Camptodactyly-fibrous tissue hyperplasia-skeletal dysplasia syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Camptodactyly-fibrous tissue hyperplasia-skeletal dysplasia syndrome. ORPHA:1321

Camptodactyly - fibrous tissue hyperplasia - skeletal dysplasia syndrome is an extremely rare chondrodysplastic malformation syndrome that is characterized by the combination of arachnodactyly, becoming evident at around the age of 10, camptodactyly (hammertoes) and scoliosis. A mild facial dysmorphism including a broad nose and flaring nostrils, and a mild intellectual disability were also noted. Camptodactyly - fibrous tissue hyperplasia - skeletal dysplasia syndrome has been described once in 3 siblings and is suspected to follow autosomal recessive transmission. There have been no further descriptions in the literature since 1972.