

## Opis choroby \*

### Definicja

Camptobrachydactyly is an extremely rare brachydactyly syndrome, characterized by short broad hands and feet with brachydactyly associated with congenital flexion contractures of the proximal and/or distal interphalangeal joints of the fingers, as well as syndactyly of feet. Polydactyly, septate vagina and urinary incontinence were also occasionally reported. Camptobrachydactyly has been described in 18 members of 1 family, suggesting an autosomal dominant inheritance. There have been no further descriptions in the literature since 1972.

### Dane

### Klasyfikacja

Zespół wad wrodzonych

#### Kod ORPHA

1319

#### Kod OMIM

114150

#### Kod ICD10

Q74.8

#### Kod ICD11

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### \*Źródło

orphanet