Pachyonychia congenita with woolly hair in a ten month old infant

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ABSTRACT

A 10-month-old female presented with severe progressive wedge-shaped thickening and discoloration of all twenty nails. Further evaluations revealed palmoplantar keratoderma along with recurrent acral blisters causing residual crusted ulcers which were present during the past six months. Other findings included scalp kinky hair and dental caries. Past medical and family history had remarkable findings such as natal teeth and similar skin lesions in her older brother since his infancy. The patients' clinical presentations and history are compatible with pachyonychia congenita presenting with concomitant features of both subtypes 1 and 2.

Key Words: Keratinization disorder, Natal teeth, Pachyonychia congenita, Woolly hair

INTRODUCTION

Pachyonychia congenita (PC) is a group of rare genodermatoses first described by Jadassohn and Lewandowsky in 1906.[1] PC is characterized by hypertrophic nail dystrophy and associated ectodermal features for which, based on the clinical features, numerous subdivisions have been suggested so far. There are two main PC subtypes, which can be readily distinguished under their variable clinical presentations and etiologic gene mutations. Furthermore, both autosomal dominant and recessive forms, as well as phenotypic variations, have been reported.[2]

CASE REPORT

A 10-month-old female infant presented with severe progressive wedge-shaped thickening and discoloration of all 20 nails. These changes appeared after her neonatal period [Figure 1]. Her mother also noticed blister formation due to minor trauma on her legs, along with residual ulceration and hyperpigmented scar [Figure 2]. On examination severe yellowish brown hypertrophic nails with wedge-shaped subungual hyperkeratosis were noted. Her palmoplantar hyperkeratosis showed progression during the previous 5 months. Other signs included woolly hair on scalp [Figure 3] and several small intact bullae surmounted by crusts found in her foot and thigh region. She was a healthy baby with physical and mental growth criteria appropriate for her age. The patient's history revealed natal teeth and similar blister formation in her elder brother's legs since infancy, which disappeared after puberty. Other family members were free of this clinical condition. The clinical findings were consistent with the diagnosis of pachyonychia congenita in this case.

DISCUSSION

The term pachyonychia congenita refers to a group of rare variable phenotypic presentations, frequently with autosomal dominant pattern of inheritance. Characteristic nail changes in this syndrome include distal onycholysis, subungual hyperkeratosis, and variable discoloration. PC type I (Jadassohn-Lewandowsky, PC-I) consists of palmoplantar hyperkeratosis, follicular hyperkeratosis, and

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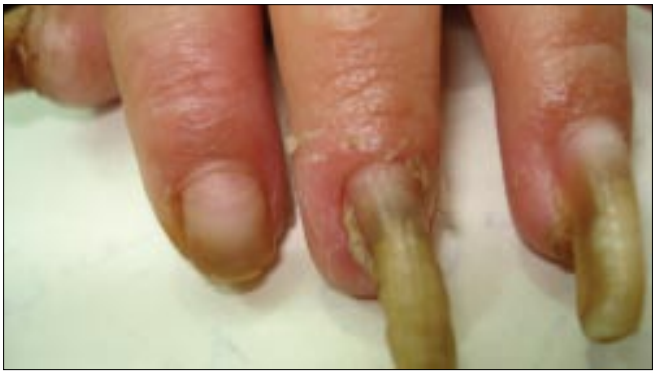

Figure 1: Severe wedge-shaped hypertrophic nails

Figure 3: Woolly hair on scalp

oral leukokeratosis. Occasionally, bullous lesions, hoarse voice due to laryngeal involvement, warty lesions on knee and elbow, and hyperhidrosis may occur.

In PC type II (Jackson-Lawler, PC-II) the palmoplantar keratoderma and oral changes are of less importance or are absent. In addition, history of natal teeth and the development of epidermal cysts or steatocysts are remarkable.[3]

PC type III (Schafer-Brunauer, PC-III) has the features of PC-I in addition to corneal leukokeratosis. In the fourth type, isolated typical nail changes begin in the second and third decades of life, and it has been described as pachyonychia congenita tarda. It is however important to mention that there have been reported cases which do not fulfill the criteria of a particular type based on the current

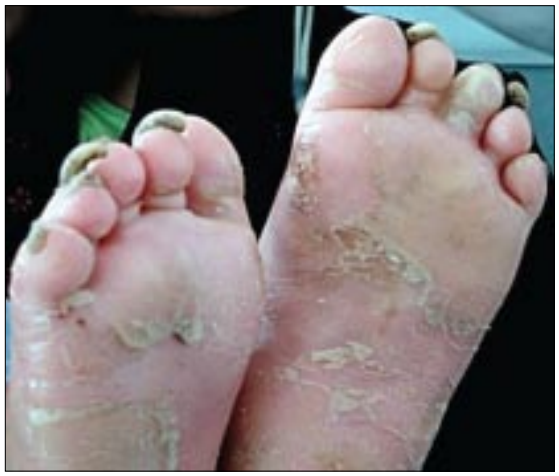

Figure 2: Acral blisters with consequent ulceration and scar

classification.[4] Histology and ultrastructural evaluations of PC lesions suggest that PC is a keratinization disorder. Its different presentations are currently known to be due to mutations in variable genes encoding one of the paired epidermis keratins, K6a/K16 in PC-I and K6b/K17 in PC-II.[5] The case presented here demonstrates typical PC type I clinical features, along with some other presentations which are characteristic of PC-II, such as natal teeth and woolly hair. These findings suggest putative multiple mutations in K6, K16, and K17 genes as the underlying cause of varying clinical presentations of this case and should prompt further genetic investigations of the disease.

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