Journal of Alzheimers Disease & Parkinsonism

Ubaldo R, J Alzheimers Dis Parkinsonism 2022, 12:3

Opinion Article Open Access

A Short Note on Spinal Muscular Atrophy

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Received: 18-Mar-2022, Manuscript No. JADP-22-60515; **Editor assigned:** 24-Mar-2022, PreQC No. JADP-22-60515 (PQ); **Reviewed:** 07-Apr-2022, QC No. JADP-22-60515; **Revised:** 12-Apr-2022, Manuscript No. JADP-22-60515 (R); **Published:** 19-Apr-2022, DOI: 10.4172/2161-0460.1000543.

Citation: Ubaldo R (2022) A Short Note on Spinal Muscular Atrophy. J Alzheimers Dis Parkinsonism 12: 543.

About the Study

Spinal muscular atrophy is a genetic disorder characterized by muscle weakness and atrophy (wasting) (skeletal muscles). It is caused by the degeneration of motor neurons, which are specialized nerve cells that control muscle movement. The weakness is more severe in muscles near the center of the body (proximal) than in muscles further away from the center of the body (distal). Muscle weakness typically worsens as people age. Changes in the same genes cause several types of spinal muscular atrophy. The age of onset and severity of muscle weakness different between the types; however, there is overlap between the types.

Mutations in other genes cause other types of spinal muscular atrophy and related motor neuron diseases, such as spinal muscular atrophy with progressive myoclonic epilepsy, spinal muscular atrophy with lower extremity predominance, X-linked infantile spinal muscular atrophy, and spinal muscular atrophy with respiratory distress type 1. Spinal muscular atrophy type 0 is the rarest and most severe form of the condition, manifesting itself before birth. Affected infants move less in the womb, and as a result, they frequently have joint deformities when they are born (contractures). At birth, they have extremely low muscle tone (hypotonia). Their respiratory muscles are extremely weak, and they frequently die before reaching the age of one due to respiratory failure. Some infants with SMA type 0 also have heart defects that are present from birth (congenital).The most common type of the condition is spinal muscular atrophy type I (also known as Werdnig-Hoffmann disease). It is the most severe form of the disorder, with muscle weakness evident at birth or within the first few months of life. The vast majority of affected children are unable to control their head movements or sit without assistance. This type of child may have swallowing issues, which can lead to feeding difficulties and poor growth. They may also experience breathing difficulties due to respiratory muscle weakness and an abnormally bell-shaped chest, which prevents the lungs from fully expanding. Because of respiratory failure, the majority of children with spinal muscular atrophy type I do not survive past childhood.

Spinal muscular atrophy type II (also known as Dubowitz disease) causes muscle weakness in children aged 6 to 12 months. Children of this type can sit without assistance, but they may require assistance getting into a seated position. However, as the muscle weakness worsens later in childhood, those who are affected may require assistance sitting. Spinal muscular atrophy type II patients are unable to stand or walk unaided. They frequently have involuntary tremors in their fingers, a curved spine (scoliosis), and life-threatening respiratory muscle weakness. Individuals with spinal muscular atrophy type II live into their twenties or thirties on average, but this condition can cause death.

Spinal muscular atrophy type III (also known as Kugelberg-Welander disease) typically causes muscle weakness after childhood. Individuals with this condition can stand and walk unaided, but walking and climbing stairs may become increasingly difficult over time. Many affected people require wheelchair assistance later in life. People with spinal muscular atrophy type III have a normal life expectancy. Spinal muscular atrophy type IV is uncommon and usually begins in early adulthood. Affected individuals typically experience mild to moderate muscle weakness, tremors, and mild breathing difficulties. People with spinal muscular atrophy type IV have a normal life expectancy. One in every 8,000 to 10,000 people worldwide suffers from spinal muscular atrophy. The most common type of spinal muscular atrophy is type I, which accounts for roughly half of all cases. Types II and III are the most common, while Types 0 and IV are uncommon.

All of the above-mentioned types of spinal muscular atrophy are caused by mutations in the SMN-1 gene. The number of SMN-2 gene copies affects the severity of the condition and helps determine which type develops.

Both the SMN-1 and SMN-2 genes encode a protein known as the survival motor neuron (SMN) protein. Normally, the majority of functional SMN protein is produced by the SMN1 gene, with a small amount produced by the SMN-2 gene. The SMN-2 gene produces several different versions of the SMN protein, but only one is functional; the others are smaller and quickly degraded. The SMN protein is part of a protein complex known as the SMN complex, which is essential for the maintenance of motor neurons. Motor neurons send signals from the brain and spinal cord to tell skeletal muscles to tense (contract), allowing the body to move.

The majority of people with spinal muscular atrophy lack a portion of the SMN-1 gene, which impairs SMN protein production. A lack of SMN protein causes motor neuron death, and as a result, signals between the brain and muscles are not transmitted. Muscles cannot contract unless they receive signals from the brain; so many skeletal muscles weaken and waste away, resulting in the signs and symptoms of spinal muscular atrophy.

In most people, each cell contains two copies of the SMN-1 gene and one to two copies of the SMN2 gene. The number of copies of the SMN-2 gene, on the other hand, varies, with some people having up to eight copies. Having multiple copies of the SMN-2 gene in people with spinal muscular atrophy is usually associated with less severe symptoms of the condition that develop later in life. The SMN protein produced by the SMN-2 genes can assist in compensating for the protein deficiency caused by SMN-1 gene mutations. People with type 0 have one copy of the SMN-2 gene in each cell, while those with type I have one or two copies, those with type II have three copies, those

with type III have three or four copies, and those with type IV have four or more copies.