Radial deficiency-tibial hypoplasia syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Radial deficiency-tibial hypoplasia syndrome. ORPHA:1121

Radial deficiency-tibial hypoplasia syndrome is a rare, genetic dysostosis syndrome with combined reduction defects of upper and lower limbs characterized by bilateral radial aplasia, absent thumbs and bilateral tibial hypo/aplasia. Additional bone anomalies (including partial toe hypo/aplasia, short fibula and clubhand) may be associated. There have been no further descriptions in the literature since 1996.