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Symptomatology and Rehabilitation of Dysphagia in Two Neuromuscular Disorders (NMDs): Spinal Muscular Atrophy (SMA) versus Dystonia

Jeanelle V. Leonce
SUNY Plattsburgh, jleone004@plattsburgh.edu

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Plattsburgh STATE UNIVERSITY OF NEW YORK

Jeanelle V. Leonce

Introduction

Spinal Muscular Atrophy (SMA) and Dystonia are two rare neuromuscular disorders that have been given more attention during the past decade. They can significantly alter one's quality of life depending on their severity and age of onset.

Definitions and Background Knowledge

SMA is defined as an autosomal recessive condition caused by a deletion or mutation of the survivor motor neuron (SMN1) gene (Chen, Shih, Chen, Kuo, & Jong, 2012; Mesfin, Sponseller, & Leet, 2012). A lack of the survivor motor neuron protein leads to a degeneration of skeletal muscles. Therefore, a high index for suspicion for SMA is hypotonia. Granger, Buschang, Throckmorton, and Iannaccone (1999) affirmed that the gene is linked to the disappearance of the anterior horn cells (lower motor neurons) of the spinal cord. SMA is one of the genetic causes of infant mortality and the second most lethal genetic disorder after cystic fibrosis (Granger et al., 1999; Mesfin et al., 2012).

Lee (2007) described dystonia as a "persistent posture from cocontractions of agonists and antagonists and is generally part of the spectrum of dyskinesias" (p. 491). According to Lee, it manifests clinically as continuous involuntary contractions with twisting or repetitive movements or abnormal postures. Dystonia consists of a variety of presentations (Figure 1) depending on musculature affected, severity, and distribution. It is the third most common movement disorder after essential tremor and Parkinson's disease (Charous & Comella, 2011; Granger, et al., 1999).

Clinical Classifications

Lee (2007) explained that "dystonias can be classified by age of onset and anatomically in region of distribution as well as into the affected body parts" (p. 492). According to Charous and Comella (2011), the younger the age of onset, the more likely it is for dystonia to be generalized, whereas the older the age of onset, the more likely dystonia will remain focal. Lee also posited that this mechanism is not well understood and dystonia could originate as a result of a "defect in the basal ganglia, especially in the sensory motor regions of the putamen" (p. 492). Dystonias can also be grouped as idiopathic, genetic, or acquired.

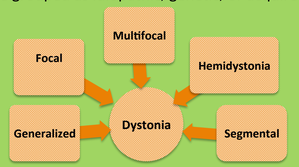

Figure 1: Clinical Classifications of Dystonia

Table 1: Clinical Classification of SMA

SMA Type	Age of Onset	Highest Function	Natural Age of Death
1 (Severe)	0 – 6 months	Never sits	< 2 years
2 (Intermediate)	7 – 18 months	Never stands	> 2 years
3 (Mild)	> 18 months	Stands and walks	Adult
4 (Adult)	2nd and 3rd decade	Walks during adult years	Adult

(Wang et al., 2007, p. 1030)

Wang et al. (2007) also classified SMA patients to address 5 care areas into the 3 functional levels: nonsitters, sitters, and walkers.

Table 2: Dysphagic Signs and Symptoms occur in ONLY First Three SMA Types

SMA Type	Signs and Symptoms
I	Patients experience impaired head control, weak cry and cough, respiratory difficulties, swallowing and feeding difficulties, difficulty with handling oral secretion before the age of one, intercostal muscle weakness, and tongue atrophy and fasciculation.
II	Bulbar weakness with swallowing difficulties may lead to poor weight gain. Patients have difficulty coughing and clearing tracheal secretions. Kyphoscoliosis develops and bracing or spinal surgery is needed.
III	Swallowing, coughing, and hypoventilation issues are less common, but may occur. Scoliosis can also develop. Messina et al. (2008) stated that scoliosis plays a role in dysphagic symptoms.

(Mesfina et al., 2012; Wang et al., 2007)

Two Most Common Dystonias with Dysphagic Symptoms

1. Cervical (Spasmodic Torticollis): studied more extensively and described as abnormal head postures brought about by involuntary, sustained, or repetitive neck muscle contractions (Plowman-Prine, Rosenbek, & Jones, 2009).
2. Oromandibular (OMD): involves abnormal repetitive contractions of the masticatory, facial, and lingual musculature as well as the presence of orobuccolingual dyskinesias leading to significant dysphagia (Papapetropoulos & Singer, 2006).

Table 3: Mastication and Deglutition Difficulties

SMA	Cervical Dystonia	OMD
- Debilitating malocclusions - Complete posterior crossbites - Anterior open bites - Severe arch discrepancies - Hyperdivergent facial patterns - Weak jaw muscles - Difficulty conveying food in mouth - Chewing difficulties - Dysphagia	- Pharyngeal swallow delay - Postswallow vallecular residue - Decreased tongue based retraction - Abnormal bolus preparation - Pharyngocele (pharyngeal outpouching) - Aspiration - Discomfort during swallowing - Odynophagia (pain during swallowing)	- Drooling - Mastication difficulty (placing food in mouth; manipulating food and liquid during oral stage) - Tongue protrusion and rotation - Choking - Tongue biting - Lip pursing and biting - Platysma contractions - Bruxism - Chewing fatigue

(Chen, 2012; Ganger et al.,1999; Papapetropoulos & Singer, 2006; Plowman-Prine et al., 2009)

In SMA II, feeding problems in patients who are younger than 20 years old are considerably high with a predominance of problems in the pre-oral phase such as limited mouth opening and self feeding (Chen 2012; Messina et al., 2008). Chewing difficulties are as a result of craniofacial abnormalities (Granger et al., 1999; Messina et al., 2008) and poor head control may preclude compensatory postures to make swallowing more manageable. Bulbar movement resulting in pharyngeal swallowing difficulties is more prevalent in SMA I, but has also been identified in type II (Messina et al., 2008). Chen identified compensatory head postures as the major determinant of dysphagia in SMA. On the other hand, Plowman-Prine et al. (2009) revealed that dysphagia and feeding difficulties occur across several forms of dystonia in both children and adults. Moreover, swallowing difficulties are dependent on the structures affected. The pharyngeal phase, however, is impacted more severely although patients experience oral challenges. Choking may be linked to aspiration and consequently aspiration pneumonia (Chen, 2012; Messina et al., 2008).

Management of Disorders

Managing these complex disorders is necessary to make patients more comfortable. The literature stresses the need for a multidisciplinary team (Figure 2) and that early detection, monitoring, and management may prevent unfavorable consequences. Tilton, Miller, and Khoshoo (1998) posited that the team must be able to identify risks and implement specific interventions. Chen (2012) indicated that treatment should be based on current motor status rather than the types of SMA.

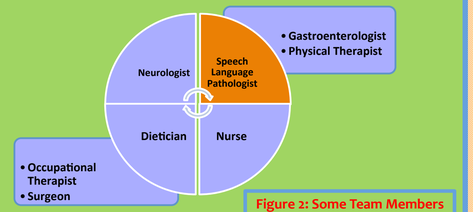

Figure 2: Some Team Members

Role of the Speech Language Pathologist

Speech therapy is occasionally useful as adjunctive therapy, but its role is vital to both disorders (Figure 4). Plowman-Prine et al. (2007) stated that "the scope of practice generally entails assessment and treatment of the oral and pharyngeal phase (or oropharyngeal function)" (p. 96). In SMA, oral structures influencing feeding efficiency are examined and the effect of positioning and head control on feeding and swallowing are considered important (Wang et al., 2007). Plowman-Prine et al. (2009) also established that an evaluation of a swallow typically begins with a clinical swallowing exam (CSE). All authors agreed that if there are concerns about swallowing, a videofluoroscopy (VFSE) should be done to determine presence or absence of penetration or aspiration.

In a VFSE, clinicians utilize a penetration aspiration scale (PAS) with scores from 1 – 8. The first score shows "material does not enter airway," whereas the last (most life-threatening) shows "material enters airway, passes below vocal folds , and no effort is made to eject" (Plowman-Prine et al., 2009, p. 99). The authors emphasized that behavioral swallowing techniques (rehabilitative or compensatory) are the most common treatments (Figure 2) and should be patient specific. Research has yielded that sensory tricks provide tactile stimulation of affected dystonic muscles, thus relaxing them; sensory tricks are a hallmark of dystonia (Plowman-Prine et al., 2009).

Rehabilitative Swallowing Techniques

For e.g., sensory therapies (geste antagonists or oral sensory feedback devices), Masako maneuver, Showa maneuver, Mendelsohn maneuver, shaker head raise, supraglottic swallow

Compensatory Swallowing techniques

For e.g., use of adaptive eating utensils/orthotic devices (Neater Eater, elbow support, & valved straw), head turn, cut food into smaller pieces, chin tuck, liquid wash, double swallow, dietary changes, postural stabilization

Figure 3: Behavioral Techniques
Plowman-Prine et al. (2009)

Dysphagia in SMA and Dystonia

In the pre-oral and oral phases, patients with SMA experience a reduction in muscular effort that contributes to craniofacial anomalies, whereas in dystonia, patients have very rigid muscles, which is debilitating. Both groups of patients require modifications to their diets, but the oral trauma causes rapid wear or early loss of teeth due to bruxism and tongue biting in those with dystonia (Lee, 2007). Semisolid diets are used as compensation for poor chewing and reduce length of mealtimes; thickened liquids are used to protect patients against aspiration of thin liquids and are more palatable (Tilton et al., 1998; Wang et al., 2007).

In addition to the physical abnormalities of the orofacial structures, bolus preparation and propulsion are tedious tasks in both disorders. However, patients with dystonic symptoms have more frequent discomfort during the pharyngeal phase of swallowing. As a result, SLPs use behavioral techniques, but compensatory strategies are more useful to patients with dystonia due to difficulties performing rehabilitative maneuvers. Instead, postural adjustment and geste antagonists such as a light touch to chin or lips, or applying an oral sensory feedback device is used to relax muscles (Charous & Comella, 2011; Verma & Sinhu, 2009). In spite of the differences, several similarities are noted in both SMA and dystonia, specifically in OMD, involving difficulties in mastication. Both disorders may also lead to aspiration if patients are not carefully monitored by SLPs or other feeding specialists.

Conclusions

- Dysphagia correlates with poor head control and patient's level of motor function in SMA.
- Patients with weak swallow or poor oral muscular control seem to do better with semisolids, which can be swallowed in a single bolus.
- Chen (2012) emphasized that the most frequent feeding and swallowing difficulties were chewing difficulties, conveying food to mouth with difficulty, and choking in SMA I and II.
- Speech, chewing, swallowing, and facial expressions can all be affected in dystonia causing considerable functional disability (Lee, 2007).
- Wang (2007) established that "treatment should be aimed at reducing the risk of aspiration during swallow, optimizing efficiency during feeding, and promoting enjoyable mealtimes."
- Long term management is needed; if behavioral treatments are ineffective or unsafe, enteral nutrition must be considered.

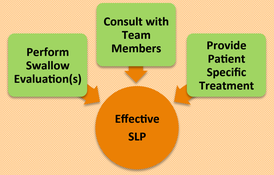

Figure 4: Synopsis of SLP's Roles

References available on separate page