Liebenberg Syndrome: Case Report and Insight Into Molecular Basis

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We report a case of Liebenberg syndrome in a 6-year-old girl, including the clinical, radiological, angiographic, and operative findings. We note that the forearm and hand malformations have similarities to leg and foot anatomy. Our observations may help provide insight into the etiology of this unusual condition. *(J Hand Surg 2013;38A:459–465.*

Key words Liebenberg syndrome, homeotic transformation.

Liebenberg syndrome is a rare condition characterized by dysplasia of the bony components of elbow, forearm, and hand. Most previous reports described different affected families and pedigrees and shared the same clinical and radiological description of dysplasia of the elbow and forearm, abnormally shaped carpals, and brachydactyly.[1–3] Spielmann et al[4] mentioned that the upper extremities undergo partial homeotic transformation and acquire features of the lower limbs in this syndrome. In this report, we describe a case of Liebenberg syndrome, including detailed clinical, radiographic, angiographic, and intraoperative findings that demonstrate a series of anomalies resembling lower extremity features in the elbow, wrist, and hand.

FIGURE 1: General appearance of the patient with Liebenberg syndrome, with deformities limited to elbows, forearms, and hands.

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Received for publication September 26, 2012; accepted in revised form December 11, 2012.

No benefits in any form have been received or will be received related directly or indirectly to the subject of this article.

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0363-5023/13/38A03-0005$36.00/0
http://dx.doi.org/10.1016/j.jhsa.2012.12.015

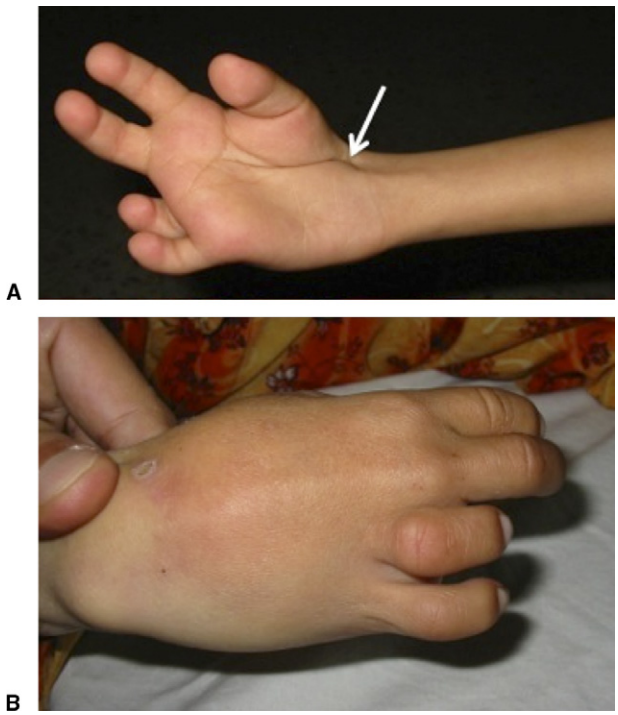

FIGURE 2: The wrist is fixed in radial deviation. **A** A volar bony prominence (arrow) is visible at the level of the wrist, with the tendon attached to it. **B** Clawing of the ulnar digits described in previous reports as camptodactyly or brachydactyly.[1–3]

FIGURE 3: A X-ray and 3-dimensional computed tomography of the elbow. The long arrow points to the bony prominence of the radius resembling the tibial tuberosity. The short arrow points to the proximal end of the ulna. **B** X-ray of normal knee and elbow. The long arrow points to the tibia and its counterpart in the upper limb (the radius). The short arrow points to the fibula and its counterpart in the upper limb (the ulna). This reflects the similarities between the forearm bones in **A** and the leg bones rather than the normal forearm bones in **B**.