Hypomyelination neuropathy-arthrogryposis syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hypomyelination neuropathy-arthrogryposis syndrome. ORPHA:2680

Hypomyelination neuropathy-arthrogryposis syndrome is a rare, genetic, limb malformation syndrome characterized by multiple congenital distal joint contractures (incl. talipes equinovarus and both proximal and distal interphalangeal joint contractures of the hands) and very severe motor paralysis at birth (i.e. lack of swallowing, autonomous respiratory function and deep tendon reflexes), leading to death within first 3 months of life. Fetal hypo- or akinesia, late-onset polyhydramnios and dramatically reduced, or absent, motor nerve conduction velocities (<10 m/s) are frequently associated. Nerve ultrastructural morphology shows severe abnormalities of the nodes of Ranvier and myelinated axons.