PAPILLON-LEFEVRE SYNDROME

A J Kanwar, Surrinder Kaur, Rajeev Sharma and Vijay Bakaya

A 13-year-old girl having Papillon-Lefevre syndrome presented with palmo-plantar keratoderma, early loss of primary dentition and gingivitis. Repeated skin infections were a striking feature. There were however, no associated immunological abnormalities.

Key word : Papillon-Lefevre syndrome.

Papillon-Lefevre syndrome (PL syndrome) is characterized by a diffuse palmo-plantar hyperkeratosis and periodontopathy which results in loss of both the deciduous and permanent teeth.[1,2] It is a rare disorder with autosomal recessive inheritance and equal sexual predilection.[3] Approximately 160 cases have been reported in the literature and in only a few cases a marked predisposition to skin infections due to impaired lymphocyte functions has been observed.[4,5] We report a case of this syndrome in which repeated skin infections were a striking feature. However, no immunological abnormalities were detected.

Case Report

A 13-year-old female student had well-demarcated, erythematous hyperkeratotic and scaly lesions on the palms and soles since the age of 3 months. She was the second child of healthy, non-consanguineous parents with no family history of similar disorder. The hyperkeratotic lesions had been progressive till the age of 3 years when she developed swelling of the gums associated with foul smell from the mouth. This was followed by the loss of teeth over a period of 2 months. The swelling of the gums subsided and she remained edentulous till the age of 9 years when permanent teeth started erupting. Since 3 years she had been having recurrent furunculosis of the extremities for which she received multiple courses of various antibiotics with improvement.

The palms and soles had diffuse erythema and hyperkeratosis with extension to the dorsa of hands and feet as well as to the skin overlying the Achilles tendon. The borders of all the affected areas were sharply demarcated and there were numerous fissures on the palms and soles. Psoriasiform plaques were present on the knees and elbows. In addition, there were multiple 2-5 cm sized depressed scars on the lower extremities. Most of the permanent teeth were intact. However, the gingivae were inflamed and a few of the teeth were loose. The nails of feet were thickened, discoloured and showed excessive ridging. The patient also had deep-seated tender swellings on the dorsa of hands and feet with a few of them showing ulcerations on the surface. Hemoglobin, total and differential leucocyte counts, peripheral blood film, urinalysis, liver and renal function tests, serum calcium, phosphorus, sodium, potassium and serum protein levels were within normal limits. The ESR was 35 mm. Culture from the ulcer on the dorsum of hand grew *Staphylococcus aureus*, susceptible to erythromycin. Cultures for *Mycobacterium tuberculosis* and fungus were negative. The serum and immuno electrophoresis showed normal patterns. The lymphocyte functions (B and T cell counts, lymphocyte transformation test using PHA), and neutrophil functions (chemotaxis, phagocytic activity against candida) were normal. Tuberculin, candidin and aspergillin tests were negative at 72 hours. Roentgenograms

From the Department of Dermatology, Post-Graduate Institute of Medical Education and Research, Chandigarh-160012, India.

Address correspondence to : Dr A J Kanwar.

of the skeletal system showed no abnormality. Chest skiagram was normal. Skin biopsy from the palm revealed irregular hyperkeratosis, marked acanthosis and a perivascular infiltrate of lymphocytes and histiocytes.

She was treated with 10% salicylic acid ointment topically to which she showed a fairly good response. The pyodermic lesions healed after a course of antibiotics.

Comments

An early onset of keratoderma involving the palms and soles with extension to the sides of hands and feet and up the Achilles tendon, psoriasiform lesions on the knees and elbows and early periodontopathy which led to the loss of primary dentition were the features of PL syndrome in our patient. Permanent teeth in the patients of PL syndrome erupt normally but are lost like primary dentition following gingivitis.[6] Our patient had already started showing clinical signs of gingivitis following a normal eruption of permanent teeth. Other less consistent findings in the PL syndrome include sparse hair, intracranial calcifications of falx cerebri and choroid plexus,[7,8] and mental retardation. None of these features were observed in our patient. A rather interesting feature which has been observed in some cases of PL syndrome is repeated skin infections.[5,9] These most often manifest as furunculosis and other pyodermas with associated deficient neutrophil phagocytosis[10] or impaired reactivity to T and B cell mitogens.[11,12] However, it was not possible to demonstrate any impairment of immunological mechanisms in our patient as evidenced by normal immunoglobulins, neutrophil and lymphocyte function tests. It could thus have been a chance occurrence only.[13] The nails in PL syndrome have been reported to be normal.[3] Our patient had marked nail changes in the form of discoloration, thickening and ridging.

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