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Murray-Jackson-Lawler Syndrome

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Abstract

Pachyonychia congenita (PC) is a rare autosomal dominant genodermatosis. It is of four types, type I due to mutation in genes 6a and 16, and 6b and 17 in type II. A 2 yr old male child presented in our OPD with hypertrophy of nails, hyperkeratotic papules over body, lusterless and sparse hair and natal teeth since childhood. Microscopy nail clippings and scrapping were done to rule out fungal infection. No evidence of any associated malignancy was found after thorough workup. He was diagnosed as PC Type 2. This case is being reported because of its rarity.

Index terms— pachyonychia congenita, genodermatosis, autosomal dominant, subungual hyperkeratosis, hyperkeratotic papules.

1 Introduction

achyonychia congenital (PC) is a rare genodermatosis having autosomal dominant pattern of inheritance with high penetration. PC mainly affects a number of ectodermal structures including nail bed, skin, teeth, oral mucosa and pilosebaceous unit. Nail involvement is commonest feature of PC which is usually present at birth or develops soon after birth. Muller and Wilson described the first case of PC 1 II.

2 Case Report

. On the basis of clinical features found in addition to the nail changes, PC has been classified into four types. Here, we report a rare case of PC type-2, known as Murray-Jackson-Lawler syndrome.

A 2-year-old male child born of a nonconsanguinous marriage presented with thickened, yellowish brown discoloration of all the nails and multiple papules all over his body and ill formed lower incisor since birth [Figure 1]. Initially, at the time of birth, parents noticed natal tooth (ill formed lower incisor) and thickening of few nails of toes and fingers, which gradually increased over months and involved total twenty nails [Figure 2 a, b]. Later, at the age of 3 months parents also noticed small hyperkeratotic papules over both knees [Figure **??**], which over a period of year increased and involved bilateral upper limb, bilateral lower limb, trunk and buttocks. On examination, the nails showed thickened, lusterless nail plate of all the nails with upward growth of its distal portion. The nails also showed subungual hyperkeratosis and the nail plate was adhered to the underlying nail bed. There was increased curvature of the transverse axis of all nail plates giving a "pinched shape" to the free edge of the nail plate. Multiple hyperkeratotic follicular papules were present over bilateral upper limb, trunk, lower limbs and buttocks. He also had lusterless, sparse hair over scalp [Figure **??**]. Systemic examination revealed no abnormality. The routine investigations were within the normal limits. Skin scraping for potassium hydroxide mount was negative for fungal elements. Patient's throat, and ophthalmological examination were normal. With all these findings the patient was diagnosed as PC type-2 and was started on topical retinoid and emollient for 1 month.

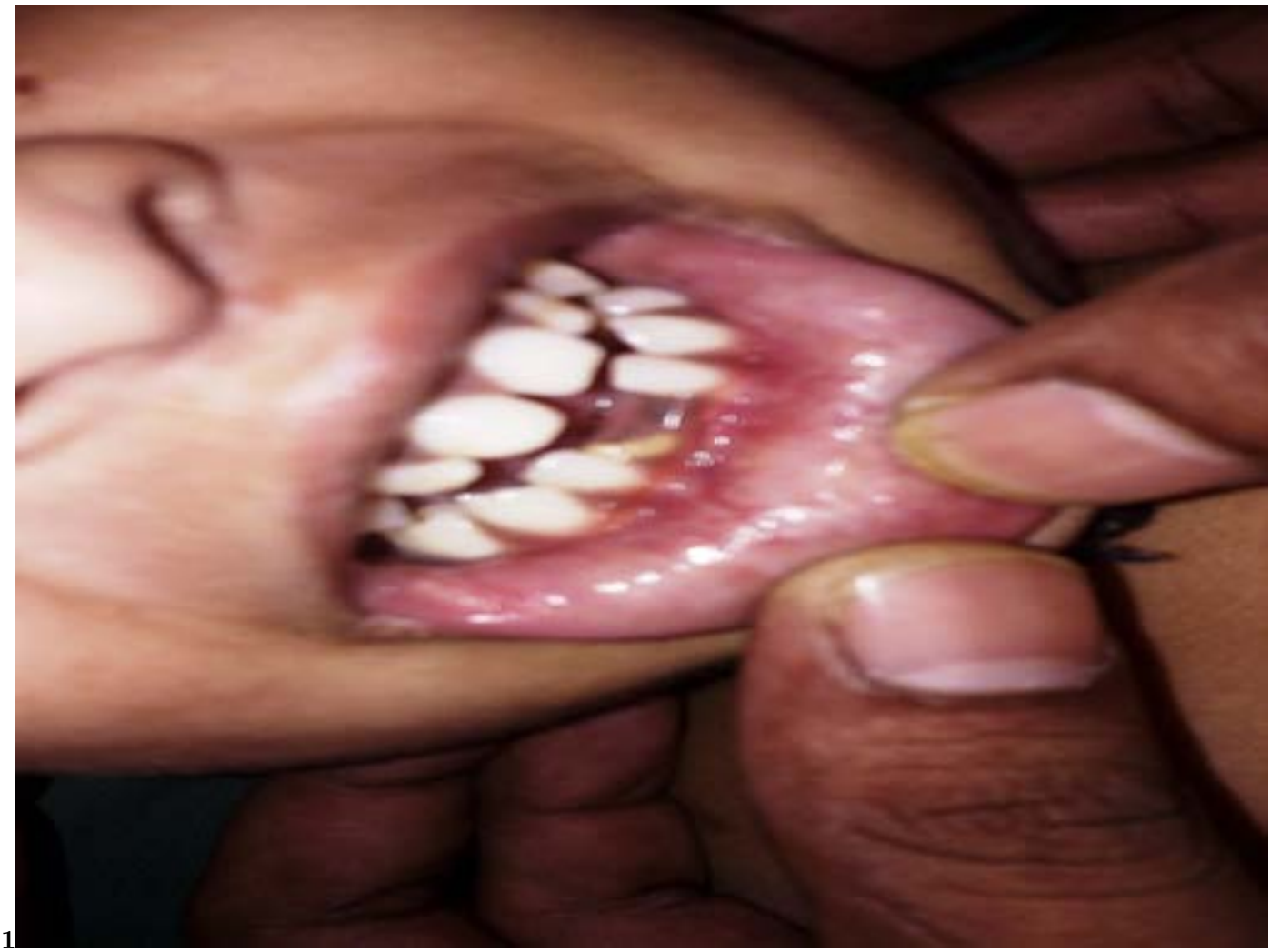
3 III.

4 Discussion

Pachyonychia congenita is a rare group of disorder characterized by inherited ectodermal dysplasias transmitted in an autosomal dominant fashion. Some autosomal recessive cases also detected 2 . Sporadic case has been also detected with spontaneous mutation 3 P . Pachyonychia congenita have four variants identified depending on the genetic mutation and clinical correlation 3 F age of onset has been reported and termed as PC tarda 4 . The majority of mutations are missense mutations with a smaller number of deletions, insertions and splice site mutations in affected genes which leads to deleterious effects on protein structure as it interferes with the assembly of polypeptides forming the keratin structure of epidermal cell 5 .

The main characterstic symptom of this syndrome is hyperkeratosis of the nail bed. This type of subungual hyperkeratosis leads to the elevation and increased transverse curvature of the nail plate and also associated with discoloration, thickening and friability of nail plate 6 , which may sometimes fail to reach the distal fingertip. Generally all the 20 nails are affected but more severely thumbs, index fingers and toes nails are involved 7 The treatment in this is usually unsatisfactory. Topical application of salicylic acid, urea and 5-fluorouracil can be advice to the patient. The other treatment modalities include systemic therapy like oral retinoids (acitretin, retinoic acid) and surgery . 8 . A long term use of retinoids may results in to some degree of flattening of the nails and other complications such as periosteal hyperostosis, increased sensitivity and fragility of the underlying epidermis, and this limits their usefulness. 9 Our patient belongs to PC type-2 as he had classical nail deformity along with hyperkeratotic papules over bilateral upper limb, bilateral lower limb, trunk and buttocks with natal teeth, and sparse lusterless thin hair over scalp. The patient did not show any of the other findings associated with other types of PC. This case is being reported because of its rarity.

A genetic counselor also plays an important role as this gene has an autosomal dominant inheritance pattern and that PC can affect one half of his or her progeny, so counselor should inform the carrier. Nail findings persist for long, but other features may become less severe later in life. [1]



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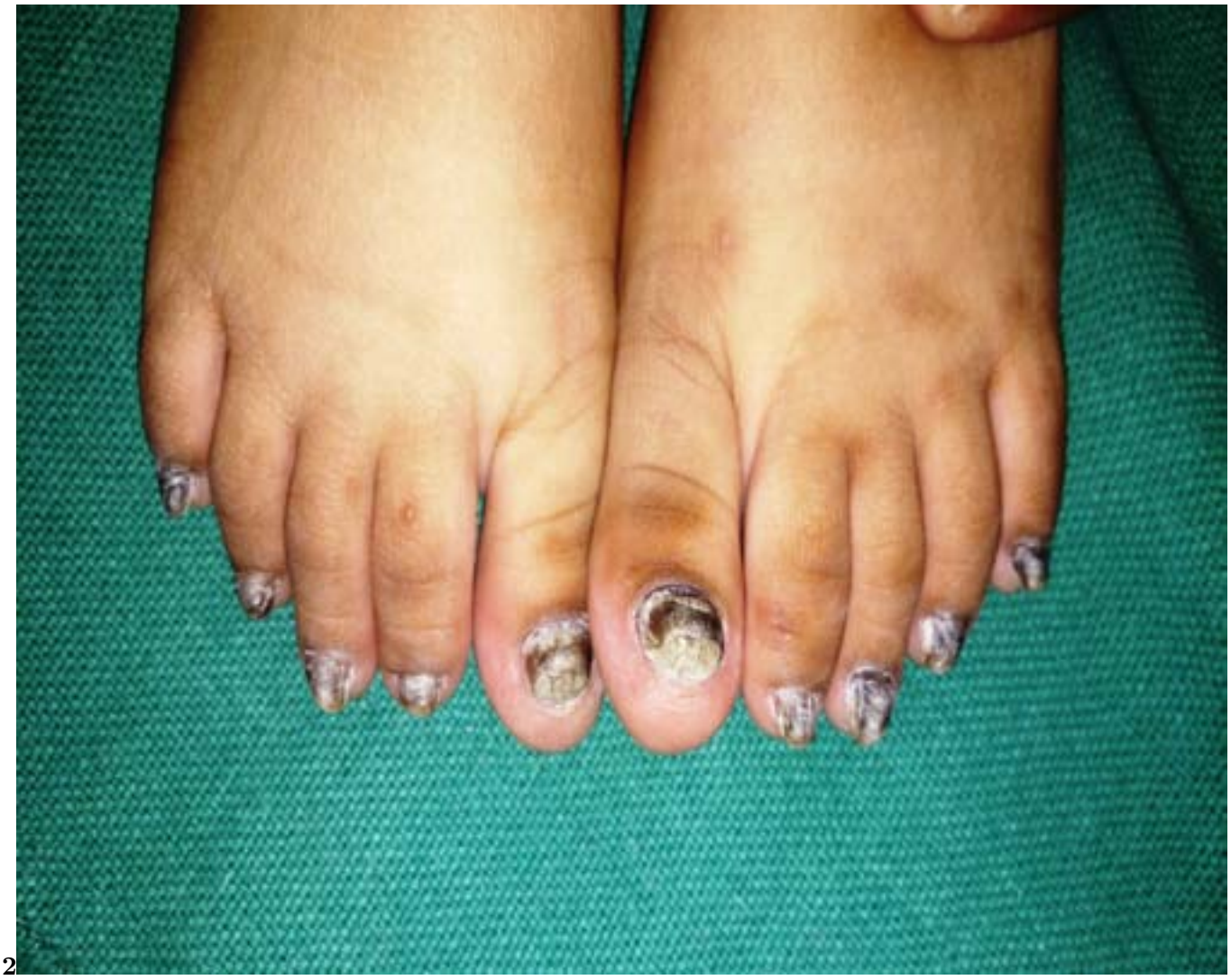

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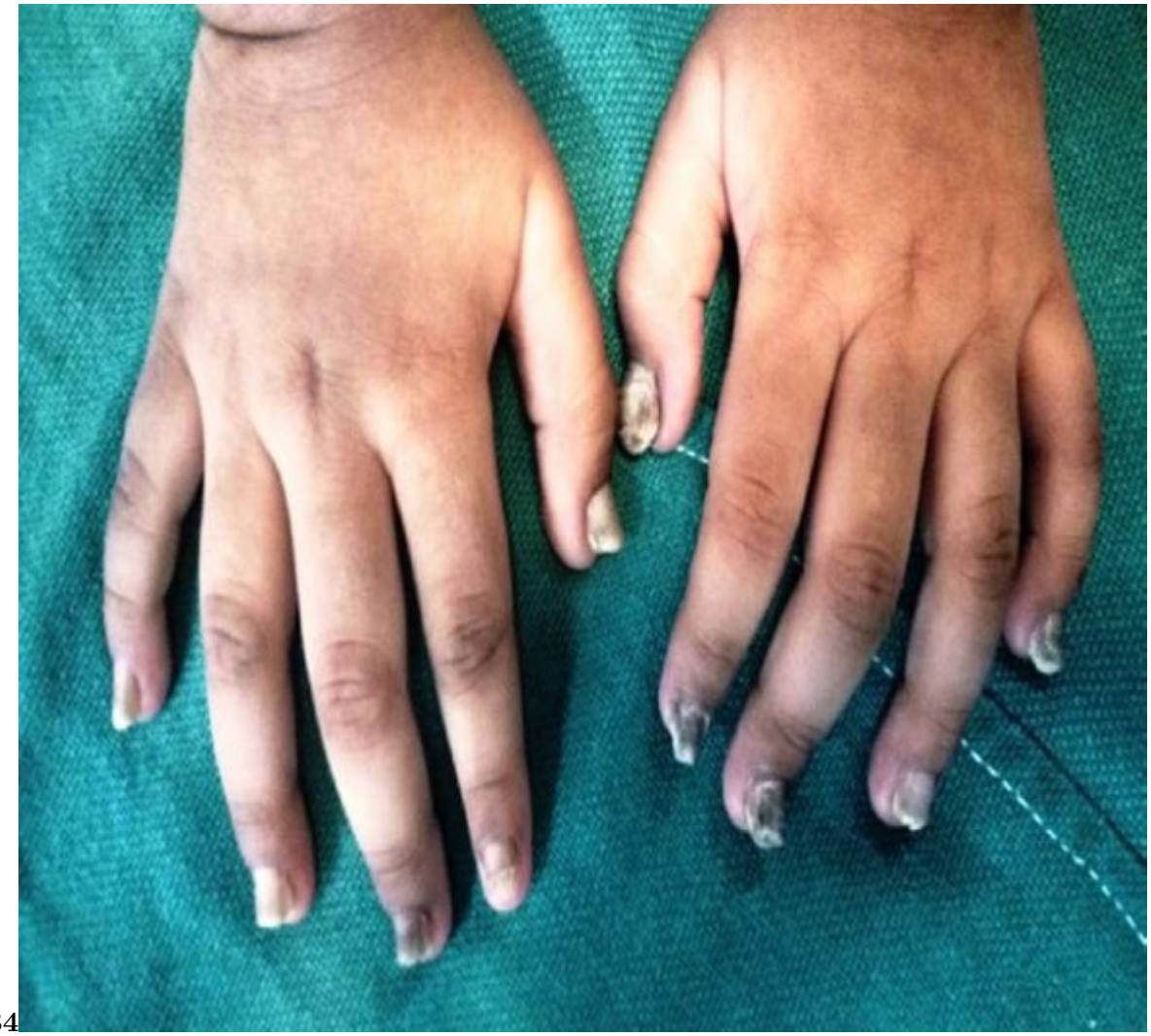
34 

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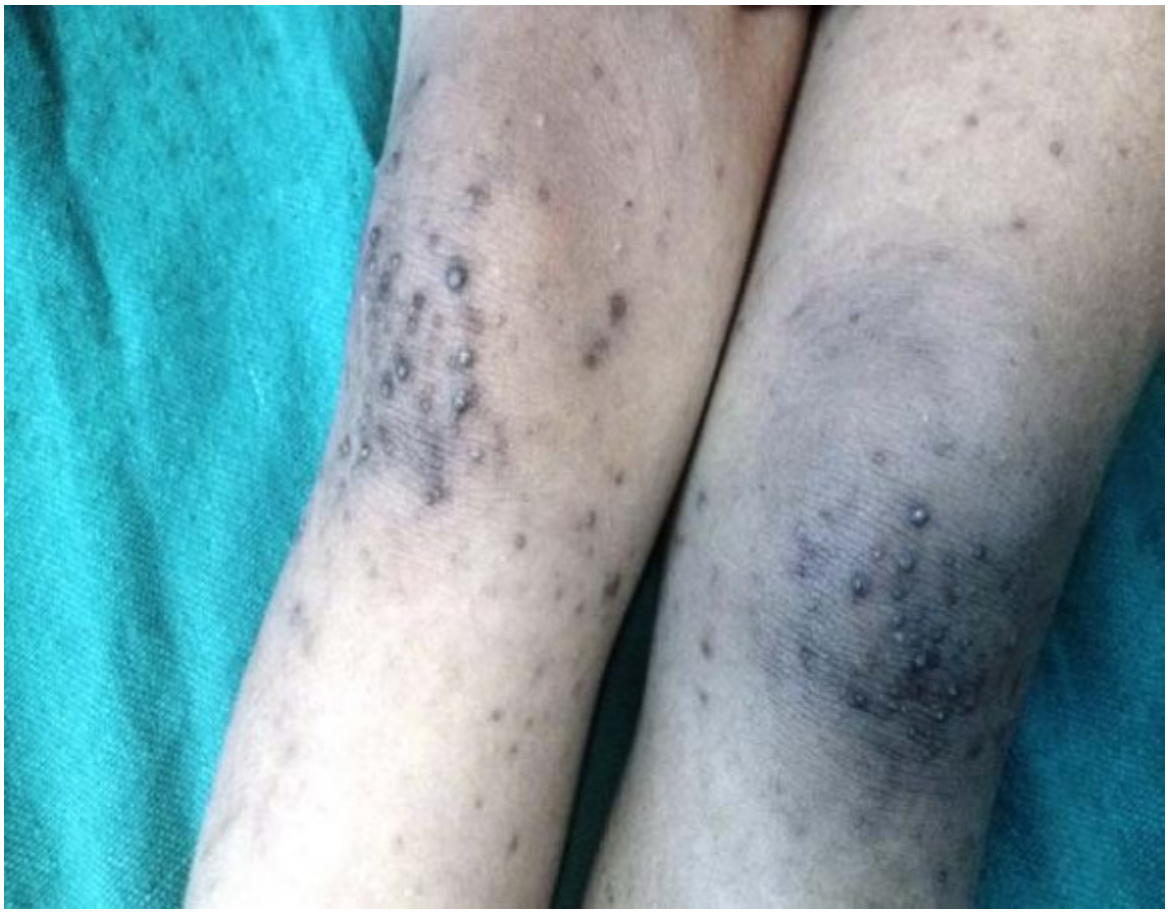

Figure 4:

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