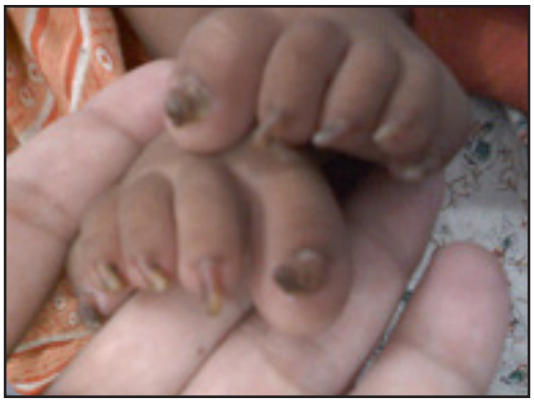
SPOT DIAGNOSIS (IMAGE GALLERY)



NAIL DISORDER

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A 14 months old male child youngest of 2 siblings, product of non consanguineous parentage presented with abnormality of all fingernails and toenails since birth. There were no similar nail changes in any other family member in two generations. There was no history of natal teeth. Anthropometrically and developmentally the child was within normal limit age. On examination, all the fingernails and toenails were dystrophic, discolored, and thickened, with subungual hyperkeratosis causing distal elevation of nail plates with wedge-shaped deformity of the nails {Fig}. Oral cavity was normal. On investigation complete hemogram, renal function test, liver function test and urine routine were normal. Parents were couselled regarding available treatment options and asked for regular follow-up.

What is the diagnosis?

Pachyonychia congenita affecting only nails.

Pachyonychia Congenita {PC} is a rare genodermatosis which is characterized by nail dystrophy, palmar and plantar hyperkeratosis, leukokeratosis of the mucous membranes, follicular keratosis, and occasional hyperhidrosis of palms and soles {1}. PC has predominantly autosomal dominant inheritance with incomplete penetrance, although Haber and Rose described cases with autosomal recessive transmission {2}. The presence of thickened wedge shaped nails is the diagnostic clinical feature of Pachyonychia congenita. PC has been divided into two main subtypes, Pachyonychia congenita type 1 {PC-1, Jadassohn-Lewandowski syndrome} and Pachyonychia congenita type 2 {PC-2, Jackson-Lawler syndrome}, which can usually be distinguished by clinical examination. Pathogenic mutations in keratin K6a, K16 are associated with PC-1 phenotype whereas K6b and K17 mutations are associated with the PC-2 phenotype. Jadassohn-Lewandowski syndrome {PC-1} shows focal palmoplanter hyperkeratosis and localized foot blistering, follicular hyperkeratosis and oral leukokeratosis.

Jackson-Lawler syndrome {PC-2} shows cutaneous cyst, hair abnormalities and presence of teeth at birth but less severe nail lesion and neither oral lesion nor keratoderma {3}. Very few cases of Pachyonychia congenita only with nails involvement are reported from India {4,5}. Emoliants, keratolytic agents, topical retinoids and oral retinoic acids derivatives are used to treat palmo-planter hyperkeratosis {6}. The only effective treatment for nail lesions is surgery with radical excision of the nail, nailbed, and nail matrix, and skin implantation at the site of the removed nail {7}.

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