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PAPILLON-LEFEVRE SYNDROME: A CASE REPORT AND REVIEW OF LITERATURE

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ABSTRACT

Papillon-Lefevre syndrome (PLS) is an autosomal recessive condition characterized by dermatological manifestations and early onset periodontitis. The pathogenesis of PLS is secondary to mutation of the cathepsin C gene. Hence, the manifestations are expressed on the areas of the body covered by epithelium, such as palms, soles, knees and keratinized oral gingiva. PLS and can occur in siblings born of consanguineous marriages. This case report discusses the literature review of pathogenesis, clinical features and treatment options in a 10 year old child.

INTRODUCTION

Papillon-Lefèvre Syndrome (PLS) is a rare autosomal recessive disorder characterized by palmar-plantarhyperkeratosis, severe periodontitis, and premature loss of deciduous and permanent teeth [1]. In addition to the dermatologic and oral findings, patients may have decreased function of neutrophils, lymphocytes, or monocytes and an increased susceptibility to bacterial infection leaing to liver abscess.. Another feature of PLS is the presence of radiographic evidence of intra-cranial calcification in the choroid plexus and tentorium [2]. Studies state that PLS is caused by mutations in the cathepsin C (CTSC) gene [3-6].

This syndrome was first described by Papillon and Lefèvre in 1924 on a brother and a sister with palmoplantar hyperkeratosis associated with severe early-onset periodontitis and premature loss of primary and permanent teeth .The prevalence of PLS is 1 to 4 per million with no sex predilection and no racial predominance, and the disease becomes apparent by 2 to 3 years of age. Carriers are thought to be present in 2 to 4 per thousand individuals. PLS is a rare condition, characterized by autosomal recessive transmission. Because of autosomal recessive inheritance pattern, the parents are not typically affected. Consanguinity is noted in approximately one third of cases. The soles of the feet are severely affected, and erythema always precedes hyperkeratosis. The hands are also affected, but to a lesser degree. The sharply demarcated, erythematous keratotic plaques involve the entire surface of the palms and soles, sometimes extending onto the dorsal surface of the hands and feet. The lesions are punctate and diffuse, with dry scaly skin, and vary in thickness from one to several millimeters. Psoriasiform plaques may also be seen on the elbows and knees. The symptoms may worsen in cold weather and be associated with painful fissures [7].

Severe periodontitis associated with PLS starts at the age of 3 to 4 years. The primary dentition is usually exfoliated prematurely by the age of 4-7 years The eruption of primary teeth occurs at the expected ages and in the normal sequence, with the teeth being of normal form and structure, but their eruption is associated with severe gingival inflammation in the absence of any local etiologic factor. With the eruption of permanent dentition, the entire process of gingivitis and periodontitis is repeated and there is subsequent premature exfoliation of the

permanent teeth by the age of 12 to 15 years. Later, the third molars undergo the same process.[2,3]

A multidisciplinary approach is necessary in the management of patients with PLS [8]. The periodontal disease may be arrested by improving oral hygiene, extraction of severely diseased teeth, scaling, systemic antibiotics, and longterm antimicrobial irrigation [9,10]. This article presents a case report involving the diagnosis and management of a young patient with PLS.

Case report

A 10-year-old boy presented with mobility of his permanent teeth. He has also persistent thickening, flaking and scaling of the skin of his palms and soles associated with recurrently swollen and friable gums since age of 3. He also had of premature shedding of his deciduouous teeth . The patient's family history revealed that he is a child born of consanguineous marriage and his elder brother did not show any signs of similar systemic illness.

Extraoral examination of the patient did not show any notable changes. Intraoral examination showed gingiva were edematous, friable, upper and lower central incisors, upper and lower molars were of grade 3 mobility.

Physical examination reveals symmetric, well-demarcated, yellowish, keratotic plaques affecting the skin of his palms ,knees and soles and extending onto the dorsal surfaces.(figure 1,2)

Panoramic radiograph showed generalized horizontal bone loss extending upto the apices of the teeth and 'floating molars'. x ray also shows developing tooth buds of all canines, premolars and second molars(figure3).. Lateral cephalogram was taken rule out any dural calcifications. Laboratory data showed normal blood count, liver function transaminase levels, total bilirubin, and alkaline phosphatase .Patients parents declined for histopathology .Based on the above findings, a diagnosis of PLS was made and the patient was put on Metronidazole tablets 400mg thrice daily for 5 days and chlorhexidine mouthwashes to be used 2-3 times daily followed by oral prophylaxis .Patient was also seen by dermatologist and the patient was advised oral acitretin tablets 25 mg once daily with topical 12% salicylic acid combination of urea ointment for her skin lesions.

Figure 1. keratotic plaques affecting the and soles and extending onto the dorsal surfaces

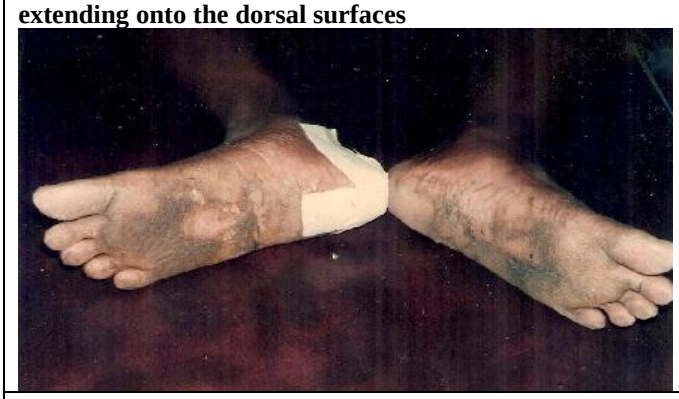

Figure 2. keratotic plaques affecting the skin of palms and knee

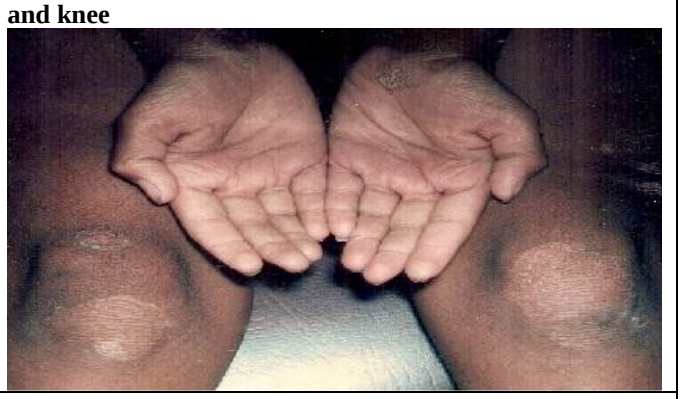

Figure 3. orthopantamofram showing bone loss in incisors and molars

DISCUSSION AND CONCLUSION

In the case presented, the dermatological and periodontal features strongly suggested the diagnosis of PLS. An extensive alveolar bone loss was noted, giving the teeth a "floating-in-air" appearance which is also characteristic of PLS [10]. Although the case reported here was associated with consanguinity of the parents, the parents were phenotypically healthy. PLS being an autosomal recessive inheritance pattern, the parents are not typically affected [2]. PLS is caused by mutations in the CTSC gene. Several mutations have been reported in this gene,. This gene is important in the structural growth and development of the skin, and is critical for appropriate

immune response of myeloid and lymphoid cells. Loss of appropriate function of the CTSC gene is thought to result in an altered immune response to infection. An increased susceptibility to infection has also been reported in approximately 25% of PLS patients. In addition, the altered gene may affect the integrity of the junctional epithelium surrounding the tooth [3-7].

Although there is a hereditary component and leukocyte dysfunction can be demonstrated, it appears that there must be an infection with a specific, potent bacterium, such as Actinobacillus actinomycetemcomitans, for the periodontitis to develop [1,2,]. PLS needs to be differentiated from other conditions showing similar oral and cutaneous clinical features, such as acrodynia, hypophosphatasia, histocytosis X, leukemiacyclic neutropenia, Haim-Munk syndrome and Takahara syndrome, which are also associated with periodontitis and premature loss of teeth [11,12].

The skin manifestations of usually treated with emollients .Salicylic acid and urea may be added to enhance their effects [1,2,10]. Oral retinoids including acitretin, etretinate, and isotretinoin are the mainstay of the treatment of both the keratoderma and periodontitis associated with PLS [11,12]. Rigorous oral hygiene, chlorhexidine mouthrinses, frequent professional prophylaxis, and periodic appropriate antibiotic therapy are necessary for long-term maintenance of the periodontium Treatment may be more beneficial if it is started during the eruption and maintained during the development of the permanent teeth [8-10,12] .

Some authors [10,13] have recommended the elimination of periodontal pathogenic flora through premature extraction of all primary teeth combined with antibiotic coverage in an attempt to minimize the possibility of infection of the unerupted permanent dentition. They claim that if the permanent teeth erupt in an oral environment free from periodontal disease, it is likely that they can remain periodontically healthy for a longer period.

If the patient has been diagnosed with PLS prior to the eruption of the permanent dentition, this therapy may be attempted as the treatment of choice. Selecting the ideal treatment is not as clear-cut when the patient seeks dental care in the mixed dentition stage or later. In these patients, the treatment plan may include either extraction of all erupted teeth followed by a period of antibiotic coverage" or extraction of only the hopelessly affected teeth combined with periodontal therapy for the remaining teeth with antibiotic coverage.

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