Papillon-Lefèvre syndrome of Iranian girl: Case report and review of the literature

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Abstract

Papillon- Lefevre syndrome (PLS) is one of rare autosomal recessive disorder which characterized by hyperkeratosis of palms and soles and diagnosed in both sexes and it may have severe destructive periodontal disease affecting the primary and permanent teeth. The exact patho-mechanism of these clinical syndrome events mainly remains speculative. This is transmitted as a recessive autosomal condition and consanguinity of parents has apparent in about one-third of all cases .This paper describes classic clinical features and briefly reviews the relevant current literature.
An 11 years old female presented with keratotic plaques over the skin of her palms and soles extending on the dorsal surface and swollen gums since the age of 4 with subsequent loss of most of his permanent dentition.

Key words: palmar-plantar keratoderma, Papillon-Lefevre syndrome (PLS), periodontitis

Introduction

The first Papillion–Lefebvre Syndrome (PLS) was described by two French physicians, Papillon and Lefevre in 1924.[1] It is an extremely rare genodermatosis inherited as an autosomal recessive trait that is mainly ascertained by dentists because of the severe perioodontitis that afflicts patients.[2] PLS varies from mild psoriasiform scaly skin to overt hyperkeratosis, typically develops within the first 3 years of life. Most patients display both periodontitis and hyperkeratosis.[3] This syndrome (PLS) is one of rare autosomal recessive disorder of keratinization and locus has been mapped to chromosome 11q14-q21. The prevalence of PLS has been reported as 1 to 4 per million population both sexes are equally

affected with a carrier rate of 2–4 per 1000.The likely mechanisms relating genetics and periodontal disease include virulent infection, immune response and underlying tissue pathology.[4,5]

It may manifests "between" 1-5 year of life. Another component of this syndrome is asymptomatic ectopic calcification in choroid plexus and tentorium. Although this has been taken as a cardinal feature, but being inconsistent it is not considered important for the diagnosis. About 20% of these patients also show an increased susceptibility to infections due to some dyes-function of lymphocytes and leukocytes.[6] The diagnosis is mainly clinical. We describe here case of PLS with classic clinical features.

Case Report

An 11-year-old girl reported to the department of Periodontics for periodontal assessment and discomfort in chewing. The family history revealed to marriage of the parents. The parents and other family members were not affected. Pregnancy period and delivery were normal. The mother had noticed of skin lesions on the palms and soles of the child when they were five months old. On general examination, the patient had overall normal physical and mental development. Routine hematological examination revealed Hb was 10.0 g/dl, total white blood cell was 9200 and ESR was 20 mm/hour. On dermatological examination there were multiple fissured erythematous hyperkeratotic plaques on the soles. Xerosis and erythema on the palms and dispersed erythematous papules on the trunk.

In extra oral examination were seen hyperkeratosis of the palms, soles, and the knees of both the limbs; the affected skin was well demarcate from adjacent normal skin but the hair or nail anomaly can be observe. (Fig 1-4).

Intraoral examination

On oral cavity examination the patient had permanent dentition with no primary teeth, normal gingival and periodontum, mild deposits of plaque and calculus, were present (Figure 5).

Orthopantomograph examination

Showed no destruction of alveolar bone. Laboratory investigation assessment was carried out, which included hematological and biochemical test. The results were within normal limits. In view of the findings, the cases were diagnosed as PLS. Treatment was planned to restore masticatory function with space maintainer to keep the teeth in correct position and to restore masticatory function.

Discussion

The Papillon-Lefèvre syndrome, described by two French physicians Papillon and Lefèvere for first time in 1924,[1] Papillon-Lefèvre syndrome (PLS) is an autosomal recessive disorder[29],and extremely rare genodermatosis inherited as an autosomal recessive trait, its affect children between the ages 1-4 years.[7,8] It has a prevalence of 1-4 cases per million persons.[9] Both sexes are affected equally and there is no racial predominance.[10] Disorder is characterized by diffuse palmoplantar keratoderma and typically has its onset between age 1 and 4 years. It may have premature loss of both deciduous and permanent teeth years.[11] PLS involve the entire surface of the palms and soles and may be extending on to the dorsal surfaces of the hands and feet.[12] Hyperhidrosis of the palms and soles may result in a foul-smelling odor.[8] Well-demarcated psoriasiform plaques occur on the elbows and knees.[12] The clinical findings may worsen in winter and be associated with painful fissures. Severe periodontitis is second major feature of PLS, which starts at age 3 or 4 years. [12] The eruption of the deciduous teeth proceeds normally, but sometimes the process is associated with appearance of gingival inflammation and subsequent rapid destruction of the periodontium.[13,4] The level of periodontal infection may not be related to degree of dermatologic involvement.[15] In advanced cases nail changes may appear and in this case, manifested by transverse grooving and fissuring.[16] In addition to the skin and oral findings, decreased neutrophil, lymphocyte, or monocyte functions and an increased susceptibility to bacteria, of the patients may have associated with recurrent pyogenic infections of the skin.[17] One of complication of PLS is associated with impairment of the immune system and pyogenic liver abscess is increasingly recognized. [17] Another feature of this syndrome may be radiographic evidence of intracranial calcification.[18]

In the literature histopathological findings of affected skin have not been well described. But those reported findings have consisted of hyperkeratosis, occasional patches of parakeratosis, acanthosis, and a slight perivascular inflammatory infiltrate.[19] PLS causes is not well understood, but recently, 2 research investigator have reported that loss of function mutations affecting both the alleles of the cathepsin-C gene, located on chromosome 11q14.1-q14.3, were associated with this syndrome.[13,20] Prevalence increase of parental consanguinity has been reported in PLS patients.[20] The palmoplantar keratodermas (PPKs) are a heterogeneous group and more than 40 different types of PPK have been described.[21] PLS, HMS has been described as allelic variants of prepubertal periodontitis. And described consider only HMS and premature periodontitis in the differential diagnosis of this syndrome. Haim Munk syndrome has been mentioned as an autosomal-recessive genodermatosis characterized by congenital palmoplantar keratoderma. In 1965 Haim and Munk first reported in Jewish families from Cochin, India on the Malabar Coast. [23] In Haim-Munk syndromes affecting a highly conserved amino-acid residue, and demonstrating that PLS and are allelic disorders .The cutaneous findings in HMS have been described to be more severe and extensive. [24]

An important multidisciplinary approach for the care of patients with PLS. The skin manifestations of PPK are usually treated with emollients.[12] It may add Salicylic acid and urea to enhance their effects.[17] Treatment might be started during the eruption and maintained during the development of the permanent teeth.[22,25] The periodontitis usually PLS is usually difficult to control. Effective treatment are includes extraction of the primary teeth combined treatment with oral antibiotics and professional teeth cleaning.[17,26] Antibiotics tried to control the active periodontitis in an effort to preserve the teeth and to prevent bacteremia and subsequently pyogenic liver abscess.[17] Noack et al in their studies mentioned that PLS should be considered in all children suffering from severe aggressive periodontitis, particularly in the deciduous dentition, although the type and location of Cathepsin C (CTSC) mutations do not predict the severity, progression or therapy outcome of the disease.[27]

Conclusion

We have described an 11 years Old Iranian girl diagnosed as having highly suspected PLS and slight palmoplanter keratosis of hands and soles, together with knee pigmentation.

Conflict of Interest: None declared.

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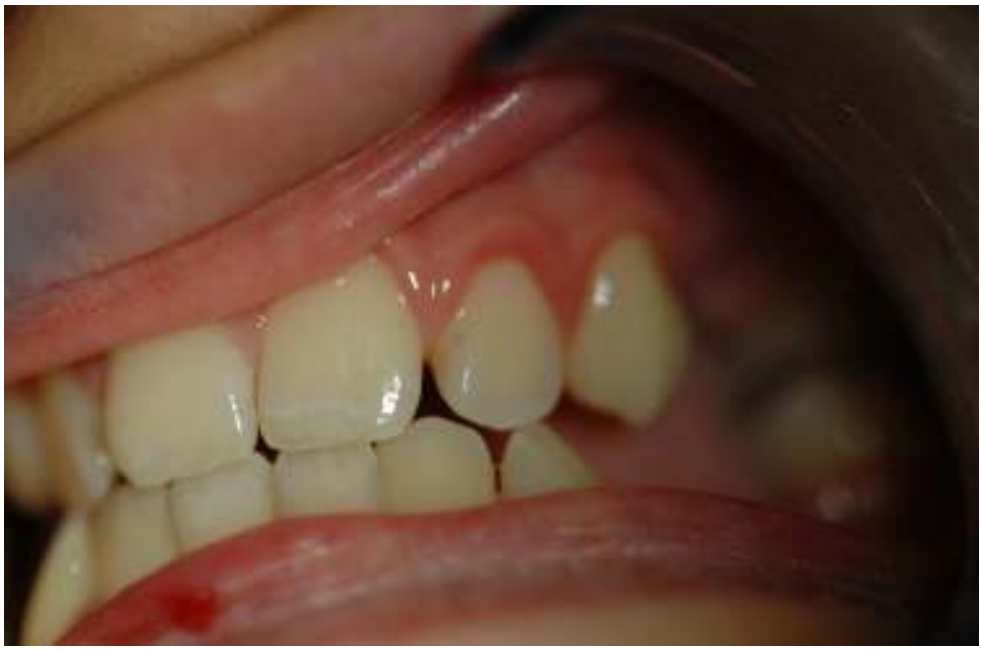

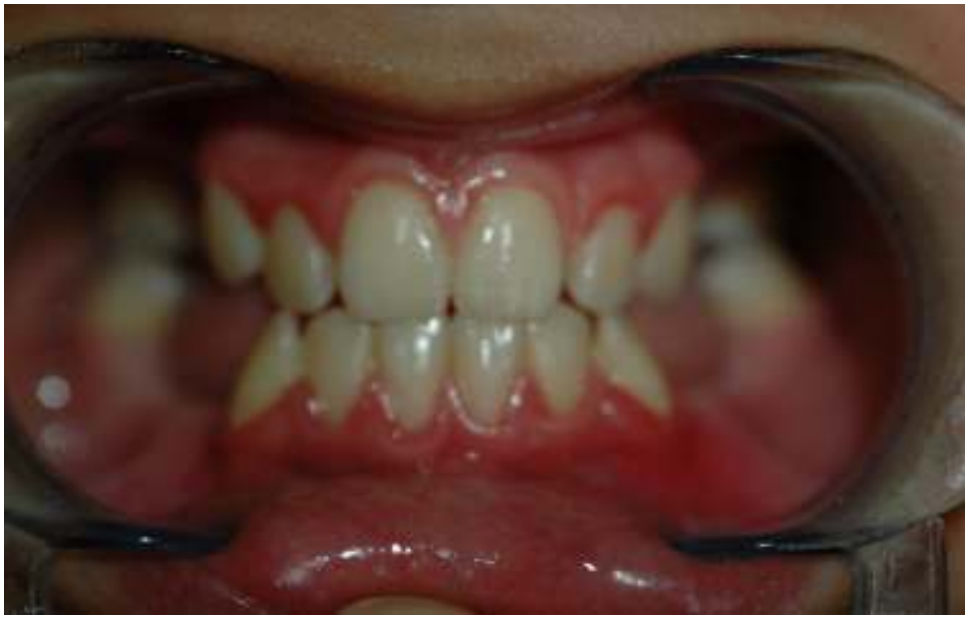

Figure 1: Absence of first and 2nd premolar in upper and lower jaw

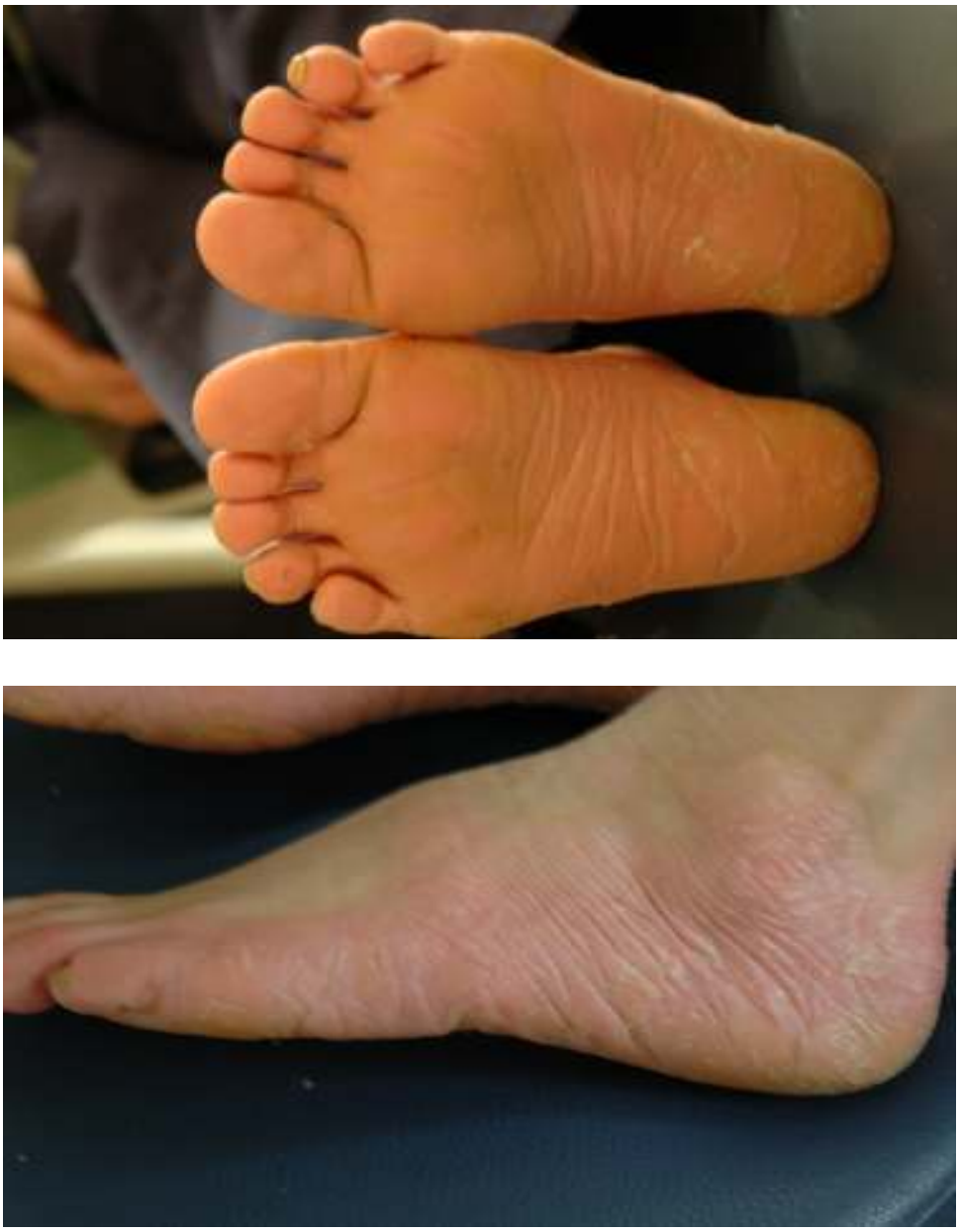

Figure 2: Yellow colored hyperkeratotic areas on sole

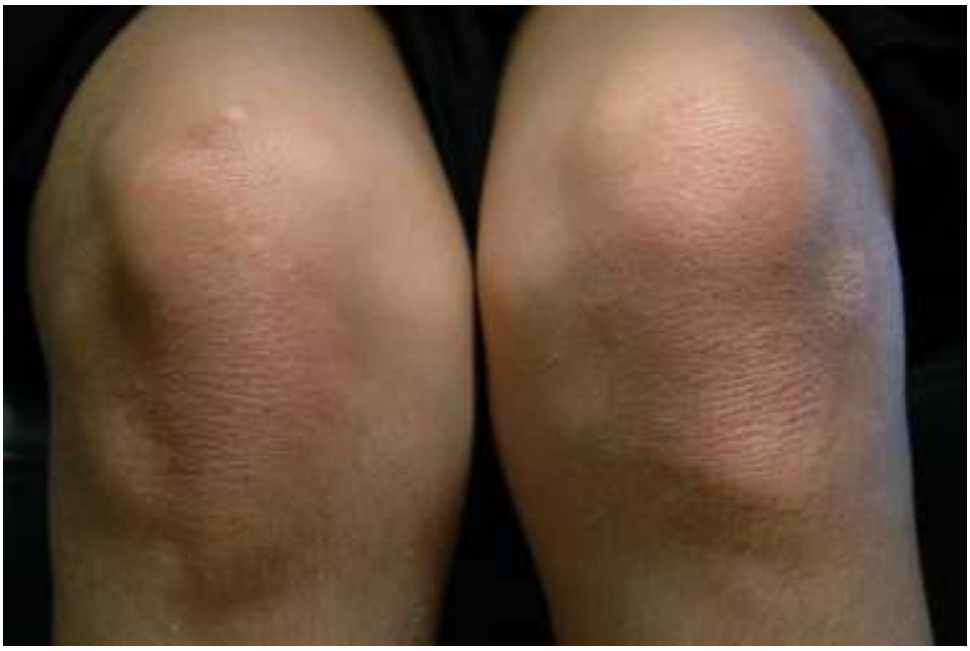

Figure 3: Photograph showing hyperkeratosis of the knees, which is well demarcated from adjacent normal skin

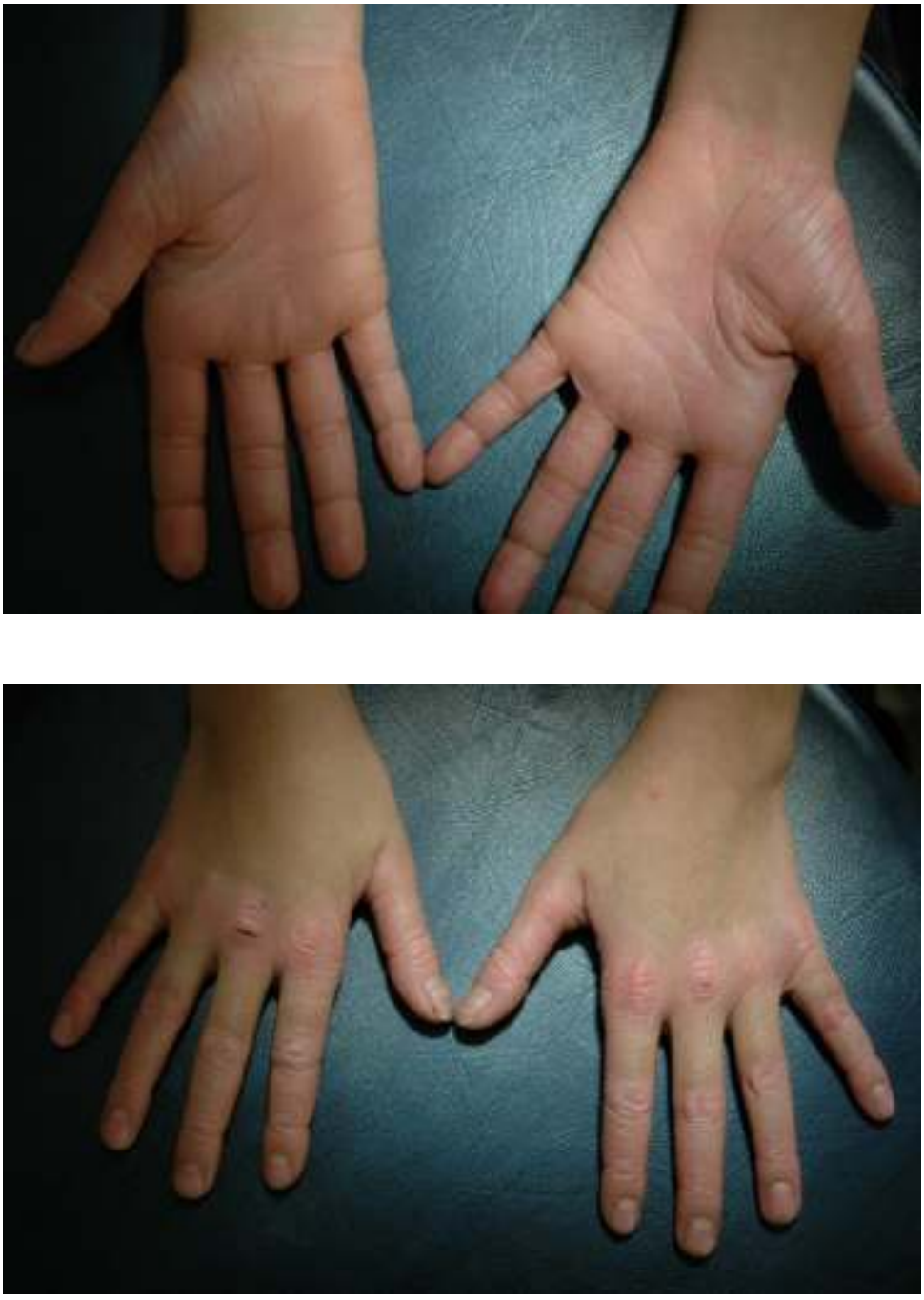

Figure 4: Photograph showing hyperkeratosis of the palms

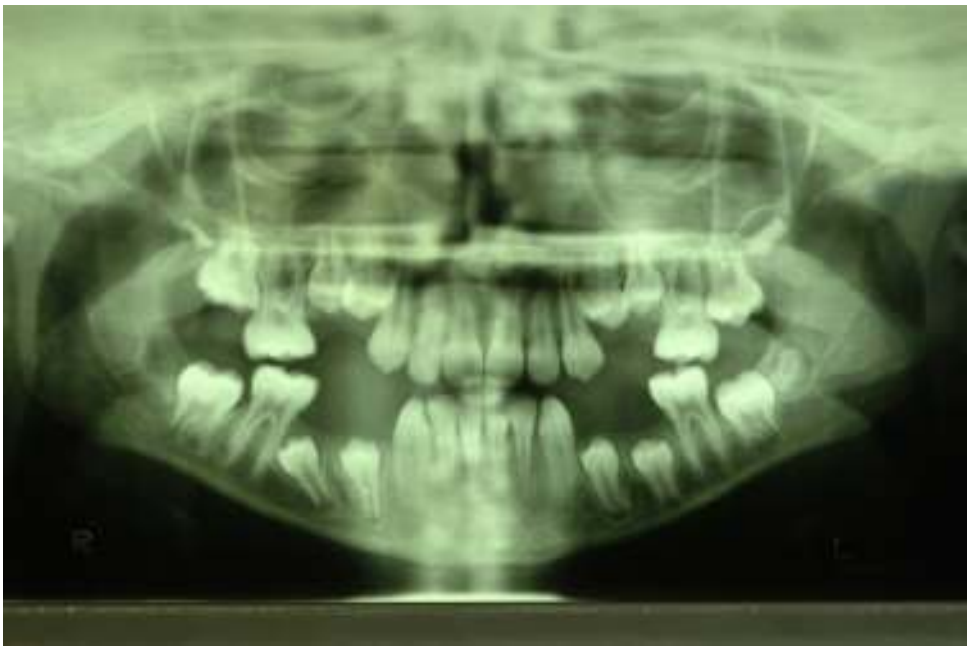

Figure 5: Orthopantomograph showing missing of first and 2nd premolars in mandible and maxilla