Cleidorhizomelic syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Cleidorhizomelic syndrome. ORPHA:1453

Cleidorhizomelic syndrome is a rhizo-mesomelic dysplasia characterized by rhizomelic short stature/dwarfism in combination with lateral clavicular defects. Additional manifestations include brachydactyly with bilateral clinodactyly and hypoplastic middle phalanx of the fifth digit. X-ray demonstrated an apparent Y-shaped or bifid distal clavicle. Cleidorhizomelic syndrome has been reported in one family (mother and son) and is suspected to be transmitted in an autosomal dominant manner. There have been no further descriptions in the literature since 1988.