

Review Article

Myotonic Dystrophy Type 1 (DM1) and Speech Problems

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Submitted: 23 March 2017

Accepted: 07 June 2017

Published: 09 June 2017

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Abstract

Myotonic Dystrophy type 1 (DM1), also called Steinert syndrome, is a multisystemic disorder transmitted in an autosomal dominant manner, characterized by myotonia. Muscles involved in voluntary movement are highly affected by myotonia especially distal muscles of upper limbs. Patients with DM1 present a myopathic face and oropharynx weakness. Reduced motor mobility and saliva flux can lead to gingival inflammation and periodontal disease together with other oral manifestations like disturbances at the temporomandibular articulation. Main causes of death are pneumonia and cardiac arrhythmias.

Although the etiology of this syndrome is well known, a specific treatment for this disease is still not available. Nowadays, treatments consist on the relief of existing symptoms, in an attempt to give a better life quality to patients. It is very important to implement actions that can prevent complications and this is why treatments should be applied in an early stage of the disease.

It is the aim of this paper to clarify the etiology, systemic characteristics of the syndrome and in particular discuss how myotonia can lead to speech disturbances and present strategies to deal with this particular problem.

Keywords

- Myotonic dystrophy
- Steinert disease
- Speech problems
- Lip force in DM1
- Tongue pressure in DM1
- Muscular weakness and dysarthria in DM1

ABBREVIATIONS

DM1: Myotonic Dystrophy Type 1; DM2 - Myotonic Dystrophy Type 2; *DMPK*: *Dystrophia myotonica Protein Kinase gene*

INTRODUCTION

Muscular dystrophies are a heterogeneous group of genetic neuromuscular disorders characterized by necrosis and progressive muscular weakness [1]. There are diverse types of muscular dystrophies and myotonic dystrophies are the most common form in adults, having an incidence that varies among different populations: from 1/475 in some populations in Canada [2] to very rare in Africans, with only one Nigerian family being reported with this type of dystrophy [3].

Myotonic dystrophies have an autosomal dominant heritage and can be divided in Myotonic Dystrophy type 1 (DM1) or Steinert Syndrome and Myotonic Dystrophy type 2 (DM2) earlier known as Proximal Myotonic Myopathy [4,5]. These dystrophies have different genetic causes: DM1 results from a triplet repeat while DM2 results from a CCTG expansion present in intron 1 of the gene coding zinc finger protein 9 [4,5]. Both forms show incomplete penetrance, variable expression and anticipation [6]. DM1 is divided in five subtypes according with the severity of symptoms [6,7].

DM1 mutation consists of an expansion of a CTG triplet repeat in the *dystrophia myotonica protein kinase gene (DMPK)*

that codes for myotonin [8]. This protein is expressed in smooth, skeletal and cardiac muscle [8] and at lower levels in brain and endocrine system [9]. *DMPK* alleles present in DM1 patients have a number of CTG repeats between 50 and 4000 [10]. Pathogenicity of myotonic dystrophy is still not clear. Bhagavati et al. [11], suggested that myotonin low levels could lead to apoptosis of muscular cells. But, in fact, it is difficult to establish a relationship between this protein and the disorder since its interaction with other proteins is not yet known.

DM1 and DM2 share some clinical phenotypes like muscular weakness, myotonia, cataracts, multiorgan involvement with cardiac conduction defects, insulin resistance and gonadal atrophy. Dysphagia (for liquids and solid food), abdominal pain and with constipation, are also present in both dystrophies, however, these symptoms are milder in DM2 when compared with DM1 [4,5]. Muscular damage is the main phenotypic characteristic of DM1. Together with it, patients also show several systemic traits being the most common muscular, cardiac, respiratory, CNS, ocular, gynecological, digestive, orthopedical, cognitive and psychological symptoms with different degrees of severity [12].

Muscles of voluntary movement especially upper limbs are the most affected by myotonia. DM1 patients also show changes in face, chewing and pharynx muscles, resulting in swallowing difficulty and dysphagia that can provoke food aspiration and suffocation [13]. Patient's decreased motor mobility and reduced

saliva flux contribute to poor oral hygiene, gingival inflammation and periodontal disease [14]. Myotonia of face and tongue muscles leads to changes in tongue pressure [15] and frequently results in speech problems [13].

In DM1 patients the central nervous system may also be affected leading to sleep disturbances and learning capacity difficulties that directly interfere with patient's life quality [16] and some patients may develop an autism spectrum disorder [17]. The main causes of death of DM1 patients are pneumonia and cardiac arrhythmias [13,18]. Due to the systemic picture of DM1 manifestations, the main goal of this work is to perform a systematic review of main manifestations of the disorder, concentrating on face muscular weakness and how this affects cognitive functions and speech, since these are fundamental in communication with the patient in order to evaluate multiple symptoms and diagnose the stage of the disease.

MATERIALS AND METHODS

For this work, several searches were performed using the websites: PubMed, B-On, SciELO, Science Direct, as well as libraries of O'Porto Medicine Faculty (FMUP) and Fernando Pessoa University. The following keywords were used: "Myotonic dystrophy", "Steinert disease", "Speech problems", "Lip force in DM1", "Tongue pressure in DM1", "Muscular weakness and dysarthria in DM1" that were interconnected in several ways. Inclusion criterion used was papers written in English, Portuguese, French or Spanish with content pertinent for the study. Another criterion used was that selected papers were new literature about the chosen theme.

SPEECH DISTURBANCES IN DM1 PATIENTS

Speech disturbances in DM1 patients have been frequently reported and there are several factors contributing to it. These problems can result from velopharyngeal insufficiency and may be the first and only presenting symptom in DM1 patients [19]. Hillarp et al. [19], stated that the analysis of speech and swallowing capacity by video radiography can reveal the presence of DM1 without prior suspicion of the disorder.

Myotonia of face muscles is the main factor leading to changes in speech often observed in DM1 patients [6]. This myotonia, together with muscle weakness, culminates in the frequent atrophy of face and neck muscles of these individuals that contributes to their speech difficulties [6,20].

According to Swart et al. [21], myotonia hinders the start of articulation and reduces speech rate while muscular weakness leads to flaccid dysarthria [22]. Swart et al. [21], stated that as myotonia onset is unpredictable, patients cope very poorly with it. However, they are more aware and deal better with muscular weakness since it is constantly present.

As mentioned, myotonia normally interferes with the moment the individual starts to speak, but some patients also experience problems during the conversation. Myotonia seems to affect the time, manner, and place of articulation [21]. As a result, some patients have more problems with bilabial sounds, and others with alveolar or velar sounds. Usually, plosive sounds seem to cause more difficulties than fricative sounds, and talking

loudly and shouting seem to increase myotonia [23]. In the same way, Sancho et al. [24], also detected speech problems in 75% of analyzed patients reflected as weakness of plosive phonemes and slow/inaccurate speech due to nasal voice [24].

Muscular weakness also leads to reduced lip force that is often observed in DM1 patients (children and adults) and contributes to the development of speech disturbances. A study of Sjögreen et al. [25], observed six patients having no speech at all between 56 children and adolescents with DM1, together with impaired facial expression, reduced intelligence, eating problems and drooling. In this study, lip force was measured and the majority of analyzed patients had very weak lips compared to healthy controls [25].

A third frequent factor also contributing to the evident speech problems is the observed atrophy of tongue muscles that leads to changes in tongue pressure [15]. Some patients also exhibit disturbances of the temporomandibular articulation that provoke tiredness and pain during mouth opening [26-28]. These changes together with the previously mentioned disturbances, contribute to speech changes. Moreover, but less frequent, maxillary bone deformities can be observed. These deformities were detected by Peñarrocha et al. [29], in two DM1 patients that, among other symptoms, exhibited speech alterations as a consequence of those changes.

TREATMENT OF SPEECH PROBLEMS

Some studies have been performed in order to test treatments to reduce speech problems developed by DM1 patients. Table 1 summarizes the goals and observed results of discussed treatments. Some studies [30,35] recommend physiotherapy stating that it does not increase the risk of muscular damage. However, it is still not clear if physical exercise can improve strength and motor function and postpone the progression of muscular atrophy in DM1 patients [30]. As discussed previously, one of the phenotypes exhibited by DM1 children and adolescents is weak lips and impaired lip functions that interfere with speech. Although several studies have been done on the use of oral motor exercises to improve lip strength, there is still no consensus on

Goal	Treatment Used	Results
Improve strength and motor function	Physiotherapy	Not clear [30]
Reduce or eliminate myotonia during speech	Warming up	Increased speech rate [21]
Increase lip strength	Exercises with an oral screen	Substantial increase in lip force [31]
		Decrease in orofacial muscle activity [32]
		Improved lip force [33]
		Improved lip force and lip force endurance but no effect on lip articulation [34]

patient's profit.

In order to increase lip strength, work of Owman-Moll and Ingervall [31] studied the effect of exercises with an oral screen normally used for orthodontic treatment in developing children. These researchers observed a substantial increase in lip force after one year of treatment of children with incompetent lip function [31].

Another study also used an oral screen in the treatment of children with lip and/or tongue dysfunction [32]. The goal was to determine the effect of this treatment on orofacial muscle activity and possible changes in craniofacial morphology. Their results indicated a decrease in orofacial muscle activity during oral functions but no significant change in tooth position [32]. Moreover, Hägg et al. [33], found that self-training with an oral screen could improve lip force and swallowing capacity in stroke patients with oropharyngeal dysphagia.

In the same way, Sjögreen et al. [34], applied the use of regular training with an oral screen in children and adolescents with DM1 in order to determine if it could strengthen lip muscles and, if so, to see if this could have an immediate effect on lip functions such as lip mobility, eating and drinking ability, saliva control, and lip articulation [34]. This study saw an improvement of lip force and lip force endurance in tested patients, however increased lip strength by itself did not have an effect on lip articulation, saliva control or eating ability [34]. These researchers concluded that lip strengthening exercises can be used as a complement of speech therapy and dysphagia treatment but cannot replace those therapies [34,35].

Another study tried to determine if warming up could reduce or eliminate myotonia during speech production or, on the other hand, would lead to adverse effects due to fatigue or exhaustion caused by intensive speech activity in patients with adult onset myotonic dystrophy [21]. It is documented that patients show myotonia of the hands at the start of activities but diminishes after warming up by repeated muscle contractions [36]. Swart et al. [21], observed that warming up increased speech rate and decreased speech variability. This prolonged and intensive use of speech musculature did not cause fatigue or exhaustion of DM1 patients. Researchers observed that after warming up, DM1 patients were able to achieve speech rate similar to healthy individuals [21]. However, the increase in speech rate after warming up varied according to the sound sequences used. This study observed no significant increase in speech rate when using (pa) sound sequences while the (ka) sequences showed higher increase in speech rate [21]. Swart et al. [21], explain this observation based on more muscle activity and tension required in the (ka) sounds production, resulting in a greater influence of the myotonia.

In the same study, researchers also aimed to determine if flaccid dysarthria symptoms increase after intense speech activity [21]. The reason for examining this is that even if warming up is effective, it is not valuable if it leads to fatigue or exhaustion. They verified that flaccid dysarthria does not increase after prolonged and intensive use of speech musculature [21].

DISCUSSION & CONCLUSION

Muscular weakness and myotonia are the main causes of

the phenotypic characteristics of DM1 patients that exhibit a group of symptoms like face and tongue muscular atrophy, changes in tongue pressure, lip weakness and disturbances of the temporomandibular articulation, directly related to the frequent speech problems observed in these individuals.

Some studies report the use of physical exercises to improve strength and motor activity but there is still no consensus on patient's profit. Only a few studies have addressed the problem of speech disturbances, using strategies to improve speech production by decreasing myotonia.

In order to increase lip strength, Sjögreen et al. [33], used an oral screen in DM1 children's training. Results of this study showed an increase in lip strength that did not result in increased lip articulation [33].

Another study detected that the speech of patients with adult onset of DM1 shows signs of flaccid dysarthria and myotonia [21]. According with this study, myotonia has a greater influence at the start of a speaking episode but it decreases after warming up during a continuous speech. However, the observed effect depends on the type of sound being produced. In this study, they observed that warming up increased the number of syllables per second that the patient could produce achieving a higher speech rate and a more regular performance [21]. So researchers conclude that warming up is advantageous for patients with adult onset DM1 since it contributed to reduction or even elimination of the effect of myotonia on speech muscles [21]. It is possible to conclude that lip exercise and face muscle warm-up can help but are not sufficient to correct speech disturbances of DM1 patients. These strategies should be used to complement speech therapy.

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Cite this article

Cardoso IL, Baptista H (2017) Myotonic Dystrophy Type 1 (DM1) and Speech Problems. *JSM Communication Dis* 1(1): 1003.