Opis choroby *

Definicja

A rare congenital limb malformation syndrome characterized by a highly variable combination of congenital anomalies of the femur, fibula, and/or ulna, which can appear along with finger/toe anomalies at the ulnar/fibular side. Limb defects are asymmetrical, with upper limbs more often affected than lower limbs, and the right side of the body more often affected than the left. Abnormalities of the upper limb include amelia, hypoplasia of the humerus, humero-radial synostosis, and malformation of the ulna and ulnar rays. Abnormalities of the lower limb include absence of the proximal part of the femur and absence of the fibula. Axial skeleton, internal organs and intellectual function are usually normal.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
FFU complex
PFFD
Kompleks FFU
Zespół udowo - strzałkowo - łokciowy
Dyzostoza udowo - strzałkowo - łokciowa
Femur-fibula-ulna dysostosis
Femur-fibula-ulna syndrome

Kod ORPHA
2019

Kod OMIM
228200

Kod ICD10
Q74.8

Kod ICD11
LD26.0

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*Źródło

orphanet