



# POSNA

## The Core Curriculum

### Spinal muscular atrophy

#### Objectives

1. Describe the spectrum of the age of onset, severity, and clinical features of spinal muscular atrophy
2. Discuss the prognosis for the differing types of spinal muscular atrophy
3. Describe the genetic transmission of spinal muscular atrophy

#### Discussion

Although the orthopaedist will see patients with spinal muscular atrophy from time to time, spinal muscular atrophy is one of many conditions which are being intensively investigated at a molecular level; thus when reviewing recent literature on spinal muscular atrophy, one realizes what a small part of the whole is related to orthopaedic management. Spinal muscular atrophy is an autosomal recessive disorder with variable clinical severity, and an overall incidence of 1/6000 - 1/10,000. The disease affects the anterior horn cells of the spinal cord, degeneration of which results in muscle weakness. Although Thompson suggested a classification based on function would be useful, the conventional classification on the basis of age at onset and severity continues to be widely used. Type I, Werdnig-Hoffman, is most severe, and usually fatal by age 2. Type II, usually is clinically evident in the first year of age, children can sit, but do not stand or walk independently. There is no eponym for Type II, it is sometimes called intermediate. Type III, Kugelberg-Welander disease, is a less severe form, independent ambulation is possible, and may even have a normal life expectancy. Weakness of respiratory muscles is linked to the fatal forms.

Children with spinal muscular atrophy may be seen by the orthopaedic surgeon at the outset for delayed developmental milestones or weakness. In patients with established disease, contracture, hip dislocation (which has been reported as difficult to treat), and spinal deformity are the usual reasons for orthopaedic intervention. Results of treatment of spinal deformity are spotty, although the most recent work by Bridwell reported a high degree of patient satisfaction with the results of surgery. A recent work reports thyrotropin-releasing hormone improved strength and velocity of peripheral nerve conduction.

The genetic defect is on chromosome 5, the critical region contains several genes including the survival motor gene, the neuronal apoptosis inhibitory protein gene, and the p44 gene which encodes a transcription factor subunit. Detection of the NAIP gene is associated with the more severe forms of SMA. Patients with type II disease have a

much higher amount of SMN protein. This field of research is being intensively studied at present.

## References

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