AMYOTROPHIC LATERAL SCLEROSIS
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SPINAL MUSCULAR ATROPHY (SMA)

Fact Sheet

What is Spinal Muscular Atrophy?

- Spinal Muscular Atrophy (SMA) is a Motor Neuron Disease.
- It is caused by the mutation of the Survival of Motor Neuron (SMN) gene.
- It occurs due to the loss of motor neurons within the spinal cord and brain.
- It results in the progressive wasting away of muscles (atrophy) and muscle weakness.
- SMA can affect people of all ages, races or genders, but the majority of cases occur in infancy or childhood.
- There are four types of SMA.

Forms of SMA

Type I (Acute Infantile)

- Also called Werdnig-Hoffman Disease.
- Most severe form of SMA.
- Usually diagnosed before 6 months of age.
- Those affected cannot sit without support, lungs may not fully develop, swallowing and breathing may be difficult and there is weakness of the intercostal muscles (muscles between the ribs).
- 95 per cent fatal by 18 months due to respiratory failure.

Type II (Chronic Infantile)

- Usually diagnosed before the age of two, with the majority of cases diagnosed by 15 months.
- May be able to sit without assistance or even stand with support.
- Increased risk for complications from respiratory infections.
- Usually do not live past 30 years of age.

Type III (Chronic Juvenile)

- Also known as Kugelberg-Welander or Juvenile Spinal Muscular Atrophy.
- Usually diagnosed before the age of three, but can strike as late as adolescence.
- Able to walk, but experience noticeable weakness.
- Most patients eventually need to use a wheelchair.
- Usually survive well into adulthood with normal life expectancy.

Type IV (Adult Onset)

- Much less common than the other forms.
- Symptoms tend to surface after the age 35, but sometimes earlier.
- The bulbar muscles - used for swallowing and respiratory function - are rarely affected.
- Progression is slow and those affected have a normal life expectancy.

Symptoms in Infants

- Muscle weakness.
- Muscle atrophy (wasting).
- Poor muscle tone.
- Areflexia (delayed reflexes).
- Weak cry.
- Difficulty sucking or swallowing.
- Feeding difficulties.
- Weak cough.
- Lack of developmental milestones (inability to lift head or sit up).
- Limpness or a tendency to flop.
- Accumulations of secretions in the lungs or throat.

Symptoms in Adults

- Muscle weakness.
- Muscle atrophy (wasting).
- Weak tongue.
- Stiffness.
- Cramps.
- Fasciculations (twitching).
- Clumsiness.
- Dyspnea (shortness of breath).

Diagnosis

- A diagnosis can be made by an SMN gene test which determines whether there any traces of the SMN 1 gene.
- When the SMN gene test is not possible or shows no abnormality, an electromyography (EMG) or muscle biopsy can be done.

Other Facts and Characteristics

- Lifespan depends on the severity of SMA in the patient.
- Intellectual ability is unaffected.
- Sexual response and reproductive functions are also unaffected.

Treatment

- Physical and occupational therapy.
- Ventilation is crucial as many infants with severe forms of SMA succumb to respiratory disease due to weakness of breathing muscles.
- Assistive devices such as ventilators, power wheelchairs help those living with SMA live longer and fuller lives.

For more information on SMA, visit:

www.fightsma.org

www.smafoundation.org

www.mda.org