Myotonic dystrophy (DM1) and dysphagia: The need for dysphagia management guidelines and an assessment tool

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Abstract
Myotonic dystrophy (DM1) is the most prevalent muscular dystrophy occurring in adulthood. DM1 is a multi-systemic disorder resulting in early-onset cataracts, cardiac rhythm problems, muscle weakness, ptosis, and cognitive and psychiatric manifestations. Dysphagia is one of the most problematic symptoms of DM1 because it may cause weight loss, aspiration pneumonias or sudden death. The purpose of this review is to describe the characteristics of DM1 that make dysphagia management problematic, and to address the need for disease-specific guidelines and a clinical tool to aid in diagnosing and managing dysphagia in this population.

Introduction
Myotonic dystrophy (DM1) is the most prevalent form of muscular dystrophy in adulthood and it is a multi-systemic, inherited disorder that affects the central nervous system, the endocrine system, skeletal, cardiac and smooth muscle, and the visual system (Machuca-Tzili, Brook, & Hilton-Jones, 2005). Patients with DM1 may also experience disordered sleep, intellectual impairment, emotional problems (Antonini et al., 2006; Rubinsztein, Rubinsztein, Goodburn, & Holland, 1998), and difficulty swallowing. The most frequent causes of morbidity and mortality in the DM1 population include sudden death, chronic respiratory failure, arrhythmias, and pneumonia (de Die-Smulders et al., 1998; Mathieu, Allard, Potvin, Prevost, & Begin, 1999).

There are currently no treatments to cure or slow progression in DM1, and management strategies are meant to prevent complications such as aspiration pneumonias and sudden death secondary to choking. Individuals with DM1 are typically cared for in an ambulatory care unit by a neuromuscular team. Thus, patients are rarely admitted to a neurological ward in a hospital. Instead, complications usually arising from chronic respiratory failure often lead to admission in an intensive care unit.

Health care professionals share concerns that individuals with DM1 may be reluctant or unable to address due to apathy or cognitive impairment. For example, individuals with DM1 may not report swallowing problems, even though researchers and clinicians consider dysphagia one of the most problematic DM1 symptoms due to the propensity to cause aspiration pneumonia (Garrett, DuBose, Jackson, & Norman, 1969, as cited in Bellini et al., 2006) and sudden death from choking. Therefore, it is essential for health care professionals to assess the swallowing function of a DM1 individual who presents to hospital. In particular, early identification and management of dysphagia by nurses may prevent negative outcomes (Ramruti, Finlayson, Mitchell, & Croft, 2001) like choking or aspiration pneumonia.

Unfortunately, there are no disease-specific clinical guidelines or dysphagia evaluation tools to aid health care professionals in assessing or managing swallowing problems in this population. Thus, the purpose of this review is to discuss the symptoms of DM1 that may impact dysphagia identification and management, and to illustrate the need for disease-specific clinical guidelines and a dysphagia clinical evaluation tool.

Methods
CINAHL, EMBASE, PubMed, and SCOPUS were searched combining the subject heading “myotonic dystrophy” and the keywords DM1, dystrophia myotonica, Steinert*, and myotonic dystrophy type 1 with the subject headings “cognition,” “dysphagia/swallow,” and “personality,” resulting in three separate searches for each database. SCOPUS, being a keywords database, was searched using keywords only. Articles searched for this review were limited to work published within the past 15 years. However, articles published prior to 1995 were included if they enhanced the topic. Titles and abstracts were analyzed by the first author for content describing the symptoms, assessment, or management of dysphagia in DM1, in addition to information regarding the cognitive and personality profiles of this population. Articles were excluded if they were not peer reviewed, were not written in English, and did not pertain to adults with DM1. This overview is not meant to synthesize the literature pertaining to the musculature and swallowing evaluation studies of DM1, nor is it an exhaustive search of the literature regarding DM1 symptoms. Further, an assessment of the quality of the articles contained in this review is beyond the scope of this paper. Rather, this paper is meant to discuss the complex symptoms of DM1, particularly those that may make diagnosis and management of swallowing problems difficult.

Myotonic dystrophy type 1
DM1 is an autosomal dominant disorder characterized by muscle weakness and stiffness (myotonia). DM1 is caused by an expansion in the number of cytosine-thymine-guanine (CTG) repeats in an untranslated region of the myotonic dystrophy protein kinase (DMPK) gene. However, DM1 is characterized by anticipation; that is, an increasing length of the CTG repeat size occurs with each generation, resulting in an earlier age at onset and increasing severity of the disease (Machuca-Tzili et al., 2005). Not infrequently, a parent with a milder form of DM1 may serve as a caregiver for his or her more affected children.

DM1 may be present at birth (congenital form), develop in childhood or, more commonly, manifest in adulthood. Unlike most other muscular dystrophies, DM1 is a multi-systemic disease that can cause cardiac conduction deficits, early-onset cataracts, and sleep, endocrine, and respiratory problems (Meola & Sansone, 2005).
2007). Affected individuals with DM1 may have mild to profound facial weakness, variably affecting their ability to manipulate food orally. For a comprehensive review of the molecular and clinical characteristics of DM1, see Machuca-Tzili, Brook, and Hilton-Jones (2005) and Turner and Hilton-Jones (2010).

**Dysphagia and DM1**

The National Institute of Deafness and other Communication Disorders (NIDCD, 2010) defines dysphagia as difficulty swallowing with or without pain that may result in malnutrition or dehydration. The prevalence and incidence of dysphagia in the general population is unknown, but it is estimated to be 22% in those 50 years of age or older (American Speech-Language-Hearing-Association, 2011). Dysphagia may occur as a result of a central nervous system disorder that disrupts the swallowing reflex (NIDCD, 2010) or it may be a consequence of progressive neuromuscular disease (a disease of the motor neuron, nerve, neuromuscular junction or muscle). Clinical indicators of dysphagia include vocal changes (i.e., hoarseness), coughing, food sticking in the throat, shortness of breath and drooling (Boczkó, 2006). It is a common symptom of several neurological disorders including Parkinson’s disease, multiple sclerosis, stroke, dementia, myasthenia gravis, amyotrophic lateral sclerosis, and DM1 (National Institute of Neurological Disorders and Stroke, 2011).

Reports of the prevalence of dysphagia in DM1 patients range from 25% to 80% (Bellini et al., 2006; Ronnblom, Forsberg, & Danielsson, 1996). For patients with DM1, their weak facial muscles may prevent them from moving food in their mouths and chewing effectively. In particular, muscle weakness in the face, tongue, jaw, and pharyngeal muscles may cause nasal regurgitation and difficulty swallowing (Willig, Paulus, Lacau Saint Guily, Beon, & Navarro, 1994). Gastrointestinal smooth muscle involvement is common in DM1 patients (Ronnblom & Danielsson, 2004), and the most prevalent dysphagia-related complaints include heartburn, regurgitation, coughing while eating, and choking (Bellini et al., 2006; Ronnblom et al., 1996). Neuromuscular patients with dysphagia may also experience malnutrition, weight loss, aspiration pneumonia, and social embarrassment (Chaudry, Umapathi, & Ravich, 2002; Hill, Hughes, & Milford, 2008).

Not uncommonly, pneumonia develops secondary to aspiration, which highlights the importance of the diagnosis and treatment of swallowing dysfunction in this population. In a subset of DM1 patients from the Netherlands, pneumonia was the most frequent cause of death (de Die-Smulders et al., 1998), and respiratory failure and pneumonia also accounted for a high mortality rate in a cohort of patients in Quebec (Mathieu et al., 1999). All too commonly, an aspiration triggers acute or chronic respiratory failure and an ICU admission for supported ventilation. Since dysphagia is a treatable condition, early diagnosis is critical.

Unfortunately, individuals with DM1 are unlikely to report swallowing problems even when there is evidence of aspiration on videofluoroscopy (Turner & Hilton-Jones, 2010). It is unknown if individuals do not report symptoms because they do not find them bothersome, or because the disease progresses slowly and they have adapted or compensated for their swallowing difficulties (Constantini et al., 1996; Leonard, Kendall, Johnson, & McKenzie, 2001; Modolell et al., 1999). It is also possible that intellectual impairment or apathy may prevent individuals from reporting swallowing dysfunction.

Regardless, it is important that health care providers are vigilant, and that caregivers are involved in the assessment process. Interestingly, while there was a correlation between disease duration and dysphagia in 15 DM1 individuals, there did not seem to be a clear correlation between muscular disability and severity of dysphagia (Marcon et al., 1998). As a result, health professionals may not check for swallowing dysfunction, thus interfering with an accurate assessment of dysphagia. There is, however, a correlation between the number of CTG repeats and the degree of dysphagia-related musculature abnormalities seen on videofluoroscopy.

Another feature of DM1 that may impact swallowing function is hypersonmnelence. Hypersonmnelence may cause poor cognitive functioning, problems focusing, mood disorders, and accidents (Happe, 2003). Thus, excessive sleepiness, in addition to other psychological and physiological features, may impact swallowing because of its prominence and the potential to lead to choking.

**Implications of cognitive, psychiatric and intellectual symptoms on swallowing**

It is essential to consider the cognitive, intellectual and emotional symptoms of DM1 to understand why individuals with DM1 do or do not follow dietary advice and modifications. Several studies have formulated cognitive and behavioural traits and a personality profile for DM1 patients (Delaporte, 1998; Meola et al., 2003; Sistiaga et al., 2010; Winblad, Lindberg, & Hansen, 2005); in particular, “patients showed a lack of energy, motivation, and commitment in a way which suggested these were primary symptoms of the disease” (Bungener, Jouvent, & Delaporte, 1998, p. 353). From their clinical experience, Meola and Sansone (2007) report that DM1 patients tend to minimize their symptoms, may not keep clinic appointments, and generally seem indifferent about their health. Further, cognitive impairment may prevent DM1 individuals from participating fully in work or social activities (Sansone et al., 2007).

Individuals with severe forms of DM1 have cognitive and intellectual impairment, while those with a milder form may experience difficulties with attention, visual memory, perception, planning, processing and organizing new information, or executive function (Rubinsztein, Rubinsztein, McKenna, Goodburn, & Holland, 1997; Sistiaga et al., 2010; Zalonis et al., 2010). Interestingly, a study of 50 DM1 patients comparing MRI findings with neuropsychological tests found that individuals with major and global white matter lesions have significant cognitive impairment (Romeo et al., 2010). Finally, Rubinsztein et al. (1998) concluded that apathy, hypersonmnelence and cognitive impairment are core features of DM1.

Moodiness, emotional blunting and a lack of motivation were recognized even in the very early literature (Rohrer, 1916). Moreover, in a study including 21 DM1 patients, 86% demonstrated an avoidant personality, reporting reluctance to make new friends, to participate in new activities, or take risks (Meola et al., 2003). Further, in a cross-sectional study of
Few studies have explored dysphagia management for individuals with intellectual impairments (Chadwick, Jolliffe, & Goldbart, 2002; 2003). Determining if intellectual and cognitive impairments affect patient self-report or their willingness and ability to participate in behavioural or dietary modifications is important since these limitations may impact diagnosis and treatment for the DM1 population. One potential way of increasing the reliability of self-report measures and improving care for DM1 patients with dysphagia is to educate family members and caregivers by including them in the assessment and management process. In particular, caregivers may be responsible for facilitating safe swallowing by instituting special techniques such as modifying the texture of food and liquids, adhering to food restrictions, and prompting patients about chewing techniques.

Case report

A 59-year-old married female diagnosed with myotonic dystrophy type 1 has been followed in the neuromuscular clinic for the past 13 years. Her son, brother and five half siblings have also been diagnosed with this autosomal-dominant disorder. Her presenting symptom was the inability to release a cup with her hand (myotonia), followed by muscle weakness in the arms and hands and pervasive fatigue. She is now bothered by dysphagia, neck pain, morning headaches and balance difficulties. Two years ago, she experienced a choking episode at a family picnic while eating steak on a bun. Despite her husband’s requests, she refused to attend the local emergency room until the following day. At that point she still perceived something lodged in her throat and she required bronchoscopy to remove the aspirated food. Both the patient and her husband were advised to undergo CPR training for the Heimlich manoeuvre. The patient continues to prop herself up with pillows while she is sleeping because she chokes on her saliva when lying flat. She does not follow recommendations for safe eating despite multiple assessments by a speech language pathologist and modified barium swallows. The swallowing profile was summarized as follows: slow and reduced mastication with difficulty clearing solid foods from the oral cavity, mild to moderate oral and pharyngeal dysphagia with a risk for aspiration for thin fluids. It was recommended that a pureed/blended diet with repeat swallows to clear residue be instituted.

Current dysphagia management guidelines and DM1-specific management dilemmas

A group of 26 experts across seven clinical specialties composed a National Guideline Clearinghouse panel (U.S. Department of Health and Human Services, 2010) and met to recommend guidelines for the diagnosis and management of dysphagia. Unfortunately, the only neuromuscular conditions used in creating the guidelines include amyotrophic lateral sclerosis and myasthenia gravis. It is unknown if DM1 individuals are grouped in the neurologial impairments listed under “degenerative disease”. Regardless, the recommendations suggest that individuals presenting with dysphagia should undergo a videofluoroscopic swallowing evaluation (VSE) or a fiberoptic endoscopic evaluation of swallowing (FEES). The VSE and FEES will determine if treatment is required, namely if a special diet or compensatory strategies are needed to facilitate safe swallowing. Those presenting with muscular weakness may benefit from strength training the swallowing muscles (with or without electromyographic feedback). In extreme cases, surgical intervention may be necessary. For patients presenting with a cough, patients and caregivers should be questioned about perceived swallowing problems and their fears about choking. A multidisciplinary team including a physician, nurse, dietitian, speech language pathologist, PT and OT should be used in assessing and managing patients with dysphagia.

Unfortunately, these guidelines are not specific to individuals with DM1. In addition, while VSE is considered the gold standard, it has several limitations in that it exposes patients to radiation, it may not be standardized at each testing centre in regard to food texture and consistency, and it is labour intensive (Perry & Love, 2001). As such, a clinical evaluation may be important for dysphagia identification (Perry, 2001). However, a clinical evaluation of individuals with DM1 may be difficult because they commonly deny swallowing problems when questioned during routine follow-ups.

If family members are present, a clear history of coughing with eating, frequent choking and near misses with choking episodes of concern is often elicited. Even with this information, patients will continue to downplay the severity of the problems. In some instances it may be lack of insight into their health problems because “they have always been this way”. It is also not unusual for patients to recall or continue to be in close contact with other family members who have the same problems and are managing despite obvious symptoms of dysphagia.

In clinical practice, especially if the individuals do not attend school or work, the sleep/wake cycle is altered. Therefore, it is not unusual for patients to start their day at 12 noon or later, and return to bed after midnight. From a practical perspective, many miss breakfast and often admit to eating only one or two meals a day. Esophageal dysmotility also contributes to increased aspiration risk in this population. Therefore, treatment of reflux is also important. Nutritional concerns need to be addressed at every clinical encounter including objective assessments of weight, and BMI calculations.

The importance of a DM1-specific guidelines and dysphagia evaluation tool

Given that dysphagia, a treatable condition, is one of the major causes of morbidity and mortality in DM1, specific guidelines for diagnosing and managing it are crucial. Unfortunately, management for DM1 patients and their families is challenging for a number of reasons including time needed for proper assessment, variability in disease presentation and course,
Arguably, dysphagia management may reduce the incidence of pneumonias, thus reducing health care costs and improving quality of life for patients. It is critical to develop a questionnaire or other clinical evaluation tool to identify swallowing dysfunction in DM1 patients to begin to develop methods to reduce morbidity and mortality in this population.

Conclusion

Individuals with DM1 manifest a multitude of symptoms. Arguably, one of the most troubling is dysphagia, and individuals presenting to hospital for any reason need to be assessed for swallowing dysfunction and provided with education and recommendations for safe swallowing. As discussed, however, the complexity of DM1 renders diagnosis and management of dysphagia problematic. The absence of disease-specific evaluation tools or management guidelines for DM1 is likely a contributing factor. We propose that disease-specific diagnosis and management guidelines would facilitate earlier identification with the goal of improving quality of life and reducing complications that lead to increased morbidity and mortality. Research investigating the creation of disease-specific assessment tools is warranted. In particular, a mixed methods approach using qualitative and quantitative strategies is likely required to fully appreciate the many facets of this condition. In the meantime, vigilance of all health care professionals involved in the care and management of those living with DM1 is necessary to minimize dysphagia-related complications.

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