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PACHYONYCHIA CONGENITA TYPE I

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A case of pachyonychia congenita Type I, a rare disorder is presented. There was no oral leucokeratosis and the family history was also negative.

Key words : Pachyonychia congenita.

Introduction

Pachyonychia congenita is a rare genodermatosis first described by Jadassohn and Lewandowsky in 1906.[1] It has an autosomal dominant inheritance with incomplete penetrance. However a recessive pattern of inheritance has also been described.[2] It is included among the first subgroup (1-2-3-4) of the ectodermal dysplasias.[1] Based on the clinical findings, four distinct types are described albeit with several features in common.[2] We describe a case who had features of the Type I syndrome.

Case Report

A 22-year-old male born out of a non-consanguineous marriage presented with thickening of all the nails since birth and palmoplantar keratoses of 6 years duration.

Examination revealed brownish discoloration and marked thickening of the nail plate with subungual hyperkeratosis of all 20 nails (Fig 1). The palms and soles showed marked hyperkeratosis of the pressure areas and scaly keratinous plaques along the medial border of the great toes and sides of the finger (Fig 2). There were multiple, skin-coloured, follicular papules distributed over the extensor surface of the elbows, knees, buttocks, hands and feet. Systemic examination did not reveal any abnormality.

There was no history of palmoplantar hyperhidrosis, bullous eruptions, mucosal lesions, premature eruption of teeth.

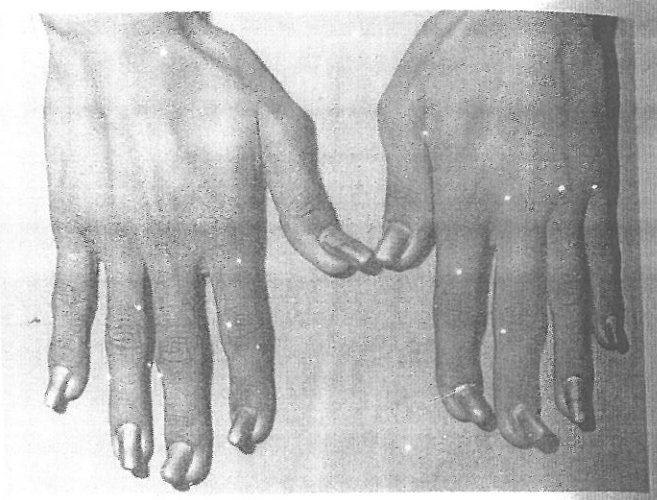

Fig. 1. Marked thickening and discolouration of nails.

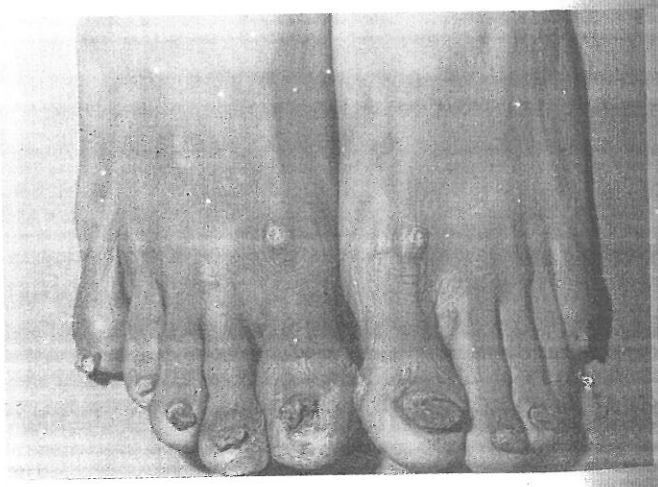

Fig. 2. Scaly plaques along the sides of great toes.

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epidermal cysts or macular pigmentation. The teeth, hair and eyes were unaffected. There was no family history of a similar disorder.

A potassium hydroxide examination of the nail clippings for hyphae and culture for fungus were negative. Histologic examinations of follicular papules from the knee showed hyperkeratosis, follicular plugging and mild acanthosis. The dermis was uninvolved. These features were suggestive of keratosis pilaris.

Discussion

Nail thickening due to orthokeratotic hyperkeratosis of the nail bed is the main feature of pachyonychia congenita.[2] It is often seen in association with several other diseases.[3-5] Because of the protean expressions of the syndrome, several classifications have been proposed.[6] Feinstein et al[7] based on a study of 168 patients have classified it into 4 types : Type I which consists of thickening of nails, palmoplantar keratosis, follicular keratosis and oral leukokeratosis with a relative prevalence of 56.2%. Type II in which in addition to type I changes, palmoplantar bullae, hyperhidrosis, natal or neonatal teeth and steatocystoma multiplex are seen. It has a relative prevalence of 24.9% Type III has a relative prevalence of 11.7% and clinical findings of type II with angular cheilosis, corneal dyskeratosis and cataracts in addition. Type IV has laryngeal lesions, hoarseness, mental retardation, hair anomalies and alopecia in addition to findings of the type III syndrome. The relative prevalence of this type is 7.2%.

Our patient had features which best fit those of type I pachyonychia congenita. Besides the rarity of the disorders, the point of interest in our case are non development of oral leucokeratosis uptil the third decade of life and negative familiy history of a similar disorder.

References

1. Norton LA. Genetic Nail Disorders. In : Genetic Disorders of the skin (Alper JC, ed), 1st edn. St. Lousis: Mosby Year Book, 1991; 195.
2. Dawber RPR, Baran R. Disorders of Nails. In: Text book of Dermatology (Champion RH, Burton JL, Ebling FJG, eds), 5st edn. Oxford : Blackwell scientific publications 1992; 2497.
3. Sarojini PA, Gopalakrishnan Nair TV, Basheer AM, Khaleel S. Pachyonychia congenita with abnormalities of the hair. Ind J Dermatol Venereol Leprol 1977; 43 : 168-9.
4. Ramesh V, Saxena U, Misra RS. Pachyonychia congenita and mental deficiency. Ind J Dermatol Venereol Leprol 1988; 54 : 150-2.
5. Sharma VK, Sharma R, Kaur S. Pachyonychia Congenita with Tuberous Sclerosis. Int J Dermatol 1989; 28 (5) : 332-3.
6. Iraci S, Bianchi L, Gatti S, Carrozzo AM, Bettini D, Nini G. Pachyonychia Congenita with late onset of nail dystrophy- a new clinical entity ? Clin Exp Dermatol 1993; 18 : 478-80.
7. Feinstein A, Friedman J, Schewach-Mellet M. Pachyonychia congenita. J Am Acad Dermatol 1988; 19 : 705-11.