Case Report

Pachyonychia Congenita Tarda

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ABSTRACT

A 50 year old woman presented with progressively thickening and discoloration of all nails since age of 14 years. Initially with thick, lusterless nails and progressed with yellow brown discoloration and distal elevation of nail plates. At presentation she had hypertrophy and warty growth of right great toe and right third toe. There was no systemic or mucocutneous involvement.

Key Words : Pachyonychia congenita tarda, onycholysis, subungual hyperkeratosis.

Introduction :

Pachyonychia congenita (PC) is a group of rare genodermatosis with autosomal dominant mode of inheritance, first described by Jadassohn and Lewandowsky in 1906[1]. It is a keratinization disorder associated with characteristic nail changes and various other skin and mucocutaneous changes.

Four types of PC are described, from PC I to PC IV. To the best of our knowledge, only two case of the fourth type, Pachyonychia Congenita Tarda (PCT) has been reported from India [2,3].

The Case :

A 50 year old female presented in the outpatient department with symptoms of upper respiratory tract infection. On examination, the most striking feature was presence of hypertrophic nail changes in all fingers and toe nails. On asking she revealed that she had progressive thickening and discoloration of nails, which started at the age of 14 years. Initially the nails became thick and lusterless. Over the years all her nails became disfigured.

Examination revealed thickening and hardening with yellowish brown discoloration of nails. All fingers and toes were involved. There was transverse over curvature of nails Subungual hyperkeratosis was present with onycholysis of overlying nails ***(fig. 1).*** Nail plates of right great toe and right third toe were completely destroyed with cauliflower like subungual hypertrophy ***(fig 2).***

Fig. 1 **- Onycholysis & subungual hyperkeratosis in finger nails.**

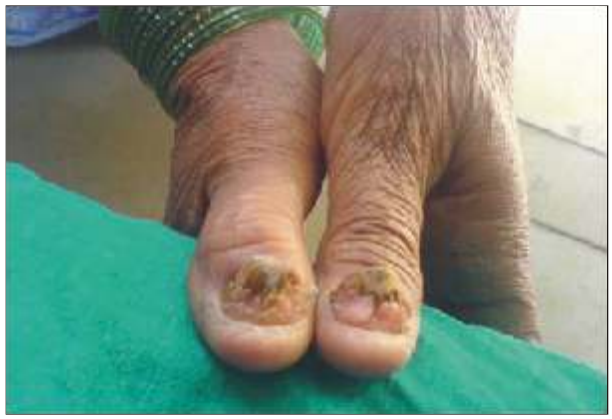

Fig 2. **- Warty subungual hyperkeratosis in toe nails with complete destruction of nail plate.**

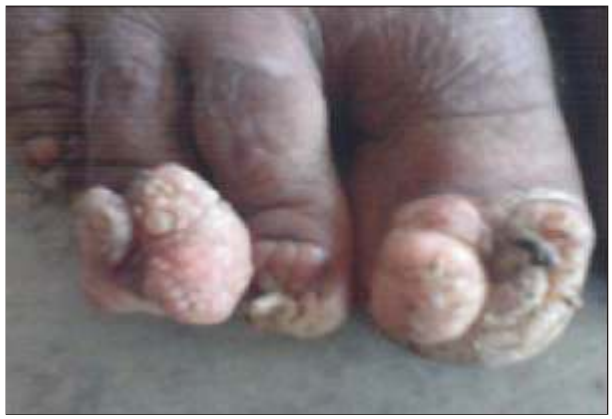

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Fig. 3 -

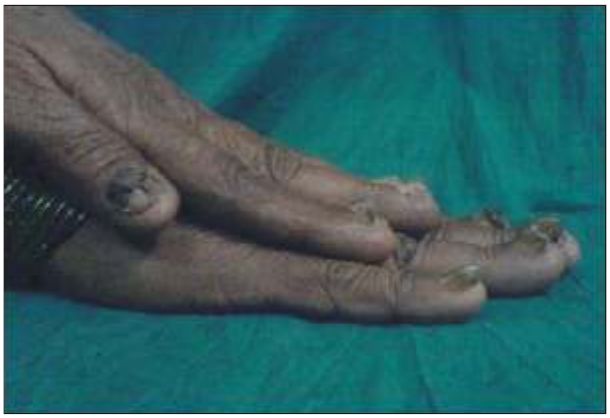
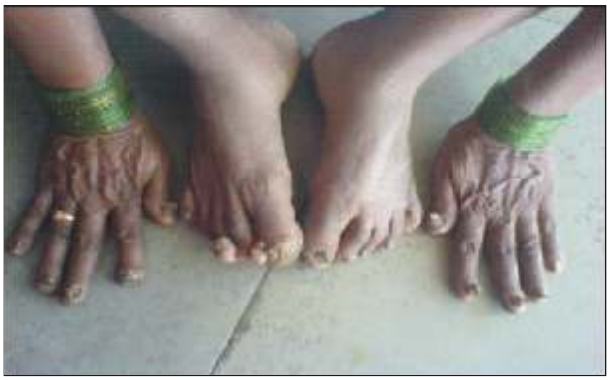


Fig. 4 -



Her palms and soles were normal. There was no evidence of hyperhidrosis, palmoplantar blistering, palmoplantar hyperkeratosis, oral leukokeratosis, alopecia, corneal abnormalities or epidermoid cysts. There was no systemic or mucocutaneous involvement.

There was no history of consanguinous marriage between her parents. Her daughter who was 30 years old also had started developing thick yellow nails with subungual hyperkeratosis. Rest of the family members had normal nails.

The nail growth was subjected for biopsy and histopathological examination hyperkeratosis and acanthosis of the stratified squamous epithelium with elongated rete pegs underneath fibrocollagenous tissue with lymphatic infiltrate, conforming the diagnosis of PC.

Since the characteristic nail changes began in the second decade of life, this is a case of PCT (PC IV).

Discussion :

PC is rare keratinization disorder with variable phenotypic presentations, having predominantly autosomal dominant mode of inheritance, although autosomal recessive inheritance has also been described[4]. All forms of this disease have characteristic nail changes including nail thickening, discoloration, subungual hyperkeratosis and distal onycholysis.

PC 1 (Jadassohn Lewandowsky syndrome) is the most frequent variant. Nails are thickened with brown grey discoloration and subungual hyperkeratosis. All fingers and toes are involved. Other features include palmoplantar hyperkeratosis, oral leukokeratosis, and follicular hyperkeratosis[5].

In PC 2 (Jackson lawler syndrome) nail changes are associated with epidermal cysts and steatocysts, natal teeth and hypotrichosis[5].

In PC 3 (Schafer - Branauer syndrome) corneal leukokeratosis is present.

PC usually presents either at birth or within the first year of life. In the fourth type, PCT, the nail changes occur in the second or third decade of life. This type was first described by Paller et. al. in 1991.[6]

Mutations in the keratin gene cluster in chromosome 17 are responsible for PC. PC 1 is caused by mutation in gene encoding keratin 6a or keratin 16, while PC 2 is caused by mutation in keratin 6b or keratin 17[7].

Keratin genes K6 and K16 are expressed in mucosal epithelia, follicular keratinocytes and palmoplantar epidermis whereas K17 is constitutively expressed in pilosebaceous units and basal appendage keratinocytes, thus explaining the features of PC 1 and PC 2.

Pachyonychia congenita tarda (PCT) is thought to represents a delayed form of PC type 1 and may have similar underlying genetic defects affecting the keratin 6a/16 pair[8].

Cases which show features of more than one clinical subtype have been reported. Around 300 cases of PC have been described till now. Pachyonychia congenital tarda with features of PC 1 as well as PC 2

have been described.

The most recent literature refers to descriptions of about 250 cases of PC[9]. Since it was first described in 1991, only around 10 cases of PCT have been described [10].

In this case patient had nail manifestations only.

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