Camptodactyly syndrome, Guadalajara type 3

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Camptodactyly syndrome, Guadalajara type 3. ORPHA:488434

Camptodactyly syndrome, Guadalajara type 3 is a rare, genetic bone development disorder characterized by hand camptodactyly associated with facial dysmorphism (flat face, hypertelorism, telecanthus, symblepharon, simplified ears, retrognathia) and neck anomalies (short neck with stricking pterygia, muscle sclerosis). Additional features include spinal defects (e.g. cervical and dorso-lumbar spina bifida occulta), congenital shortness of the sternocleidomastoid muscle, flexed wrists and thin hands and feet. Brain structural anomalies, multiple nevi, micropenis and mild intellectual disability are also observed. Imaging reveals increased bone traveculae, cortical thickening of long bones and delayed bone age.