CASE REPORT

Papillon- Lefevre Syndrome with Liver Abscess

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An 8 year old boy presented with fever of unknown origin in whom the diagnosis of liver abscess was made. He-also had palmoplantar keratoderma and premature loss of teeth, consistent with the diagnosis of Papillon Lefevre syndrome.

Key words: *Liver abscess, Papillon-Lefevre syndrome.*

Papillon Lefevre syndrome (PLS) is rare autosomal recessive disorder characterized by palmoplantar keratoderma (PPK) and juvenile periodontitis. In addition to these cardinal features, frequent pyogenic infections, hyperhidrosis and nail dystrophy have been described.The association of skin and oral lesions differentiate this unusual genodermatosis from other forms of palmoplantar keratoderma. The diagnosis is mainly clinical.

CASE REPORT

An eight year old boy was referred to us with fever and pain in the right hypochondrium of two months duration .There was no history of vomiting or jaundice. He was the third born of second degree consanguineous parents.

On examination he was febrile and had tachycardia. There was mild pallor but no icterus. Well demarcated symmetrical psoriasiform plaques were present over his elbows and knees. He also had diffuse palmoplantar keratoderma with transgradiens extending to the dorsae of hands and feet. The nails showed transverse grooves with dystrophic changes (***Fig.* 1a**). The skin lesions worsened in winter. Examination of the oral cavity revealed an edentulous upper jaw with just two molars (***Fig.* 1b**). The lower jaw had four incisors and two molars. All teeth had grade1 mobility except the right upper molar which had grade 3 mobility with surrounding gingival inflammation.The mother gave a history of early shedding of deciduous as well as permanent teeth. There was no history of recurrent infections or excessive sweating. No other family member suffered from similar problems. Abdominal examination showed a bulge on the lateral aspect of the right hypochondrium with tender hepatomegaly. The spleen was not palpable.

Hematologic and biochemical investigations were normal except for leucocytosis and neutrophilia. Abdominal ultrasonography (USG) showed a well defined hypoechoic lesion (abscess) measuring 6.4× 4.5×4.2 cm in the right lobe of the liver lateral to the gall bladder.

In view of the skin lesions and early loss of teeth, dermatological and dental consultations were sought and the diagnosis of PLS was established. Considering the association of pyogenic liver abscess and PLS, the patient was started on

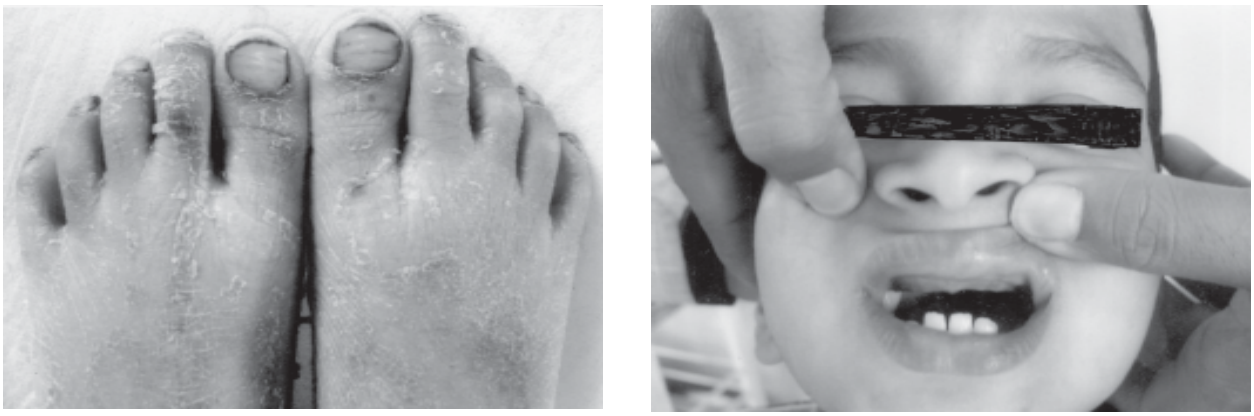

FIG.1 *Patient with papillon-Lefevre syndrome showing (a) transverse grooves with dystrophic changs of nails and (b) absence of age-appropriate dentition.*

broadspectrum antibiotics (cefotaxime with sulbactum and amikacin). Percutaneous aspiration under USG guidance was planned but was withheld as the patient started showing clinical improvement . He responded well to conservative management, with disappearance of the abdominal bulge and was afebrile by the fifth day. Antibiotics were continued for 4 weeks and repeat USG showed complete resolution of the abscess. The patient was started on Isotretinoin and there was marked improvement in skin lesions.

Skin biopsy was suggestive of chronic keratodermatitis. The *X*-ray skull showed no evidence of intracranial calcification. An Ortho-pantogram showed typical ''tooth in air'' appearance of the right upper molar.

DISCUSSION

Papillon Lefevre syndrome is a rare disorder of keratinization affecting children between the ages of 1-5 years(1). The reported incidence of this disorder is 1-4 per million(2). A major gene locus for PLS has been mapped on chromosome 11q 14 and mutations of cathepsin C gene is found in homozygotes of PLS(3).

Palmoplantar keratoderma usually starts within the first 4 years of life with sharply demarcated erythematous keratotic plaques involving palms and soles, sometimes extending on to the dorsal surfaces of the hands and feet. The cutaneous lesions have a tendency to worsen in winter(4). The other characteristic feature of PLS is progressive early onset periodontitis which starts by the age of 3 or 4 years, affecting both deciduous as well as permanent teeth. The teeth erupt normally but are soon lost and the patients usually become edentulous in their early teens(4). Although periodontal inflammation subsides after exfoliation of primary teeth, the same cycle is repeated with the eruption of permanent teeth. However, the third molars may be spared(3). Severe resorption of alveolar bone gives the teeth a "floating in air" appearance on dental *X*-ray. Our patient presented with the above two features of PLS.

Increased susceptibility to infections has been reported in about 20% of these patients, possibly due to some dysfunction of leukocytes . Another feature of PLS may be radiological evidence of intracranial calcification(1). The nails may show onychodys-trophy and transverse grooving(5).

Patients with PLS are predisposed to develop pyogenic liver abscess(6). Few reports of this interesting association have been published so far, with only two reports from India(1,6-10). Bacteremia resulting from periodontitis coupled with neutrophil dysfunction appears to be responsible for the development of liver abscess(9).

Early recognition of this condition and a multidisciplinary approach may help in improving the prognosis of these patients. Skin manifestations are usually treated with emollients, salicylic acid and urea. Oral retinoids including acitretin and isotretinoin are the main stay of treatment of both kertoderma and periodontitis(9). Effective treatment

of periodontitis includes extraction of primary teeth, prompt institution of antibiotics and maintenance of dental hygiene. There is no consensus regarding prophylactic antibiotics. However antibiotics should be given to treat periodontitis.

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