Respiratory Management in Spinal Muscular Atrophy

TERRENCE SHENFIELD MS, RRT-ACCS, RPFT, NPS, AE-C

Objectives

Analyze genetic roots

Identify classifications

Evaluate pathophysiology

Standardize assessment

Implements the secretion care

Select ventilation modes

Synthesize RT interventions

Section 1: Understanding SMA

Etiology, Genetics, Pathophysiology, and Clinical Subtypes

The Role of SMN1

The SMN1 gene (Survival Motor Neuron 1) is the primary gene responsible to produce the **survival motor neuron (SMN) protein**.

In the context of Spinal Muscular Atrophy (SMA), it is the central genetic component of the disease

The **survival motor neuron (SMN) protein** is a critical protein found in all eukaryotic cells, essential for the health and survival of motor neurons—the nerve cells responsible for controlling voluntary muscle movement.

Protein production

- Healthy people the SMN 1 gene provides the essential instructions to create a functional protein
- This protein is vital for the survival of motor neurons

SMA patients have a mutation that limits production of SMN1 gene

Who Gets SMA?

Genetic inheritance by autosomal recessive disorder

- When an individual inherits two mutated copies of a specific gene—one from each parent.

SMA affects both males and females equally

Often diagnosed in infancy or childhood

Carrier frequency is approximately 1 in every 50 people in the general population who have a mutated SMN 1 gene

SMA occurs in one in every 10,000 live births

If both parents our carriers there's a 25% chance that the child will have SMA

What is Spinal Muscular Atrophy?

Spinal Muscular Atrophy (SMA): The most well-known condition associated with this protein is SMA. It is caused by mutations in the *SMN1* gene, which result in insufficient levels of functional SMN protein.

Pathology: As SMN protein levels drop, motor neurons in the spinal cord begin to degenerate and die. This loss disrupts the signaling between the brain and the muscles, leading to muscle weakness, atrophy (wasting), and, in severe cases, respiratory failure.

Genetic neuromuscular disease

- Autosomal recessive disorder caused by a mutation or deletion in the *SMN1* (survival motor neuron 1) gene

Signal failure

- As motor neurons die due to the lack of SMN protein, the "cables" connecting the central nervous system to the muscles are severed.

Critical risk

- One of the most severe risks in SMA is the weakening of the intercostal muscles (those between the ribs) and the diaphragm, which can lead to life-threatening respiratory failure.

Genetics of SMA: SMN1 and SMN2

The Primary Driver: SMN1 Gene

- 95% of cases

The Backup Modifier: SMN2 Gene

- Only produces 10% of SMN protein

The Splicing Difference (Exon7)

- Critical segment of the survival motor neuron (*SMN*) genes

Copying number determines severity

- *SMN2* is the only source of SMN protein in an SMA patient, it acts as a "backup" system. The more copies of *SMN2* a patient has, the more "backups" they have to potentially produce the protein, which directly impacts how severe the disease is

SMA Type	Age of Onset	SMN2 Copies	Highest Motor Milestone	Natural Survival (No Care)
Type 1 (Werdnig-Hoffmann)	0 – 6 months	1 – 2 copies	Never sits independently	< 2 years of age
Type 2 (Intermediate)	7 – 18 months	3 copies	Sits, never stands independently	Adulthood (highly variable)
Type 3 (Kugelberg-Welander)	> 18 months	3 – 4 copies	Stands and walks independently	Normal life expectancy
Type 4 (Adult-onset)	2nd or 3rd decade	> 4 copies	Normal adult milestones	Normal life expectancy

Clinical Classifications of SMA

Section 2: Respiratory Manifestations

Neuromuscular Weakness, Anatomy, and Deformity Trajectories

Neuromuscular Weakness and Anatomical Impact

- Selective muscle movement
- Diaphragm dependent breathing
- Bulbar weakness
- Impaired cough mechanism
- Bell shaped chest
- Progressive scoliosis
- Restrictive lung disease
- Paradoxical breathing
- Sleep disordered breathing

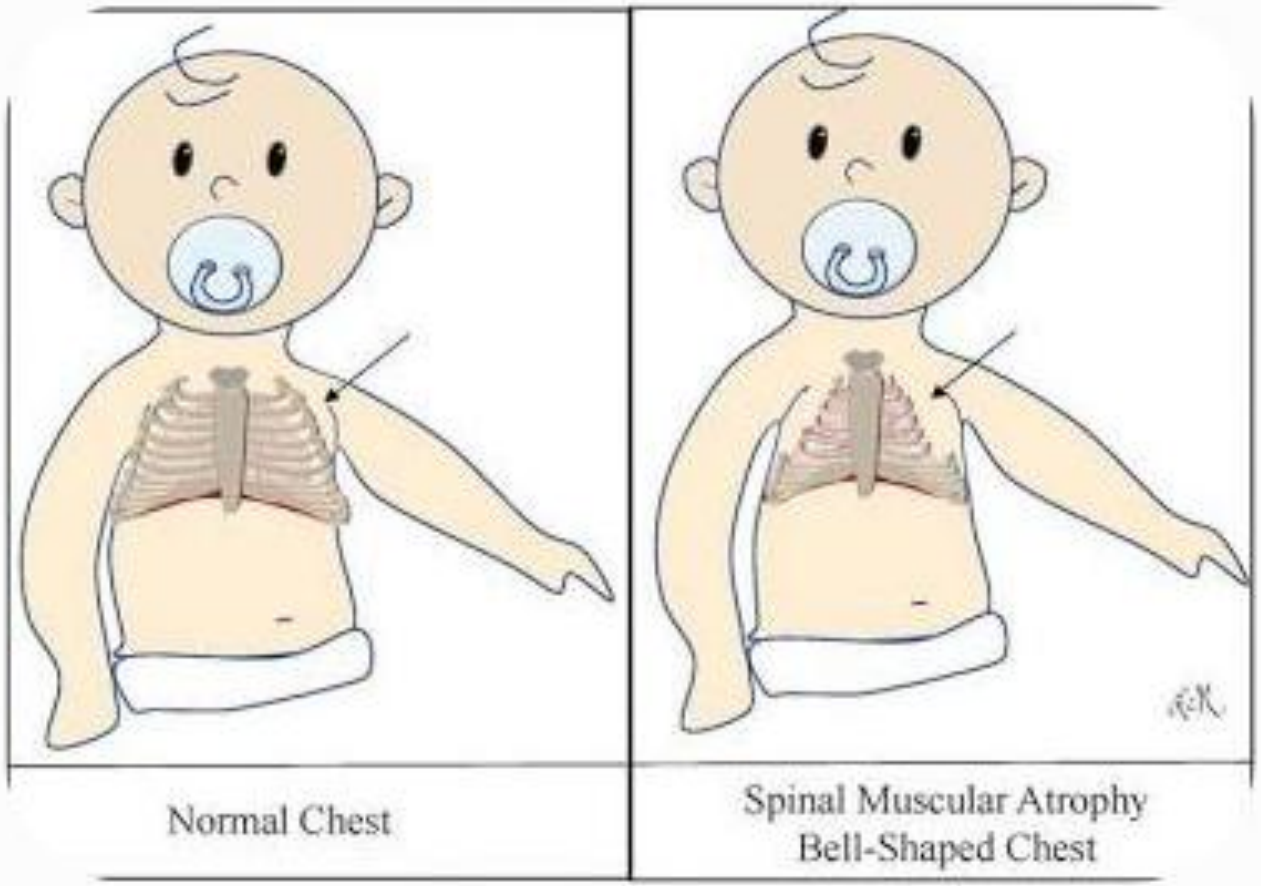
The Pathological Cascade

Primary respiratory muscle failure

Restricted Physiology

Impaired secretion defense

High risk of aspiration



Anatomical development of the bell-shaped chest deformity in SMA. Source: ResearchGate

Anatomical Impact: Paradoxical Breathing

- Mechanism of sparing
- Bell shaped chest deformity
- The mechanism of sparing
- Mechanics of paradoxical breathing
- Anatomical consequences

Section 3: The Respiratory Therapist

Core Competencies, Diagnostics, Assessments, and Critical Measures

Specialized RT Care Responsibilities

Advanced diagnostics

- Secretion evaluation
- Gas exchange trends
- Bulbar evaluation

Proactive secretion defense

- Insufflation-Exsufflation (MIE)
- HFCWO (High-Frequency Chest Wall Oscillation)
- Intrapulmonary Percussive Ventilation (IPV)

NIV management

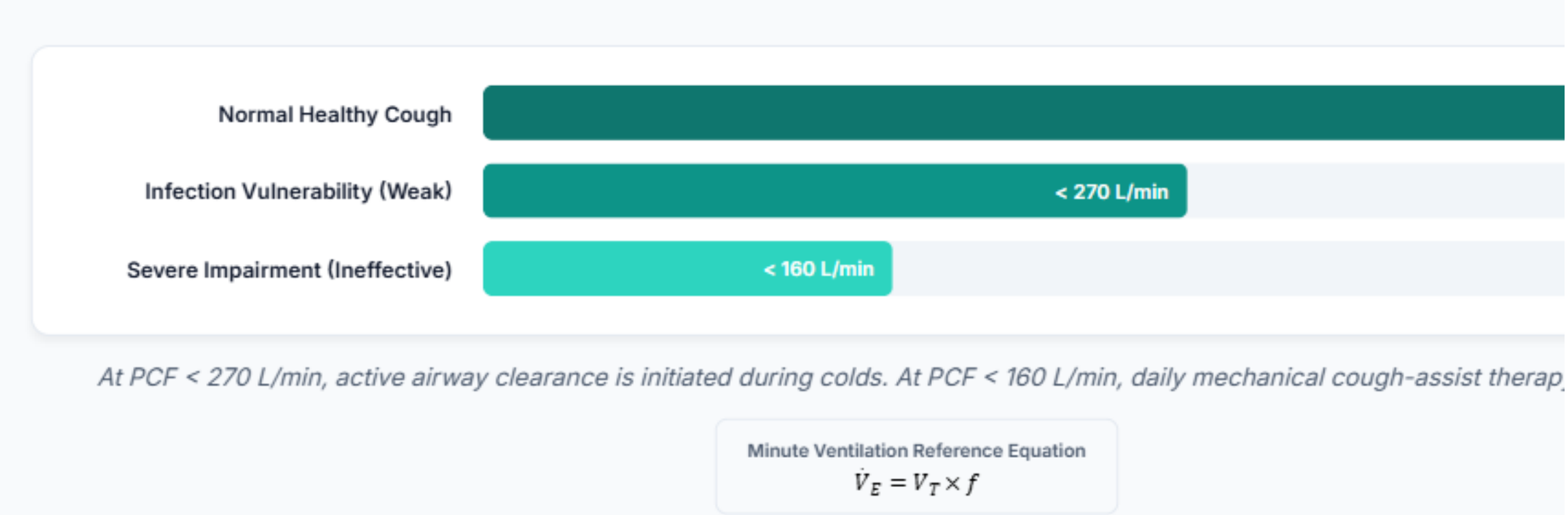
- Continuous High-Span Pressure Support
- Anatomical Interface Selection

Pulmonary Assessment Protocols

Forced Vital Capacity (FVC): Golden standard metric tracked in sitting and supine positions. A drop of >15% in supine confirms diaphragmatic weakness.

Peak Cough Flow (PCF): Direct metric of expiratory muscle power, indicating ability to clear thick mucus plugs.

Nocturnal Capnography: Continuous tracking of CO2 to catch hypoventilation long before daytime symptoms present.



Peak Cough Flow (PCF) Thresholds

Section 4: Airway Clearance

Techniques, Mechanical Insufflation-Exsufflation, and Titration Protocols

Cough-Assist (MIE) Therapy

Mechanical Insufflation-Exsufflation

This critical therapy mimics a natural cough by delivering positive pressure to expand the lungs (insufflation), then rapidly reversing to negative pressure (exsufflation) to drag secretions up.

RTs titrate parameters starting at ±15-20 cm H2O, scaling up to ±35-40 cm H2O.

This provides the shear force needed to mobilize thick plugs without compromising fragile airways.

Comparing HFCWO and Manual CPT

High-Frequency Chest Vest (HFCWO)

Manual Chest Physiotherapy

Comparison Summary Table

Feature	High-Frequency Chest Wall Oscillation (HFCWO)	Manual Chest Physical Therapy (Manual CPT)
Primary Mechanism	Global air-pulse chest wall oscillations	Localized manual percussion & postural drainage
Chest Wall Impact	Can restrict breathing/FRC in weak chest walls; risk of hypoxemia	Minimal restriction; easily customized to avoid skeletal deformities
Adaptability to Scoliosis	Poor; vest fits poorly over severe spinal curves	High; caregiver can avoid bony prominences and rods
Caregiver Burden	Low; automated device operation	High; physically demanding and technique-dependent
Core Requirement	**Must** be paired with MIE/suction for central clearance	**Must** be paired with MIE/suction for central clearance

Acute Secretion Clearance Timeline

Section 5: Ventilatory Support

Non-Invasive BiPAP, Device Interfaces, and Invasive Transitions

Non-Invasive Ventilation

Nocturnal BiPAP

Targeting Nocturnal Hypoventilation

Why BiPAP, Not CPAP

Preserving Chest Geometry

Impact of Disease-Modifying Therapies (DMTs)

Sizing & Support

Anatomical Sizing Vulnerabilities

Home Invasive Vent Integration

Tracheostomy indications

Failure of noninvasive ventilation

Severe bulbar dysfunction

Inability to clear secretions

Preparing for the Home Transition

Caregiver network

Home environment assessment

Emergency infrastructure

Lung Volume Recruitment (LVR)

Air-Stacking Technique: Uses a manual resuscitator bag and a one-way valve to stack multiple deep breaths, expanding the chest wall.

Chest Wall Compliance: Daily deep stretching of intercostal cartilage prevents joint stiffness, helping the rib cage grow.

Micro-Atelectasis Defense: Opens collapsed alveolar units to improve ventilation-perfusion matching and prevent pneumonia.

Target Demographics: Highly recommended twice daily for all cooperative patients with FVC < 60% predicted.

Modern Treatment Breakthroughs

Splicing Modifiers (Spinraza / Nusinersen)

- Approved for all ages and SMA types, it dramatically stabilizes or improves motor function, reduces the need for mechanical ventilation, and improves survival rates when initiated early.

Gene Therapy (Zolgensma)

- It addresses the root genetic cause of the disease in a single dose. When administered pre-symptomatically or early in the disease course, children frequently achieve major motor milestones—such as sitting unassisted or walking—that were completely unachievable in the natural history of the disease.

Oral Small Molecule (Evrysdi)

- Offers non-invasive systemic protection, improves or stabilizes motor function, enhances bulbar functions (like swallowing and speech), and slows down respiratory decline across a highly diverse demographic.

Supportive/Experimental Care

- Combination Therapies

Summary

- Genetic Basis
- Intercostal Paralysis
- Airway Clearance Standard
- Non-Invasive Ventilation
- Diagnostic Strategy
- Medical Integration
- Therapist & Caregiver Team

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