

Opis choroby *

Definicja

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.

Dane

Klasyfikacja	Synonimy
Wada morfologiczna	Acheiropody

Kod ORPHA	Kod OMIM	Kod ICD10
931	200500	Q74.8

Kod ICD11
LB9B

*Źródło

orphanet