

Kamptobrachydaktylia

Kod Orpha: 1319 Kod OMIM: 114150

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

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

Dostępna na stronie www.orphanet.pl