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INTERNATIONAL JOURNAL OF ADVANCED RESEARCH (IJAR)

Article DOI: 10.21474/IJAR01/16019
DOI URL: http://dx.doi.org/10.21474/IJAR01/16019

RESEARCH ARTICLE

A RARE CASE OF PAPILLON-LEFEVRE SYNDROME: A CASE REPORT

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Manuscript Info

Manuscript History
Received: 10 November 2022
Final Accepted: 14 December 2022
Published: January 2023

Key words:-
Papillon Lefevre Syndrome, Cathepsin C, Consanguinity, Systemic Retinoids

Abstract

Papillon Lefevre syndrome is an extremely rare autosomal recessive disorder characterized by diffuse palmoplantar hyperkeratosis, rapidly progressive and devastating periodonitis, and pyodermas. The etiopathogenesis of the disorder is multifactorial, with genetic and immunological factors playing a major role. Consanguinity is a contributing factor. Genetic mutations of the gene 11q14- q21 encoding for cathepsin-c, a lysosomal protease that activates enzymes involved in a variety of inflammatory and immune processes. Management directed towards halting periodontal destruction using conventional periodontal treatment, systemic antibiotics, oral hygiene instructions, antiseptic mouth rinses. Palmoplantar hyperkeratosis usually treated with topical application of emollients, keratolytic agents, topical steriods and systemic retinoids.

Introduction:-

Papillon Lefevre syndrome (PLS) was first described by two French physicians, Papillon and Lefèvre in 1924, described palmoplantar hyperkeratosis (PPK) and severe early onset periodontitis in a brother and sister (born to a consanguineous couple/ 1st cousin matching)[(1)]. Later, Gorlin et al. reviewed 46 Papillon-Lefevre syndrome cases and they added calcification of the falx cerebri to the above manifestations, converting it into a triad: palmoplantar hyperkeratosis (PPK), severe early onset periodontitis, and calcification of the falx cerebri[(2)]. It is an autosomal recessive inherited disorder of keratinization, caused by mutations in cathepsin-c (CTSC) gene, a lysosomal protease that activates enzymes vital to the body defense process.

Case Report:

A 22 years old male, ITI student presented with thickening of skin over palms and soles since 6 months of age. The lesions initially started as hyperkeratotic scaly plaques over plantar and palmar surfaces, gradually extended to dorsum of feet and hands with bilateral knee and elbow joints. History of loss of teeth at the age of 9 years, initially started as enamel loss leads to loss of teeth, now patient using artificial dentures. He is the second child born to a consanguineous couple delivered at full term through normal vaginal delivery. No history of similar complaints in the family. On examination, diffuse well defined hyperkeratotic scaly plaques with fissures noted over the bilateral palmo plantar and dorsal aspect of hands and feet extended to lateral surfaces involving Achilles tendon. Well defined psoriasiform, hyperkeratotic scaly plaques noted over bilateral knee and elbow joints. Examination of the oral cavity showed artificial dentures. Finger nails showed clubbing. Xerosis was seen all over the body. No other lesions were noted elsewhere on the body.

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Figure 1:- Hperkeratotic scaly plaques with fissures over bilateral palms and sole.

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Figure 2:- Well defined scaly plaques over dorsal aspect of hands and feet extended to lateral surfaces involving Achilles tendon.

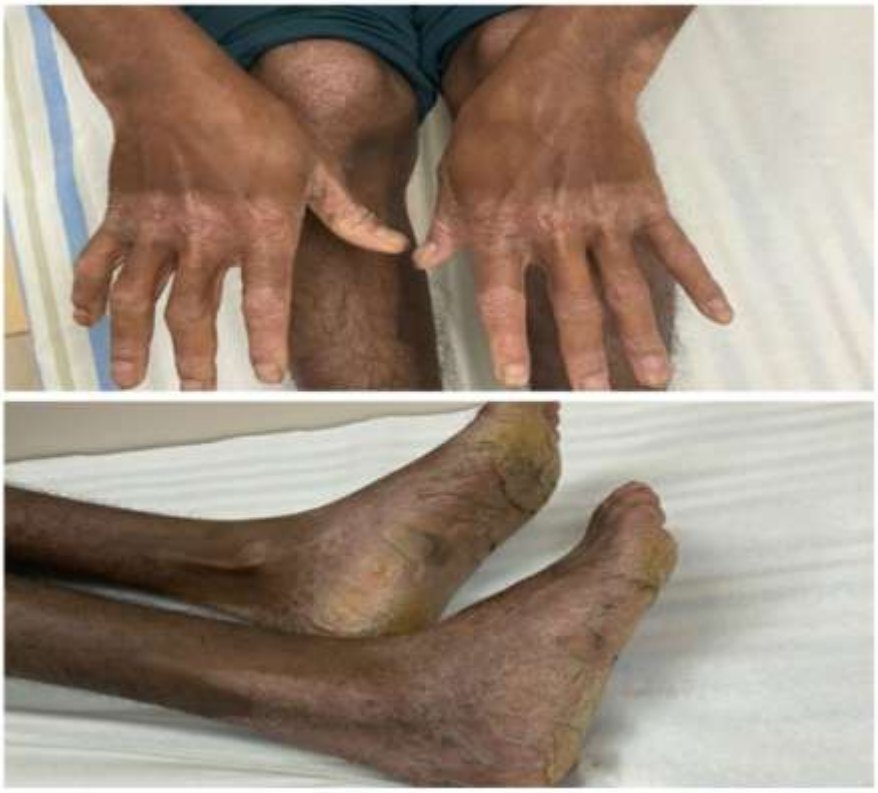
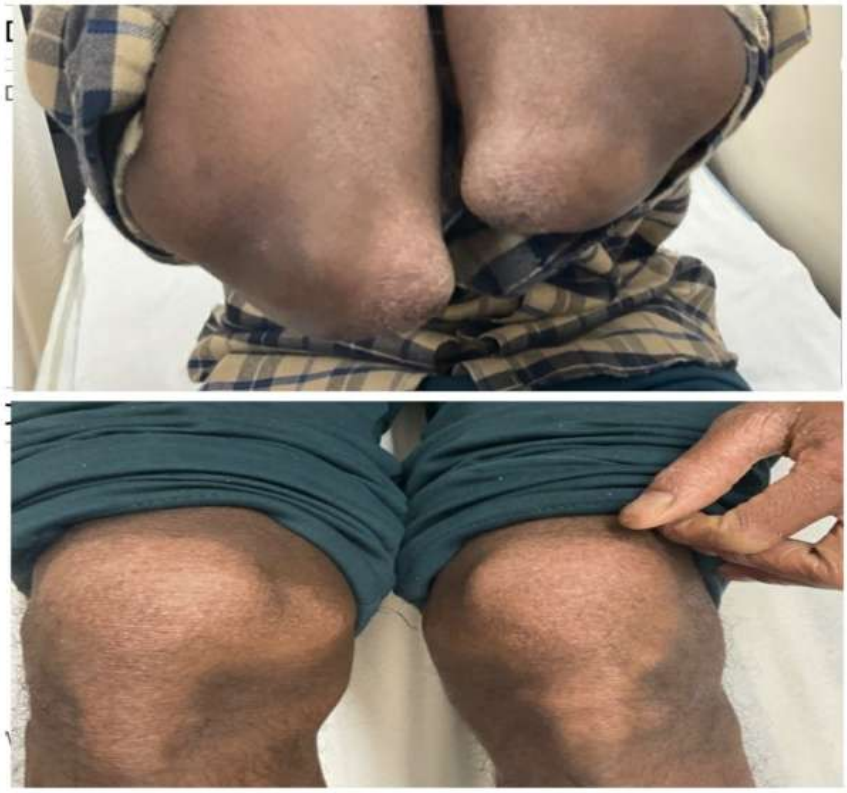


Figure 3:- Psoriasiform hyperkeratotic scaly plaques over bilateral knee and elbow joints.



Discussion:-

Papillon Lefèvre syndrome is also known as keratoderma with periodontitis. This syndrome transmitted as autosomal recessive trait, characterized by diffuse transgradient PPK, periodontosis, and frequent pyodermas, with a prevalence of 1-4 cases/million. Males and females are equally affected. 75% of the Arab probands were products of close parental consanguinity. Consanguinity seen in 1/3rd of cases.[2,3] The aetiology is homozygous genetic mutations (loss of function) encoding cathepsin-c (CTSC) enzyme; known as dipeptidyl peptidase-1. CTSC is expressed at high levels in polymorphonuclear leukocytes, alveolar macrophages, skin-epidermis of extremities and gingiva, kidney, and placenta. It plays an important role in intracellular degradation of proteins. CTSC is lysosomal cysteine protease that removes dipeptides from the free N- terminal of protein and peptides and involves a wide variety of immune and inflammatory responses by activating serine proteinases from cytotoxic T- cells, NK cells, mast cells and neutrophils. In general, children with PLS are moderately built and their physical and intellectual development levels are within normal limits. A sub-type of PLS is Haim Munk syndrome characterized by PLS along with onychogryphosis, arachnodactyly and acro-osteolysis with out pyoderma and falx cerebri, seen in Jews of South Indian origin[4].Diagnosis is based on clinical signs and symptoms, patient/family history, genetic tesing. Urine CTSC analysis in infants (complete absence) and absent of azurophilic granules in mature neutrophils but not in progenitor cells, indicating CTSC promotes protease degradation in immune cells.[4] Treatment goal is mainly halting the rapid devastating periodontal destruction and reducing the hyperkaratotic skin lesions by topical emollients and keratolytic agents and oral retinoids 0.5-1 mg/kg/day.[5]

Conclusion:-

We are reporting this case as it is rare with limited publications, 200 cases until 1995; 250 cases until 2005; and 300 cases until 2019[1], and early identification is necessary to prevent devasting complications like periodontosis.

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