

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

Wada morfologiczna

Synonimy

Acheiropody

Kod ORPHA

931

Kod OMIM

200500

Kod ICD10

Q74.8

Kod ICD11

LB9B

*Źródło

orphanet