Camptobrachydactyly

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Camptobrachydactyly. ORPHA:1319

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.