Spinal Muscular Atrophy

Spinal muscular atrophy (SMA) is a genetic, motor neuron disease. It is caused by progressive degeneration of motor neurons in the spinal cord. The disorder causes weakness and wasting of the voluntary muscles. Weakness is often more severe in the legs than in the arms.

The childhood SMAs are all autosomal recessive diseases. This means that they run in families. More than one case is likely to occur in siblings or cousins of the same generation. Parents usually have no symptoms but still carry the gene. The gene for SMA has been identified and accurate diagnostic tests exist. There are many types of SMA. Some of the more common types are described below.

SMA type 1 is also called Werdnig-Hoffmann disease. It is evident before birth or within the first few months of life. There may be a reduction in fetal movement in the final months of pregnancy. Symptoms include:

- Floppy limbs and trunk.
- Weak movements of the arms and legs.
- Swallowing and feeding difficulties.
- Impaired breathing.

Affected children never sit or stand. They usually die before the age of 2.

Symptoms of *SMA type 2* usually begin between 3 and 15 months of age. Children may have:

- Respiratory problems.
- Floppy limbs.
- Decreased or absent deep tendon reflexes.
- Twitching of arm, leg, or tongue muscles.

These children may learn to sit. But they will never be able to stand or walk. Life expectancy varies.

Symptoms of *SMA type 3 (Kugelberg-Welander disease)* appear between 2 and 17 years of age. They include:

- Abnormal manner of walking.
- Difficulty running, climbing steps, or rising from a chair.
- Slight tremor of the fingers.

Kennedy syndrome or *progressive spinobulbar muscular atrophy* may occur between 15 and 60 years of age. Features of this type may include:

- Weak muscles in the tongue and face.
- Difficulty swallowing.
- Speech impairment.
- Overly developed male mammary glands.

The course of the disorder is usually slowly progressive. Kennedy syndrome is an X-linked recessive disorder. This means that women carry the gene, but the disorder only occurs in men.

Congenital SMA with arthrogryposis (persistent contracture of joints with fixed abnormal posture of the limb) is a rare disorder. Symptoms include:

- Severe contractures.
- Curvature of the spine.
- Chest deformity.
- Respiratory problems.
- An unusually small jaw.
- Drooping upper eyelids.

Adult SMA may begin between 40 and 60 years of age and progresses rapidly. It has an average life expectancy of about 5 years. Most cases prove to be variants of ALS (commonly called Lou Gehrig's disease). Symptoms include progressive limb weakness and weakening of the muscles, difficulty speaking and swallowing, and respiratory problems.

TREATMENT

Treatment of all forms of SMA is symptomatic and supportive. It includes treating pneumonia, curvature of the spine, and respiratory infections, if present. Also, physical therapy, orthotic supports, and rehabilitation are useful. Genetic counseling is necessary.

WHAT WILL HAPPEN

The prognosis for individuals with SMA varies depending on the type of SMA and the degree of respiratory function. Condition tends to deteriorate over time, depending on the severity of the symptoms.

RESEARCH BEING DONE

The NINDS supports research to study gene function in SMA. Researchers have found the specific gene that, when mutated, causes SMA. Several animal models of the disease have been developed as well as tests that can determine SMA gene function. This allows scientists to screen drugs that may be useful in treating SMA.

Research is continually being done to learn more about this problem.

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