Arthrogryposis-oculomotor limitation-electroretinal anomalies syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Arthrogryposis-oculomotor limitation-electroretinal anomalies syndrome. ORPHA:1154

Distal arthrogryposis type 5 is an inherited developmental defect syndrome characterized by multiple congenital contractures of limbs, without primary neurologic and/or muscle disease that affects limb function, and ocular anomalies (ptosis, external ophtalmoplegia and/or strabismus). Intelligence is normal.