

Egyptian Dermatology Online Journal

Volume 6 Number 2

Pachyonychia congenita type 1- Jadassohn Lewandowsky syndrome

Tejinder Kaur; Rajesh Rani Gupta; BB Mahajan; Rajiv Sachdeva

Department of Dermatology and Venereology, Institute- Guru Gobind Singh medical College and Hospital, Faridkot, Punjab, India

Egyptian Dermatology Online Journal 6 (2): 15

Corresponding Author: Dr. Tejinder Kaur,

E-Mail: tejinderkaurdr@gmail.com

Submitted: July 25, 2010

Accepted: Oct. 17, 2010

Abstract

Pachyonychia congenita (PC) is a rare, but well characterized autosomal dominant disorder of keratinization in which different mutations affect the genes of keratins K6a, K6b, K16 and K17. A three year old male child presented with over curvature of nails and spiny papules over the body since early infancy. Further examination revealed small area of leukokeratosis in the mouth and palmoplantar keratoderma. There was no hair and laryngeal involvement which can be due to incomplete penetrance of genes. The case is being reported for its rare occurrence.

Introduction

Pachyonychia congenita (PC) is a group of inherited ectodermal dysplasias in which the most prominent feature is hypertrophic nail dystrophy [1]. Muller and Wilson described the first case of pachyonychia congenita (PC) and one year later similar case was reported by Jadassohn and Lewandowsky [2]. There are mainly two types of pachyonychia congenita (PC): PC1 (Jadassohn Lewandowsky syndrome) and PC2 (Jackson Lawler syndrome). Here we report a case of three-year-old male child with features suggestive of Jadassohn Lewandowsky syndrome.

Case report:

A three-year-old male child born to non-consanguineous parents presented with papular, scaly lesions over the trunk with thickening of all nails and soles at pressure points since early infancy. Patient had burning sensation in the tongue and mild hoarseness of voice, for the last six months. History of mild hyperhidrosis was present and developmental milestones were normal. There was no history of similar complaint in the family. Systemic examination revealed no abnormality. On cutaneous examination: generalized, follicular, erythematous, papular lesions

were present mainly over the upper trunk, elbows, knees, buttocks, and anterior ankles (Fig. 1). Thick, yellowish, hyperkeratotic plaques were present over the pressure points of the soles (Fig. 2). Palms also showed thick, hyperkeratotic papules. On nail examination: all the twenty nails were wedge shaped, yellow, and thickened with subungual hyperkeratosis (Fig.3). In the oral cavity: angular cheilitis and whitish plaque was present over the dorsum of the tongue extending on to the lateral margins (Fig.4). Otolaryngeal, ophthalmic and hair examination was normal.

All routine investigations were within normal limits. KOH examination of nails was negative for fungus. Biopsy of the lesions could not be done due to unwillingness of the parents. With all these clinical findings, diagnosis of PC1 (Jadassohn Lewandowsky syndrome) was made and patient was put on oral vitamin A, keratolytic agents and emollients.



Fig 1: Showing follicular keratosis on the knees.



Fig 2: Showing focal keratoderma on soles.



Fig 3: Showing pachyonychia in hands and feet.



Fig 4: Showing leukoplakia on tongue and angular cheilitis of mouth.

Discussion

Pachyonychia congenita (PC) is a rare, autosomal dominant disorder characterized by triad of subungual hyperkeratosis with accumulation of hard keratinous material beneath the distal portion of the nails, lifting the nails from the nail bed, keratosis palmaris et plantaris with thick callosities, especially on the soles and thick white areas on the oral mucosa[3]. Other associated features which may occur include keratosis pilaris, hyperkeratotic follicular papules on the sites of friction, hair abnormalities and hyperhidrosis of the palms and soles. These disorders have been suggested to be due to mutations in paired keratins, K6a/K16 (in PC1) and K6b/K17 (in PC2) [4,5]. According to these mutations, various clinical variants have been described [6]:

Type 1- PC1: Jadassohn Lewandowsky syndrome which is characterized by focal palmoplantar keratoderma (PPK), follicular keratosis mainly on the trunk and oral leukokeratosis.

Type 2-PC2: Murray Jackson Lawler syndrome is the most common form associated with mild focal PPK, pili torti, natal teeth and multiple epidermal cysts. Angular cheilitis, hoarseness, follicular keratosis may be present in both types,

Type 3: includes combined features of types 1 and 2 with angular chielitis, corneal dyskeratosis, and cataracts.

Type 4: includes features of type 1 and type 3 with laryngeal lesions, hoarseness of voice with mental retardation, hair abnormalities and alopecia.

Other rare variants include pachyonychia congenita with only nail involvement and pachyonychia tarda that is pachyonychia congenita with onset in teenage years [7,8]. Complications like respiratory distress due to laryngeal leukokeratosis and acro osteolysis, malignant changes in palmoplantar lesions can occur in pachyonychia congenital [6].

Treatment options for PC fall into four broad categories:

- (1) Non- invasive (mechanical) e.g. abrasion with some hand tool
- (2) Invasive (surgical) e.g. electrofulgration, excision
- (3) Chemical methods using urea, propylene glycol, alpha hydroxy acid
- (4) Pharmacological (vitamin A, retinoids), all basically targeted at reducing the hyperkeratosis involving different sites [9].

When the familial mutation is known, genetic counseling can be done and if required, prenatal diagnosis can be done at early stage of pregnancy by chorionic villi biopsy [10].

For all these reasons, a patient with pachyonychia congenita should be thoroughly investigated and treated accordingly.

References

1. Irvine AD, McLean WH. Human keratin diseases: the increasing spectrum of disease and subtlety of the phenotype-genotype correlation, *Br J Dermatol* 1999; 140: 815- 828.
2. Feinstein A, Friedman J, Schewach-Millet M, Pachyonychia congenita, *J Am Acad Dermatol* 1998; 19: 705- 711.
3. Johnson BL Jr, Yan AC. Congenital diseases (Genodermatosis). In: Elder DE (eds). *Lever's Histopathology of the Skin*, 10th edition. Philadelphia, Lippincott Williams & Wilkins, 2009: P138.
4. McLean WH, Rugg EL, Lunny DP et al., Keratin 16 and keratin 17 mutations cause pachyonychia congenita, *Nat Genet* 1995; 9: 273- 278.
5. Leachman SA, Kaspar RL, Fleckman P et al., Clinical and pathological features of pachyonychia congenita, *J Invest Dermatol Symp Proc.* 2005; 10: 3- 17.
6. Muruges SB, Reddy S, Ragunatha S et al., Acro-osteolysis: a complication of Jadassohn-Lewandowsky syndrome. *Int J Dermatol* 2007; 46: 202- 205.
7. Bansal A, Sethuraman G, Sharma VK. Pachyonychia congenita with only nail involvement, *J Dermatol* 2006; 33(6): 437- 438.
8. Paller AS, Moore JA, Scher R. Pachyonychia congenital tarda: a late onset form of pachyonychia congenita, *Arch Dermatol* 1991; 127: 701- 703.
9. Milstone LM, Fleckman P, Leachman SA et al., Treatment of pachyonychia congenita, *J Invest Dermatol Symp Proc.* 2005; 10(1): 18- 20.
10. Smith FJ, McKusick VA, Nielsen K et al., Cloning of multiple keratin 16 genes facilitates prenatal diagnosis of pachyonychia congenita type 1, *Prenat Diagn.* 1999; 19(10): 941- 946.