Autosomal recessive lower motor neuron disease with childhood onset

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive lower motor neuron disease with childhood onset. ORPHA:206580

Autosomal recessive lower motor neuron disease with childhood onset is a rare, genetic, neuromuscular disease characterized by proximal muscle weakness with an early involvement of foot and hand muscles following normal motor development in early childhood, a rapidly progressive disease course leading to generalized areflexic tetraplegia with contractures, severe scoliosis, hyperlordosis, and progressive respiratory insufficiency leading to assisted ventilation. Cranial nerve functions are normal and tongue wasting and fasciculations are absent. Milder phenotype with a moderate generalized weakness and slower disease progress was reported.