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HEREDITARY ONYCHO-OSTEO DYSPLASIA SYNDROME

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We report a two and a half year old male child with dystrophy of all the nails, absent patellae and iliac horns. In addition he had dysmorphic facial features, sparing of lunula and bilateral hallux valgus deformity, hitherto not reported earlier.

Key Words : Hereditary onycho-osteo dysplasia, Dystrophic nails, Iliac horns, Dysmorphic face

Introduction

Hereditary onycho-osteo dysplasia syndrome (HOODS) or nail-patella syndrome (NPS), is a rare autosomal dominant disorder affecting tissues of both ectodermal and mesodermal origin. The diagnostic triad comprise of dystrophic nails, aplasia or hypoplasia of patella and iliac horns.[1-4] It has been found to be strongly linked to ABO blood group locus.[2]

The clinical profile of this syndrome is so variable that with each new family reported some additional features come to light. We report a case which had dysmorphic facial features and hallux valgus deformity.

Case Report

A two and a half-year-old male child was brought with complaints of improper development of nails since birth. He was the first child to consanguineous parents who were second degree cousins. The mother suffered from seizure disorder for which she received phenobarbitone throughout the pregnancy. The natal and the postnatal periods were uneventful. No other family member in the last three generations had similar defects. His development has been normal.

The examination revealed a dysmorphic face with hypertelorism (interinnercanthic distance 3.5 cm), bilateral epicanthic folds, depressed nasal bridge, high arched palate, low set ears and a protruding chin. There was no midline cleft. The eyes, ears and hair were normal.Both the nipples were inverted. The weight, height and head circumference were within 50-60th percentile for age. Blood pressure was normal.He had mild genu valgus and bilateral hallux valgus. The upper limbs were normal. The patellae were absent on both sides. The examination of respiratory, cardiovascular system, abdomen and central nervous system did not reveal any abnormality except for great difficulty encountered in eliciting knee jerks.

The nails of the thumbs and index fingers of both hands were small and hypoplastic. The middle finger nails revealed koilonychia with horizontal splitting. The nails of the ring fingers had platynychia with vertical splitting and ridging. The nails of little fingers were small with vertical splits and ridges (Fig.1). The lunula in all the nails was normal.

The toe nails of right foot were small and hypoplastic with a horizontal split in the fifth. The left great toe nail revealed pits with discolouration while the second had pits with koilonychia while the third and the fourth toe had platynychia. The fifth left toe nail was small and hypoplastic (Fig.1)

The investigations revealed normal haemogram and normal levels of blood sugar, urea, and serum creatinine, sodium,

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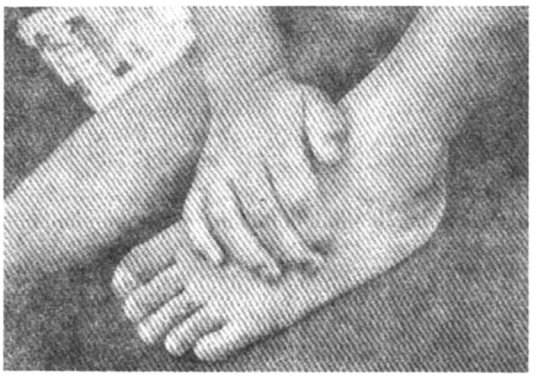

Fig. 1. Dystrophic nails of left foot and right hand.

potassium, bicarbonate, proteins, lipids, GOT, GPT, bilirubin, C_3, C_4, ANF and rheumatoid factor. Examination of urine and stool did not reveal any abnormality. X-ray of the chest and elbow were normal. X-ray of pelvis revealed bilateral iliac horns. Chromosomal analysis revealed a normal karyotype.

Discussion

Hereditary onycho-osteodysplasia is characterized by dysplasia of nails and patella. Nails have been found to be affected in all cases of HOODS, especially of thumbs and index finger. Nails might be small, absent, flat spongy with ridges and pits.[1-2] The lunula of the nails might be tented or triangular.[2] This has been considered by some authors as a sensitive screening test for HOODS.[2] In the present case, in contrast to the cases reported earlier, all the finger and toe nails were affected and strikingly the lunula was normal in all the nails.

The common skeletal abnormalities reported with HOODS include, absence or hypoplasia of patella and iliac horns, hypoplasia of radial head, capitulum and epicondyle.[1-4] Patella may be small, absent, subluxated, fractured or partitioned.[1-3] Iliac horns are triangular bilaterally symmetrical bony crescents arising from the posterior surface of the iliac bones unrelated to the muscular attachments. These are detected in X-ray of pelvis and known as "Fong's sign" These have been found to be present at birth in 8% of cases.[2,4]

A distinct progressive lethal nephropathy has been found to develop in one third of cases of HOODS with proteinuria, haematuria, hypertension, inability to concentrate urine, and uremia over 5-25 years.[2,4] The pathological features reported in such case include thickening of glomerular basement membrane with lucent areas containing fibrils of cross-striated collagen bundles, increased mesangial matrix, atrophic tubules with IgM and C_3 deposits on glomerular basement membrane.[3] There have been other occasional associations reported with HOODS like shortening of muscles, mental retardation, ocular abnormalities,[1-3] vascular dysplasia of cerebral and aortic vessels.

In the present case, consanguinity of parents suggested a recessive mode of inheritance, however, a definite mode may be obvious only on further follow-up of future generations.

The present case is distinct,in involvement of all the nails with sparing of lunula, presence of dysmorphic facial features and hallux valgus deformity. Moreover, the significance of phenobarbitone in the development of NPS in our case is difficult to ascertain in the absence of any previous report implicating phenobarbitone in inducing changes in vivo in ectodermal and mesodermal tissues.

In our opinion, HOODS may be included under subgroup B-3, of Freire-Maia's classification of ectodermal dysplasia; a large

heterogenous group of disorders of structures of ectodermal origin, since under this subgroup another condition with onycho-osteo dystrophy is already included; DOOR Syndrome (deafness, onychoosteo dystrophy and mental retardation).

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