*SPINAL MUSCULAR ATROPHY: Considerations for Care*

Prepared by:
Valerie Anderson, MSN, RN & Lisa Minahan, BSN, RN

**Objectives**
- Discuss Spinal Muscular Atrophy (SMA) including etiology & risk factors, pathophysiology, clinical manifestations, diagnostic criteria and differential diagnoses
- Identify recommended treatment patterns including mobility support, orthopedics, nutrition, respiratory care, and pharmacology
- Describe the anatomy and function of the respiratory system including differences between the child and adult airway
- Explain the role of tracheostomy and ventilation in the SMA child

**Outcomes**
- Learner will pass a posttest with a score of 90% or better at the conclusion of the presentation
- Learner will be able to identify at least two treatment goals for the SMA patient that they can incorporate into their current practice
- The learner will be able to state an increase in knowledge about the difference between child and adult airways
- The learner will be able to identify four patient teaching objectives to ensure a safe environment for the SMA child at home

**Introduction**

Spinal Muscular Atrophy (SMA) Type 1 is the leading genetic cause of death in infancy and early childhood.
The SMA patient and family will need lifelong care and support.
The role of the advanced practice nurse is integral in meeting the family’s needs.

**SMA: Overview**

Neuromuscular disorder
Degeneration of the anterior horn cells in the spinal cord and motor nuclei in the lower brain stem
Classified as Type 1 through Type 4 depending on age of onset and clinical course
Incidence: 4 to 10 per 100,000 live births

**SMA: Etiology & Risk Factors**

SMA is an autosomal recessive condition caused by a deletion or mutation in the SMN1 gene
When both parents are carriers, there is a 1 in 4 chance they will have a child with SMA
Personal or family history of SMA in the patient or father of baby
ACMG and ACOG recommend parental carrier screening for all at-risk pregnancies

**Prognosis**

Figure 1. Kaplan-Meier survival curves and survival probabilities for SMA 1, 2, and 3.

Reference:
Prognosis

* Significant associations between age of onset and long-term outcome, specifically survival in SMA type 1
* 95% of SMA 1 patients in this study died from respiratory failure
* Cause of death in SMA 2 patients included sepsis, acute respiratory infection, and chronic respiratory failure
* All SMA 3 patients were still alive at the conclusion of the study

SMA: Pathophysiology

* Mutation in the survival motor neuron gene
* Gene is normally inactive during fetal period and becomes active in the mature fetus to stabilize the neuronal population
* Absence of gene results in programmed cell death
* Mechanism and timing of abnormal motor neuron death remains unknown
* Maximal therapeutic window presents early

SMA: Clinical Manifestations

* All SMA patients
  * Diffuse symmetric muscle weakness; Greater in lower limbs
  * Absent or markedly decreased DTRs
  * Restrictive, progressive respiratory insufficiency
* Type 1 Infants
  * Severe symmetric flaccid paralysis
  * Unable to sit unsupported
  * Alert expression, furrowed brow, normal eye movements
  * Weak cry, poor suck and swallow reflexes, pooling of secretions, aspiration and fasciculations of the tongue

SMA: Diagnostic Criteria

* Molecular genetic testing with targeted mutation analysis
* Electromyography and nerve conduction studies
* Muscle biopsy

SMA: Differential Diagnoses

* Arthrogryposis multiplex congenita
* X-linked infantile SMA
* SMA with respiratory distress type 1
  * Congenital myasthenic syndromes
  * Congenital myopathies
  * Hypoxic-ischemic myelopathy
  * Lysosomal acid maltase deficiency
  * Prader-Willi syndrome
  * Traumatic myelopathy
  * Zellweger syndrome

SMA: Recommended Treatment Patterns

Supportive treatment
  * Mobility Support
  * Orthopedics
  * Nutrition
  * Respiratory Care

WE MUST PREVENT COMPLICATIONS OF WEAKNESS
*Physical Therapy
  * May be beneficial

*Spinal Bracing
  * Delays progressive scoliosis
  * Expiratory volume decreased while sitting

*Growth Rod Placement
  * Helps straighten and support spine

*Spinal Fusion Surgery

*Muscle-Release Operation
  * Releases stiffness and pain from prolonged wheelchair use

*SMA: Mobility Support

*SMA: Orthopedics

*Reciprocating Gait Orthosis (RGOs)
* Knee Ankle Foot Orthosis (KAFOs)
* Wrist or Hand Splints
* Manual Wheelchair
* Power Wheelchair

*SMA: Nutrition

*Tube Feeding is commonly offered
  * Nasogastric (NG) Tube
  * Nasoduodenal (ND) Tube
  * Nasojejunal (NJ) Tube
  * Gastrostomy (G) Tube
  * Gastrostomy-jejunostomy (G-J) Tube

*Reflux Medication may be necessary
* Avoid excess weight gain, as it can make mobility more difficult

*SMA: Nutrition

References:

CASE STUDY

What next?
Short-term goals?
Long-term goals?

Case Study Follow-up Question

Case Study Answer

This infant underwent a gastrostomy and was supported by noninvasive pressure ventilation as needed. With satisfactory gastrostomy feeding, her weight increased considerably.
**SMA: Respiratory Care**

- Secretion mobilization and clearance
- Manual or mechanical chest physiotherapy
- Manual cough assist
- Mechanical insufflation/exsufflation

**RESPIRATORY SUPPORT IS ESSENTIAL IN THE SMA PATIENT**

**Respiratory Considerations: Differences Between Adult and Child**

- Full Term Newborn Airway
  - 1mm of edema, the diameter will be 44% of normal.
- Adult Airway
  - 1mm of edema, the diameter will be 81% of normal.

- Airway Resistance

  - If radius is halved, resistance increases 16-fold
  - Resistance increases 3x in an adult and 16x in an infant.

\[
R = \frac{8 \pi l}{\pi r^4}
\]

**Respiratory Needs of the SMA Child**

- Restrictive and progressive respiratory insufficiency
- Muscle weakness leads to progressive respiratory failure
- Intercostal muscles more affected than diaphragm → paradoxical breathing → bell-shaped chest
- Patients and families must decide course of care

**Invasive vs. non-invasive ventilation**

- What does the research say?

  - How does respiratory care affect survival time?

**Tracheostomy and Ventilation in the SMA Child**
Tracheostomy and Parent Teaching

- Reason for tracheostomy
- How to maintain proper ventilation
- Secretion removal for airway patency
- Site and tube care
- Knowledge of use of home care and monitoring equipment
- Managing airway emergencies
- Potential complications
- Community resources

Future treatments that may have a potential therapeutic role:
- Gene therapy
- Intracerebroventricular or systemic injection
- Selective modification of SMN2 messenger RNA gene drug

SMA: Are There Pharmacology Options?

- SMA Foundation
- Cure SMA
- Muscular Dystrophy Association
- Pediatric Neuromuscular Clinical Research (PNCR) Network

SMA Drug Development

<table>
<thead>
<tr>
<th>Therapeutic target</th>
<th>Approaches</th>
<th>Clinical trial</th>
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</thead>
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<tr>
<td>Antisense SMN transcript</td>
<td>HbAT602, sodium diphosphopyridine nucleotide</td>
<td>Phase 1, NCT01993054</td>
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<tr>
<td>neuroprotection</td>
<td>Paliperidone, Amitriptyline</td>
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<tr>
<td>SMN2 rescue / inclusion</td>
<td>Nusinersen, Spinraza</td>
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<td>Induction of SMN protein</td>
<td>Agomelatine, Amifostine</td>
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<tr>
<td>Neuroprotection</td>
<td>Neurontin, Levetiracetam</td>
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<tr>
<td>Cell therapy</td>
<td>Stem cell, Gene therapy</td>
<td>Phase 1/II, NCT01993054</td>
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SMA: Are There Pharmacology Options?

Patient Resources

- SMA Foundation
- Cure SMA
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Role of Interdisciplinary team

- Neurologists, RNs/APRNs, Pulmonologists/Intensivists, orthopedics, genetic counselors, PT/OT, Dietary, Social Work
- Goal Planning, Individual Plan of Care

Implications for Practice

- Considerations for individual and population
- Cognitive development
- Patient advocacy in an academic setting
- Interdisciplinary approach to care
- Effective communication with patient and family

References:
**Summary**

Provided learner with an overview of Spinal Muscular Atrophy (SMA) including etiology & risk factors, pathophysiology, clinical manifestations, diagnostic criteria and differential diagnoses

Identified recommended treatment patterns including mobility support, orthopedics, nutrition, and respiratory care

Provided overview of the anatomy and function of the respiratory system including differences between the child and adult airway

Reviewed the role of tracheostomy and ventilation in the SMA child

Future of pharmacology

Implications for practice

**References**


Ryan, E. (2014). Respiratory disease. Children's Hospital and Medical Center, Omaha, NE.