

## Pachyonychia Congenita With Unusual Features

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Pachyonychia congenita is a rare hereditary disorder characterized by gross thickening of all finger and toenails. We report an infant who had clinical features consistent with pachyonychia congenita type II, with unusual features of microcephaly, seizures, electroencephalogram abnormalities, failure to thrive, and heterochromia iridis.

**Key words:** Congenital dyskeratosis, Nail dystrophy, Pachyonychia congenita

**P**achyonychia congenita (PC) is a rare hereditary disorder characterized by nail dystrophy, palmar and plantar hyperkeratosis, leukokeratosis of the mucous membranes, follicular keratosis, and occasional hyperhidrosis of palms and soles(1). We report an infant with PC type II, who also had unusual features.

### CASE REPORT

An 11-month old male infant, 1<sup>st</sup> born of 2<sup>nd</sup> degree consanguineous parentage presented with abnormal nails and skin lesions since birth, along with delayed motor and mental milestones and poor weight gain. There was history of seizures at 3<sup>rd</sup> month of life. There was no similar nail and skin changes in any other family member in two generations. There was no history of natal teeth. Anthropometric measurements revealed grade II protein-energy malnutrition (IAP classification) and microcephaly (head circumference 42 cm). The infant had sparse and thin hair over the scalp, heterochromia of iris, low set ears, angular cheilosis, with a hoarse voice and stridor. All the fingernails and toenails were dystrophic, discolored, and thickened, with massive subungual hyperkeratosis producing distal elevation of nail plates with wedge-shaped deformity of the

nails (**Fig. 1** and **2**). He also had pin-head sized follicular papules over the elbows, knees, popliteal and gluteal areas. Marked hyperhidrosis was observed in the palms and soles. Oral cavity was normal with 4 primary teeth. Neurological examination revealed spasticity of all the limbs, with scissoring of lower limbs, dystonia and pseudo-bulbar palsy. A complete hemogram, serum biochemistry and liver enzymes were normal. Urine metabolic screening tests were negative. Karyotyping was normal. Electroencephalogram was abnormal with evidence of seizure activity from left hemisphere. Computed tomography (CT) of head revealed cerebral atrophy. Direct laryngoscopy showed moderate degree of laryngomalacia with mild congestion of vocal cords. He was treated with haloperidol, sodium valproate, emollients and parents were counseled regarding rehabilitation.

### DISCUSSION

Pachyonychia congenita has both autosomal dominant and autosomal recessive forms of inheritance, reflecting heterogeneity(2). PC encompasses a range of inherited ectodermal dysplasias, divided into two main subtypes, Pachyonychia congenita type 1 (PC-1, Jadassohn-Lewandowski syndrome) and Pachyonychia



**FIG. 1** Nails grown to full length with an upward slant caused by the predominant distal hyperkeratosis giving 'wedge-shaped' nails.



**FIG. 2** Thickened and dystrophic toenails.

congenita type 2 (PC-2, Jackson-Lawler syndrome), which can usually be distinguished by clinical examination. Considerable overlap can occasionally exist between PC-1 and PC-2, which can make diagnosis difficult. This most recent classification of PC is based on clinical findings and by molecular genetic testing(3).

The predominant and most common clinical feature in PC is hypertrophic nail dystrophy. Other findings common to PC-1 and PC-2 are: focal palmoplantar keratoderma, blistering, oral leukokeratosis, and palmoplantar hyperhidrosis, follicular keratosis on trunk and extremities, and pilosebaceous cysts. Other findings observed in PC-2 are steatocystoma which normally develops at puberty, pili torti (hair anomalies) and natal teeth(3). The pattern of Pachyonychia congenita correlates well with the keratin gene mutation. The two keratin genes known to be associated with PC-1 are KRT6A and KRT16 and the two keratin genes known to be associated with PC-2 are KRT6B and KRT17(3). PC should be differentiated from traumatic thickening of nails and from congenital onychogryphosis, which are easy to recognize because they do not involve all nails and are not associated with dyskeratotic skin lesions. Based on the clinical manifestation, our case appear to be type II, as per recent classification(3). Hoarseness is a feature of PC-1 but may also be seen in PC-2 along with unruly hair. The relationship of mental retardation to Pachyonychia congenita is not clear. It has been suggested that the fetal ectodermal

lesions that affect the skin may also affect the central nervous system(4). Our case in addition had other unusual features namely microcephaly, seizures(1), electro-encephalogram abnormalities(5) and failure to thrive(4), which have been described in few previous isolated case reports. However, a combination of all these unusual features has not been described earlier. The presence of heterochromia iridis may be another new association of pachyonychia congenita or may just be a chance occurrence.

Treatment of PC primarily provides symptomatic relief. Emollients, keratolytics, topical retinoids and oral retinoic acid are used to treat palmoplantar hyperkeratosis(6). The only effective treatment for nail lesions is surgery with radical excision of the nail, nail bed, and nail matrix, and skin implantation at the site of the removed nail(7).

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