Tibial hemimelia-polysyndactyly-triphalangeal thumb syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Tibial hemimelia-polysyndactyly-triphalangeal thumb syndrome. ORPHA:988

Tibial hemimelia-polysyndactyly-triphalangeal thumb syndrome is a rare, genetic dysostosis syndrome, with marked inter- and intra-familial variation, typically characterized by triphalangeal thumbs, hand and/or foot polysyndactyly and/or absent/hypoplastic tibiae (associated with duplication of fibulae in some cases), although isolated triphalangeal thumbs have also been reported. It is often accompanied with remarkable short stature and additional features may include radio-ulnar synostosis and hand oligodactyly, as well as abnormal carpal and metatarsal bones.