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Case Report

Papillon-Lefèvre Syndrome - A Rare Case Report

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Keywords

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Abstract

Papillon-Lèfevre is a rare syndrome, autosomal recessive, which shows dermatological and periodontal tissue manifestations. The main oral manifestation is aggressive periodontitis that happen even in primary or permanent dentition. Radiography shows the loss of bone insertion identified as floating teeth. Skin lesions are presented as palmoplantar keratosis, plaques and white, light yellow, brown or red spots that develop crusts, crevices or deep fissures. The aim of this study is to present a rare case report of a child-juvenile patient with Papillon-Lefevre syndrome and its ways of recognition and treatment.

INTRODUCTION

Papillon-Lèfevre syndrome was first described in 1924 by Papillon and Lefevre [1,2] as variant of "Mal de Meleda" [2]. It is a rare syndrome affecting 1-4 people per 1 million inhabitants, autosomal recessive [1-6], belonging to a group of pathologies associated with palmoplantar keratosis, and with dermatological and periodontal characteristics [2,6]. Its etiology is unknown and there is no predilection for gender [1,2,5,6]. Around 1% of cases are based on genetic issues besides its association to immunological causes.

The main feature of this syndrome is the aggressive periodontitis associated to the palmoplantar keratosis [1,2,4-6], but these characteristics may be separated initially. Transgressive and diffuse palmoplantar keratosis is the main dermatological manifestation [1,6], showing plaques and white, light yellow, brown, or red spots which develops crusts, crevices or deep fissures. Other keratinocyte lesions may appear in other sites such as skin, knees and elbows and the lesion severity predominates during winter [1,2,6].

Aggressive periodontitis appears as soon as teeth eruption leading to a quick loss of bone insertion and support [1,2,4-6]. Defects in neutrophil function and multiple immune-mediated mechanisms apparently trigger this phenomenon [2].

Radiographically, it appears as floating teeth in soft tissue because of the rapid alveolar bone resorption [5]. Papillon-Lèfevre syndrome makes differential diagnosis with Haim-Munk syndrome in virtue of the clinical findings [1,3,5,6].

The administration of retinoid shows the best treatment result for skin lesions. However, due to many adverse reactions [1], in milder cases are used topical lubricants, keratolytic agents, corticosteroids or antibiotics [3]. In oral manifestations, the treatment involves antibiotic therapy and intense periodontal therapy, but with different applicabilities [1,2,5,6].

The aim of this study is to present a rare case report of a child-juvenile patient with Papillon-Lefevre syndrome and its ways of recognition and treatment

CASE PRESENTATION

A 12-year-old male patient (Figure 1) went, student, with his mother, to the Oral and Maxillofacial Surgery Service of Universidade Federal da Bahia (UFBA), in Salvador, Bahia, Brazil referring severe toothache, halitosis, and recurrent gingival bleeding. He mentioned softening and falling teeth about three years ago.

During clinical examination was observed a hyperemic gingiva (Figure 2), an easily bleeding at touch, and a considerable mobility in almost all teeth. Hematology tests and periapical radiography were requested.

The hematology tests had no alterations and radiographies showed (Figure 3-5) generalized bone loss in all sextants. A more detailed anamnesis was done and findings of palmoplantar keratosis (Figure 6,7) and complaints ignored by the genitor because she judged less important in a dental consultation and therefore it was not previously informed were decisive for the diagnosis of Papillon-Lèfevre syndrome. The patient reported that the keratoses lesions were asymptomatic, but they were frequent foci of infections. Faced with this finding, the marriage's history of the parents was investigated, who were not consanguineous.

The patient was referred to Periodontics Service at Dentistry College of Federal University of Bahia and was diagnosed with

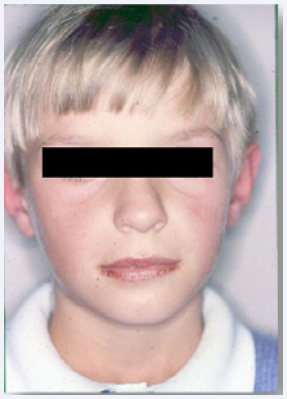

Figure 1 Patient in frontal photo.

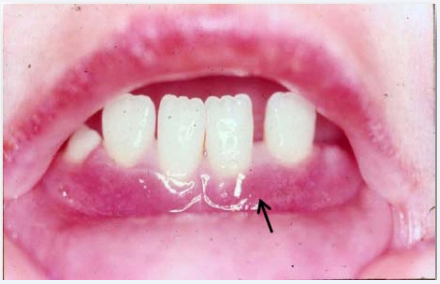

Figure 2 Gingival hyperemia.

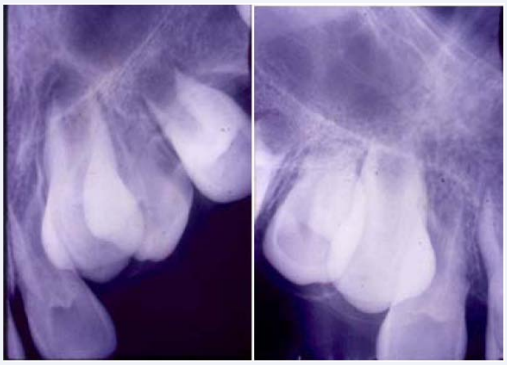

Figure 3 Generalized bone loss at sextants 1,3.

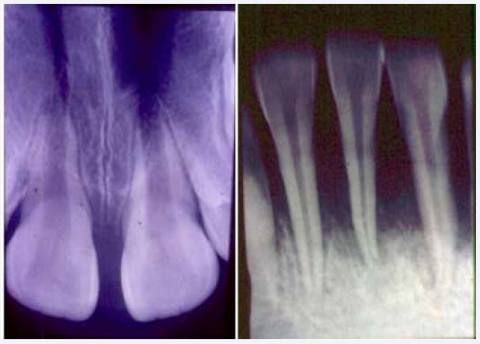

Figure 4 Generalized bone loss at sextants 2,5.

general aggressive periodontitis. Extractions of remains of primary teeth, scaling, root planning, plaque control, and oral hygiene instructions were planned. Antibiotic therapy was started with tetracycline 20 mg/kg/day orally for ten days. The dose may be repeated as judged by the need assessed by the professional.

The patient and his mother were enlightened about the disease and he was also referred to the doctor to evaluate and treat cutaneous lesions.

DISCUSSION

Papillon-Lèfevre syndrome is autosomal recessive in which both parents are carries of the defective gene and the risk of affected children is approximately 25%. It is also a keratinization disorder characterized by redness, thickening of the plants and palms of the hands and severe and destructive periodontal disease [7,8].

Besides its unknown etiology, at a genetic level, the gene 11q

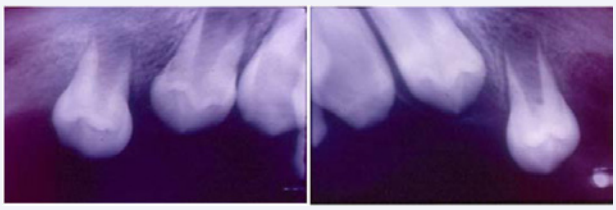

Figure 5 Generalized bone loss at sextants 3,6.

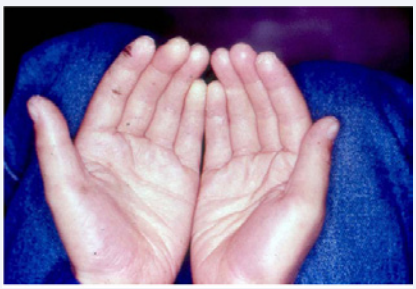

Figure 6 Palm keratosis.

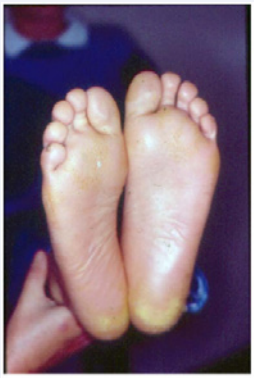

Figure 7 Plantar keratosis.

and mutation at Cr 12 and 17, which produce cytokeratin, have been associated to [1,2]. Studies such as that of Toomes et al. [7], point out that one of the causes involved in the development of the syndrome is also the gene gene mutation of cathepsin C found in commonly affected epithelial regions.

Other factors are associated: anatomic, viral, and microbial agents and host response. In the sub gingival plaques of periodontal pockets, the most frequent microbial agent, which covers all age groups, is *Actinomyces actinomycestemcomitans* [1], considered as one of the suspected periodontal pathogens [7-9].

Papillon-Lèfevre syndrome is restricted, for the most part, to study cases because of it is a rare pathology [6,10]. Few studies demonstrate alterations in saliva and gland function, severe periodontal bone destruction caused by neutrophil dysfunction in addition to deregulatory factors of the cells of immune system [1,2,10].

The differential diagnosis includes acrodynia, hypophosphatasia and cyclic neutropenia. Although these illness are associated to palmoplantar hyperkeratosis, the periodontitis is not seen in them [7,10].

In order to investigate the syndrome, laboratory tests such as urine analysis, alkaline phosphatase, hormone tests, blood count, neutrophil function test, may be asked. However, all of them are within the normal range for Papillon-Lefèvre Syndrome, being important to rule out other pathologies [7-10]. As observed in the case report, the patient in question had no alterations in laboratory tests, and in view of the clinical evidence of plantar palmar keratosis and the early inflammation of the mixed dentition, there was no doubt about the Papillon-Lefèvre diagnosis. Therefore, more specific exams such as biopsy of gingival or keratotic lesions, chromosome analysis, are necessary.

Skin lesions usually start in the first four years of life. The treatment in milder cases is the use of topical lubricants and keratolytic agents [1], corticosteroids or antibiotics [3]. The administration of retinoid has showed the best treatment results for skin lesions in both epidermis and oral cavity, but it presents many adverse effects [1]. Normally the recommended starting dose for acitretin is 0.5mg/kg/day5 and 1.5mg/kg/day for isotretinoin [7,10].

The patient was referred to the dermatologist so that specific and individualized treatment of the skin lesions could be done. He did not return to the team that made the diagnosis and therefore it was not possible to follow the progress of the treatment.

In oral manifestations, the treatment is, at most, palliative and involves antibiotic use associated to extractions of primary dentition, intense periodontal therapy, and oral hygiene, which have showed effective results to maintenance and preparation of the oral cavity for the permanent dentition [1,2,5,6].

The commonly used antibiotics are tetracycline and erythromycin, but in some cases systemic antibiotics such as amoxicillin (20-50 mg/kg/day) plus metronidazole (15-35 mg/kg/day) in divided doses of 8 hours are used as adjunctive treatment to the conventional treatment. In the present report, tetracycline was used, which shows good results in the periodontal treatment, already based on the literature [8].

It is concluded that the Papillon-Lefèvre syndrome can be easily diagnosed by dentists, due to the association of clinical and radiographic findings related to generalised aggressive periodontitis, as well because of the characteristics of palmoplantar keratosis. It is necessary a cautious look, giving importance to lesions that are far from the oral cavity, and that can be crucial for the closing of the early diagnosis, leading to an optimization of the treatment.

CONFLICT OF INTEREST

The author(s) declare(s) that there is no conflict of interest regarding the publication of this paper.

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