Autosomal recessive myogenic arthrogryposis multiplex congenita

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive myogenic arthrogryposis multiplex congenita. ORPHA:319332

Autosomal recessive myogenic arthrogryposis multiplex congenita is a rare inherited neuromuscular disease characterized by prenatal presentation (usually in the second trimester) of reduced fetal movements and abnormal positioning resulting in joint abnormalities that may involve both lower and upper extremities and is usually symmetric, severe hypotonia at birth with bilateral club foot, motor development delay, mild facial weakness without opthalmoplegia, absent deep tendon reflexes, normal motor and sensory nerve conduction velocities, no cerebellar or pyramidal involvement, and progressive disease course with loss of ambulation after the first decade of life.