Fibular aplasia-ectrodactyly syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Fibular aplasia-ectrodactyly syndrome. ORPHA:1118

Fibular aplasia-ectrodactyly syndrome is characterized by fibular aplasia and ectrodactyly. Less than 50 familial and sporadic cases have been reported in the literature. Shortening of the femur, a curved tibia, severe foot anomalies and pathologies of the hip, knee and ankle may also be present. The disorder is probably inherited as an autosomal dominant trait, with reduced penetrance, especially in females.