

DISEASE:
Acheiropodia

NAME:	Acheiropodia
DESCRIPTION:	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.
ORPHACODE:	931
SYNONYMS:	Acheiropody
XREF(S):	Orphanet OMIM MeSH ICD-10
ANALYTE(S):	LMBR1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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