Iso-Kikuchi syndrome with absence of ring fingers and metacarpal bone abnormality

Sir,

Iso-Kikuchi syndrome (I-K S), congenital onychodysplasia of the index fingers (COIF), is a rare condition characterized

by various forms of nail dysplasia commonly involving the index fingers. Not infrequently, the neighboring fingers such as the middle fingers and thumbs are also affected.[1] A case of COIF in a 34-year-old female patient is reported here for its rarity.

A 34-year-old female patient, born of a consanguineous marriage, presented with generalized bodyache. On examination she was found to have 4 digits bilaterally and absent nail on the right index finger, while the left index finger showed a rudimentary nail. There was micronychia on the middle fingers bilaterally, being more marked on the right side [Figures 1-2]. Lunula was absent in all nails. Palmar creases were normal. She could not flex her right index finger. All toe nails were normal. Her hair and mucous membranes were normal. She had normal dentition. Systemic examination did not reveal any abnormality. An X-ray of both the hands showed normal carpal bones and five metacarpal bones. The phalanges of the ring fingers were absent. The third and fourth metacarpal bones were found articulating with a single large proximal phalanx of the middle finger. Except for an elder brother, who had syndactyly bilaterally, her 3 other siblings were normal. Our patient had 2 children, who were unaffected. The patient gave a history that her mother had consumed an abortifacient while she was in utero.

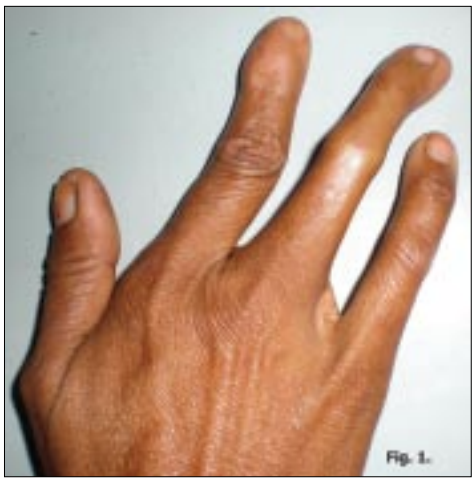

Figure 1: Photograph showing right hand with four digits, absent nail on the index finger, with marked micronychia of the middle finger on the right side

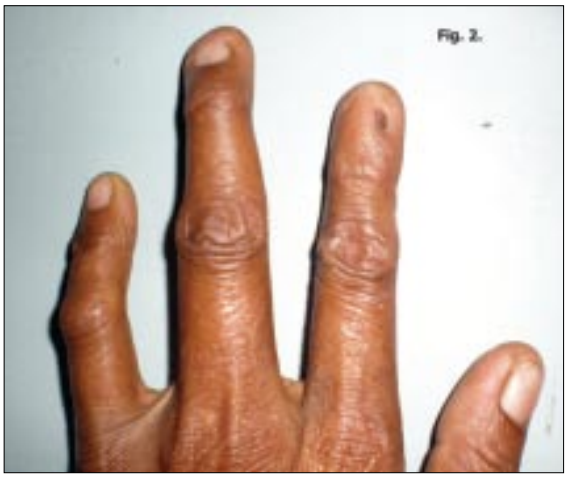

Figure 2: Photograph of left hand showing four digits, a rudimentary nail on left index, and micronychia on the middle fingers

Ichiro Kikuchi *et al.,*[2] in 1974, coined the term 'congenital onychodysplasia of the index fingers' (COIF) and identified a clinical syndrome consisting of nail dysplasias of the index fingers associated with underlying bone abnormalities. The first case report of this condition was by Kamei. Later, Iso collected a series of patients and defined the clinical syndrome. The term Iso-Kikuchi syndrome was introduced in 1980 by Robert Baran, a French dermatologist in Cannes.[1]

Iso-Kikuchi syndrome is rarely reported outside Japan, with an international incidence of 4.2 cases per 100,000 live births.[2]

The nails of COIF include the full spectrum of nail dysplasia, from irregular lunula, malalignment, micronychia (hypoplastic and rudimental), polyonychia (split rudimental), to anonychia, specifically affecting the index fingers. In our patient, there was micronychia of the middle fingers, while micronychia and malalignment of the left second toenail has been described by other authors.[3]

The five criteria characterizing COIF include the following: (i) congenital occurrence; (ii) unilateral or bilateral index finger involvement; (iii) variability in nail appearance; (iv) possible familial involvement; and (v) frequently associated bone abnormalities.[3] Our patient satisfied all the five of the above-mentioned criteria. The configuration of the lunula is supposed to play an important role in shaping the free edge of the nail plate.[4] In our patient, lunula was absent in all nails. Hemi-onychogryphosis of both index fingers and bifurcation of the distal phalanges are the other reported anomalies.[5] Our patient had two metacarpal bones articulating with the proximal phalanx of the middle finger, in addition to the absence of ring finger on both sides. The proximal phalanx of the middle finger was large and could be suggestive of syndactyly. The absence of alopecia and palmoplantar keratoderma and normal dentition ruled out ectodermal dysplasia.

In utero, ischemia of the palmar digital artery and a dysplastic

change in the crescent-shaped cap of the distal phalanx are the two main candidate pathogenetic mechanisms that have been proposed.[6] Exposure to teratogens, especially antiepileptic drugs in utero, in mothers with epoxide hydrolase deficiency is another supposed cause of COIF. This probably could explain our patient's predicament, as her mother had consumed an abortifacient when our patient was in utero. Due to patient's noncompliance and lack of facilities, arteriographic studies could not be undertaken.

Iso-Kikuchi syndrome has also been associated with discoid lupus erythematous. As there were filiform arteries of the fingers and slow blood circulation on angiographic studies, the authors considered vascular pathogenic mechanisms to be responsible for this syndrome.[7] However, there was no clinical or laboratory evidence of any connective tissue disease in our patient.

Transmission of COIF can be either hereditary as autosomal dominant or sporadic. In our patient, a positive family history involving her brother could be suggestive of some hereditary involvement. Due to unavailability, chromosomal studies could not be undertaken.

Since COIF is only of cosmetic significance and has not interfered with her day-to-day activities, our patient was reassured and advised physiotherapy for the limitation of movements of the fingers.

The present case is being reported for its rarity and the hitherto unreported abnormality of the metacarpal bones, in addition to the absence of one digit (ring finger) on both sides.

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