Proximal spinal muscular atrophy type 1

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Proximal spinal muscular atrophy type 1. ORPHA:83330

Proximal spinal muscular atrophy type 1 (SMA1) is a severe infantile form of proximal spinal muscular atrophy (see this term) characterized by severe and progressive muscle weakness and hypotonia resulting from the degeneration and loss of the lower motor neurons in the spinal cord and the brain stem nuclei.