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Case Report

Papillon–Lefèvre Syndrome: A Rare Case Report

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Abstract

Papillon-Lefevre syndrome (PLS) is an autosomal recessive disorder that begins in early childhood and is extremely rare. It is characterized by aggressive periodontal disease, palmar plantar hyperkeratosis, a tendency for dry and chapped skin, and thin sparse hair. Patients may exhibit early tooth loss between the ages of 2 and 4, followed by premature loss of permanent teeth during adolescence. The etiopathogenic factors that are associated with PLS includes genetic, immunological, and microbial factors. A 4-year-old boy presented with a chief complaint of multiple losses of teeth and an unsatisfactory aesthetic appearance. His gingiva appeared edematous, bright red, and bled readily upon probing, and the teeth were mobile. Hyperkeratosis was observed on both the palms and soles. These findings are consistent with Papillon-Lefèvre syndrome. Early detection and diagnosis of Papillon-Lefevre syndrome play important roles in improving the physical and mental well-being of the patients.

Keywords: Cathepsin C, consanguinity, palmar-plantar hyperkeratosis, Papillon-Lefèvre syndrome, premature tooth loss

Introduction

Papillon-Lefevre syndrome (PLS) is an extremely rare genodermatosis of autosomal recessive inheritance manifesting as palmar-plantar hyperkeratosis with periodontitis.[1] In 1924, two French physicians named Papillon and Lefevre were the first to describe it.[2] Both males and females are equally affected with no racial predominance. It has a prevalence of approximately 1–4 per million.[3] The exact etiologies and pathogenesis of PLS are still controversial and difficult to understand.[4] Three factors have been proposed to underlie the progression of PLS.[5] First-factor mutation of the lysosomal protease cathepsin C gene (CTSC, or DPPI; 602365) which is located on chromosome 11q14.1-q14.3. The bacterial flora in PLS patients is resemble to the flora presented in patient with chronic periodontitis such as aggregatibacter actinomycetemcomitans which is considered the second most common factor associated with progression of PLS.[6,7] The third factor is that approximately 20% of patients with PLS have an increased susceptibility to infection and a reduced immunological response due to the reduced chemotactic and phagocytic functions of neutrophils and other granulocytes.[4,8,9]

Approximately one-third of the cases are linked to consanguineous marriages.[1,6]

This syndrome is characterized by diffuse palmoplantar hyperkeratosis, premature loss of deciduous and permanent teeth, and a tendency for recurrent pyogenic infections of the skin.[5] Palmoplantar hyperkeratosis can spread to other parts of the body such as the knees, elbows, back, and fingers.[6] The hair is typically normal, but in advanced cases, it may become thin and sparse.

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The nails may display transverse grooving and fissuring in advanced cases. The onset of palmoplantar keratosis of PLS typically occur between the ages of one and four years, coinciding with the eruption of primary teeth.[8]

The second major feature of PLS is severe periodontitis, which results in the premature loss of both deciduous and permanent teeth, leading to edentulism in adolescence.[3,6] Girls tend to exfoliate their teeth earlier than boy , but the tooth loss sequences remain consistent in both groups.[10] The gingiva is bright red, edematous, and easily bleeds. Gingival inflammation is associated with the eruption of primary teeth in the absence of any etiological factors.[6] The periodontal pockets deepen rapidly, leading to severe loss of alveolar bone and marked fetor exoris.[3] After exfoliation of the primary teeth, the gingiva returned to normal. The aggressive periodontal inflammatory process retriggers itself after the eruption of permanent teeth, with subsequent premature exfoliation of the permanent teeth by the age of 13–16 years.[3,6]

Radiographic features are characterized by calcification of the falx cerebri and severe generalized loss of alveolar bone giving the teeth a 'floating-in-air' appearance.[3,6] In some cases, the unerupted teeth take an unusual posture and have partially developed roots.[11] Histologic examination of the affected skin revealed hyperkeratosis, acanthosis, tortuous capillaries in thinned dermal papillae, and slight perivascular inflammatory infiltrate.[3,12] In periodontal pockets, the inflammatory response leads to exocytosis of inflammatory cells.The underlying connective tissue shows increased vascularity, which facilitates the influx of immune cells such as plasma cells, lymphocytes , histocytes and polymorphonuclear neutrophils.[11]

Several other disorders can be differentiated from papillon Lefever syndrome (PLS) such as Feer's syndrome (mercury intoxication), Palm Haim-Munk syndrome, Howel-Evans syndrome, Greithers's syndrome, and Vohwinkel syndrome.[11] Haims-Munk Syndrome (HMS) is a variant of Papillon-Lefevre Syndrome. Clinical features include palmoplantar keratoderma, loss of dentition, arachnodactyly, and acroosteolysis.[13]

The prognosis of this disease is poor, and the course is unpredictable therefore the management of PLS is challenging to dentists. A team of dermatologists, pediatricians, and dental surgeons (periodontists, pediatric dentists, and prosthodontists) should work together to manage the case. The present report represents a suspected case of PLS in a 4-year-old boy.

Case Report

A 4-year-old boy presented to the pediatric dentistry department in Al Mushrif Children's Specialty Center, part of the Ambulatory Healthcare Services (AHS) in Abu Dhabi, UAE, with a chief complaint of multiple tooth loss and an unsatisfactory aesthetic appearance. Proper informed and photographic consent was obtained from the patient's father. According to his family history, his parents were in a consanguineous marriage. His parents were not affected and had a normal healthy pregnancy and delivery. According to the patient's father, his deciduous teeth erupted normally but started exfoliating at the age of three. There was no previous dental checkup or treatment, and no medical history that influenced the outcome.

Clinical examination

During the general examination, it was observed that the patient's physical and mental health were within normal limits. Extraoral examination revealed normal face symmetry with loss of vertical facial height and an edentulous appearance due to premature loss of teeth (Fig. 1).

Cutaneous examination revealed symmetrical, well-demarcated, rough, scaly accentuation with eczematous changes affecting both the palmar surface of his hands and the plantar surface of his feet (Fig. 2).

Intraoral examination revealed the presence of multiple deciduous teeth with grade III mobility. The teeth included primary maxillary canines and second molars, mandibular left canines, and second molars. The gingiva surrounding these teeth appeared erythematous and edematous associated with thick plaque accumulation and deep periodontal pockets which is evidence of severe gingival recession in some teeth. Generalized alveolar bone resorption and halitosis were also evident. (Fig. 3).

Radiographic examination

Panoramic radiography confirmed generalized alveolar bone resorption with a floating-in-the-air appearance of the present teeth due to their increased spacing and gingival recession extending to the apical third of the roots. Permanent tooth buds are evident radiographically in the alveolar bone of both the maxillary and mandibular arches (Fig. 4).

Treatment

After considering the patient's clinical and radiographic features, a diagnosis of PLS was made. A genetic test was recommended to confirm the diagnosis, but the parent could not afford it. Its management is challeng-

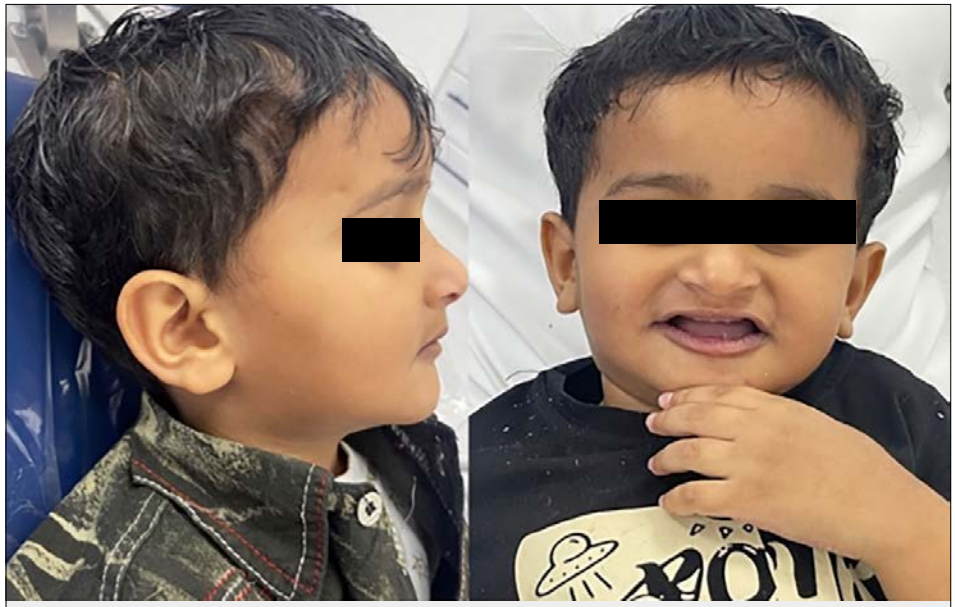

Figure 1. Showing lateral and frontal views of the patient's face reveals a normal face symmetry with loss of vertical facial height and edentulous appearance due to early loss of teeth

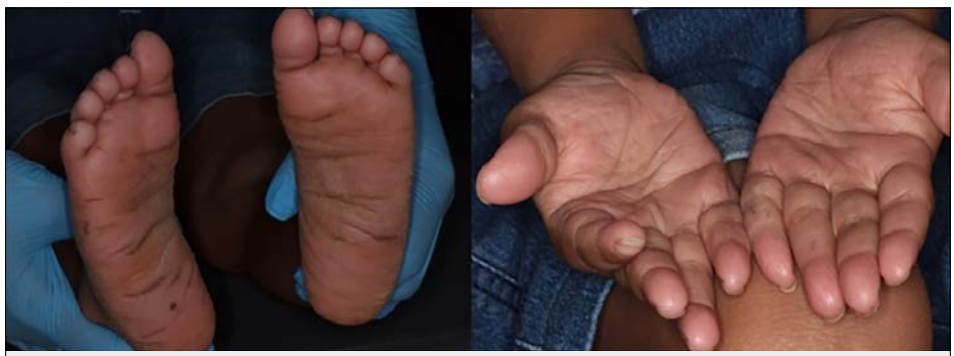

Figure 2. Physical examination of hands and feet revealed palmoplantar keratosis present as rough, scaly, symmetric, well-demarcated plaque with eczematous changes in both hands and feet

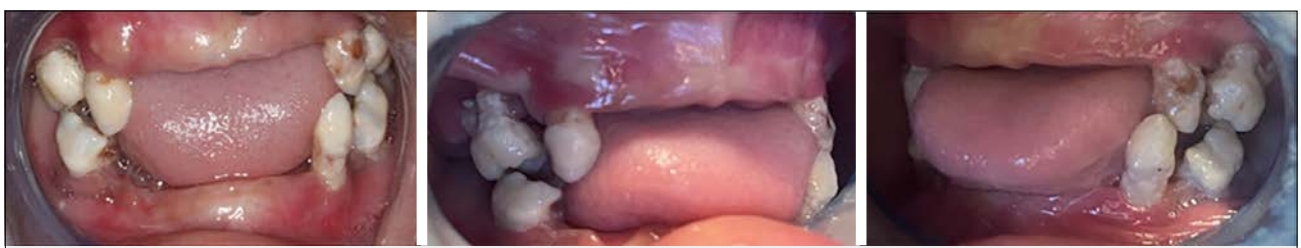

Figure 3. Intra-oral exam revealed presence of several mobile primary teeth. Severe gingival inflammation with evidence of thick periodontal plaque and gingival recession associated with present teeth. Generalized alveolar bone resorption was also evident

ing due to its poor prognosis and unpredictable disease course. A multidisciplinary team of healthcare professionals. including a pediatric dentist, a prosthodontist, a periodontist, a dermatologist, a pediatrician, and a psychologist collaborated to provide comprehensive care for the patient. The patient was referred to a pediatric clinic for medical attention and to a dermatology clinic for skin treatment. Dermatologist prescribed several topical medications to manage symptoms, enhance skin function, and prevent complications from arising.

Collaboration among a pediatric dentist, a periodontist, and a prosthodontist leads to a comprehensive full-mouth rehabilitation that aims to improve the physical

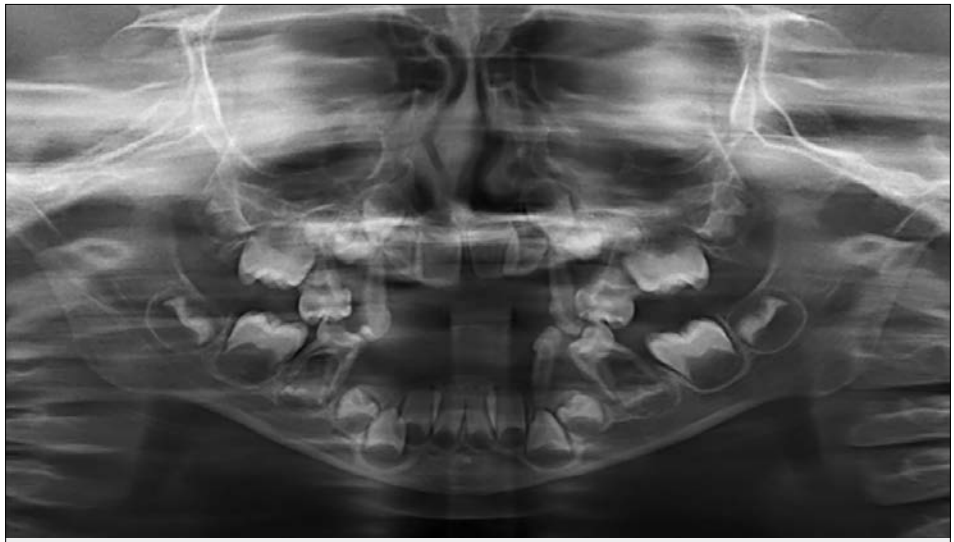

Figure 4. Orthopantomograph showed several missing teeth with generalized alveolar bone resorption; teeth seemed to be ''floating in air''. Permanent teeth buds evident in both arches

and mental well-being of the patient. A treatment plan was discussed with and agreed upon by the parent, which included extraction of the remaining teeth due to the risk of inhalation, and to alleviate the child's discomfort. Maxillary and mandibular complete dentures were fabricated for the patient as part of the rehabilitation plan. Fabricating dentures for a child with Papillon-Lefèvre syndrome presents several challenges due to the specific characteristics associated with this condition. These challenges include significant bone resorption in the jaws, which makes it difficult to ensure proper denture stability and fit. Additionally, limited cooperation from young children can make it challenging to sit still for the necessary impressions and fittings, which are crucial for accurate denture fabrication. Children's gums and oral tissues may also be more sensitive than those of adults, making it necessary to ensure that the dentures fit comfortably without causing discomfort. Furthermore, children's jaws are still developing, and this growth factor needs to be considered during the fabrication process to ensure that the dentures will accommodate future changes. In all processes of denture fabrication, psychological support and patience from dental professionals are always needed.

The denture process involved multiple steps. First, primary impressions were taken using stock trays with silicon putty impression material followed by final impressions using special acrylic trays with fast-set silicon impression material (Fig. 5a, b). The third step involved wax try-in and bite registration to achieve an accurate relationship between the maxillary and mandibular jaws which is crucial for the denture functionality. Wax try-in and bite registration technique were achieved through following steps 1) wax rim construction on record bases created from final impression. 2) Trimming and fitting the wax rim to the approximate bite height of the child, ensuring they were low enough to allow comfortable closure without strain. 3) Vertical dimension and retention check with the wax occlusion rim in place. For the upper occlusal rim, we evaluated its position relative to the upper lip and the level of occlusal plane in both the anterior and posterior regions. For lower occlusal rim we examined its position in relation to the corners of the mouth anteriorly, the retromolar pads posteriorly and the lateral border of the tongue to ensure the proper height of the occlusal rim. Later we asked the patient to smile to check the visibility of the upper wax rim in relation to the upper lip and occlusal plane from anterior to posterior. Pronouncing the letter 'S' was used to confirm the adequate speaking space. 4) Guided bite registration in centric relation using verbal guidance and gentle support, we helped the child to close comfortably in centric relation, continually monitoring for facial muscle relaxation to confirm the correct vertical dimension. We took care to avoid excessive pressure to ensure accuracy and comfort. 5) Final bite registration after confirming satisfaction with the vertical dimension, we applied softened wax to the occlusal rims to capture the bite in stable centric relation. To minimize discomfort or movement, we recorded the bite as promptly as possible. 6) Verification and support; we removed the wax rims and checked the occlusal registration for accuracy and stability. A dental assistant and the child's father were present to help keep

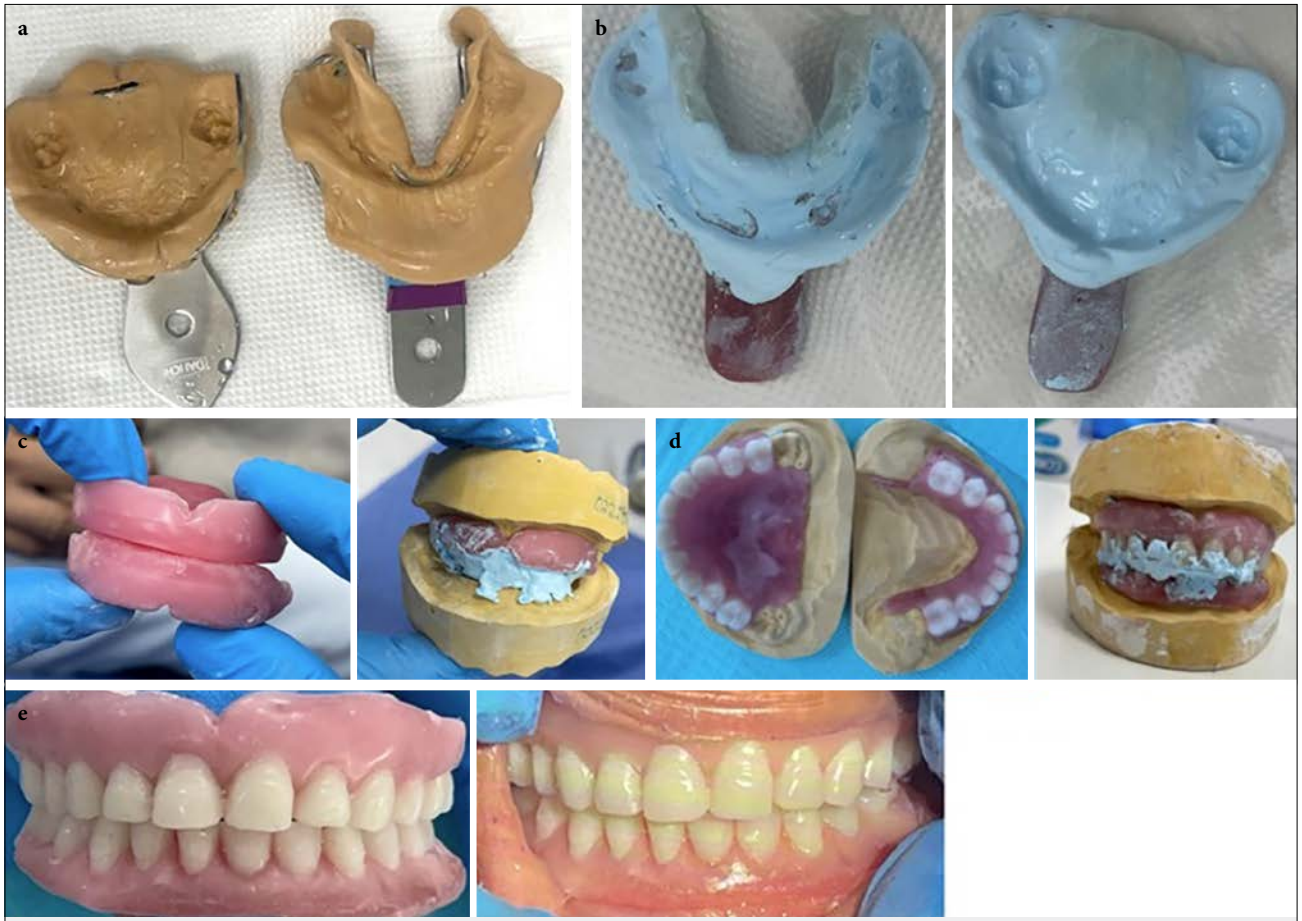

Figure 5. (a) Primary alginate impressions. (b) Final impressions. (c) Wax try-in and bite registration. (d) Dentures try in and bite registration. (e) Delivery of the dentures

the child calm and distracted during the procedure (Fig. 5c). The fourth step involved denture try-in and bite registration. This step checked the vertical dimension, centric relation, and aesthetics. Adhesive glue was used to stabilize the acrylic base and ensure its stability and retention. After the dentures were fitted, adjustments were made to ensure proper fit, comfort, and functionality. At this stage, the patient and her parents were very happy (Fig. 5d). Finally, the upper and lower dentures were delivered, and parents were educated on proper denture care and hygiene. Regular follow-up visits are necessary to monitor the dentures' fit and address any issues that may arise (Fig. 5e). The installation of complete dentures improved the patient's facial appearance and functionality, as shown in (Fig. 6).

Discussion

In this case report, a 4-year-old boy displayed classic features of PLS, which were sufficient for a clinical diagnosis of this syndrome. These included palmoplantar hyperkeratosis and severe aggressive periodontitis which resulted in the premature loss of deciduous teeth. [1,14] Consanguinity was observed between his parents. According to the previous literature, approximately one-third of the cases are associated with consanguineous marriage.[1,6] The majority of the literature revealed that mutation in the CTSC gene which one of the factors associated with PLS. The exact etiologies and pathogenesis of PLS are still controversial and difficult to understand.[4] Clinically, the cutaneous examination of this patient revealed symmetrical, well-demarcated, rough, scaly accentuation with eczematous changes affecting both the palmar surface of his hands and the plantar surface of his feet. Intraorally, the findings were remarkable with characteristic features of aggressive periodontitis. The patient had multiple remaining deciduous teeth with grade III mobility. The gingiva surrounding these teeth exhibited all signs and symptoms of gingiva inflammation with evidence of generalized alveolar bone resorption. Additional characteristics included the presence of gingival abscesses, purulent exudate, and offensive halitosis.[15] Radiographically, the patient showed severe generalized alveolar bone resorp-

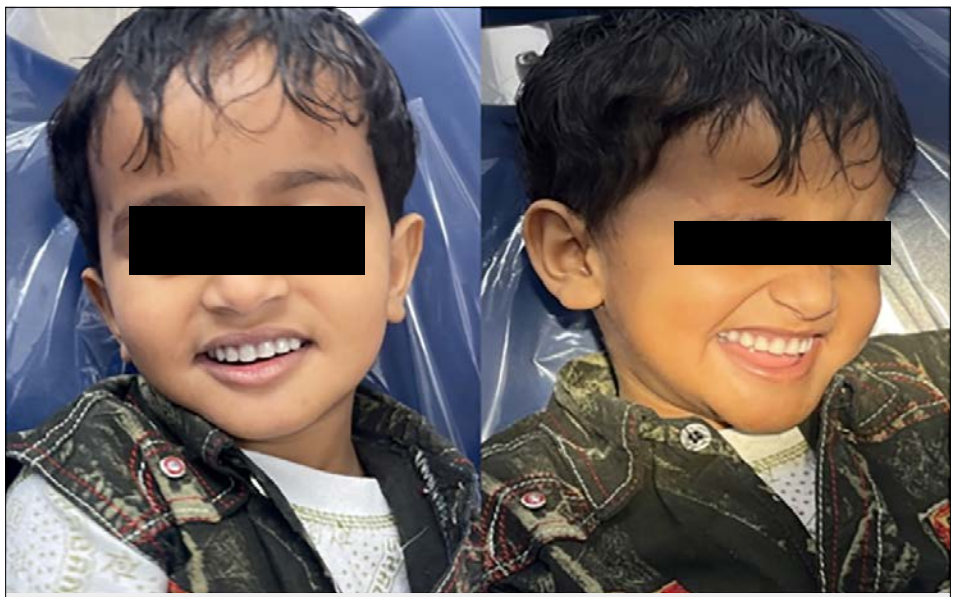

Figure 6. Improvement of patient facial height after installation of the dentures

tion with a floating-in-the-air appearance of the present teeth with evidence of a permanent tooth bud in the alveolar bone of both arches. All these cutaneous and dental findings are consistent with the classic characteristic features of PLS. The prognosis of this disease is very poor, and the course of the treatment is unpredictable.[1] A multidisciplinary team involving dermatologists, pediatricians, and pediatric dentists is essential for the comprehensive treatment of patients with PLS.[16] In our case, a periodontist, prosthodontist, and psychologist were included with the team for further support. In previous case reports of clinical dental management were limited to mixed and permanent dentition, and only a few reports have described the management of primary dentition in younger children aged 3–4 years.[6,17] In general, the early management of this syndrome involve non-surgical and conventional periodontal treatment in the form of scaling and root planning in addition to antibiotic coverage.[15] In this case, full dental rehabilitation including the extraction of the remaining teeth and the fabrication of complete dentures, was deemed the best treatment option. The patient had concerns about aesthetics and function and the presented teeth were considered hopeless. Fabricating the prosthesis was particularly challenging due to the child's limited cooperation. However, early diagnosis and prompt management of the patient were highly beneficial. Both aesthetics and function were significantly improved, and the child's cooperation with dental treatment gradually improved. Periodic recalls may be necessary for prosthesis modification or replacement due to ongoing growth and development.[18]

Conclusion

Consanguinity in families is considered a key etiological factor of PLS, especially in Arab populations. Early detection of this syndrome is very important for improving a person's general well-being, as a multidisciplinary team of professionals works together to provide the best possible care to PLS patients.

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