Acheiropodia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Acheiropodia. ORPHA:931

Acheiropodia is an extremely rare developmental disorder characterized by bilateral, congenital and complete amputation of the distal extremities (amputation of distal epiphysis of the humerus, distal portion of the tibial diaphysis, aplasia of the radius, ulna, fibula) and aplasia of hands and feet (aplasia of carpal, metacarpal, tarsal, metatarsal and phalangeal bones). Rarely, an ectopic bone can be found at the distal end of the humerus. No other systemic manifestations have been reported and the disorder follows an autosomal recessive pattern of inheritance.