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Role of Oral Physician in Early Diagnosis of Papillon Lefevre Syndrome – A rare case report

[1]Dr. Hemcle Shalma G, MDS, Senior Lecturer, Department of Oral Medicine and Radiology, Sri Ramakrishna Dental College and Hospital, Coimbatore, Tamil Nadu.

[2]Dr. Akshita A, CRI, Sri Ramakrishna Dental College and Hospital, Coimbatore, Tamil Nadu.

[3]Dr. Sidra Bano, MDS, Reader and Head of the Department, Department of Oral Medicine and Radiology, Sri Ramakrishna Dental College and Hospital, Coimbatore, Tamil Nadu.

[4]Dr. A. Arun Prasath, Senior Lecturer, Department of Oral Medicine and Radiology, Sri Ramakrishna Dental College and Hospital, Coimbatore, Tamil Nadu.

Corresponding Author: Dr. Hemcle Shalma G, MDS, Senior Lecturer, Department of Oral Medicine and Radiology Sri Ramakrishna Dental College and Hospital, Coimbatore, Tamil Nadu.

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Abstract

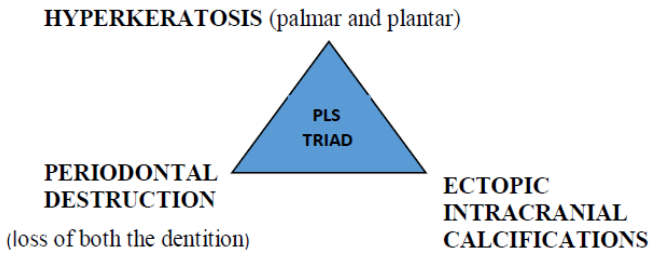
The Papillon Lefevre Syndrome (PLS) is an atypical form of Genodermatosis, which is of autosomal recessive inheritance with a remarkable feature of diffuse palmo-plantar hyperkeratosis and juvenile periodontitis. This disease is rare with an equal gender predilection , by affecting both boys and girls who are at the age between 6months to 4years with no racial predominance. Its prevalence is one to four in a million population. The etiopathogenesis of the disorder is multifactorial with genetic , immunological and microbial factor playing a major role. Other significant factors involved are environmental and decreased host defense mechanism. Mutation of the gene 11q14-q21 encoding for cathepsin-C,an enzyme involved in case of inflammatory and immune responses. The individual may experience an early shedding of primary and permanent teeth with periodontal destruction. Early diagnosis is indispensable to prevent the edentulism at the young age and to avoid the upcoming obscure pathologies. This article will enlighten, about a clinical case report of an 18-year-old female patient presented with aggressive periodontitis and palmoplantar hyperkeratosis. It provides a detailed audit about various possible etiology, clinical features, differential diagnosis and treatment protocols of PLS.

Keywords: PLS, PPK, CTSC

Introduction

Papillon Lefevre Syndrome (PLS) otherwise known as keratoris palmoplantar is (PPK) with periodontopathic,

is an atypical form of Geno keratosis and is auto so mally recessive [1].It was first described by two French physicians named , Papillon and Lafe / vre in the year 1924[2].PLS usually has an onset between 6months to 4years of age. It's prevalence is of 1-4 cases/million population and till date only about 300 cases have been reported with a higher racial predilection in the Arabs, Indians and Africans. There has always been an quandary in the etiology, but it is believed that this may be due to CTSC mutations and Vitamin A deficiency [2,5]. Recent evidences prove that these populations maybe be affected due to parental consanguinity, which is common in the above geographic regions. This syndrome is usually characterized by the development of well-demarcated dry scaly patches in palms and soles along with erythema , and with precocious juvenile periodontitis which may lead to early exfoliation of the primary and permanent teeth along with halitosis and plaque accumulation in the deep crevices due to infection caused by gram negative anaerobic rods(A.actinomycetemcomitans).The syndrome may also involve pyogenic abscess, nail dystrophy(transverse grooving) , hyperhidrosis and floating in air appearance of teeth[4].About 20% of the individuals have suffered from dural calcifications. Radiographic features depicts the generalized alveolar bone loss. Histologically, hyperkeratosis, occasional patches of parakeratosis, acanthosis and slight perivascular inflammatory infiltration are detected [4,3,5]. The early slump of the dermatological and dental vigour may stir up the lifelong functional, aesthetic, social and psychological impacts on the growing child. Therefore, early treatment may lead to the preservation of the health by elimination of the causative factor and may provide a lighting future to the affected child [6].



Flowchart 1

Case report

An 18year-old female patient came to the Department of Oral Medicine and Radiology with the chief complaint of fractured prosthesis. On eliciting the history, patient revealed that there was normal emergence of the deciduous teeth at 7months of age, but it started to slack at 3years and were eventually lost by 5years of age. Patient was not sure about the time of eruption of the permanent teeth, but described about the gingival bleeding during brushing and eating after the eruption of permanent teeth. There was loosening of the permanent teeth from 10years of age, where eventually most of the teeth were lost by the age of 15. The mother noticed the skin lesions on the palms and soles of the child.

On probing the history deeper, patient disclosed that she had undergone extraction of the mobile deciduous teeth 13 years back and the extraction of mobile permanent teeth before 3 years. She had also undergone prosthetic rehabilitation (Removable partial denture for upper arch and Fixed partial denture for lower arch), since 3years.Patient had skin lesions on hands and feet and was under medication. Patient mentioned that the early loss of teeth made it very difficult for her to chew food and led to loss of appetite. The family history reveals the consanguineous marriage of the parents.

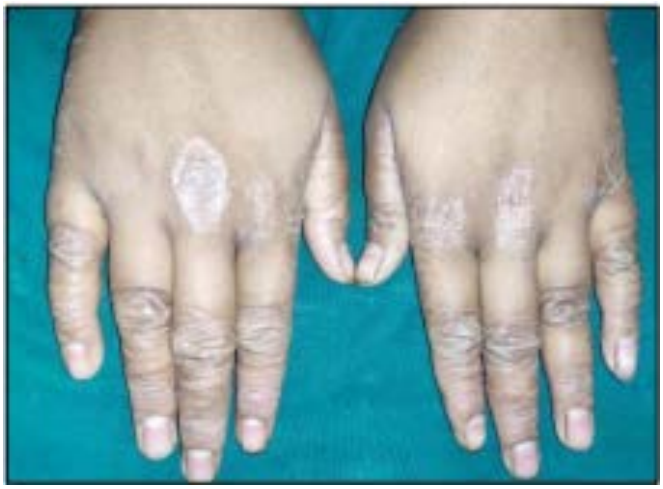
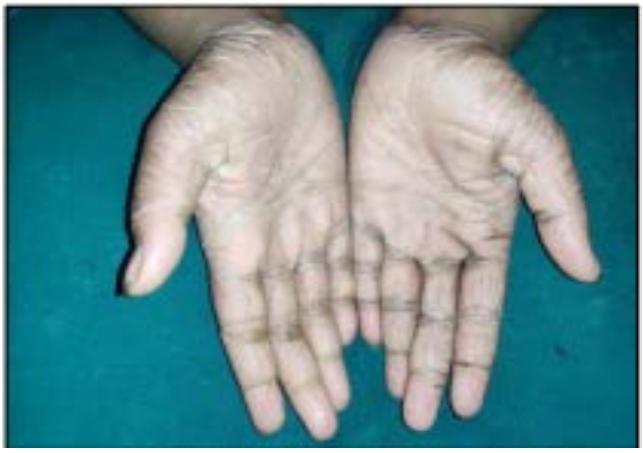


Figure:1



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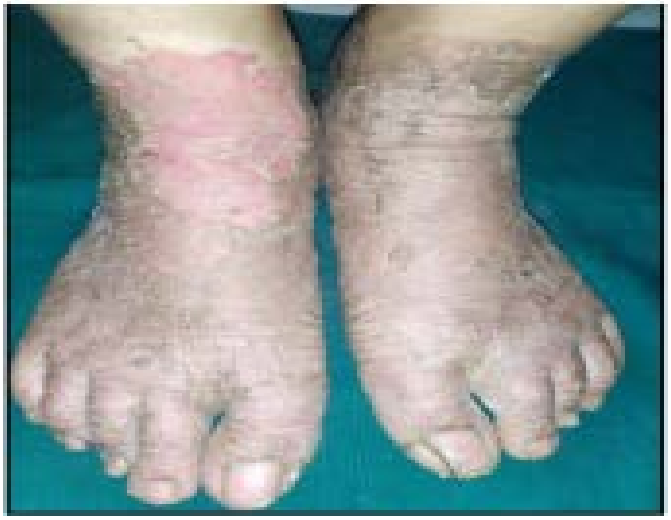

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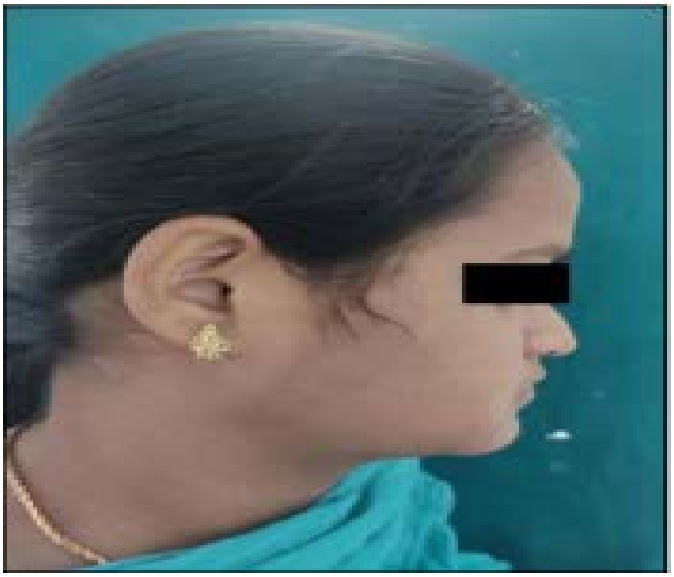

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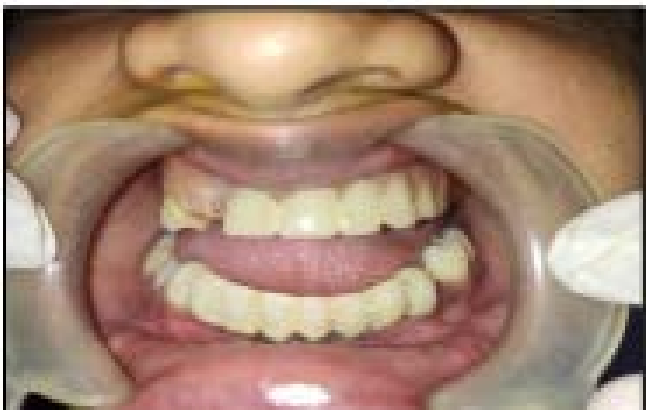

Figure: 6

On Clinical examination, the patient seemed to be of normal built, moderately nourished, with normal vitals.

On clinical examination she was broached with increased keratinization of the skin of the palmar and plantar surfaces as well as the skin overlying the dorsal surfaces of the joints of hands with multiple fissured, erythematous and hyperkeratotic plaques on the foot and sole[fig.1,2,3].On Extraoral examination patient appeared to have reduced facial height due to resorption of the alveolar ridge[fig.4,5].Intraorally, the patient presented only with the following teeth 17,16,23,27,33,34,36,37,43,44,46,47 ,which were mobile and drifted ,rest of the teeth were missing. Mild deposits were noted. RPD in relation to 11,12,13,14,15,21,22,24,25,26 and FPD in relation to 31,32,41,42 [fig6]. The gingiva appeared to be pink and pigmented which was firm and resilient. The tooth 23 appeared to have a periodontal pocket of 5mm depth. The provisional diagnosis was PLS was rendered with a differential diagnosis of HAIM-MUNK syndrome and Prepubertal periodontitis.

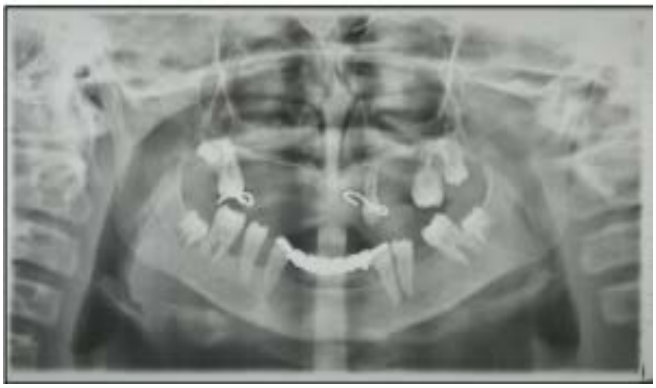

Figure: 7

The provisional diagnosis was confirmed by running few investigations. Radiographic, Biochemical investigations and Genetic mapping were requested to confirm the syndrome. The outcome of the biochemical investigations were normal. [Complete blood count (CBC), Liver Function Tests (LFT), urine analysis and alkaline phosphatase levels]. Onperpetuatingthe panoramic radiograph presently [at 18years of age] it was perceived as generalized alveolar bone loss and migration of all the teeth[fig.7]. Patient had “FLOATING TEETH APPEARANCE” when she was 13years old. Result of the genetic mapping revealed pathogenic homozygous mutations in exon6 of CTSC gene. Based on the history, clinical findings and Investigations, it was authenticated that the patient is under the tribulation of PAPILLON LEFEVRE SYNDROME. In our scenario, we have referred the patient to a dermatologist to hamper the skin lesions and the dental treatment provided to the patient is total extraction of the remaining permanent teeth and rehabilitation with complete denture [fig.8] and later according to patient’s need and affordability implants-supported overdentures would be planned.

Figure: 8

Discussion

This paper throws a light on PLS which has been inherited as an autosomal recessive disorder. Haneke et, al., considered three criteria to detect PLS PPK, Autosomal Recessive Inheritance, Loss of Deciduous and Permanent Dentition. The main cardinal features of

PLS is aggressive periodontitis with premature and extensive loss of teeth and alveolar bone in young individuals and psoriasis in palm and soles were seen discerned in this case.[11]

The etiopathogenesis of the syndrome is multifactorial with immunogenic, genetic or bacterial etiologies being the main factors[1,2,4].Other consequential factors are environmental and compromised host defense mechanism owing to decreased function of lymphocytes, PMNs , or monocytes[2,10,11].There is 25% chance for offspring to get affected from phenotypically healthy consanguineous parents who carry an autosomal gene[1].Until today nearly one-third of the PLS cases were due to the above etiology. Laa et al were the 1st that mapped PLS on chromosome 11q14-q21.CathepsinC(CTSC) and lysosomal cystein protease involves in wide variety of immune and inflammatory response by activating serine protease, an enzyme implicating in activation of phagozytic cell sand t-lymphocytes. The mutation of the cathepsin C gene is usually expressed in soles and knees and in keratinized oral gingiva and also in high levels of PMN and macrophages [1,2,4,9,11]. The complete absence of cathepsin C is responsible for the occurrence of PLS. It results in the activation of Serine protease and suppresses the immune system. A range of functional neutrophil defects in PLS, arising secondary to CTSC gene has been demonstrated. Some studies prove that the periodontal destruction may occur due to the alteration in the saliva i.e., because of the destruction of neutrophils dysfunction. Albander et.al., examined the subgingival plaque from 13 PLS patients and found a total of 170 bacterial species had a high frequency of Actinobacillus actinomycetemcomitans (Aa), Campylobacter, Capnocytophagegranulosa, Streptococcu s and Trannerella forsythia. Aa [7]. Plays a significant role in initiation and progression of periodontitis seen in PLS. Hence, in this syndrome the immune, genetic and microbial factors are correlated.

Dermatological manifestations involve sharp, demarcated, well defined keratotic plaques, which was dry, scaly and erythematous bilaterally involving the palmar and plantar regions. Cutaneous lesions develop in pressure areas such as palm, soles, knuckles, ankle, elbows and knees. Intraorally, it clearly establish the features of aggressive periodontitis with innumerable missing teeth and the other remaining teeth were mobile and appeared drifted with periodontal pocket with respect to 23.

Certain PLS patients may also exhibit intracranial calcifications (mainly, Choroid plexus and tentorium), hyperhidrosis, gingival inflammation, gingival bleeding, tenderness, gingival abscess, purulent exudates, susceptible to infections and mental retardation [12]. But, none of these findings were contemplated in our case. The differential diagnosis that provides a contrary to this syndrome is [5],

Table 1

Haim munk syndrome	Here, arachnodactyly, Acro osteolysis, onychogr yosis, recurrent abscess, ple-planus, will be seen.
Pre-pubertal periodontits	No PPK, not all cases are associated with CTSC gene mutation. Generalized or localized periodontitis- rapid destruction.
Acrodynia	Dusky pink discolouration in the hands and feet, loss of teeth, hair and nails, red lips cheeks and nose. Transientrashes,

	photophobia, peripheral neuropathy and neuropsychiatric symptoms.
Hypophosphatasia	Knock knees, bowing of femur and tibia, enlarged wrists, hypoplastic teeth, an increased amounts of phosphoethanolamine in urine.
Cyclic neutropenia	No PPK, periodontal destruction.
Takahara syndrome	Progressive gangrenous lesion in gingiva and alveolar bone which leads to the exfoliation of the teeth

Hence, in order to overcome the sceptical between provisional diagnosis and differential diagnosis, the investigations are done.

Although the laboratory test in patients with this syndrome are all essentially within normal limits, they are particularly valuable in distinguishing this syndrome from other conditions causing gingival inflammation and early loss of deciduous and loss of permanent teeth and PPK. An important multidisciplinary approach for case of the patient with PLS. The skin manifestations of PPK are beaten with emollients. The periodontitis associated with PLS is usually difficult periodontitis associated with PLS is usually difficult to control. Effective treatment includes extraction of mobile primary teeth which eliminates periodontal pathogens and improves future prognosis of the permanent teeth. Extractions must perform under antibiotic coverage. Prosthetic replacement in such patients is an age specific specially treatment involving initial replacement with complete or partial denture and future considerations for an implant supported prosthesis. This case had been followed up for past 14years and appropriate prevention and treatment modalities have been carried out until now.

Conclusion:

PLS is a rare Geno dermal condition which is severely associated with early-onset of periodontitis along with premature loss of both dentition and PPK. Usually, genetic mapping is a definitive investigation to confirm the diagnosis. In dental aspect, comprehensive treatment can be provided at the early stages of detection. The periodontal destruction can be stumbled by elimination of the reservoir of the causative agents and by providing appropriate treatments like oral prophylaxis, oral hygiene instructions, antibiotic therapy. A multidisciplinary approach is done with a correlation of dentists, dermatologist and pediatrician to provide an overall care to the PLS patients in order to avoid the psychological and social trauma to the patient.

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