Open Peer Review on Qeios

Distal arthrogryposis type 10

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal arthrogryposis type 10. ORPHA:251515

A rare, genetic, distal arthrogryposis syndrome characterized by plantar flexion contractures, typically presenting with toe-walking in infancy, variably associated with milder contractures of the hip, elbow, wrist and finger joints. No ocular or neurological abnormalities are associated and serum creatine phosphokinase levels are normal.