Infantile Spinal Muscular Atrophy

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What is Spinal Muscular Atrophy?

Spinal muscular atrophy is a disorder that affects the control of muscle movement. It is caused by a loss of specialized nerve cells, called motor neurons, in the spinal cord and the part of the brain that is connected to the spinal cord (the brainstem). The loss of motor neurons leads to weakness and wasting (atrophy) of muscles used for activities such as crawling, walking, sitting up, and controlling head movement. In severe cases of spinal muscular atrophy, the muscles used for breathing and swallowing are affected. Spinal muscular atrophy is divided into subtypes based on the severity of the disease and the age when symptoms appear.

Four types of spinal muscular atrophy affect children before the age of 1. Type I spinal muscular atrophy (also called Werdnig-Hoffman disease) is a severe form of the disorder that is evident at birth or within the first few months of life. Typically, affected infants have difficulty breathing and swallowing and are unable to sit without support.

Type II spinal muscular atrophy is characterized by muscle weakness that develops in children between ages 6 and 12 months. Children with type II can sit without support, although they cannot stand or walk unaided.

X-linked infantile spinal muscular atrophy has features that are very similar to type I, except that children with this type are typically born with joint deformities (contractures) that impair movement. In severe cases, affected infants are born with broken bones. Poor muscle tone before birth may contribute to the contractures and broken bones seen in these children.

The fourth type of spinal muscular atrophy that appears in infancy is called distal spinal muscular atrophy type 1. This form of the disorder is characterized by progressive muscle weakness in the hands and feet that eventually spreads to the limbs. Affected individuals also develop paralysis of the muscle that separates the abdomen from the chest cavity (the diaphragm), which leads to respiratory failure. The signs and symptoms of distal spinal muscular atrophy type 1 typically appear between ages 6 weeks and 6 months. Symptoms are usually evident at an earlier age. On rare occasions, people with this condition can begin to show symptoms during late childhood or adolescence.

Three other types of spinal muscular atrophy can affect people in early childhood and adulthood. Type III spinal muscular atrophy (also called Kugelberg-Welander disease or juvenile type) is a milder form of the disorder than types I or II, or the X-linked form. Symptoms appear between early childhood and early adulthood. Individuals with type III spinal muscular atrophy can stand and walk unaided, but usually lose this ability later in life. Two types of spinal muscular atrophy, type IV and Finkel type, usually occur after age 30. Symptoms of these adult-onset types of spinal muscular atrophy are typically mild to moderate and include muscle weakness, tremor, and twitching.

Infantile spinal muscular atrophy (SMA) is an inherited genetic condition and is also known as Werdnig-Hoffman disease. The nerve cells that service the muscles don't work properly, causing muscle weakness and withering. There is no cure. Treatment can ease complications including pneumonia and breathing difficulties. A child with SMA rarely lives beyond three years.

Infantile SMA is the most severe form. The symptoms include:

- Muscle weakness
- Poor muscle tone
- Weak cry
- Limpness or a tendency to flop
- The legs tend to be weaker than the arms
- Feeding difficulties
- Increased susceptibility to respiratory tract infections
- Developmental milestones, such as lifting the head or sitting up, can't be reached

A child with infantile SMA is prone to respiratory infections. Pneumonia is a type of lung infection where the smallest airways, called the alveoli, are blocked with mucus and secretions. In healthy people, pneumonia can be simply treated with antibiotics. However, a child with infantile SMA is already in a weakened and vulnerable state. Pneumonia is the cause of death in the majority of cases.

There is no cure

Infantile SMA is a fatal disorder and there is no cure. Treatment can only ease any associated complications. For instance, since a child with infantile SMA is prone to respiratory infections and pneumonia, treatment focuses on trying to maintain the child's lung function and health. Usually, a team of professionals - pediatricians, physiotherapists, neurologists, and respiratory physicians and therapists - work together to help improve the child's quality of life.

Developmental Areas

Children may display some of the following characteristics

Social and Emotional Development

- May have low or limited understanding of social interaction with peers
- May have difficulty in making friends due physical appearance and language delays
- Social development can also be delayed due to health issues resulting in the child being absent from care or school
- Child may have limited peer initiated social experiences

Physical Development

- May be short in stature making activities difficult
- May have visual impairment
- May have hearing impairment
- May have gross and fine motor difficulties such as catching and throwing balls
- May have unusual weight gain through hypothyroidism
- May have bouts of feet and hand swelling due to faulty lymphatic drainage

Language and Communication Development

- May have frequent bouts of Otitis media thus affecting hearing and communication
- Speech may slow with a horse voice
- May have difficulty with conversations

Cognitive Development

- May have had limited concrete experience with the environment
- May have learning disabilities particularly in the cognitive understanding of word comprehension and mathematical concepts

Health and Safety Issues

- Be aware of health implications that may affect participation in aspects of the program
- Short stature can be increased with medication however may present educational problems, particularly with physical education
- Otitis Media should be adequately monitored and treated
- May be under medication for the hypothyroidism
- Weight gain can also be an issue
- May suffer from coarctation (narrowing) of the aorta

Reference

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