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Case Report

Infantile Papillon-Lefèvre Syndrome: About a Case

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Abstract

Papillon-Lefèvre syndrome (PLS) is an autosomal recessive disorder characterized by palmoplantar keratoderma and early and severe periodontitis. We report the case of a 3-year-old child with erythematous palmoplantar keratoderma with diffuse involvement of the palms and soles. The father reported that his older son had the same symptoms. On examination, lesions of palmar-plantar hyperkeratosis were well limited. Endobuccal examination showed premature loss of temporary teeth, severe and generalized gingival inflammation, recessions and mobilities on residual teeth. The panoramic radiograph had shown lysis of the alveolar bone around the residual teeth. The diagnosis of LPS was made on the basis of clinical and radiographic findings. The treatment consisted of odontological care by teaching the child the means and methods necessary for oral hygiene, followed by the extraction of non-preservable teeth. And for the dermatological manifestations they were treated with emollients and salicylic acid at first. LPS is a rare hereditary pathology associating cutaneous and dental disorders. The diagnosis of certainty is established by genetic analysis. The treatment is multidisciplinary between pediatrician, dermatologist and dentist.

Keywords: *Papillon-Lefèvre Syndrome; Genetic Disorder; Palmoplantar Keratoderma; Periodontitis*

Introduction

Butterfly-Lefèvre syndrome (PLS), an inherited genetic disease with autosomal recessive inheritance, characterized by a total loss of cathepsin C activity. This disease has an estimated prevalence of 1 - 4 cases per million people, has no gender bias but has a higher incidence in countries with a high rate of consanguineous marriages. Early diagnosis of SPL is usually based on clinical signs. Generally speaking, keratoderma and periodontitis begin simultaneously between the 6th month and the 4th year of life of the patient, coinciding with the eruption of the first teeth.

It is characterized by palmoplantar keratoderma and early and severe periodontitis.

We report the case of a patient with Lefèvre butterfly syndrome discovered at an early age.

Observation

A 3-year-old male child from a first-degree consanguineous marriage had erythematous palmoplantar keratoderma with diffuse palm and plant involvement. The father reported that his eldest son has the same symptomatology. On clinical examination lesions of palmo plantar hyperkeratosis well limited (Figure 1 and 2).

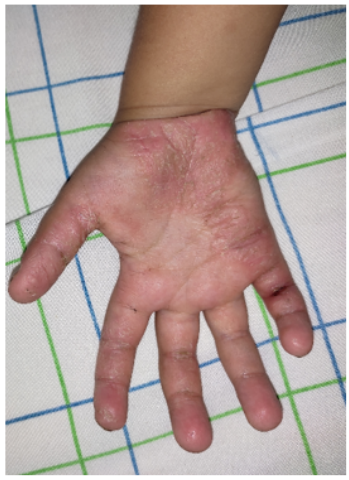

Figure 1: *Palmar hyperkeratosis.*

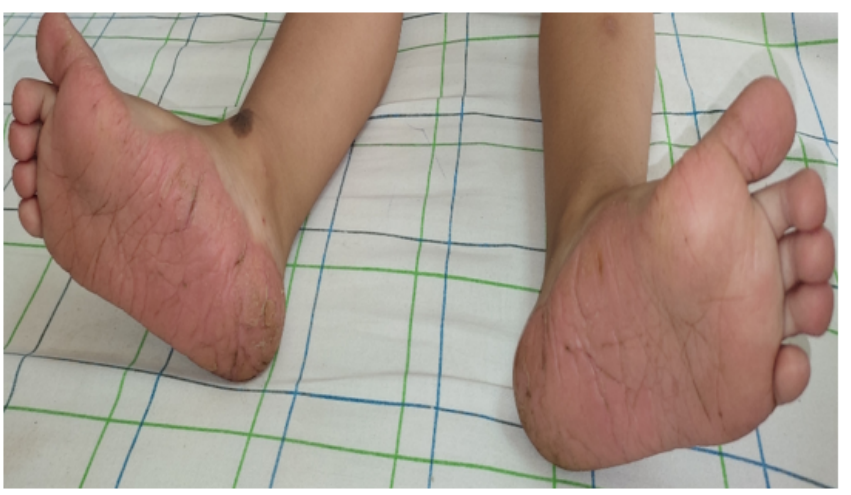

Figure 2: *Plantar hyperkeratosis.*

Endobuccal examination showed premature loss of temporary teeth, severe and generalized gum inflammation, recessions and mobilities on residual teeth (Figure 3).

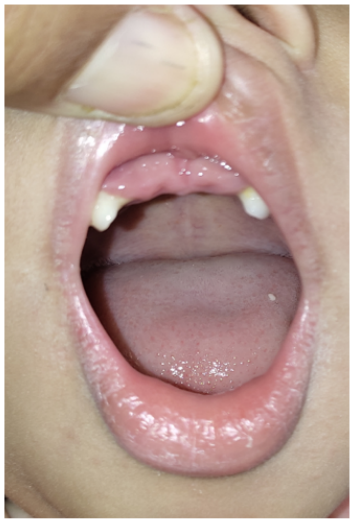

Figure 3: *Odontological anomalies.*

The panoramic X-ray had objectified a lysis of the alveolar bone around the residual teeth.

The diagnosis of SPL was made based on clinical and radiographic signs. The genetic study was not done because of the high cost and non-availability in Morocco.

The treatment consisted of odontological care by teaching the child the means and methods necessary for oral hygiene first and then the extraction of non-conservable teeth.

Regarding the dermatological manifestations they were treated with emollients and salicylic acid at first.

Discussion

Butterfly-Lefèvre syndrome (SPL) was first described in 1924 by two French dermatologists Papillon and Lefèvre. It is a rare genotypic condition with an incidence of 1 to 4 cases per million [1-3].

This is lysosomal ectodermal dysplasia. Cathepsin C is a lysosomal protease involved in epidermal differentiation and desquamation also playing a minor role in immunity explaining the susceptibility of patients to infections, especially cutaneous and respiratory [4].

SPL constantly combines constant dermatological and dental signs.

The main odontological features consist of gingivostomatitis and severe periodontitis. The eruption of baby teeth occurs at the expected age, in normal order, with normal structure and shape, although microdontia, root resorption and incomplete root formation have been reported in some cases. In the first year after the eruption of the baby teeth, the gum becomes inflamed, followed by rapid destruction of the periodontium, which is manifested by a visibly red and swollen gum, associated with significant bone resorption and deep periodontal pockets from which pus flows at the slightest pressure. Chewing is very painful due to the mobility of the teeth. Usually, a foul smell from

the mouth is present. Features of dental manifestations are loosening, mobility, drifting, migration and exfoliation of teeth without signs of root resorption [5].

In our case the odontological manifestations were premature loss of temporary teeth, severe and generalized gingival inflammation, recessions and mobilities on residual teeth c which joins the literature.

For dermatological manifestations, they were characterized by hyperkeratosis diffuse palmoplantar and erythematous [1] and usually manifest themselves at the same time as oral manifestations between the age of 6 months and 4 years, coinciding with the eruption of milk teeth. At first, a thickening of the skin is observed; later, the skin lesions are well delineated and extend to thenar and hypothenar eminences of the palms, the Achilles tendon and the external malleolus of the feet. The dorsal side of the fingers and toes as well as the elbows, knees, legs and thighs are less severely affected, and the trunk is rarely affected. Lesions can vary in color, texture and manifestation; they come in the form of white, brown, red and scaly patches that undergo crusts, cracks and deep cracks, Skin lesions worsen in cold weather and patients feel pain when walking. An infection can overlap with defective skin, leading to the formation of abscesses [5]. The butterfly-Lefèvre type keratoderma is clearly limited by a red band; it evolves by flare-ups and is accompanied by partial, sometimes almost total, remissions [1]. Our patient had experienced red and well limited palmo plantar desquamation with thickening of the skin without other dermatological manifestations.

Genetically, transmission is autosomal recessive and there is no predilection according to sex, a high incidence of SPL is observed in consanguineous marriages [1,5]. The gene responsible for the syndrome was localized in 11q14/q21, encodes cathepsin C (CatC), lysosomal cysteine protease responsible for the activation of many proinflammatory serine proteases of immune cells, including neutrophilic serine proteases (NHP), elastase, proteinase 3 and cathepsin G [1,7]. The CTSC gene is expressed in epithelial regions affected by butterfly lip syndrome, such as palms, plants, knees, and keratinized gums. Several mutations have been reported in the CTSC gene in individuals of various ethnic groups [5].

These were the cases for our patient given the unavailability of the genetic study we retained the diagnosis on the clinical and radiological elements.

Multidisciplinary care involving a dermatologist, a pediatrician and a dental surgeon.

There are no cures for SPL, so it is important to diagnose this disease as early as possible to be able to treat patients in prophylaxis, even before clinical signs appear, thus improving the quality of life of these patients.

Dermatological manifestations of SDP are usually treated with emollients, to which salicylic acid and topical steroids can be added to enhance their effect. Oral retinoids such as acitretin, etretinate and isotretinoin have been shown to be beneficial in the treatment of dental and skin lesions of SDP. Retinoid therapy is usually started at the time of eruption of the succession teeth and is followed until the end of the normal developmental process. These retinoids are metabolites of vitamin A that are involved in regulating the growth and differentiation of epithelial cells, and they are known to have a profound effect on keratinization by decreasing the total keratin content of keratinocytes [5,6,8], our patient was put on emollients and salicylic acid at first, until the moment we did not resort to retinoids.

The main goal of dental treatment in the case of PLS is to remove the bacterial reservoir in order to control the destruction of the periodontium. Several treatment modalities have been suggested, such as: conventional periodontal treatment in the form of scaling and root planing; oral hygiene lessons and mouthwashes with 0.2% chlorhexidine gluconate; and antibiotic therapy, which is suggested to control active periodontitis with the aim of preserving teeth and preventing bacteremia and subsequent pyogenic liver abscess, which is a complication of PDS due to impaired immune system [5]. Oral hygiene measurements with mouthwashes with 0.2% chlorhexidine gluconate, was set up for our patient with conventional periodontal treatment in the form of scaling and root planing is planned [9].

A restorative phase consisting of the prosthetic replacement of the absent teeth by a temporary prosthesis aimed at restoring aesthetics, especially to maintain the psychological profile of the child. A phase of supportive periodontal therapy aimed at maintaining the results obtained and controlling the course of the disease [1,9].

Conclusion

Papillon-Lefèvre syndrome is a rare hereditary pathology that must be thought of in the context of the association of skin involvement with a type of palmoplantar hyperkeratoderma erythematosquamous and dental type of severe periodontitis with alveolysis and premature fall of the teeth. The diagnosis of certainty is established by genetic analysis. The treatment is multidisciplinary between dermatologist and dentist and is mainly based on oral retinoids and the placement of dental prostheses. The life expectancy of these patients is not diminished.

Bibliography

1. Mustapha El Alloussi., *et al*. "Papillon-Lefèvre syndrome: a case report". *Médecine Buccale Chirurgie Buccale* 15.4 (2009): 193.
2. Shahbaz AJ and Khachemoune A. "Papillon-Lefèvre syndrome: case report and review of the literature". *Dermatology Online Journal* 10.1 (2004): 13.
3. Peter H and Susanna K. "Palmoplantar keratodermas". *Clinics in Dermatology* 23.1 (2005): 15-22.
4. N Gaslain. "Syndrome de Papillon-Lefèvre". *Annales de Dermatologie et de Vénéréologie* 147.12 (2020): A271-A272.
5. Basapogu Sreeramulu. "Papillon-Lefèvre syndrome: clinical presentation and management options". *Clinical, Cosmetic and Investigational Dentistry* 7 (2015): 75-81.
6. Kola MZ. "Prosthodontic Management of Papillon-Lefevre syndrome with special focus on its characterization and diagnosis: A Clinical Report". *Journal of Advanced Medical and Dental Sciences Research* 2.4 (2014): 97-104.
7. Brice Korkmaz. "Diagnostic précoce du syndrome Papillon-Lefèvre and Registre international des patients". International Cathepsine-C consortium (2015).
8. Zhao Yu-Lei., *et al*. "Papillon-Lefèvre Syndrome: A Case Report". *International Journal of Dermatology and Venereology* 4.2 (2021): 97-99.
9. Ahuja V., *et al*. "Papillon-Lefèvre syndrome: a successful outcome". *Journal of Periodontology* 76.11 (2005): 1996-2001.

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