IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 14, Issue 6 Ver. VIII (Jun. 2015), PP 56-58
www.iosrjournals.org

Jadassohn Lewandowsky Syndrome: A Rare Genodermatoses in Association with Metabolic Syndrome.

DR. SHISHIRA. R. JARTARKAR[1], DR. MALLIKARJUN. M[2],
DR. BUGUDE. GANGADHAR[3], DR. MANJUNATHA. P[3]
[1]Junior Resident, [2]Professor and Head of Department, [3]Assistant Professor
Department of Dermatology, K V G Medical College and Hospital, Kurunjibhag, Sullia, Dakshina Kannada, Karnataka, India
Corresponding author–Dr.Shishira.R.Jartarkar

Abstract: *Pachyonychia congenita is a rare genodermatoses transmitted as an autosomal dominant trait with only 450 cases reported since 1906. It is of four types. Pachyonychia congenita type 1 is called as Jadassohn Lewandowsky syndrome.The syndrome results from mutations in the gene encoding epidermal keratins. The patients present with classical nail hypertrophy, palmoplantar hyperkeratosis and follicular keratotic papules on the body. The case has been reported for its rarity and its rare association with metabolic syndrome.*
Keywords*: pachyonychia congenita, nail hypertrophy, palmoplantar hyperkeratosis*

I. Introduction

Pachyonychiacongenita is a rare genodermatoses transmitted as an autosomal dominant triat with high degree of penetrance. First case report was documented in 1904 by Muller[1] followed by reports published in 1905 by Wilson and in 1906 by Jadassohn and Lewandowsky.[2] Since then, only about 450 cases have been reported.[3] Skin, nails, oral mucosa, larynx, hair, teeth are involved in variable combination with nail involvement being most prominent. Four types of pachyonychia congenita have been described.[4]We report a rare case of pachyonychia congenita type 1, known as Jadassohn Lewandowsky syndrome, in association with metabolic syndrome.

II. Case Report

A 60yr old obese female patient, born of a consanguinous marriage, known case of diabetes since 5yrs and hypertension since 8yrs, presented to medicine department with exertional dyspnoea and palpitations since two weeks. On examination, patient had anaemia, high postprandial blood glucose levels and hypertriglyceridaemia. The doctor noticed her skin and nail changes which were present since childhood and referred her to us. She had hypertrophic nails, involving both finger and toe nails. Nail plate was hard, lustreless, hypertrophied with thickening of the nail plate noted maximum at the distal end. Patient also complained of palmoplantarkeratoderma since childhood with thick hyperkeratotic plaques. Follicular keratosis was noted at the anterolateral aspect of the thighs bilaterally. Multiple callosities were present over the feet. On skin biopsy, hyperkeratosis, parakeratosis with orthokeratosis and acanthosis was seen. The skin scraping for potassium hydroxide mount was negative for fungal elements. Hair, teeth, throat, oral cavity was normal. Similar lesions to a lesser degree were noted in her mother and two of her siblings. With all these findings, the patient was diagnosed as pachyonychia congenita type 1 syndrome and started on topical keratolytics for 2months. The patient had no response and then patient lost on follow up.

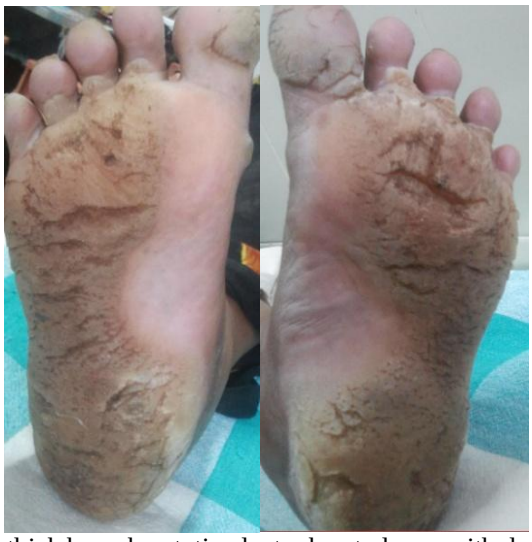

Fig 1& 2 – thick hyperkeratotic plantar keratoderma with deep fissuring

Fig 3 – thickening of the nail plate , more significant over distal end with dry lustreless nails

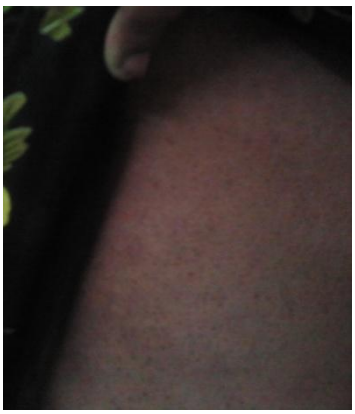

Fig 4 – multiple keratotic follicular erythematous papules present over anterolateral aspect of left thigh

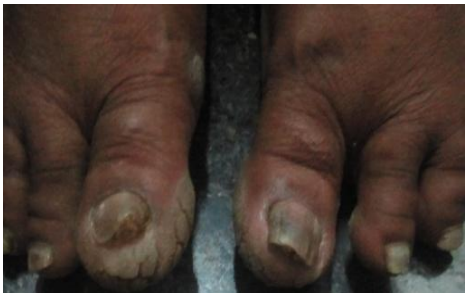

Fig 5 – multiple callosities present over medial aspect of base of right great toe and tip of the great toe.

III. Discussion

Pachyonychia congenita (PC) is a rare inherited genetic disorder transmitted in an autosomal dominant fashion. It can also occur as a sporadic case with spontaneous mutations.[4]The disease has been classified into fo ur types. PC type 1 (JadassohnLewandowsky syndrome) characterised by focal palmoplantar keratoderma and f ollicular keratotic papules over the body. PC type 2 (Murray Jackson Lawler syndrome) having natal teeth and s teatocystoma multiplex along with features of PC type 1. PC type 3 (Schafer Branauer syndrome) includes comb ined features of type 1 and 2 with angular cheilitis, corneal dyskeratosis and cataracts. PC type 4 includes featur es of type 1 to type 3 with laryngeal lesions, hoarseness of voice with mental retardation, hair abnormalities and alopecia. PC with late age of onset has been suggested by Paller*et al.* and termed as PC tarda.[5]This syndrome res ults from mutations in gene encoding epidermal keratins.[6]Type 1 has mutations in keratin 6a and 16 and type 2 h as mutations in keratin 6b and 17. The mutation has a deleterious effect on the protein structure, as it interferes with assembly of polypeptides forming keratin skeleton of epidermal cells.[4]

On histopathology, there is gross hyperkeratosis with alternating orthokeratosis and parakeratosis with acanthosis and patchy hypergranulosis with large keratohyalin granules withour gross epidermolysis.[7]

Treatment is usually unsatisfactory and includes topical keratolytics like salicylic acid, lactic acid and u rea. Others include propylene glycol with occlusion and topical retinoic acid. Intracutaneous plantar injection of botulinum toxin type A in three patients with PC has been reported. This has resulted in remarkable relief of pai n from plantar pressure sites for upto 6 months.[8]Other treatment modalities include oral retinoids like acitretin 2 5-35mg/day may be effective.[7]Few patients need radical surgical excision of the nail plate, matrix and bed with skin transplantation at that site to achieve permanent total nail removal. Vigorous curettage of nail bed and matri x is most effective and simplest treatment.

Our patient belongs to PC type 1 as she has classical nail deformity along with palmoplantar hyperkerat osis and follicular papules over thigh. This case is reported because of its rarity and also its rare association with metabolic syndrome.

References

[1]. Muller C. On the causes of congenital onychogryphosis. Mcn Med Wochenschr. 1904;49:2180-2
[2]. Jadassohn J, Lewandowsky F. Pachyonychiacongenita. Keratosis disseminatacircumscripta. Tylomata. Leukokeratosis linguae. In: Jacob's IkonographiaDermatologica 1 Berlin: Urban und Schwarzenberg; 1906:29-30
[3]. Agarwal P, Chhaperwal MK, Singh A *et al.*Pachyonychiacongenita. A rare genodermatoses. Indian Dermatol Online J. 2013;4:225-7
[4]. Agarwal SN, Kulkarni YA, Jane SD, *et al.*Pachyonychiacongenita type 1 (JadassohnLewandowsky syndrome). Indian J Paediatr Dermatol.2014:15:137-9
[5]. Paller AS, Moore JA, Scher R. Pachyonychiacongenitatarda. A late onset from of pachyonychiacongenita. Arch Dermatol.1991;127:701-3
[6]. Dahl PR, Dauoud MS, Su WP. Jadassohn Lewandowski syndrome (pachyonychiacongenita). Semin Dermatol.1995;14:129-34
[7]. Thomas DR, Jorizzo JL, Brysk MM, *et al.* Pachyonychiacongenita. Electron microscopic and epidermal glycoprotein assessment before and during isotretinoin treatment. Arch Dermatol.1984;120:1475-9
[8]. Swartling C, Vahlquist A. Treatment of pachyonychiacongenita with plantar injection of botulinum toxin. Br J Dermatol.2006;154:763-65