Spinal Muscular Atrophy

Spinal muscular atrophy (SMA) is a genetic disease affecting the second motor neuron, causing progressive muscle atrophy and weakness due to decreased expression of the survival motor neuron. Different subtypes exist, type 2 is one of the most frequent ones.

The most severe form is called Werdnig-Hoffmann disease and is usually fatal within months.

Epidemiology

A rare autosomal recessive congenital disease of childhood.

Etiology

Spinal muscular atrophy is an inherited disorder and is passed on in an autosomal recessive manner with degeneration of anterior horn cells.

The disorder is caused by a genetic defect in the SMN1 gene, which encodes SMN, a protein widely expressed in all eukaryotic cells (that is, cells with nuclei, including human cells) and necessary for survival of motor neurons. Lower levels of the protein results in loss of function of neuronal cells in the anterior horn of the spinal cord and subsequent system-wide atrophy of skeletal muscles.

Clinical features

Only rarely evident at birth (where it presents as a paucity of movement), produces weakness, areflexia, muscle and tongue fasciculations with normal sensation. Usually starts in proximal muscles and muscles of respiration. Severe cases progress over the first year or two to quadriplegia.

Spinal muscular atrophy manifests in various degrees of severity, which all have in common progressive muscle wasting and mobility impairment. Proximal muscles, arm and leg muscles that are closer to the torso and respiratory muscles are affected first. Other body systems may be affected as well, particularly in early-onset forms of the disorder. SMA is the most common genetic cause of infant death.

Fasciculations represent discharge of a group of muscle fibers (all or part of an entire motor unit), and occur most often in diseases involving anterior horn cells, including spinal muscular atrophy.

Diagnosis

Spinal Muscular Atrophy Diagnosis.

Treatment

see Nusinersen.

These patients show a high incidence of scoliosis requiring surgery.

Case series

Spinal Muscular Atrophy case series

Case report

A 16-year-old girl with SMA type 2 was referred for intrathecal nusinersen therapy. Because of severe scoliosis, spondylodesis of the segments TH7-S1 was performed at 14 years of age. The first two loading doses were given by spinal tap under sedation and computed tomography guidance, but we were unable to administer the following dose because of severe scoliotic spinal deformation. To ensure further drug therapy, an intrathecal port catheter (Celsite® Safety; Braun, Germany) was implanted via microsurgical hemilaminectomy L4. Further intrathecal nusinersen administration was uneventful.

They conclude that the implantation of an intrathecal port system in patients with SMA and profound scoliosis is a safe and feasible procedure and allows the administration of nusinersen while reducing the need for sedation and exposure to radiation [1)].

1)
Flotats-Bastardas M, Linsler S, Zemlin M, Meyer S. Nusinersen Administration Via an Intrathecal Port in a 16-Year-Old Spinal Muscular Atrophy Patient with Profound Scoliosis. Pediatr Neurosurg. 2019 Nov 13:1-4. doi: 10.1159/000504058. [Epub ahead of print] PubMed PMID: 31722365.