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Spinal muscular atrophy.

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Abstract

Spinal muscular atrophy is an autosomal recessive neurodegenerative disease characterised by degeneration of spinal cord motor neurons, atrophy of skeletal muscles, and generalised weakness. It is caused by homozygous disruption of the survival motor neuron 1 (SMN1) gene by deletion, conversion, or mutation. Although no medical treatment is available, investigations have elucidated possible mechanisms underlying the molecular pathogenesis of the disease. Treatment strategies have been developed to use the unique genomic structure of the SMN1 gene region. Several candidate treatment agents have been identified and are in various stages of development. These and other advances in medical technology have changed the standard of care for patients with spinal muscular atrophy. In this Seminar, we provide a comprehensive review that integrates clinical manifestations, molecular pathogenesis, diagnostic strategy, therapeutic development, and evidence from clinical trials.

Comment in

Carrier frequency of spinal muscular atrophy. [Lancet. 2008]

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