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Brunauer-Fuhs-Siemens palmoplantar keratoderma: A rare, striate type of focal palmoplantar keratoderma

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Abstract: *Brunauer-Fuhs-Siemens palmoplantar keratoderma, commonly known as striate palmoplantar keratoderma, is a rare, autosomally inherited disease of linear hyperkeratosis in which patient usually presents with conspicuous longitudinal hyperkeratosis on volar surface of hands and feet. Mutations in 3 genes namely desmoglein 1, desmoplakin and keratin 1, have been identified and held responsible for this type of keratoderma which can efficiently lead to the structural defects in desmosomal plaque proteins manifesting as mechanical weakness in the pressure areas of palm and soles. Apart from the cutaneous features, woolly hair and cardiomyopathy may also be assosiated with striate palmoplantar keratoderma. We herein report a unique case of striate palmoplantar keratoderma in a 18 years old male having linear hyperkeratosis over dorsal surface of hands and feet alongwith nummular keratoderma since chilhood.*

Keywords: *Brunauer-Fuhs-Siemens, desmoglein, palmoplantar keratoderma, striate, woolly hair*

I. Introduction

Striate palmoplantar keratoderma is one of the rare type of focal or areate group of palmoplantar keratoderma (PPK),also known as Brunauer-Fuhs-Siemens type striate keratoderma which is characterized by linear hyperkeratosis of palm and soles. [1] The mode of inheritance is usually autosomal dominant and age of onset is first few years of life. [2] Being a heterogeneous disease, genetic linkage has been found to 3 gene loci: desmoglein 1 gene, desmoplakin gene and keratin 1. [3] Apart from the cutaneous features, involvement of nails and hairs may be seen and cardiac abnormalities has also been seen in some cases.

II. Case Report

A 18 years old male came to the outpatient department of dermatology presenting with linear thickening over the dorsal surface of both feet and hands since childhood. The lesions were painless and non-tender. The patient was born of non-consanguineous parents and there was no family history of similar lesions. On local cutaneous examination, linear pattern of hyperkeratosis was noted along the dorsomedial border of both great toe. Nummular or areate pattern of skin thickening was found on the dorsal surface of distal phalanx of index and middle finger of both hands and linear pattern on volar surface of right index finger. Interdigital space between thumb and index finger alongwith medial border of thumb of both hands had also linear hyperkeratosis [Fig.1-5].

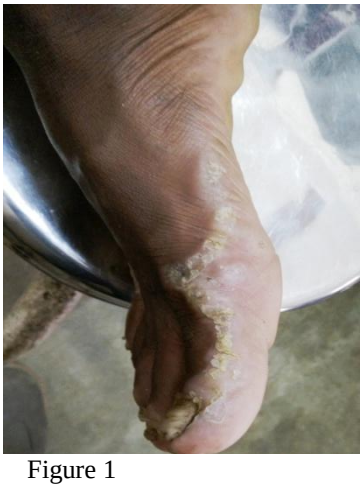

Figure 1

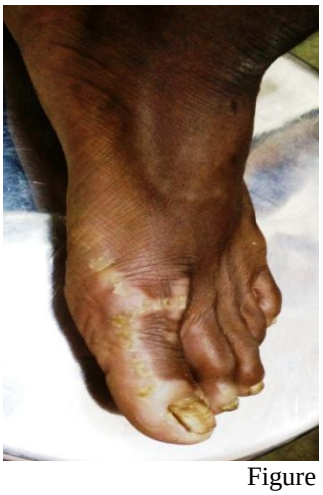

Figure 2

Figure 1 & 2 showing linear or striate hyperkeratosis over dorsomedial border of right and left great toe.

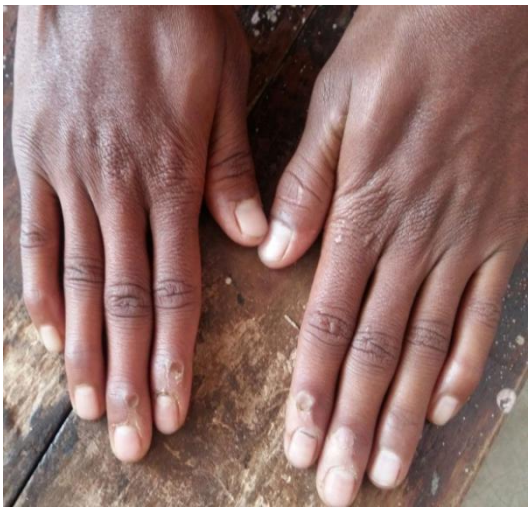

Figure 3 : Nummular huperkeratosis on dorsal aspect of fingers

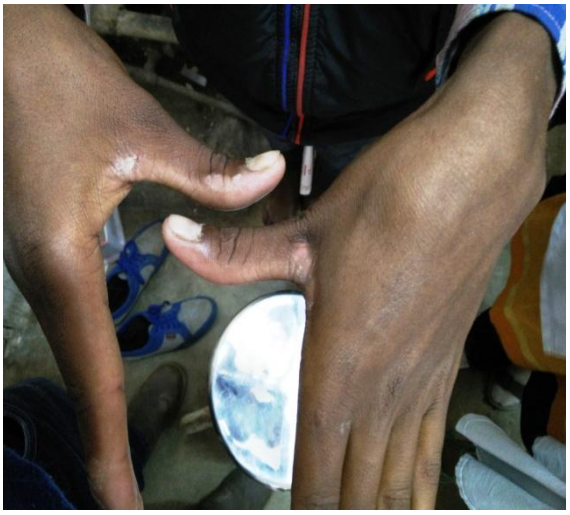

Figure 4 :focal and linear pattern of hyperkeratosis in interdigital space between thumb and index finger

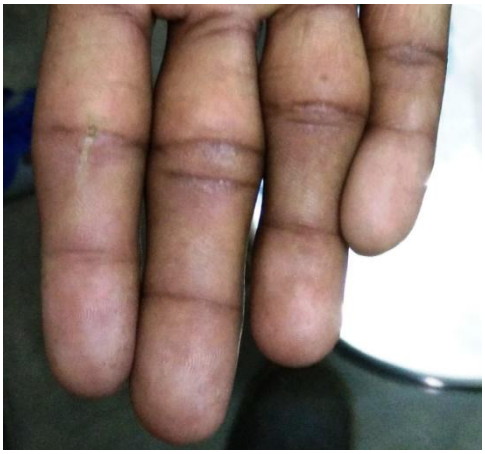

Figure 5 : linear skin thickening on flexor aspect of right index finger

On general physical and systemic examination, nothing was found to be remarkable. All routine investigations and laboratory parameters were within normal limits.

To confirm the lesion as keratoderma, sample was taken from dorsum of right great toe with punch biopsy and sent for histopathological examination.

III. Histopathological Examination

Histopathological examination was done which revealed marked compact orthohyperkeratosis, hypergranulosis, acanthosis and thickened bundles of collagen in vertical array in papillary dermis [Fig.6]. On the basis of clinical pattern and histopathology, diagnosis of striate palmoplantar keratoderma was made.

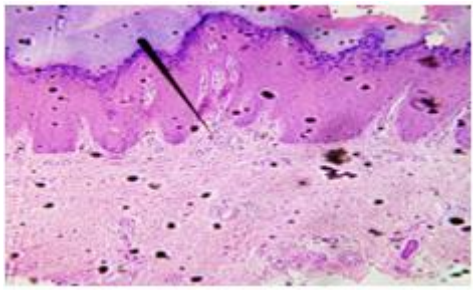

Figure 6 : marked compact orthohyperkeratosis, hypergranulosis, acanthosis, thickened collagen bundles in papillary dermis (H & E , x 40)

IV. Discussion

Palmoplantar keratoderma (PPKs) are a diverse group of hereditary and acquired disorders characterized by excessive epidermal thickening of palms and soles. [1] Over the time, a number of classifications of PPKs have been put forward but none of them could unite the three basic differentiating criteria: molecular pathogenesis, pathology and clinical presentation. On the basis of pattern of presentation, PPKs are often classified as diffuse, focal (areate), striate (linear) and punctuate or papular, but many times, it is not possible to distinctly confine the lesions of PPK as more than one pattern may frequently coexist in the same patient. [4] The demographic profile of the patient like age, gender and occupation and site of the lesion are those factors which usually influence the pattern of PPKs.

Further, focal PPKs are subclassified into nummular (Wachter keratoderma), striate or linear keratoderma and pachyonychia congenita types. Earlier due to the familial occurrence of nummular and striate type of PPK, Wachter suggested a single entity 'Keratoderma varians' for both of these two pattern. [6] Now both have been distinguished clinically and striate type PPK was named 'Brunauer-Fuhs-Siemens type striate PPK' after three dermatologist and physician. They were Stefan Robert Brünauer (an Austrian physician), Herbert Fuhs (an Austrian dermatologist) and Hermann Werner Siemens (a German dermatologist). [7]

The lesions of striate type are typically longitudinal, particularly on the palm and soles overlying flexor tendons while nummular type presents as focal painful calosities on pressure areas of palm and soles. However, lesions of striate type may frequently overlap with Wachter keratoderma.

Striate PPK is usually inherited as an autosomal dominant trait and is caused by defects in at least 3 different loci of genes. It has been linked to the desmosomal cadherin cluster on 18q12.1 encoding desmogleins (Dsg) and desmocollins (Dsc), 6p21 encoding desmoplakins (Dp) and keratin 1 gene in very few cases. [3] The most common mutations in these genes which have been identified, are nonsense mutation or missense mutation arising due to haploinsufficiency. The majority of mutations in striate keratoderma are seen in Dsg1 or Dp1 but in two cases frameshift mutations in the V2 tail domain of keratin 1 gene were found which are important in cross-linking to cornified envelope proteins. [2] [8] Apart from the keratoderma and skin fragility, such kind of mutations in these identified genes may also be responsible for some characteristic hair changes like woolly hair, nail changes and dilated cardiomyopathy as trichological and cardiac conduction abnormalities can be seen with or without keratoderma. [1]

On electron microscopy of involved skin in patients with keratoderma associated with Dp mutations, abnormal perinuclear aggregation of keratin filaments and reduction in the peripheral keratin network, with loss of connections with desmosomes are seen. [9] [10] Desmosome number was also found to be reduced in Dsg and Dp mutations but desmosome size was reduced only in the Dsg mutation forms. [10].

Clinically, linear pattern of skin thickening on the palms and flexor aspects of the fingers and soles develop during infancy or in first few years of life.

Plantar skin may be found to be more severely and early affected than palmar skin. Rarely, other sites such as elbow and knee may be involved but dorsum or dorsomedial aspects of feet has been reported in only one case in previous literature. So our case is the second case with such presentation of striate type of PPK. Skin fragility can also be seen as a distinct cutaneous manifestation. As in all other types of keratoderma, keratolytics, topical and oral retinoids are the only modalities with variable results.

V. Conclusion

As previously mentioned, owing to the same underlying genetic mutations, skin fragility, woolly hair and dilated cardiomyopathy or cardiac arrhythmia forms a triad which are found in Naxos disease and Carvajal syndrome. In the view of such possible coexisting conditions, patients presenting with striate type of PPK should always be investigated in this way.

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