

Case Report

PACHYONYCHIA CONGENITA– CASE REPORT

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ABSTRACT

This is a congenital nail disorder with an autosomal dominant inheritance in which misshapen, hypertrophic nails are present from birth. There may also be associated mucous membrane abnormalities. At areas prone to surface friction in the lower limb, hyperkeratosis is a common sequel. Pachyonychiacongenita affects the skin (especially palms and soles), nails, hair and mucous membranes. Specific features depend on which keratin gene is involved. We are reporting a case of Pachyonychiacongenita in an infant.

Keywords: *Pachyonychiacongenita, Hypertrophic Nails, Autosomal Dominant*

INTRODUCTION

Pachyonychiacongenital (PC) is a rare genodermatosis due to mutations in one of 4 keratin genes. It is characterized by dystrophic, thickened nails and painful palmoplantarkeratoderma. Müller made one of the first documented observations of pachyonychiacongenita in 1904 (Muller, 1904). The next reports were published in 1905 by Wilson (1905) and in 1906 by Jadassohn and Lewandowsky (1906). Although the exact frequency of pachyonychiacongenita is unknown, it appears to be rare. An estimated 5,000–10,000 cases have been reported worldwide (Kaspar, 2005).

CASES

A 3 month old female child born as a second issue to second degree consanguineous parents presented with complaints of hurried breathing since 2 days and refusal of feeds for 1 day. On admission child was in distress with a heart rate of 190 per minute and respiratory rate of 66 per minute. Capillary refill time was prolonged and SpO₂ at room air was 78%. Investigations revealed hemoglobin-5.4 gm/dl, Total leucocyte count – 38,300 cells/cu mm, Polymorphs – 84%, Lymphocytes- 16%, Platelets – 78,000/cumm. Liver function tests were normal, BUN – 96 mg/dl, Serum creatinine – 1.2 mg/dl. Serum electrolytes were normal. Blood culture isolated *Klebsiella*. Urine culture has shown significant bacteriuria (*E.coli*). Pus culture from the lesion isolated multiple varieties of species of bacteria. KOH mount of pus from lesion isolated *Candida albicans* and *Cryptosporidium*.



Figure 1: The most prominent feature is a substantially thickened, brownish gray nail plate with a rough surface

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Figure 2: Hyperkeratotic lesions of the sole

DISCUSSION

Pachyonychia congenita is characterized by hypertrophic nail dystrophy, painful palmoplantar-keratoderma and blistering, oral leukokeratosis, pilosebaceous cysts (including steatocystoma and vellus hair cysts), palmoplantarhyperhydrosis, and follicular keratoses on the trunk and extremities.

The disorder has historically been divided into 2 main subtypes. Pachyonychia congenita type 1, or the Jadassohn-Lewandowsky type (Mendelian Inheritance in Man (MIM entry 167200), was attributed to mutations in genes encoding keratin 6A (KRT6A) or keratin 16 (KRT16) and constituted the most common form of the disorder (McKusick, 1994). Pachyonychia congenita type 2, or the Jackson-Lawler type (MIM entry 167210), was attributed to mutations in keratin 6B (KRT6B) or keratin 17 (KRT17) and could be distinguished from type 1 by the development of natal teeth, widespread steatocystomas, and occasionally pili torti.

PC is diagnosed by clinical findings and by molecular genetic testing. Mutations in the keratin genes KRT6A, KRT6B, KRT16, and KRT17 are known to cause PC. Like most genodermatoses, no specific treatment or cure is known for pachyonychia congenita. Therapy is generally directed towards symptomatically improving the most bothersome manifestations of the disease and, because of the rarity of pachyonychia congenita, is based largely on anecdotal findings. A clinical report of 254 patients from the International Pachyonychia Congenita Research Registry (IPCRR) represents the largest phenotype-genotype study of the disorder to date (Eliason *et al.*, 2012). We had an infant with PC with secondary infection and infant died of complications of PC.

Conclusion

Due to rarity of this condition recognition of the same is very important to prevent the complications.

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