Papillon Lefèvre Syndrome

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Abstract
Papillon-Lefèvre syndrome is a rare disease characterized by skin lesions caused by palmar-plantar hyperkeratosis, and severe periodontal destruction involving both the primary and permanent dentitions. It is transmitted as an autosomal recessive condition and consanguinity of parents is evident in about one-third of cases. Pyogenic liver abscess is an increasingly recognized complication. We report a new case of this association and review the current literature.

INTRODUCTION
Papillon-Lefèvre syndrome (PLS) is a rare, autosomal recessive disorder. The syndrome is believed to affect 1 to 4 persons per million. Till date more than 200 cases have been reported worldwide. PLS is characterized by the development of dry scaly patches on the skin of the palms and the soles of feet (palmar-plantar hyperkeratosis) in association with severe inflammation and degeneration of the structures surrounding and supporting the teeth (periodontium). The primary (deciduous) teeth frequently become loose and fall out. Without treatment, most of the secondary (permanent) teeth may also be lost. Additional symptoms and findings may include frequent pyogenic skin infections, nail dystrophy, and hyperhydrosis. Patients typically have an underlying disease associated with functional or quantitative neutrophil abnormalities, and 50% are immunocompromised. Pyogenic liver abscess is an uncommon presentation. Patients with PLS seem to be particularly predisposed to develop pyogenic liver abscess.

CASE REPORT
An eighteen-year-old boy presented with history of right sided chest pain, breathlessness and dry cough of one-week duration. He gave history of recurrent skin infections and two admissions for liver abscess. On examination vitals were normal. General examination revealed thick, dry scaly, shiny patches of skin on his palms and soles (Fig. 1), present since his childhood, and loss of teeth (Fig. 2) except one or two molar teeth. He informed that he had malformed teeth since childhood which fell off one by one. Chest examination revealed signs suggestive of right sided pneumothorax. On investigations, chest radiograph revealed loculated pneumothorax. Complete blood count, blood chemistry
profile, and liver function tests were within normal limits. Immunologic studies revealed low (CD₃⁺ CD₄⁺) count. A diagnosis of Papillon-Lefèvre syndrome (PLS) was made. Patient was treated with antibiotics (ceftriaxone 1gm intravenously 12hrly for 5 days and gentamycin 60mg intravenously 12hrly for 5 days) and retinoids. Keratolytic preparations containing 20% salicylic acid were prescribed for the skin lesions, and a dental opinion was taken. His parents were consanguineous. His brother and two cousins had similar skin lesions and were edentulous.

**DISCUSSION**

Papillon Lefèvre syndrome was first described by Papillon and Lefèvre in 1924. The disease is characterized by diffuse palmoplantar hyperkeratosis and juvenile periodontitis.¹³

PPK usually manifests during the first 4 years of life with sharply demarcated hyperkeratosis, more pronounced on the soles of feet and possibly extending to the dorsa of the hands and feet. Erythematous hyperkeratotic plaques may also be present at the elbows, knees, and trunk. The second major feature of PLS is severe periodontitis, which starts at the age 3 or 4 years and affects both the deciduous and permanent teeth. The teeth erupt normally but are soon lost, and by the age of 14 years, patients are usually edentulous.³ The underlying cause of the juvenile periodontitis is not well understood but is now thought to be related to an abnormal immune system and to invading bacteria in the cementum of the teeth. Various immunologic defects have been described.⁶ A decreased peripheral T-lymphocyte subpopulation (CD₃⁺ CD₄⁺), noted in our patient, has been described.⁶

Defective production of superoxide radicals by polymorphonuclear leukocytes (burst test), has been described in PLS patient.⁴ Defective chemotaxis of polymorphonuclear leukocytes is also a commonly described abnormality.⁵

The association of PLS and spontaneous pneumothorax has not been described in the literature. Whether this association was just a coincidence or due to some underlying pathology is not clear.

A multidisciplinary approach is important. PPK is usually treated with topical emollients. Salicylic acid and urea can be added to enhance their effect. Systemic retinoids have proven to be effective in PPK of PLS as well as in other PPKs.¹⁵ The concern that retinoid treatment in PLS may increase the risk of pyogenic liver abscess,¹⁴,⁵ is probably unfounded, as this may occur in patients not receiving retinoids. The periodontitis is usually difficult to control. Reported effective treatment includes extraction of the primary teeth combined with oral antibiotics and professional teeth cleaning.⁵

Etretinate and acitretin have been though claimed to modulate the course of periodontitis and preserve the teeth.¹⁵ frequently do not succeed in preserving the permanent teeth. Prophylactic antibiotics use has not been studied, and there are no clear guidelines. The risk of pyogenic liver abscess should be kept in mind in evaluating these patients. Recently, the gene for PLS has been mapped to 11q14-q21. In 1999, Hart et al⁴ identified a germline missense and truncating mutations in the gene encoding cathepsin C (or dipeptidyl aminopeptidase I), a lysosomal cysteine protease that plays an important role in intracellular degradation of proteins in families with PLS. Cathepsin C is an enzyme that processes and activates several granule serine proteases critical to immune and inflammatory responses of myeloid and lymphoid cells.⁶

**REFERENCES**