Camptodactyly-tall stature-scoliosis-hearing loss syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Camptodactyly-tall stature-scoliosis-hearing loss syndrome. ORPHA:85164

Camptodactyly-tall stature-scoliosis-hearing loss syndrome is characterised by camptodactyly, tall stature, scoliosis, and hearing loss (CATSHL). It has been described in around 30 individuals from seven generations of the same family. The syndrome is caused by a missense mutation in the FGFR3 gene, leading to a partial loss of function of the encoded protein, which is a negative regulator of bone growth.