Papillon Lefevre Syndrome: A Case Report

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Abstract

Papillon-Lefevre Syndrome (PLS) is a rare autosomal recessive trait, which is transmitted with an estimated frequency of one to four per million individuals. It is characterized by palmar plantar keratosis and severe early-onset periodontitis affecting both deciduous and permanent dentition. In this report, we present a five year-old male child patient with symptoms typical of Papillon-Lefevre Syndrome.He had hyperkeratosis of palms, soles, elbows and knees. The mandibular deciduous molars of the child exhibited mobility.

Keywords

Palmar plantar keratosis, Periodontitis Papillon-Lefèvre Syndrome.

Introduction

The Papillon-Lefevre syndrome (PLS) is a rare genodermatosis of autosomal recessive inheritance manifesting as palmer plantar hyperkeratosis with periodontitis. It was first described by two French physicians, Papillon and Lefevre, in 1924(1). It has a prevalence of 1-4 cases per million persons and both males and females are equally affected with no racial predominance(2). The disorder is characterized by diffuse palmoplantar keratoderma and premature loss of both deciduous and permanent teeth(1,3). The palmoplantar keratoderma typically has its onset between the ages one and four years. The sharply demarcated erythematous keratotic plaques may occur focally, but usually involve the entire surface of palms and soles(4). PLS is caused by mutations in cathepsin C gene located on chromosome 11q14.1-q14.3 The cathepsin C gene encodes a cystine lysosomal protease also known as dipeptidyl peptidase I. It is expressed in epithelial regions commonly affected by PLS such as palms, soles, knees and keratinized gingiva. It is also expressed at high levels in various immune cells including polymorphonuclear leucocytes, macrophages and their precursors(5,6) The exact cause of the periodontal disease has not been found but it has been attributed to decreased neutrophil phagocytosis, bacterial infection and impaired reactivity to T and B cell mitogens. The exact mechanism of the increased susceptibility to infections is also unknown but some investigators have demonstrated a dysfunction in neutophil motility and bactericidal function(7).

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Case Report

A 5-year-old male patient presented to the department of Pedodontics and Preventive Dentistry, with a chief complaint of the pain and mobility of the lower posterior teeth. Clinical history revealed that he had normal emergence of deciduous teeth at 8-9 months of age, which started loosening at the three years of age.Intraoral examination revealed severe gingival inflammation, deep periodontal pockets,and mobility of all the molars in the maxillary as well as the mandibular arch. Calculus particularly involving the molars and halitosis, was also present (figure-1).

The patient was moderately built with a steady gait. His physical and mental development was also normal. Extraoral examination revealed hyperkeratosis of the forehead(figure-2) palms, soles, and the knees of both the limbs;

the affected skin was well demarcated from adjacent normal skin. There were symmetric, well-demarcated, keratotic, and confluent plaques affecting the skin of his palms and soles, also extending onto the dorsal surfaces of hands and feet(figure-3,4).

Medical history was noncontributory. Parents were not of consanguineous marriage. The parents and other family members were not affected. Pregnancy and delivery of the child were normal. The mother had noticed skin lesions on the palms and soles of the child when he was 7 months old.

The mobile deciduous molars were extracted as they were not in the restorable condition and was patient was having difficulty in mastication.The patient has been put on the regular dental check up. Patient also reported that the skin lesions remained refractory to any therapy administered by the dermatologist.

Discussion

Papillon - Lefevre syndrome is an inherited disorder of keratinization which is probably inherited as an autosomal recessive fashion(8). It is generally characterized by redness and thickening of palms and soles, associated with periodontosis and tendency to frequent pyogenic skin infection. In our patient there was no history of consanguinity. Lesions started at the age of 7 months in the child, which has been well described.

Some of the other conditions where patients present with severe periodontitis and dermatological lesions are prepubertal periodontitis, and Haim-Munk syndrome (HMS).

Further, these conditions have been described as allelic variants of cathepsin C gene. Hence, we need to consider only HMS and prepubertal periodontitis in the differential diagnosis of PLS.

HMS has been described as an autosomal recessive genodermatosis characterized by congenital palmoplantar keratoderma and progressive early onset periodontitis(9). In addition to palmoplantar keratosis and periodontitis, other clinical findings in this condition include recurrent pyogenic skin infections, acro-osteolysis, atrophic changes of the nails, arachnodactyly, and a peculiar radiographic deformity of the finger consisting of tapered, pointed phalangeal ends, claw-like volar curve, and pes planus. In contrast to PLS, the cutaneous findings in HMS have been reported to be more severe and extensive(10). The periodontium in HMS may be less-severely affected than in PLS, but gingival inflammation and alveolar-bone destruction are present and severe. Although the palmoplantar findings and periodontitis are suggestive of HMS, the absence of other distinct clinical features, like nail deformities and arachnodactyly prevented us from diagnosing it as HMS.

Prepubertal periodontitis is another rare genodermatosis with an etiology attributed to a cathepsin-C gene mutation(10). It is characterized by rapidly progressive early onset periodontitis with destruction of the periodontium of deciduous and permanent teeth. Prepubertal peridontitis may be localized or generalized. Also pyogenic infection, a common feature of PLS was seen in both the legs of the present case.

A multidisciplinary approach is important for the care of patients with PLS. PLS threatens children and their parents with the prospect of edentulism if left untreated. Hence, early diagnosis and intervention is essential. For edentulous patients, oral rehabilitation is required; this includes partial or complete denture prosthetic replacement (according to the age of the patient). Osseointegrated implants are an option for the future and can have a great impact psychosocially by restoring esthetics as well as function. The pediatric dentist is the first member of the health team to see and treat children afflicted with unusual syndromes such as PLS and, therefore, awareness of this syndrome is essential if the dentist is to provide appropriate and comprehensive dental care. In addition, greater awareness of this syndrome will be helpful in identifying more cases for further study.

The skin manifestations are usually treated with emollients and oral retinoids. Oral retinoids including acitretin, etretinate, and isotretinoin are the mainstay of treatment of both the keratoderma and periodontitis associated with PLS Treatment may be more beneficial if it is started during eruption and maintained during the development of permanent teeth. But the periodontitis in PLS is usually difficult to control(11).

References

1. Papillon MM, Lefevre P. Two cases of symmetrical, familial (Meleda's malady) palmar and plantar keratosis of brother and sister: Coexistence in two cases with serious dental changes (in French). Bull Soc Fr Dermatol Syphiligr. 1924; 31: 82-7.
2. Haneke E. The Papillon Lefevre syndrome: Keratosis palmoplantaris with periodontopathy: Report of a case and review of the cases in the literature. Hum Genet. 1979; 51: 1-35.
3. Rathi SK. Papillon-Lefevre Syndrome in two brothers. Indian J Dermatol Venerol Leprol. 2002; 68:155-6.
4. Bach JN, Levan NE. Papillon-Lefèvre syndrome. Arch Dermatol. 1968; 97: 154-8.
5. Newman, Taket, Carranza. Carranza's clinical periodontology. Elsevier Publications. 2003; 172.
6. Cury VF, Costa JE, Gomez RS, Boson WL, Loures CG, De ML. A novel mutation of Cathepsin C gene in Papillon Lefevre Syndrome. J Periodontol. 2002; 73: 307-12.
7. Firatli E, Tüzün B, Efeoðlu A. Papillon Lefevre syndrome: Analysis of neutrophilchemotaxsis. J Periodontol. 1996; 67: 617-20.
8. Jansen LH, Dekker G. Hyperkeratosis palmoplantaris with periodontosis (Papillon-Lefevre). Dermatologica. 1956; 113: 207-219.
9. Haim S, Munk J. Keratosis palmo-plantaris congenital, with perikontosis, arachnodactyly and peculiar deformity of the terminal phalanges. Br J Dermatol. 1965; 77: 42-54.
10. Hart TC, Hart PS, Michalec MD, Zhang Y, Firatli E, Van Dyke TE, et al. Haim Munk syndrome and Papillon Lefevre syndrome are allelic mutation in cathepsin C. J Med Genet 2001; 38: 79.
11. Siragusa M, Romano C, Batticane N, Batolo D, Schepis C. A new family with Papillon-Lefevre syndrome: Effectiveness of etretinate treatment. Cutis. 2000; 65: 151-5.