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Papillon - Lefevre Syndrome: A Rare Case Report

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Abstract: ***Papillon - Lefevre syndrome is a rare condition, inherited through autosomal recessive inheritance, defined by hyperkeratosis of the palms and soles and severe, early - onset destructive periodontitis that results in the premature loss of both primary and permanent teeth. This syndrome is linked to mutations in the cathepsin C gene, located in the main gene locus of chromosome 11q14. Early diagnosis and intervention can help preserve the patient's teeth and delay premature loss. This case study discusses a 6 - year - old Saudi girl who displays all the characteristic features of Papillon - Lefevre syndrome.***

Keywords: Papillon - Lefevre syndrome, hyperkeratosis, periodontitis, cathepsin c gene

1. Introduction

Papillon - Lefevre syndrome (PLS) was first described by Papillon and Lefevre in 1924 [1]. This rare autosomal recessive condition presents clinically with palmoplantar hyperkeratosis and rapidly progressive periodontal disease, leading to premature loss of both primary and permanent teeth [1, 2]. PLS can be hereditary, acquired, or associated with other syndromes [3]. The incidence of PLS ranges from 1 to 4 per million, with no gender preference and a higher likelihood of offspring from consanguineous marriages. It typically manifests within the first 4 to 5 years of age, affecting both deciduous dentition and periodontal involvement, and often results in early loss of teeth. The dermatologic feature of PLS includes palmoplantar keratosis, which ranges from mild psoriasiform scaly skin to overt hyperkeratosis and often affects the elbows and knees [1, 2].

2. Case Report

A 6 - year - old girl, the first child of her parents, who are first - degree cousins and have no family history of significant diseases, was examined at The Pediatric Clinic of the National Guard Comprehensive Specialized Clinic in Riyadh, Saudi Arabia. She presented with persistent thickening, flaking, and scaling of the skin on her palms and soles (Figure 1). Additionally, she reported loose teeth, discomfort while chewing, recurrently swollen, friable gums, and multiple missing teeth (Figure 2). Based on these symptoms, she was diagnosed with PLS. Her laboratory examinations and an ultrasound scan of her abdomen showed normal results. Her management included extensive moisturization and a referral to dental services, which confirmed the missing teeth and offered continued follow - up for further management. Additionally, she was referred to a genetics clinic for genetic studies.

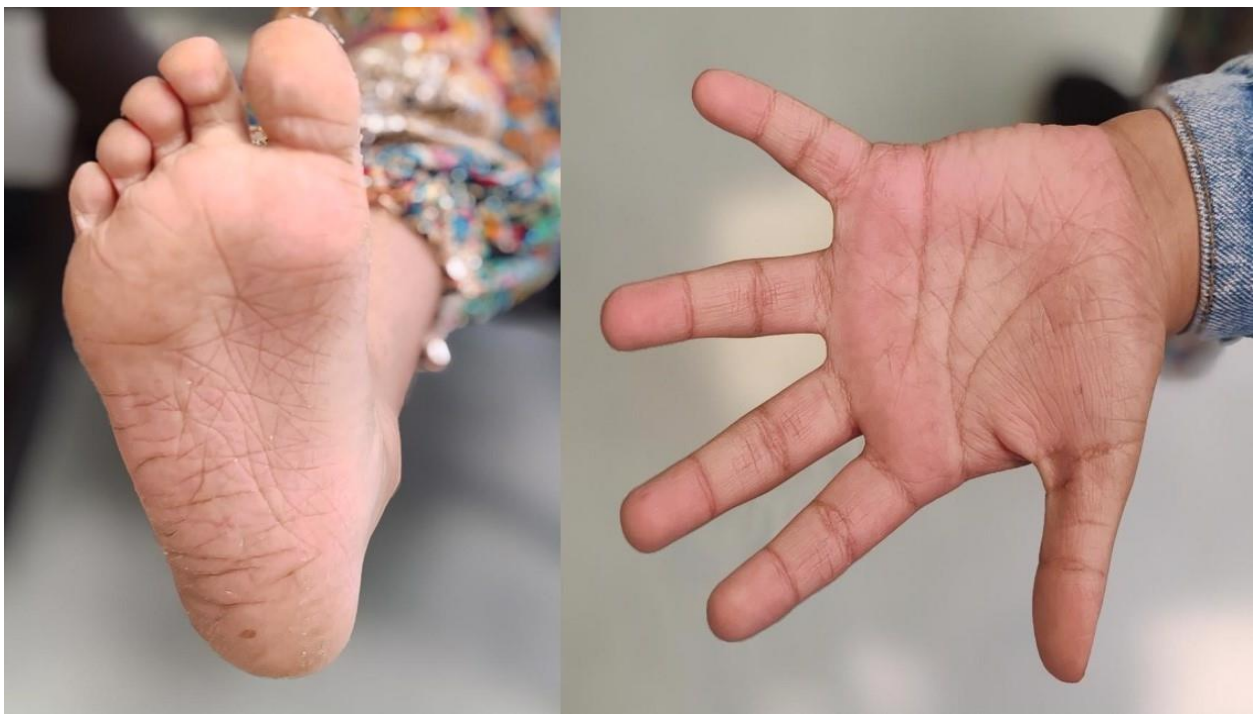

Figure 1: Palmoplantar hyperkeratosis

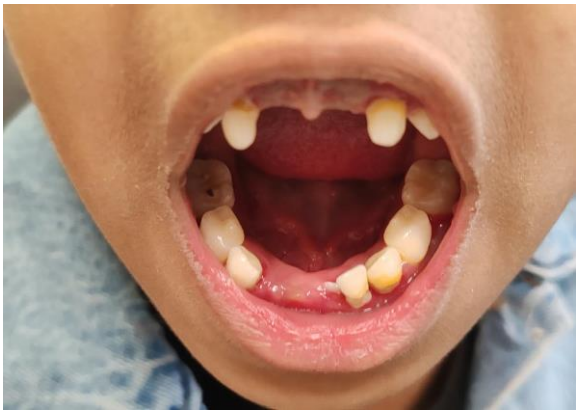

Figure 2: Intra - oral view indicating missing teeth

3. Discussion

PLS is an extremely rare genetic disorder that often remains undiagnosed until a severely affected periodontium develops, significantly affecting the social and psychological well - being of the diagnosed patient. The exact cause of PLS remains uncertain. Yet, its development and progression are thought to be influenced by microbial, immunological, and primarily genetic factors. PLS's primary gene locus is on chromosome 11q14.1 - q14.3. In PLS patients who are homozygous for cathepsin C gene mutations, a loss of function was identified [4]. This gene is highly active in immune defense cells and epithelial tissues, particularly in gingiva and the skin's ventral surfaces of hands and feet [5]. Toomes et al. argued that PLS has a strong genetic basis, noting that PLS patients exhibited mutations that affected the cathepsin C gene alleles, a lysosomal protease [6]. Cathepsin C is crucial for the production of cathelicidin antimicrobial peptide LL - 37, which was found in reduced levels in the gingival fluid and neutrophils of PLS patients [5]. Almuneef et al. found that pyogenic liver abscess could be identified as a PLS complication due to compromised immune system function [7]. No abnormal liver function or ultrasound findings were reported in the presented case.

The early detection of PLS and a multidisciplinary treatment approach can significantly improve patient prognosis. Systemic antibiotics are effective in mitigating active periodontitis, while oral hygiene practices can improve patient quality of life [5]. Pacheco et al. concluded in a case study that by combining scaling and root planning with amoxicillin - metronidazole systemic therapy, the progression of periodontal disease in PLS patients can be effectively halted. This positive result could likely be attributed to the reduction or eradication of *A. actinomycetemcomitans*. Removal of deciduous teeth with a poor prognosis, along with the elimination of periodontal infections, creates a more favorable environment for the emergence of permanent teeth [8].

The selection of rehabilitation techniques for edentulous PLS patients rests on numerous factors, such as specific patient needs and preferences, support from the surrounding bone structure, and the cost of treatment procedures. Various treatment methods, including overdentures, conventional, modified, and implant - supported complete dentures or tailored modifications of these approaches may be recommended for rehabilitation [8].

Prosthodontic rehabilitation significantly contributes to the patient's psychological well - being and their parents'. This improvement occurs because aesthetics are restored, and overall oral function is enhanced [8]. Effective treatment options include emollients, salicylic acid, and urea to address skin lesions. In addition, oral retinoids, such as acitretin and isotretinoin, have proven effective in treating keratoderma.

Genetic counseling is advised, and consanguinity should be discouraged to prevent inherited conditions that can diminish the quality of life of the offspring. Parents of a child with PLS need to be aware of the 25% chance of PLS occurring in subsequent offspring [9].

4. Conclusion

PLS is a rare, autosomal recessive inherited disorder with profound long - term impacts on a child's social, psychological, aesthetic, and functional attributes. Early detection and intervention are crucial to prevent the child patient from becoming fully toothless.

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