Autosomal dominant multiple pterygium syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal dominant multiple pterygium syndrome. ORPHA:65743

Autosomal dominant multiple pterygium syndrome is a rare distal arthrogryposis syndrome characterized by multiple pterygia (typically involving the neck, axilla and popliteal areas), joint contractures, ptosis, camptodactyly of the hands with hypoplastic flexion creases, vertebral fusions, severe scoliosis and short stature.