Fibular aplasia-complex brachydactyly syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Fibular aplasia-complex brachydactyly syndrome. ORPHA:2639

Fibular aplasia-complex brachydactyly syndrome is characterised by severe reduction or absence of the fibula and complex brachydactyly. Less than 30 cases have been described in the literature so far. The syndrome is inherited in an autosomal recessive manner and is caused by mutations in the cartilage-derived morphogenetic protein-1 gene (WCDMP1).