SMN1

Spinalmuscular 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 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.

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.

Spinal muscular atrophy is an inherited disorder and is passed on in an autosomal recessive manner (see video explanation of autosomal recessive inheritance). In December 2016, nusinersen became the first approved drug to treat SMA while several other compounds remain in clinical trials.