Papillon Lefevre Syndrome: A case series with review of literature

Kundoor V.K. Reddy[1], Kotya N. Maloth[2,*], Nayanala V. Anusha[3], Venkata S.R Thummala[4], Moni Thakur[5]

[1]Professor & Head, [2,5]Senior Lecturer, [3]PG Student, [4]Associate Professor, Dept. of Oral Medicine & Radiology, Mamata Dental College & Hospital, Khammam, Telangana

***Corresponding Author:**
Email: dr.kotyanaik.maloth@gmail.com

Abstract

Papillon Lefevre syndrome (PLS) is a rare autosomal recessive inherited genodermal disorder, caused by cathepsin C gene mutation leading to the deficiency of cathepsin C enzymatic activity and consanguinity of parents is evident in about one third of cases. The disorder is characterized by palmoplantar hyperkeratosis and periodontitis that results in premature loss of deciduous and permanent teeth. Here we report a case series of PLS with typical clinical and radiographic features.

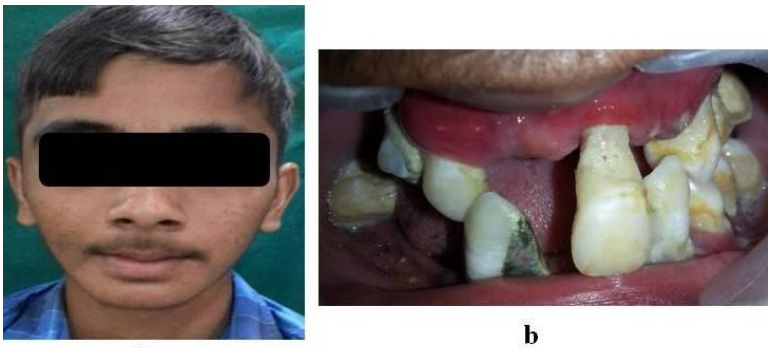
Keywords: Cathepsin C Gene, Genodermal Disorder, Palmoplantar Hyperkeratosis, Periodontitis.

Introduction

Papillon Lefevre syndrome (PLS) is a rare autosomal recessive genodermal disorder as an trait, characterized by development of dry scaly patches on cutaneous part of the palms and soles of the feet (palmo-plantar hyperkeratosis) associated with severe loss of periodontal support as well alveolar bone destruction resulting in periodontitis and premature loss of both the dentition.(1) This rare condition was first described by two French physicians, Papillon and Lefevre in 1924, thus it came to be known as Papillon Lefevre syndrome or Palmo-Plantar Keratoderma.(1,2) It affects 1 to 4 persons per million and carrier frequency results 2 to 4 persons per thousand with no sex and racial predilections.(3) Here we report a case series of PLS with typical clinical and radiographic features.

Case Report 1

A 15 year old boy came to the department with a chief complaint of multiple loosening of teeth (Fig. 1a). The patient revealed that eruption of primary teeth started at 7-8 months of age, and were gradually lost by five years of age. Patient was not sure about the time of eruption of permanent teeth, but described gingival bleeding during brushing. There was gradual loosening of permanent teeth from l0 years of age and eventually many teeth were lost by 15 years of age.



a b

Fig. 1: profile picture (a), and periodontitis with multiple missing teeth (b)

Medical history was noncontributory and the family history revealed consanguineous marriage of his parents, but patient's siblings were healthy. On general physical examination the patient was moderately built with a steady gait and his physical and mental development was normal. Cutaneous examination revealed symmetric, keratotic and well-demarcated psoriasiform plaques affecting the skin on the dorsal and ventral surfaces of soles and palms (Figure 2a, 2b), right and left elbow (Fig. 2c), right and left knees (Fig. 2d), with dry, scaly skin rough on palpation. Intra oral examination revealed multiple missing teeth along with reddish pink, soft and edematous gingiva and generalized mobility of teeth (Grade-III in relation to 15, 13, 21, 26, 36, 35, 34, 33, 46; Grdae-II irt 24,25,43,47 and Grade-I irt 37) (Fig. 1b). Panoramic view showed generalized extensive loss of alveolar bone, with multiple missing teeth (Fig. 3).

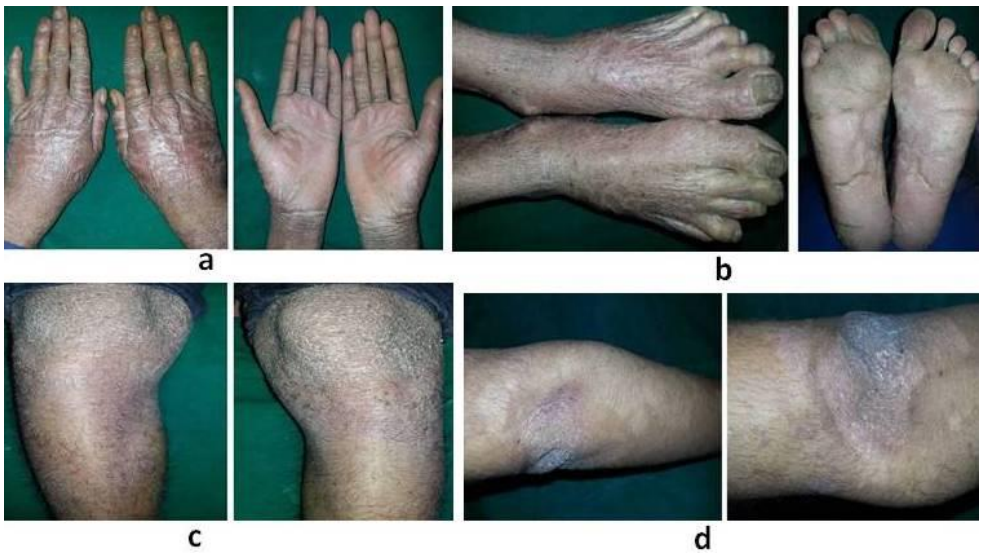


Fig. 2: keratotic and well definedplaques on the dorsal surface of hands and ventral surface (a); keratotic and well defined psoriasiform plaques on the dorsal surface of feet and ventral surface (b); keratotic and well defined psoriasiform plaques on the right knee and left knee (c); keratotic and well defined psoriasiform plaques on the right hand and left elbow (d)

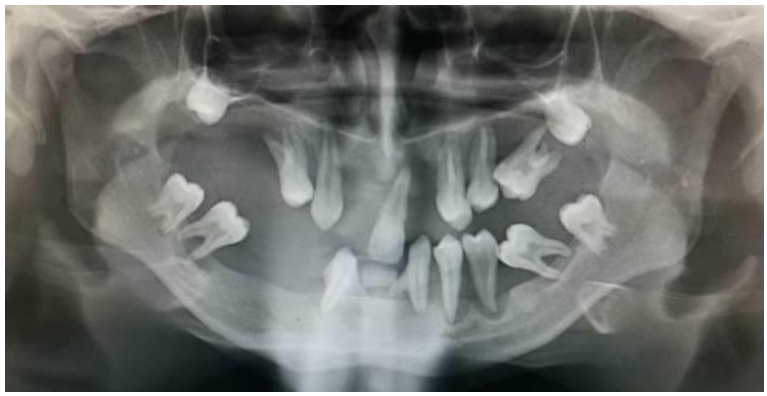


Fig. 3: OPG shows extensive loss of alveolar bone with generalized periodontitis and multiple missing teeth in the maxillary and mandibular arches

Case Report 2

A 22 year old male patient visited to the department with a complaint of missing teeth in the upper front tooth region since 2 years. Past history revealed frequent infections of the gums resulting in bad breath and loss of teeth. Medical history revealed frequent skin infections on the hands and legs. The family history was noncontributory. Cutaneous examination revealed, a well-defined hyperkeratosis affecting the cutaneous part of dorsal and ventral surfaces of hands and feet in a symmetric pattern (Fig. 4b, 4c). On intraoral examination revealed missing teeth in relation to 16, 11, 21, 22, 31, 32, 41, 46 with gingival inflammation resulting in periodontitis (Fig. 4a).

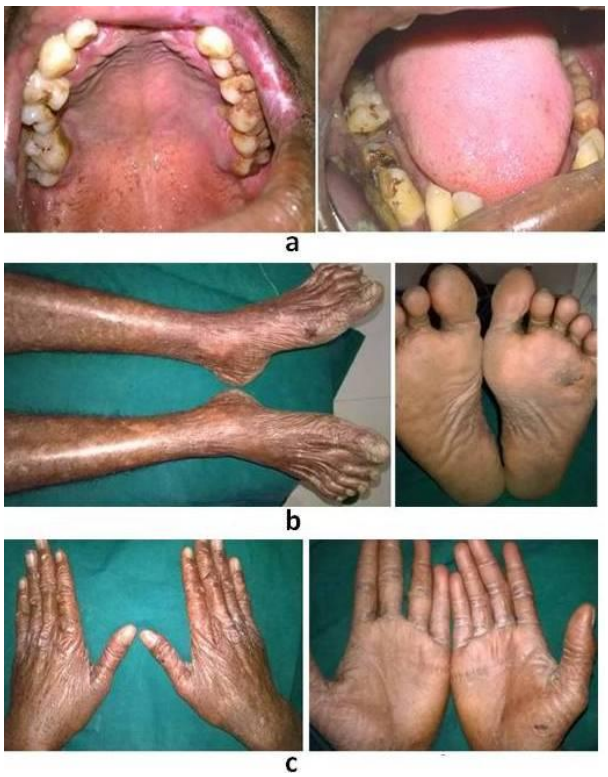


Fig. 4: Missing maxillary anterior and periodontitis (a); keratotic plaques on the dorsal surface of legs and ventral surface of the feet (b); keratotic plaques on the dorsal surface of hands and palms (c)

Case Report 3

A 23 year old female patient reported to the department with a chief complaint of loss of multiple teeth (Fig. 5a). Past history revealed that she had normal eruption of primary teeth, she also gave history of recurrent infections of skin and gums resulting in foul breath and loss of teeth. Medical history was noncontributory. The family history revealed that there was no consanguineous marriage. Cutaneous examination revealed a well-defined hyperkeratosis affecting the cutaneous part of palms and soles extending to the dorsal surface of hands and feet in a symmetric pattern (Fig. 5b), well-defined psoriasiform plaques were seen bilaterally on hands and feet. Examination of the oral cavity revealed complete missing of maxillary and mandibular teeth (Fig. 5a). Panoramic view showed complete loss of maxillary and mandibular teeth associated with alveolar ridge resorption (Fig. 5c).

Based on the clinical and radiographical findings a provisional diagnosis of Papillon-Lefevre syndrome was established in these patients.

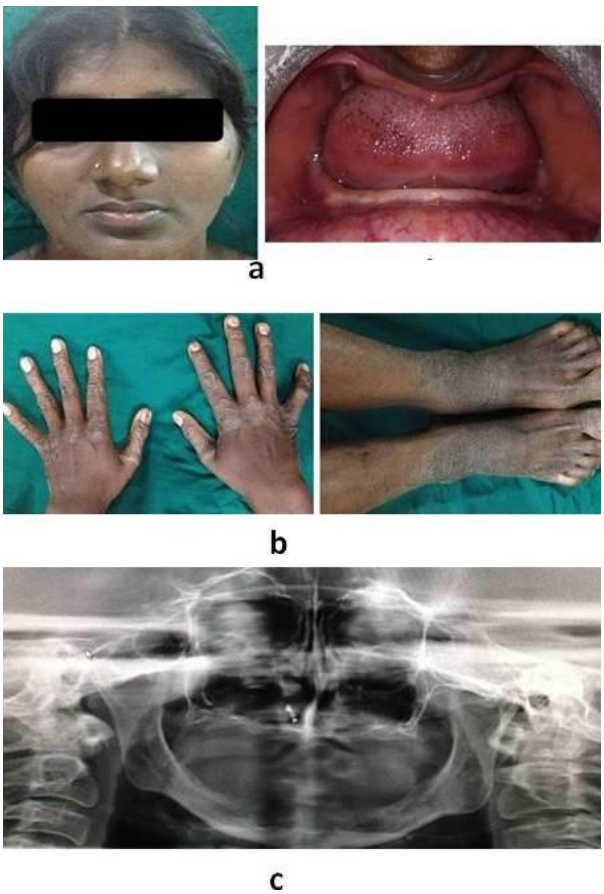


Fig. 5: profile picture and completely edentulous maxillary and mandibular arches (a); keratotic and well defined psoriasiform plaques on the dorsal surface of hands and dorsal surface of legs (b); OPG shows completely edentulous maxillary and mandibular arches (c)

Discussion

PLS is a rare genetically inherited genodermal disorder affecting 1 to 4 cases per million population, males and females are equally affected without any racial predominance.[4] The exact etiology of PLS is unknown remains relatively obscure, Hattab et al[5] classified the etiology into three types: microbiologic, immunologic and genetic factors. The immunological aspect includes an impaired chemotaxis of neutrophil, phagocytosis and impaired migration cell. The immune mediated mechanism such as lymphocyte response to pathogens helper/ suppressor T-cells ratio and monocyte function is also impaired. Microbiologically, the main trigger factors resulting in periodontitis are the presence of virulent gram negative anaerobic pathogens like Actinobacillus actinomycetemcomitans, Porphyromonas gingivalis.[6] Genetically it has been suggested that mutation in cathepsin C gene located on chromosome 11q14.1-q21 results in inactivation of the cathepsin C gene and is responsible for the abnormalities in skin and periodontal disease progression.[1,4] The etiology is presumed to be microbiological in case 1, 2

and 3 and genetical in case 1. Genetic testing could not be done in our cases because of their low socio-economic status. In case-1 patient reported consanguinity of parents, this supports genetical cause for this case.

PLS manifested as a well-defined hyperkeratosis, affecting on the dorsal and ventral surface of the hands and feet, which is seen during first 4 years of life. Erythematous to psoriasiform hyperkeratotic plaques are present on the knees, elbows and trunk. The severe periodontitis affecting both the dentition starting at the age of 3 or 4 years is considered as second major feature of PLS. The teeth erupt normally but by the age of 14 years, patients are usually edentulous.[5] The present cases are consistent with the literature.

Radiographic features show 'floating-in-air' appearance of teeth due to extensive resorption of alveolar bone as seen in case-1 and completely edentulous as seen in case-3. Other radiographic features show calcification of choroid plexus intracranially and tentorium and palmoplantar hyperhidrosis,[4,7] which were not evident in our cases.

The differential diagnoses of PLS are Haims-Munk Syndrome[8] and prepubertal periodontitis.[9,10] Haims-Munk syndrome is an allelic variant of PLS which includes additional clinical features of arachnodactyly(claw like phalanges with convex nails), and acroosteolysis.[8] In prepubertal periodontitis palmoplantar hyperkeratosis is absent.[9]

According to recent studies PLS syndrome can be manageable and the permanent teeth can be saved.[6] The guidelines put forward by Mc Donald et al. for the successful management of PLS cases, includes: i) extraction of all primary teeth, ii) construction of complete dentures three months after the removal of primary teeth, iii) prophylactic doses of tetracycline for 10 days immediately after the denture insertion, iv) adjustment of denture bases to allow for the emergence of the permanent dentition followed by another therapeutic dose of the tetracycline administered at a dosage of 250 mg four times daily for one month.[9] Medications such as retinoid (acitretin, etretinate and isotretinion) combined with higher antibiotics are used to prevent and control microbial attack and before the periodontitis exceeds the clinical therapeutic limits proper steps to be taken to conserve the periodontium and it should always be considered as first priority.[11]

Conclusion

PLS presents with diversified oral and dermatological manifestations, which an oral physician should be aware of for early diagnosis and successful management to prevent the loss of dentition by providing proper periodontal therapy and antibiotics.

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