Camptodactyly-joint contractures-facial skeletal defects syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Camptodactyly-joint contractures-facial skeletal defects syndrome. ORPHA:1323

Camptodactyly-joint contractures-facial skeletal defects syndrome is characterised by the association of camptodactyly, multiple eye defects (fibrosis of the medial rectus muscle, severe myopia, ptosis and exophthalmos), scoliosis, flexion contractures and facial anomalies (arched eyebrows, facial asymmetry with an abnormal skull shape, a prominent nose, small mouth, low-set and dysplastic ears, and a low nuchal hairline).