

Opis choroby *

Definicja

Congenital absence of both forearm and hand is a rare developmental defect during embryogenesis characterized by unilateral or bilateral arrest of proximal to distal development of the upper limb, leading to a transverse deficiency with absence of the forearm, wrist and hand. A short below-the-elbow amputation is most commonly observed and the residual limb is usually well cushioned, with rudimentary nubbins or dumpling possibly found on the end.

Dane

Klasyfikacja	Synonimy
Wada morfologiczna	Radio-ulnar terminal transverse meromelia Radio-ulnar terminal transverse meromelia

Kod ORPHA	Kod OMIM	Kod ICD10
294979	-	Q71.2

Kod ICD11
LB99.5

*Źródło

orphanet