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Spinal Muscular Atrophy  
The 4 major subtypes

Amber L. Ward, MS, OTR/L, BCPR, ATP/SM5

Objectives

- Identify the hallmark symptoms and differences between the 4 types of SMA.
- Select 3 unique pieces of equipment which might be used for someone with SMA.
- Identify 3 treatments which would be appropriate for persons with SMA of each type.

Causes

- SMA is characterized by the loss of motor neurons. It is classified as a motor neuron disease.
- The most common forms of SMA (types 1-4) are caused by a mutation in the SMN1 gene on chromosome 5.
- A mutation in the SMN1 gene leads to a deficiency of a motor neuron protein called SMN, for survival of motor neuron.
- In SMA types 1 through 4, symptoms vary on a continuum from severe to mild, based on how much SMN protein there is in motor neurons.
- The more SMN protein there is, the later in life symptoms begin and the milder the course of the disease is likely to be.
- SMN 2 makes some protein, and more in less severe cases
What Physicians Look For:

- The weakness is usually symmetrical and more proximal than distal.
- Sensation is preserved.
- Tendon reflexes are absent or diminished.
- Weakness in the legs is greater than in the arms.
- The severity of weakness generally correlates with the age of onset.
- The most severe type presents in infancy.
- The infant may appear normal at birth.

Diagnosis

- Blood test for an enzyme - Creatine Kinase (CK)
- Genetic testing
- Electromyogram - nerve conduction velocity

SMA Type 1 - Werdnig-Hoffman

- Children who have noticeable SMA symptoms at or shortly after birth usually are very weak, have difficulty breathing, sucking and swallowing, and never reach the developmental milestone of being able to sit on their own.
- Survival 2-5 years generally - bulbar/pulmonary complications
- Impaired head control, weak cry and cough
- Swallowing, feeding, and handling of oral secretion are affected. The tongue may show atrophy and fasciculation.
- Weakness and hypotonia in the limbs and trunks are eventually accompanied by intercostal muscle weakness.
- Have PEG, BiPAP or Vent
SMA Type 2
- When SMA symptoms begin in babies at approximately 7 to 18 months of age
- Learn to sit unassisted (most) but not to stand or walk independently
- At the strongest end of this category are those who can stand with a standing frame or long leg braces but are not able to walk independently.
- Bulbar weakness with swallowing difficulties may lead to poor weight gain in some children. They have difficulty coughing and clearing tracheal secretions.
- Scoliosis eventually develops, and bracing or spinal surgery is needed.
- Joint contractures commonly evolve over years.

SMA Type 3 - Kugelberg-Welander
- Muscle weakness begins in older children and teens, who learn to stand and walk but lose the ability later in life
- Although some with type 3 stop walking in adolescence, others walk well into their adult years.
- Scoliosis can develop in these patients.
- Swallowing, cough, and nocturnal hypoventilation are less common problems than in type 2 spinal muscular atrophy but may occur.
- Muscle aching and joint overuse symptoms are common.

SMA Type 4
- SMA that comes on in late teens or adulthood is called type 4, or adult-onset SMA.
- Life span is generally normal
- Symptoms as young as age 18, but generally after 35
- Mild muscle weakness, tremor, twitching, muscle aches, fatigue
- Generally maintain walking, may have waddling gait
- As much physical activity as is comfortable - swimming, adaptive sports
Research

- Gene modification therapies - Synthetic SMN1
- Make SMN 2 stronger, more productive
- Oxasemine - protecting motor neurons (http://tinyurl.com/lj5b6as)
- Stem cells - SMA mice treated had longer survival, improved mobility

SMA type 1

- Non-sitter
  - Postural management
  - Contracture/pain management
  - Feeding tube/respiratory support
  - Power mobility possibly, supportive mobility
  - Assistive technology

SMA Type 2

- Sitter
  - Power wheelchair mobility early!!!!
  - Positioning, contracture mgnt, ROM, bracing
  - Equipment for self care, AT
  - Orthopedic mgnt
SMA 1 and 2

- Optimize developmentally appropriate play
- Optimize function
- Sensory experiences
- Exploration of environment
- Control

SMA Type 3

- Walker
  - Bracing, ROM, exercise to tolerance
  - Mobility aides, safety, prolong ambulation
  - AT, environmental access, energy conservation

SMA Type 4

- Supportive exercise, ROM, remain active
- Supportive mobility, safety
- Fatigue management, energy conservation
- ADL equipment
Monitor Equipment Needs

- Car seats
- Strollers
- Power mobility
- Standers
- ADL equipment
- Mobile arm support
- Assistive technology, computers, ECU

Promote independence with functional activities

- Adaptive equipment
- Positioning
- School
- Independence

Helpful websites

- www.mdausa.org
  - Good support and information

  - Great SMA resources
Questions?

- Amber.ward@carolinashealthcare.org
- 704-355-0787