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PAPILLON-LEFEVRE SYNDROME – A RARE CASE REPORT

Dermatology

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ABSTRACT

Papillon-Lefevre syndrome is an autosomal recessive disorder characterized by transgradiens Palmo-plantar keratoderma, early onset severe periodontitis and recurrent pyogenic infections. Here we report a rare case of Papillon-Lefevre syndrome in an 18 year old female.

KEYWORDS

Papillon-Lefevre syndrome, Transgradiens Palmo-plantar keratoderma, Periodontitis, Recurrent pyogenic infections.

INTRODUCTION:

Papillon-Lefevre syndrome is a rare autosomal recessive disorder characterized by transgradiens Palmo-plantar keratoderma, early onset severe periodontitis and recurrent pyogenic infections. It is due to Cathepsin-C gene mutation leading to deficiency of Cathepsin C enzyme activity.

CASE REPORT:

An 18 year old female came with complaints of scaly lesions over B/L upper limbs, lower limbs, palms, soles, lower back and scalp. She has developed scaly lesions first over the scalp which later extended to extremities and trunk. She has history of recurrent pyogenic infections since the age of 1 year. She lost her deciduous teeth by the age of 6 years and Permanent Upper and Lower incisors by the age of 12years. History of 2nd degree consanguinity present.

O/E: Scaly plaques are present over B/L upper and lower limbs, lower back and scalp. Hyperkeratoses and fissures noted over palms and soles. Clawing is present in 3rd & 4th fingers. Nails are Hypertrophic, Onycholysis and trachyonychia noted. Post inflammatory pigmentation is seen in Inframammary area. She has upper incisor dentures gingival erythema noted over Upper lateral incisors. On genital examination, Vulvo vaginal candidiasis is present.

A 4mm punch biopsy was done on lower back lesion and sent for HPE. Histopathological findings are Hyperkeratosis with focal area of parakeratosis, acanthosis and areas of spongiosis. Basal layer is intact. Dilated blood vessels seen in papillary dermis. Inflammatory cells (Lymphocytes) are noted in Dermis.

Based on above mentioned findings we made a diagnosis of Papillon-Lefevre syndrome.

DISCUSSION:

Papillon-Lefevre syndrome is a rare genodermal syndrome described initially by two French physicians Papillon and Lefevre in 1924 in a brother and sister[1] with Palmoplantar hyperkeratosis, premature loss of teeth and periodontitis. It is autosomal recessive disorder due to mutation in Cathepsin C gene encoding for Cysteine lysosomal protease. This gene is present in epithelial regions like palms, soles, knees and oral gingiva, also highly expressed in immune cells. It is mainly seen in consanguineous marriage[2]. Both the genders are equally affected[3].

It is classified into 3 groups –Diffuse, Focal and Punctate[4]. Clinical features include Palmo-plantar hyperkertoses extending onto dorsal surface, Hyperkeratotic plaques on elbows and knees and recurrent pyogenic infections. Early onset severe periodontitis which leads to loss of both deciduous and permanent teeth[5]. Dural and choroid pleuxes calcification may be noted[6].

Histologically there is Hyperkeratosis, focal parakeratosis, marked acanthosis, tortuous capillaries in dermal papilla and mild mononuclear cell infiltrate of papillary dermis.

It should be differentiated from Haim-Munk syndrome which is an allelic variant of Papillon-Lefevre syndrome[7] and Prepubertal periodontitis.

Management includes Oral retinoids, topical emollients and salicylic acid for cutaneous lesions. Oral antibiotics for periodontitis can be given.

CONCLUSION:

Papillon-Lefevre syndrome is rare autosomal recessive genoderm atoses and reports on it are relatively sparse.

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CONFLICT OF INTEREST: The authors declare that they have no conflict of interest.

LEGENDS TO IMAGES

FIG 1: Hyperkeratoses of Palms and Upper limb with Nail changes showing onycholysis and trachyonychia

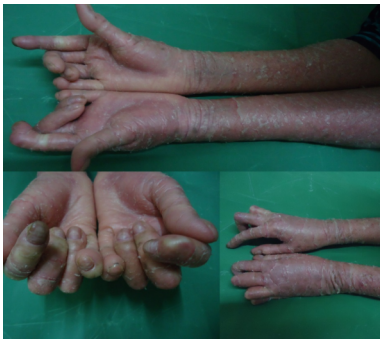


FIG 2: Hyperkeratoses over soles and lower limbs

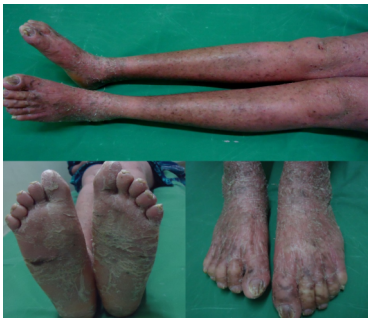


Fig 3 : Upper incisor dentures, loss of lower incisors and gingival erythema in oral cavity

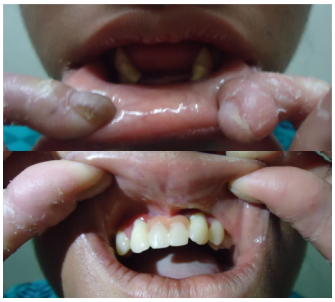


Fig 4: Hyperkeratotic lesion over back and biopsy was taken from here

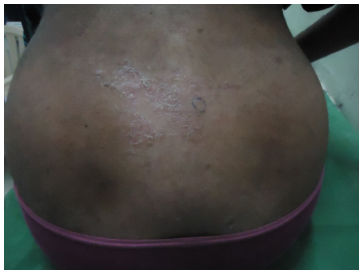


Fig 5 - Low power view showing hyperkeratosis,focal parakera toses, spongiosis with acanthosis

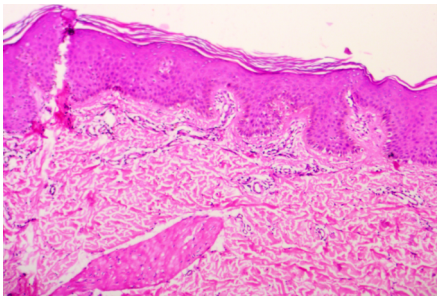
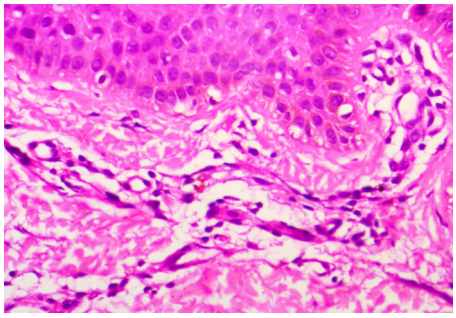


Fig 6- High power view showing dilated blood vessels with lymphocytic infiltrate in the papillary dermis



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