

Nail-Patella Syndrome: A Classic Case

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Case Presentation

A 4½-year-old boy presented with a delayed appearance of all fingernails and near absence of both thumbnails, present since birth, along with incomplete extension of the bilateral elbows. On examination, the bilateral thumbnails were hypoplastic and the index fingers of both hands showed tenting with triangular lunulae. Flexion deformity was noted at the bilateral elbows. A roentgenogram of the pelvis showed bilateral iliac horns, while that of both knees showed absent patella. The rest of the examination was within normal limits. On the basis of the presence of bilateral iliac horns, triangular lunulae, webbing of elbow, and an absent patella (Figure 1, A-D), a diagnosis of nail-patella syndrome was made.

Teaching Point

Nail-patella syndrome presents with a classic tetrad of triangular lunulae, elbow pterygium, absent patellae, and posterior iliac horns. Dermoscopy aids in better visualization of the triangular lunulae.

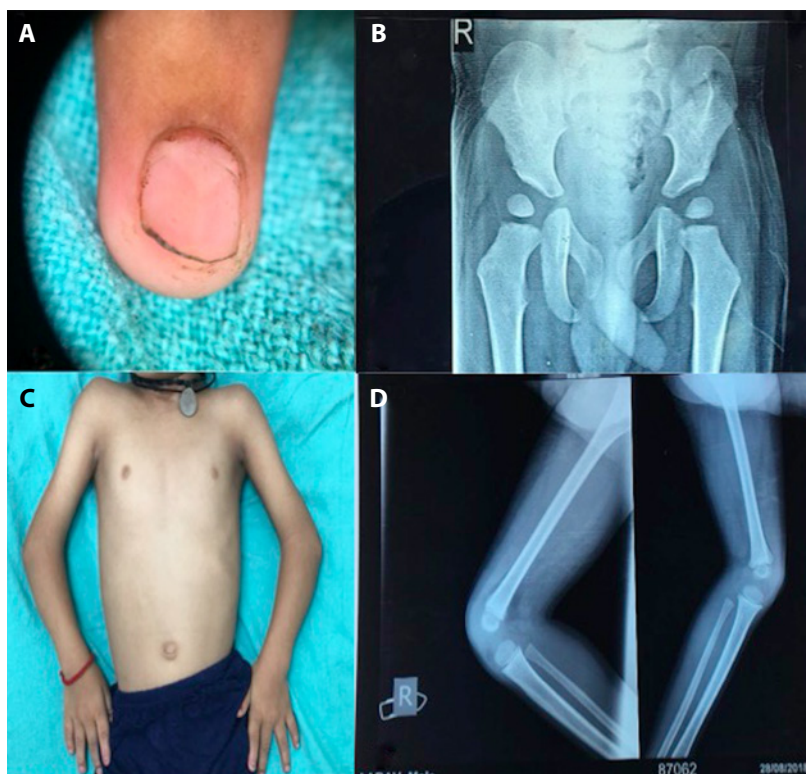


Figure 1. Nail-patella syndrome collage showing (A) triangular lunula on dermoscopy, (B) bilateral posterior iliac horns, (C) bilateral elbow pterygium, and (D) absent patella. [Copyright: ©2019 Sanke et al.]