflammation leads to premature loss of primary and permanent teeth. Clinical observations and investigations have led to various theories regarding possible etiologic mechanisms, including altered immune response.10-13

underlying tissue pathology,14-15 and virulent and aggressive periodontal flora. Actinobacillus actinomycetemcomitans is a periopathogen of key importance in periodontal infections and has often been identified in periodontal lesions in PLS patients.15-19 However, others have found flora without any particular periodontal pathogens.20-21

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).
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.

Figure 3: Orthopantomogram showing extensive alveolar bone loss.
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. 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:


