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Distal arthrogryposis type 5D

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Distal arthrogryposis type 5D. ORPHA:329457*

Distal arthrogryposis type 5D is a rare subtype of distal arthrogryposis syndrome characterized by arthrogryposis multiplex congenita affecting the hands, feet, ankle, shoulders and/or neck, with camptodactyly of the fingers and limited knee and hip extension, associated with asymmetric ptosis and, less frequently, other ocular manifestations (e.g. ophthalmoplegia, strabismus). Affected individuals frequently have a bulbous nose, furrowed tongue, micro/retrognathia, a short neck, congenital hip dislocation, club feet, scoliosis and short stature.