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Case Study

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GREITHER DISEASE- A RARE CASE REPORT

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ABSTRACT

Greither disease was first described by the Greither in 1952[1]. It is an autosomal dominant condition with marked inter- and intrafamilial variability and due the missense mutation of gene encoding Keratin 1[2]. We report a case of 19 year old female with classical features of Greither disease.

KEYWORDS: • Greither disease • Transgradiens • Keratin 1 • Autosomal dominant • Achilles tendon • Hyperhidrosis.

INTRODUCTION

Greither disease (OMIM #133200), first described by the Greither in 1952.[1] The mode of inheritance will be of autosomal dominant of inter- or intrafamilial variations and associated mutation of gene encoding KERATIN 1.[2]

It usually starts at second year of life but can also occur soon after the birth or as late as in childhood or adolescence. It is manifested as diffuse, thickened, yellowish PPK with an erythematous rim and transgradiens. The involvement of Achilles tendon is characteristic and there is gradual extension towards shin, knees, thighs, knuckles, wrists, elbows and flexural areas as patchy hyperkeratotic, erythematous and hyperpigmented papules and plaques.[3] Sometimes there is an association with hyperhidrosis.

Histopathologically there will be characteristic findings of acanthosis, marked hyperkeratosis and hypergranulosis. Other features are vacuolation of superficial keratinocytes, numerous keratohyaline granules in granular layer and focal depressions of the epidermis occupied by round foci of a compact orthokeratotic horny layer.[4]

CASE REPORT

A 19 years old female patient had attended our out patient department with chief complaints of thickening of palms first and then soles with a gap of 1month had an insidious onset and are gradually progressed. Lesions started as small raised skin coloured lesions over palms and soles later gradually progressed to involve both dorsum of hand, foot and also Achilles tendon. She also complained of excessive sweating of palms. She was born out of a consanguineous marriage of third degree. Similar lesions were present over the soles in her mother.

On cutaneous examination revealed diffuse palmoplantar keratoderma with hyperkeratotic hyperpigmented plaques and erythematous border encroaching on to the dorsa of foot and to Achilles tendon. Well defined, smooth dome shaped thickened plaques were noted over the knuckles of both the hands. Hyperhidrosis was noted over the palms.

Figure 1. Hyperkeratotic plaques over knuckles.

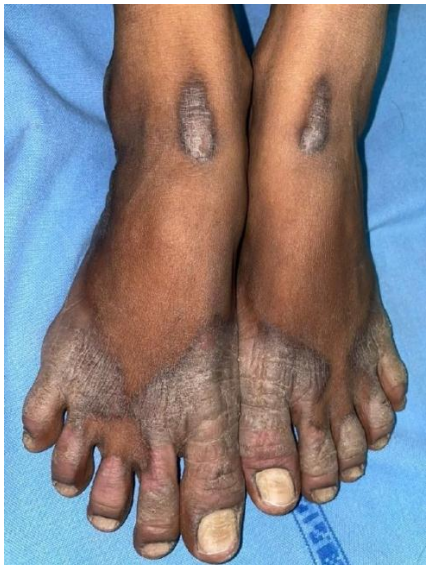

Figure 2. Transgradiens with erythematous rim.

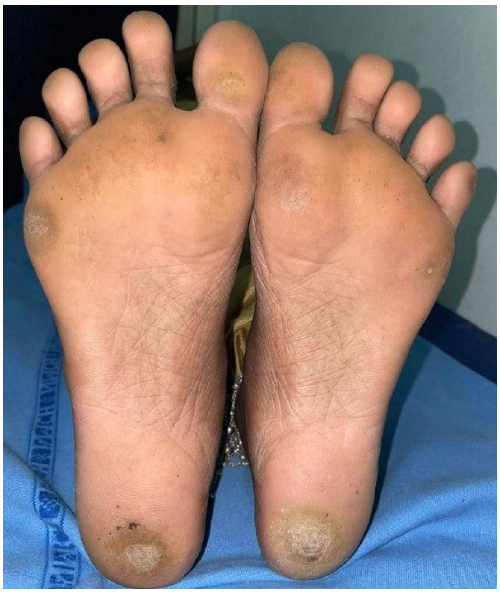

Figure 3. Plantar keratoderma.

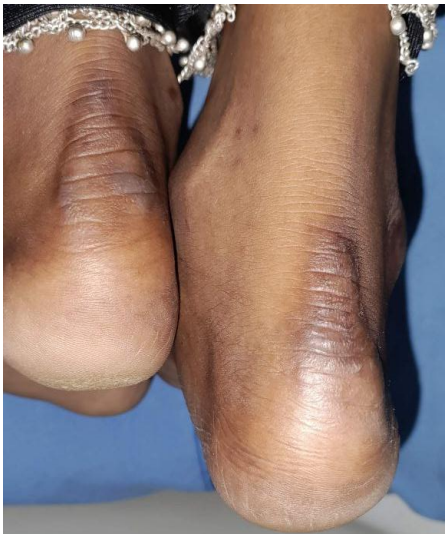

Figure 4. Hyperkeratotic plaque over Achilles tendon.

Basic routine investigations like Complete blood count, blood sugars, liver function tests, renal function tests, echocardiogram, chest Xray, ultrasound abdomen and urine routine were sent and came out to be normal.

A punch biopsy of 4mm was taken one from the left hand and other from left foot. Histopathological examination revealed epidermis showing mild to moderate acanthosis, hyperkeratosis, hypergranulosis and elongation and broadening of the rete ridges. Horny layer showed round foci of compact orthokeratosis. Papillary dermis showed dilated capillaries and perivascular infiltrate.

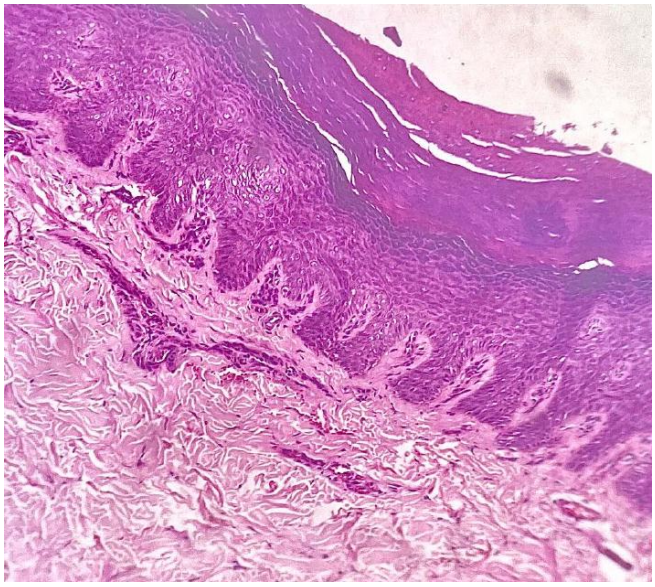

Figure 5. Hyperkeratosis, acantosis, hypergranulosis and elongation and broadening of rete ridges.

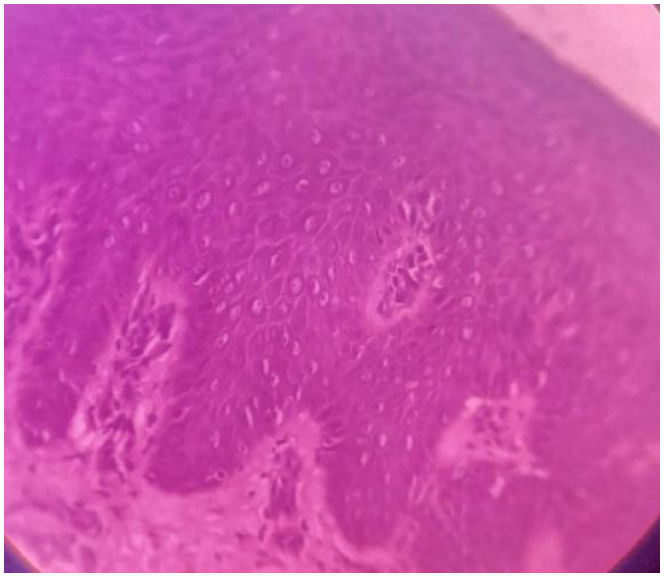

Figure 6: Round foci of compact orthokeratosis on higher magnification.

DISCUSSION

Palmoplantar keratodermas form a heterogeneous group of hereditary or acquired disorders defined by excessive epidermal thickening of the palms and soles.[5] Most of the disorders in this group are hereditary.

Greither disease is classified under diffuse type of palmoplantar keratodermas with transgradiens. Lesions usually develop after second year of life and involute after fifth decade of life. There will be diffuse PPK with erythematous rim of transgradiens involving dorsa of hands, feet, tendo achilles (characteristic), knees, elbows. There will be marked hyperhidrosis.

Associations include pitted keratolysis, neonatal blistering and erythroderma[6], atopic dermatitis[7], malignant melanoma[8] and others like incontinentia pigmenti, acrocyanosis, erythrokeratoderma varialbilis.

Complications like pseudoainhum and digital deformities may occur.

Acral keratoderma of Sybert is a variant of Greither disease which is a progressive diffuse, autosomal dominant PPK with transgradiens and autoamputation. Hyperkeratosis will be more severe when compared to the Greither disease.[9]

Greither syndrome should be differentiated from Unna-Thost syndrome. Some authors describe Greither disease as variant of Unna-Thost syndrome where as others consider it as a separate entitiy. In Unna-Thost syndrome the onset of lesions will be earlier than in Greither disease i.e. during 1st month of life and characteristic hyperkeratotic plaque over Achilles tendon, knees and elbows is less marked in Unna-Thost syndrome and there is no involution of disease after fifth decade. Küster and Becker had proposed Unna-Thost syndrome as a separate entity.[10]

It should also be differentiated from Mal de Meleda syndrome. Mal de Meleda syndrome has an autosomal recessive inheritance with onset early after birth and is a progressive condition involving different regions without spontaneous resolution. Typical nail changes may be present.

Other differentials include Vörner PPK, acrokerato elastoidosis, focal acral hyperkeratosis.

Further studies like ultrastructural studies can be done which reveals normal keratin filaments with abnormal structure and distribution of keratohyaline granules.

Treatment modalities like topical keratolytics like salicylic acid, topical corticosteroids and oral acitretin (25-35mg/ day or thrice weekly) can be used.

CONCLUSION

To conclude Greither disease is a diffuse palmoplantar keratoderma with transgradiens of autosomal dominant inheritance and due to missense mutation of Keratin 1 gene. There is a spontaneous involution after fifth decade.

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