What is SMA (Spinal Muscular Atrophy) in children?

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Introduction

SMA (spinal muscular atrophy) is a devastating hereditary illness in which muscles weaken and waste away over time. A missing or mutant gene causes this inherited neuromuscular illness, which causes a baby to lose motor neurons, which are nerve cells in the spinal cord that control voluntary muscle movement. Muscles become weak and atrophy when there is insufficient supply of motor neurons.

Spinal muscular atrophy is more common in new-borns and young children, but it can also occur in adults (though this is rare). SMA affects around one in every 8,000 to 10,000 persons on the planet. Type 1, which appears at birth or in early childhood, accounts for more than half of all cases. Spinal muscular atrophy is a life-threatening condition that reduces a baby's quality of life and shortens his or her life expectancy [1].

The symptoms, treatment, and prognosis of spinal muscular atrophy in neonates will be discussed in this article.

Spinal Muscular Atrophy (SMA) comes in a variety of forms

There are five different forms of spinal muscular atrophy, each of which can appear at any time during one's life:

Type 1: is the most prevalent type of SMA and is also the most severe. Type 1 Werdnig-Hoffman disease, also known as Werdnig-Hoffman syndrome, appears at birth or within the first six months of life. Babies with type 1 diabetes often have trouble holding their heads erect. Because of sucking and swallowing issues, the majority of people have difficulty eating. A new-born with type 1 SMA misses developmental milestones and is more susceptible to respiratory infections. Because of respiratory infections or a collapsed lung, type 1 babies seldom live through their second birthday.

Type 2: SMA, often known as Dubowitz disease, is an intermediate type of spinal muscular atrophy. Symptoms appear between the ages of 6 and 18 months, and they predominantly affect the lower limbs. This kind of SMA causes infants and young children to be able to sit up but not walk. They frequently have scoliosis and uncontrollable tremors in their fingers. This kind of SMA can cause babies to live into their 20s or 30s.

Type 3: SMA is a milder form of the disease. Symptoms of Kugelbert-Welander SMA, also known as juvenile-onset SMA, usually appear after 18 months. Muscle weakness, trouble walking, and respiratory infections are some of the symptoms that may not appear until early adulthood. Type 3 people can normally walk alone, however they may struggle with stairs. As the disease advances, people with type 3 diabetes may need to use a wheelchair. Type 3 does not, fortunately, diminish one's life expectancy [2,3].

Symptoms of spinal muscular atrophy

The following are some of the symptoms of SMA in new-borns, depending on the type 2:

- Muscle weakening develops over time.
- Muscle control loss
- Muscles nearest to the torso are weak.
- Inability to sit, move, or stand for long periods of time
- Having trouble eating
- Respiratory infections are common.

Causes

SMA is a genetic condition. This means that it is caused by a genetic aberration passed down from parents to their children. A child with spinal muscular atrophy inherits two copies of the missing or mutant motor neuron (SMN1) gene from each parent [4].

The SMN1 gene produces the SMN protein, which is required for motor neuron activity. Motor neurons shrink and die if there isn't enough of this protein. The brain is unable to govern voluntary muscular movements when this happens. Muscles in the head, neck, arms, and legs are particularly affected.

Diagnosis

A physical examination and medical history are used to diagnose SMA. The following are examples of diagnostic tests: Blood test, Genetic testing, Electromyogram (EMG)

Treatment

SMA has no known cure. Treatment options will be determined by the type, symptoms, and overall health of your kid. Babies with spinal muscular atrophy need to be treated by a multidisciplinary team that includes respiratory, orthopaedic, gastroenterological, and nutritional specialists. Occupational and physical therapists are frequently involved

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in the treatment of patients. As your child grows, assistive devices such as braces, crutches, or a wheelchair may be required [5].

Your healthcare professional may also offer drugs to treat SMA, depending on your child's age and prognosis. The following drugs are used to stimulate the production of the SMN protein:

- Spinraza (nusinersen)
- Evrysdi (risdaplam)

For new-borns and early toddlers with SMA, your medical team may investigate gene replacement therapy. Zolgensma is a medicine that is used to replace a missing or mutant SMN1 gene with a healthy one.

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