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Papillon - Lefèvere Syndrome: A Case Report

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Authors' contributions

This work was carried out in collaboration between all authors. Author MSAD wrote the first draft of the manuscript as regard to introduction and case history. Author HM wrote the oral and dental findings and author AE managed the investigations and treatment of the case. All authors read and approved the final manuscript.

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Case Study

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ABSTRACT

Papillon-Lefèvere Syndrome (PLS) is one of the rare autosomal recessive disorders of keratinization (1-4 cases per million). It is inherited as an autosomal-recessive condition with one-third of the patients showing consanguinity of the parents and diagnosed in both sexes. Lesions are characterized by palmo-plantar hyperkeratosis and severe destructive periodontal disease affecting the primary and permanent teeth. This paper reports a case of a 13 -year-old African boy presented to the outpatient clinic of Qassim University with diffuse keratotic plaques of his soles extending to the dorsal surface, localized keratotic plaques of his palm and severe inflammation of the gingiva with premature teeth loss and looseness of remaining teeth.

Keywords: Papillon-Lefevre syndrome; severe periodontitis; autosomal.

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1. INTRODUCTION

Papillon-Lefèvere syndrome (PLS) was initially described in 1924 by two French physicians, Papillon and Lefèvere, is extremely uncommon genodermatosis inherited as an autosomal recessive trait, affecting children between the ages 1-4 years [1]. The prevalence of PLS is 1-4 cases per million persons [2], without sex and racial predominance [3,4].

The cause of PLS is not well understood. The genetic basis for most PLS cases appears to be mutation affecting both alleles of the cathepsin-C gene, located on chromosome 11q14.1-q14.3 [3,5-7].

The disorder is characterized by sharply demarcated erythomatous keratotic plaques usually involve the entire surface of the palms and soles, sometimes extending on to the dorsal surfaces. Hyperhidrosis of the palms and soles, often associated resulting in a foul-smelling odor. Severe periodontitis is the second major feature, which starts at age 3 or 4 years. The development and eruption of the deciduous teeth proceed normally, but associated with gingival inflammation and subsequent rapid destruction of the periodontium. Nail changes occur only in advanced cases, manifested by transverse grooving and fissuring [3,8].

2. CASE REPORT

A 13 -year-old adolescent African male presented to the outpatient dermatology clinic, College of Medicine, Qassim University, Saudi Arabia with a history of thickening and fissuring of the skin over his palms and soles starting two weeks after delivery. The lesions were asymptomatic with the exceptional pain over the fissures. He was the second child of a consanguineous marriage, he had 2 brothers and one sister and his elder brother had the same complain while others are free. The boy had associated dental problem, including gingival inflammation, premature loss of primary and permanent teeth and looseness of the remaining teeth. Otherwise, he is healthy with no significant history for skin or any associated systemic manifestations. He was great in his studies and had a normal motor and psychosocial development history. Additionally, there was no significant history of recurrent skin or systemic infections. There was also no history of any other significant and relevant skin or systemic illness in the family.

2.1 Clinical Findings

On clinical examination hyperkeratotic yellowish plaques with fissuring and some crusting was seen over the palms and soles. The soles were almost complete, symmetrically involved, while the palms showed only localized involvement (Figs. 1 and 2). Hair and nails were normal. No abnormalities were detected on ophthalmic examination. No skeletal abnormalities or other systemic abnormalities were seen.

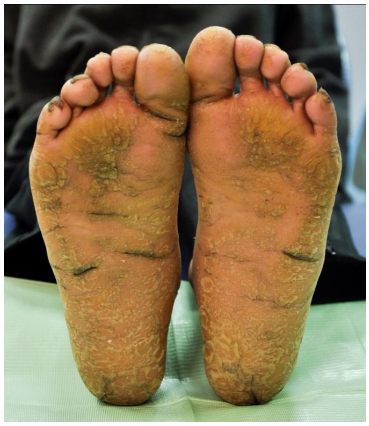

Fig. 1. Hyperkeratotic plaques involving both soles symmetrically

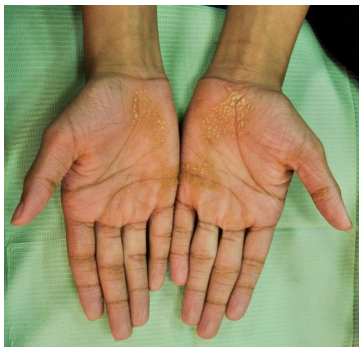

Fig. 2. Hyperkeratotic plaques over both palms

2.2 Dental Findings

Oral and dental examination showed severe gingival inflammation, premature loss of anterior permanent incisors, first molars and two of the upper premolars, grade three mobility and over eruption of permanent canine as well as the remaining teeth (Fig. 3). The panoramic radiograph showed a severe alveolar bone loss around all erupted teeth. No bone resorption around the unerupted four wisdom teeth is seen (Fig. 4).

2.3 Oral and Dental Management

In the current case, oral and dental management protocol was planned, including scaling and root planning, mouthwash and oral hygiene instruction (0.2% chlorhexidine gluconate), prescription of systemic antibiotic as an adjunctive with conventional treatment, extraction of hopeless teeth, construction of temporary partial dentures in order to restore masticatory function (Fig. 5), and finally planning for fixed prosthesis through dental implants.

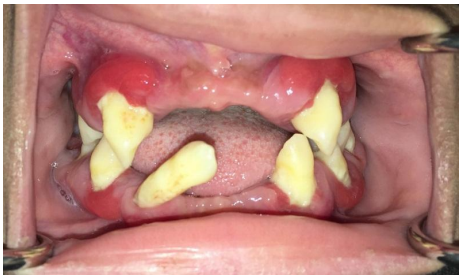

Fig. 3. Severe gingival inflammation and premature loss of permanent incisors

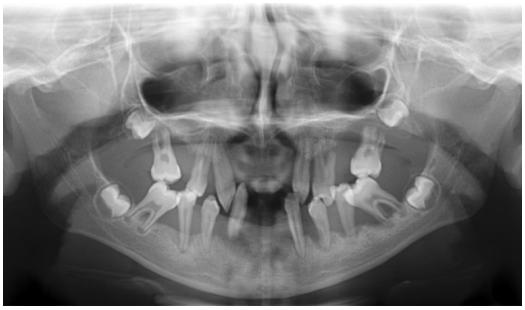

Fig. 4. Panoramic radiograph showing severe alveolar bone loss of erupted teeth and premature loss of anterior incisors, first molars and two of the upper premolars

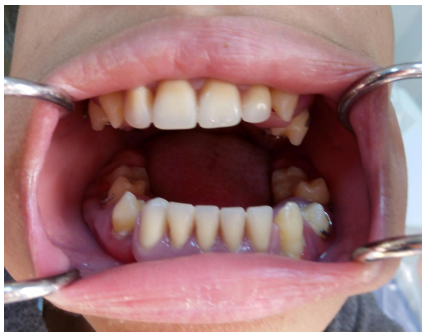

Fig. 5. The constructed upper and lower partial denture after insertion

3. DISCUSSION

PLS is an autosomal recessive disorder characterized by palmo-plantar hyperkeratosis and severe periodontitis. Papillon-Lefevre syndrome usually occur in childhood. Males and females are equally affected. Patients are normal at birth. The onset of palmo-plantar keratoderma in PLS may occur within the first 3 months of life, but usually, full picture of palmo-plantar hyperkeratosis with severe periodontitis start simultaneously between the ages of 1 and 4 years [9,10].

PLS and Haim-Munk syndromes (HMS) are two types of hereditary Palmo-planter Keratoderma (PK) which can be associated with severe periodontitis. However, Haim-Munk syndrome is associated with other characteristic features like acroosteolysis, atrophic changes in the nails, arachnodactyly, and peculiar radiographic deformity of the fingers consisting of tapered pointed phalangeal ends. In contrast to PLS, the cutaneous findings in HMS are more severe and extensive and the periodontium is less severely affected [11].

The etiology of PLS is still unknown. Factors suggested as responsible for initiation and progression of disease include (1) impairment of neutrophil chemotaxis, phagocytosis and bactericidal activity. (2) actinobacillus actinomycetemcomitans, a virulent gram negative bacteria, found in periodontal pockets may act as triggering factor (3) defect in immune mediated mechanisms [12,13].

Previous studies reported that loss of function mutations were associated with PLS affecting both alleles of the Cathepsin-C gene (CTSC), located on chromosome 11q14.1-q14.3. The CTSC gene encodes a cysteine-lysosomal protease also known as dipeptidyl-peptidase I, which functions to remove dipeptides from the amino terminus of the protein substrate. It also has endopeptidase activity. The CTSC gene is expressed in epithelial regions commonly affected by PLS such as palms, soles, knees and keratinized oral gingiva. It is also expressed at a high level in various immune cells, including polymorphonuclear leukocytes, macrophages and their precursors [3].

Clinically the keratoderma of PLS is characterized by sharply demarcated hyperkeratotic plaques usually involving the whole surface of the palms and soles, sometimes

extending to the dorsal surface of the hand and feet. Elbows, knees, and trunk, can be involved, especially in older patients. Hyperhidrosis resulting in a foul‑smelling odor and secondary infection can be associated with the skin lesions. There is severe periodontopathy and premature loss of primary and permanent dentition. Teeth are lost in ultimately the same sequence in which they are erupted. When the last tooth is lost gingiva obtained a normal appearance. The permanent dentition starts to erupt at proper time, and around 8-9 years of age, periodontal destructions are repeated in the same manner as in primary dentition. All permanent teeth are usually lost before 14-16 years of age [12].

Complications of PLS include recurrent pyogenic infections and pyogenic liver abscess due to associated impairment of the immune system in which there is decreased neutrophil, lymphocyte and monocyte functions and an increased susceptibility to bacterial infection [14].

Management of PLS requires a combined consultation from the dermatologist and the dentist. Early treatment and prophylaxis is essential for the management of periodontitis. Teeth with advanced periodontal disease should be extracted and replaced with a partial denture. The primary drug of choice for the treatment of keratoderma is oral retinoids. Topical emollients, salicylic acid and urea preparations can be used as adjuncts. It has been suggested that early retinoids treatment (before the eruption of permanent teeth) can help ensure a normal dentition in cases of PLS [15-17]. It is reported that etretinate and acitretin modulate the course of periodontitis and preserve the teeth [18,19].

4. CONCLUSION

We discussed a case of a 13 -year-old adolescent boy diagnosed of Papillon- Lefèvere syndrome. Dentist and dermatologist teams can help to save the permanent dentition if they diagnose this disease in early childhood.

CONSENT

Written informed consent was taken from the patient for performing this case study.

ETHICAL APPROVAL

This study was approved by the Research Ethics Committee of Qassim University.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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