SPINAL MUSCULAR ATROPHY

The ALS Society of Alberta

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; however, the majority of cases occur in infancy or childhood. There are four types of SMA.

FORMS OF SMA

TYPE I (ACUTE INFANTILE)

- Also called Wernig-Hoffman Disease.
- Most severe form of SMA.
- Usually diagnosed before six 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

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.
- Fasciculation (twitching).
- Clumsiness.
- Dyspnea (shortness of breath).

DIAGNOSIS

- A diagnosis can be made by an SMN gene test which determines whether there are any traces of the SMN1 gene.
- When the SMN gene test is not possible or shows no abnormality, an electromyography (EMG) or muscle biopsy can be done.

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 and power wheelchairs, help those living with SMA live longer and fuller

OTHER FACTS

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

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