Opis choroby *

Definicja

A rare genetic syndrome with limb malformations as a major feature characterized by preaxial polydactyly of the hands and feet with variable phenotypic expressivity in combination with hypertrichosis extending from the posterior hairline to the middle of the back. Reported limb malformations include triphalangeal thumbs, duplicated thumbs, preaxial extra ray, and syndactyly between digits I and II in the hands, and large or duplicated hallux and syndactyly between toes I and II in the feet.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
476119

Kod OMIM
-

Kod ICD10
Q87.2

Kod ICD11
-

---

*Źródło

orphanet