

## 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 FFU complex

#### Synonimy

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

---

#### \*Źródło

orphanet