Spinal Muscular Atrophy Type 3

National Cancer Institute

Source

National Cancer Institute. Spinal Muscular Atrophy Type 3. NCI Thesaurus. Code C118847.

A rare, autosomal recessive inherited disorder caused by mutations in the SMN1 gene. It is characterized by progressive degeneration and loss of the anterior horn cells in the spinal cord and brain stem. It is manifested with hypotonia and muscle weakness, usually in late childhood or adolescence. Affected individuals can stand and walk but walking and climbing stairs becomes progressively difficult.