

Longitudinal performance of swallowing in myotonic dystrophy type 1

Desempenho longitudinal da deglutição orofaríngea na distrofia miotônica tipo 1

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ABSTRACT

The purpose of the present study was to describe the longitudinal performance of oropharyngeal swallowing in individuals with type 1 myotonic dystrophy. A single case report of a 66-year-old man with a neurological diagnosis in 2010. He was submitted to his first clinical and objective evaluation of swallowing four years after the neurological diagnosis. Seven objective evaluations of swallowing were performed by fiberoptic endoscopic evaluation of swallowing using pureed food, thickened liquid and liquid consistencies (3, 5, and 10 ml) during the diagnosis and management of swallowing over a period of one year and two months. Laryngeal sensitivity, oral spillage and pharyngeal residues were evaluated using the Yale Pharyngeal Residue Severity Rating Scale, and laryngeal penetration and/or laryngotracheal aspiration were determined using the Penetration-Aspiration Scale (PAS). No change in laryngeal sensitivity was observed during the study period, whereas oral spillage, pharyngeal residues and laryngeal penetration were observed since the beginning of the objective evaluations. Four months after the first evaluation, the level of pharyngeal residues of pureed consistency changed from trace to moderate in piriform recess, and in the vallecula the increase in the severity index was demonstrated in the last month. There was an increase in PAS score for all consistencies tested. Laryngotracheal aspiration occurred with thin liquid in the last month. During the follow-up of oropharyngeal swallowing in myotonic dystrophy type 1, pharyngeal residues and laryngeal penetration were present since the beginning of the evaluations, but laryngotracheal aspiration occurred only in the last month of follow-up and with thin liquid.

Keywords: Deglutition; Deglutition disorders; Endoscopy; Myotonic dystrophy; Neurology

RESUMO

Este estudo teve por objetivo descrever o desempenho longitudinal da deglutição orofaríngea em indivíduo com distrofia miotônica tipo 1. Estudo de caso único de indivíduo de 66 anos, sexo masculino, com diagnóstico neurológico em 2010. Realizou a primeira avaliação clínica e objetiva da deglutição após quatro anos do diagnóstico neurológico. Foram realizadas sete avaliações objetivas da deglutição, por meio de videoendoscopia de deglutição, nas consistências pastosa, líquida espessada e líquida, com 3, 5, 10 ml, durante o processo de diagnóstico e gerenciamento da deglutição, por um ano e dois meses. Foram analisados sensibilidade laríngea, escape oral posterior, resíduos faríngeos, por meio da *Yale Pharyngeal Residue Severity Rating Scale*, penetração laríngea e/ou aspiração laringotraqueal, com aplicação da *Penetration-Aspiration Scale* (PAS). Constatou-se, durante o período de estudo, que não houve alteração na sensibilidade laríngea. Escape oral posterior, resíduos faríngeos e penetração laríngea estiveram presentes desde o início das avaliações objetivas. Após quatro meses da primeira avaliação, na consistência pastosa, o nível de resíduos faríngeos passou de vestígio residual para moderado, em recessos piriformes, já em valéculas, e o aumento no índice da gravidade evidenciou-se no último mês. Houve aumento na PAS em todas as consistências de alimento testadas. A presença de aspiração laringotraqueal ocorreu com líquido ralo, no último mês. Durante o período de acompanhamento da deglutição orofaríngea na distrofia miotônica tipo 1, os resíduos faríngeos e a penetração laríngea estiveram presentes desde o início das avaliações, porém, a aspiração laringotraqueal somente ocorreu no último mês do acompanhamento, com líquido ralo.

Descritores: Deglutição; Transtornos de deglutição; Endoscopia; Distrofia miotônica; Neurologia

Study carried out at Laboratório de Disfagia – LADIS, Departamento de Fonoaudiologia, Faculdade de Filosofia e Ciências – FFC, Universidade Estadual Paulista “Júlio de Mesquita Filho” – UNESP – Marília (SP), Brasil.

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INTRODUCTION

Oropharyngeal dysphagia is characterized by difficulties in food transport along the path from the mouth to the stomach. It is a symptom of high incidence and prevalence ranging from 25% to 80% among neurodegenerative diseases^(1,2) including myotonic dystrophy type 1 (DM1), or Steinert disease⁽³⁾.

DM1 is considered to be the most frequent form of muscular dystrophy among adults. It is a multisystemic disease of autosomal dominant inheritance due to expansion of the CGT triad (cytosine, guanine and thymine) on the long arm of chromosome 19, with an estimated prevalence of about 2.1 to 14.3/100,000 in the general population. The disease is characterized by extensive and distinct manifestations such as ocular and reproductive changes, diabetes, gastrointestinal alterations, oropharyngeal dysphagia, and even intellectual deficiency. However, it is fundamentally characterized by muscle weakness mainly involving the distal muscles, with worsening occurring when the proximal muscles are attacked⁽⁴⁾.

Although few studies have been conducted on oropharyngeal swallowing in DM1, there are reports of changes in swallowing with the presence of laryngeal penetration and laryngotracheal aspiration, pharyngeal residues, and multiple swallowing. The progressive muscle weakness affecting this population may have an impact on the oral and pharyngeal phases of swallowing, negatively affecting the efficiency and safety of swallowing⁽³⁻⁶⁾.

Thus, early screening and assessment of oropharyngeal swallowing is essential for this population, so that conducts may be adopted in order to minimize the risks of feeding safety and efficiency. Fiberoptic Endoscopic Evaluation of Swallowing (FEES) is an objective method for the assessment of the pharyngeal phase of swallowing and of the impact of muscle weakness on the physiopathology of swallowing, with the identification of posterior oral spillage and of the presence of pharyngeal residues, laryngeal penetration and laryngotracheal aspiration⁽⁷⁻¹⁰⁾.

Thus, in view of the pharyngeal muscle weakness present in DM1, and considering the absence of studies with longitudinal follow-up of oropharyngeal swallowing in DM1 describing the manifestations observed during the progression of the disease, it is relevant to report information about these aspects in order to collaborate with interdisciplinary teams so that they will adjust their conducts regarding the oral pathway and manage the nutritional and pulmonary risks as well as the quality of life of this population. On this basis, the objective of the present study was to describe the performance of oropharyngeal swallowing in subjects with myotonic dystrophy type 1.

CASE REPORT

The study was approved by the Research Ethics Committee of the Faculty of Philosophy and Sciences – FFC, São Paulo State University, Marília Campus, SP (protocol no. 0176/2009) and the patient gave written informed consent to participate.

This was a descriptive longitudinal study of a 66-year-old single adult male with a diagnosis of muscular dystrophy type 1 (DM1), or Steinert disease, conducted at the Research Center on Human Genome and Stem Cells in 2010.

In 2014, the patient was referred by a Basic Health Unit team for specific evaluation of oropharyngeal swallowing at the Dysphagia Laboratory (LADIS) of the São Paulo State University “Júlio de Mesquita Filho”- UNESP, Marília Campus, SP, due to a complaint of difficulty in swallowing.

In April 2014, the patient was submitted to clinical evaluation and to FEES for the diagnosis of oropharyngeal swallowing. The patient reported frequent choking episodes with pureed food and a weight loss of approximately 5 kg during the four months preceding the evaluation, and a current weight of 60.9 kg. He also reported difficulties with the oral phase of swallowing, a sensation of food “sticking” in the pharyngeal region after swallowing and the need for approximately 30 minutes in order to finish a meal. He denied pulmonary interurrences or other associated diseases and reported the use of antiepileptic medication twice a day. On the date of evaluation the patient breathed spontaneously and fed orally.

After speech therapy clinical evaluation he was submitted to objective instrumental evaluation by the otolaryngologist in charge according to the protocol of the institution using a Pentax® nasofibroscope and a Pentax® light source model LH-150 PC. The images were captured with Zscan 6.0® software and stored in a computer.

For the exam, the patient was instructed to remain sitting and the instrument was introduced through the more pervious nasal fossa without the use of topical anesthesia. For the dynamic swallowing study, monitored by a speech therapist, standardized pureed and thickened liquid consistencies were offered (levels 4 and 3, respectively, of the International Dysphagia Diet Standardisation Initiative (IDDSI)⁽¹¹⁾), both consistencies were prepared with peach flavored diet juice and instant food thickener consisting of modified cornstarch and maltodextrin, and thin liquid (level 0 of the IDSSI⁽¹¹⁾). A blue dye was added in order to facilitate food visualization. The consistencies were offered on disposable spoons in quantities of 3, 5 and 10 ml and the sequence was interrupted only when safety was compromised. The parameters analyzed were laryngeal sensitivity, posterior oral spillage, pharyngeal residues, and laryngotracheal penetration and aspiration, as described below:

1. Laryngeal sensitivity: glottic adduction response in the epiglottic fold: The test was conducted by touching the arythenoid region and anepiglottic folds with the distal tip of the endoscope. Data were analyzed considering presence, unilateral presence (when the cough reflex occurred with or without adduction movement of the vocal fold on the tested side), and absence (when none of the above events occurred)⁽¹²⁾. The score was: 0 = absence; 1 = presence; 2 = unilateral presence.
2. Posterior oral spillage: food spillage from the base of the tongue to different pharyngeal regions before the occurrence of the swallowing response. The score was: 1 = present, when the food bolus reached the base of the tongue, valleculae, lateral walls of the pharynx or the piriform recess before white out⁽¹³⁾, and 0 = absence.
3. Pharyngeal residues, food stasis in the region of the valleculae and piriform recess at five levels of involvement after swallowing. Data were analyzed using the Yale Pharyngeal Residue Severity Rating Scale⁽¹⁴⁾, which

classifies the residues present in the valleculae and piriform recess according to the following five levels of involvement: 0 = absence; 1 = vestigial residue; 3 = mild; 4 = moderate, and 5 = severe.

- Laryngeal penetration and/or laryngotracheal aspiration: the vocal folds were used as an anatomical marker. Laryngeal penetration was considered to have occurred when the food remained above the vocal fold and laryngotracheal aspiration was considered to have occurred when there was food passage below the level of the vocal folds. Data were analyzed by applying the Penetration-Aspiration Scale (PAS)⁽¹⁵⁾, which proposes eight levels of increasing involvement from 0 = material that does not enter the airway to 8 = food entry into the lower airway without effort for the removal of the aspirated material.

After diagnostic, clinical and objective evaluation by FEES in April 2014, the patient continued to be under swallowing management within the Management and Rehabilitation Program for Oropharyngeal Dysphagia in Neurodegenerative Diseases of the service. Oropharyngeal swallowing management was conducted over a period of one year and two months with the evaluation and rehabilitation program of the service. After the patient accepted follow-up, also for research purposes, he was submitted to seven objective evaluations of swallowing, each performed after clinical speech therapy. The intervals between sessions ranged from 30 minutes to 72 hours in order to perform longitudinal monitoring of oropharyngeal swallowing according to the protocol of the service.

The patient received 13 sessions of the traditional therapy during the first semester and 11 during the second semester of 2014, twice a week with a speech therapist and was

instructed three times for home training. The general objective of the sessions was to increase the efficiency and safety of oropharyngeal swallowing. In order to achieve specific objectives such as increasing oral propulsion, clearance of pharyngeal residues and protection of the lower airways the patient was submitted to therapeutic techniques such as effort swallowing, multiple swallowings, alternation of food consistency, and postural maneuvers of flexed head while ingesting food of liquid or thickened liquid consistency. Three control FEES were performed during the first trimester of 2014. In 2014 and 2015 the patient was kept under management of swallowing with reevaluation and instructions, with two more FEES during the second semester of 2014. The last objective instrumental evaluation was performed at the end of the first semester of 2015. Pharyngeal residues were found to be present since the initial evaluations regarding all the food consistencies and volumes tested regardless of the region, also with the observation of laryngeal penetration of pureed food and thin liquid. The increased level of severity of pharyngeal residues of pureed occurred in piriform recess with volumes of 5 and 10 ml in the last month, after the first objective evaluation. The severity score of laryngeal penetration was higher for all the food consistencies tested, with a volume of 5 ml starting during the fourth month after the first objective evaluation, whereas laryngotracheal aspiration occurred only with thin liquid on the occasion of the last objective evaluation during the first semester of 2015. After this evaluation, the previous instructions were maintained, with the discontinuation of thin liquids and with the patient being referred to pneumology and continuing to be under the management of swallowing (Tables 1-3).

Table 1. Distribution of oropharyngeal swallowing findings regarding food of pureed consistency in myotonic dystrophy type 1

LS	FEES 09/04/2014		FEES 14/05/2014		FEES 11/06/2014		FEES 18/06/2014		FEES 13/08/2014		FEES 29/10/2014		FEES 17/06/2015	
	1		1		1		1		1		1		1	
	5 ml	10 ml	3 ml	5 ml	3 ml	5 ml	3 ml	5 ml	5 ml	10 ml	3 ml	5 ml	5 ml	10 ml
POS	1	1	1	1	1	1	1	1	1	1	1	NT	1	NT
PR in VL	2	2	2	2	1	1	1	2	2	2	2	NT	3*	NT
PR in PRS	1	2	1	2	1	1	1	1	3*	3*	1	NT	2	NT
LP	0	0	0	0	0	2	0	3*	5*	5*	2	NT	3*	NT
LA	0	0	0	0	0	0	0	0	0	0	0	NT	0	NT

*More severe level

Legend: FEES: Fiberoptic Endoscopic Evaluation of Swallowing; LS: Laryngeal sensitivity; POS: Posterior oral spillage; PR: Pharyngeal residues; VL: Valleculae; PRS: Piriform recess; LP: Pharyngeal penetration; LA: Laryngotracheal aspiration; NT: Not tested

Table 2. Distribution of oropharyngeal swallowing findings regarding food of thickened liquid consistency in myotonic dystrophy type 1

LS	FEES 09/04/2014		FEES 14/05/2014		FEES 11/06/2014		FEES 18/06/2014		FEES 13/08/2014		FEES 29/10/2014		FEES 17/06/2015	
	1		1		1		1		1		1		1	
	5 ml	10 ml	3 ml	5 ml	3 ml	5 ml	3 ml	5 ml	5 ml	10 ml	3 ml	5 ml	5 ml	10 ml
POS	1	1	1	1	1	1	1	1	1	NT	1	1	1	NT
PR in VL	1	2	1	1	1	1	2	2	1	NT	1	2	2	NT
PR in PRS	1	1	1	2	1	1	1	2	1	NT	1	2	2	NT
LP	3*	3*	0	0	0	2	3*	3*	5*	NT	2	3*	5*	NT
LA	0	0	0	0	0	0	0	0	0	NT	0	0	0	NT

*More severe level

Legend: FEES: Fiberoptic Endoscopic Evaluation of Swallowing; LS: Laryngeal sensitivity; POS: Posterior oral spillage; PR: Pharyngeal residues; VL: Valleculae; PRS: Piriform recess; LP: Pharyngeal penetration; LA: Laryngotracheal aspiration; NT: Not tested

Table 3. Distribution of oropharyngeal swallowing findings regarding food of liquid consistency in myotonic dystrophy type 1

LS	FEES 09/04/2014		FEES 14/05/2014		FEES 11/06/2014		FEES 18/06/2014		FEES 13/08/2014		FEES 29/10/2014		FEES 17/06/2015	
	1		1		1		1		1		1		1	
	5 ml	10ml	3 ml	5 ml	3 ml	5 ml	3 ml	5 ml	5 ml	10 ml	3 ml	5 ml	5 ml	10 ml
POS	1	1	NT	NT	1	1	NT	1	1	NT	1	NT	1	NT
PR inm VL	1	1	NT	NT	1	1	NT	1	1	NT	1	NT	1	NT
PR in PRS	1	1	NT	NT	1	1	NT	1	1	NT	1	NT	1	NT
LP	3*	3*	NT	NT	0	2	NT	3*	5*	NT	3*	NT	5*	NT
LA	0	0	NT	NT	0	0	NT	0	0	NT	0	NT	7*	NT

*More severe level

Legend: FEES: Fiberoptic Endoscopic Evaluation of Swallowing; LS: Laryngeal sensitivity; POS: Posterior oral spillage; PR: Pharyngeal residues; VL: Valleculae; PRS: Piriform recess; LP: Pharyngeal penetration; LA: Laryngotracheal aspiration; NT: Not tested

DISCUSSION

The progressive neurological diseases that affect the neuromuscular control of the oropharynx are a source of concern for interdisciplinary teams in terms of diagnosis and mainly conduct, since conduct changes with advancing disease. In DM1, current evidence about oropharyngeal swallowing focuses on the characterization of the findings, with few papers being published in this area. Thus, there is little information contributing to the management of oropharyngeal dysphagia in the population with DM1^(3,4).

In DM1, the presence of pharyngeal residues, the need for multiple swallowings, and laryngeal penetration and/or laryngotracheal aspiration are frequent findings reported in the literature, signs that suggest impaired and reduced efficiency and safety of swallowing⁽³⁻⁶⁾. However, we do not know at what time during the progression of the disease the dysphagic symptoms will become a risk for the efficiency and/or the safety of swallowing.

In the present longitudinal study of oropharyngeal swallowing in DM1 we did not detect changes in laryngeal sensitivity as reported in the literature for this population⁽⁶⁾. We also wish to point out that no changes in laryngeal sensitivity were detected on the occasion of the initial evaluation of swallowing, or four years after the neurological diagnosis or at the final evaluation after one year and four months of follow-up. The presence of laryngeal sensitivity in the diagnosis of DM1 suggests that one of the mechanisms of airway protection, i.e., the cough reflex, is preserved and thus contributes to the expulsion of part or all the material that may be aspirated by the patient⁽⁶⁾. Although there are many mechanisms of lower airway protection, the preservation of laryngeal sensitivity in DM1 even by five years after the initial diagnosis is a finding that may contribute to the definition of the conduct regarding the fed orally.

In the current case report, posterior oral spillage was present after the intake of all the food consistencies tested in the volumes offered, regardless of the stage of assessment. This finding was also frequently reported by other authors who, when classifying the severity of posterior oral spillage, concluded that, even though it was present after intake of all food consistencies in DM1, it was more severe after the intake of liquid⁽⁵⁾. In this disease, impairment of muscle strength compromises the performance of the oral and pharyngeal phases of swallowing according to the muscle weakness present in the different stages of the disease. The impact of this muscle weakness on the organization and propulsion of the oral phase of swallowing, together with

the performance of the modulation of the pharyngeal response, promotes constant posterior oral spillage, especially of liquids.

The presence of pharyngeal residues was also detected since the first objective evaluation of swallowing in both pharyngeal regions (valleculae and piriform recess) and after the intake of all food consistencies offered. This result is expected for patients with oropharyngeal dysphagia and muscle weakness since the increased viscosity of the food bolus plus the progression of muscle weakness interfere with both the oral food propulsion and pharyngeal cleaning mechanisms. Oropharyngeal muscle weakness impacts the oral phase of swallowing and consequently also the pharyngeal phase since the reduced pressure wave reduces the propulsion of the food bolus, increases the oral transit time and reduces the swallowing response as well as the pharyngeal peristalsis, thus compromising the elevation of the larynx⁽⁴⁻⁶⁾.

Regarding the evolution of pharyngeal residues in DM1, one of the advantages of the present study compared to the few available ones was the method applied for the analysis of these residues. The scale used to classify the pharyngeal residues takes into consideration the site and quantity, permitting a more precise comparison of the initial and final stages of follow-up regarding the degree of impairment of this finding during one year of follow-up. Although there still is no consensus in the literature about the relationship between pharyngeal residues, laryngeal penetration and/or laryngotracheal aspiration in the various neurological diseases, their constant presence in DM1 should be considered to be a predictive risk factor regarding the safety of oropharyngeal swallowing.

As previously mentioned, the signs of reduction and impairment of swallowing safety in oropharyngeal dysphagia have been frequently described in the literature about DM1, although none of the available studies used a scale to classify laryngotracheal penetration/aspiration^(3,4,6). In the current case report, laryngeal penetration was present since the first evaluations and progressed to a higher level with all the food consistencies used, culminating with the presence of laryngotracheal aspiration. The increased level of laryngotracheal penetration and aspiration with the evolution of DM1 demonstrate that the advancement of the disease impairs different physiopathological mechanisms of swallowing, with an increased risk of aspirative bronchopneumonia after this period. It should also be pointed out that, in the present study, the impairment of swallowing safety with aryngal penetration and/or aspiration in DM1 was greater with food of liquid consistency, as also reported by other authors⁽⁵⁾.

For a more in-depth reflection about the present study, it should be considered that the present DM1 patient was an

older individual, a situation that also affects the biomechanics of swallowing due to the aging process. Also, the longitudinal monitoring with the investigation of swallowing revealed the details of the changes occurring in this function in terms of factors such as the disease, aging and the management of swallowing.

A limitation of the present study was the absence of raters for the analysis of FEES findings, which might have contributed to an increased reliability of the results. In addition, it was not possible to describe the performance of oropharyngeal swallowing since the beginning of the neurological diagnosis since the patient was referred to the assessment of swallowing only four years after the onset of the disease. It was also possible to observe that, during the fifth year after the initial diagnosis of DM1, the changes indicated a risk for the efficiency and safety of swallowing. On this basis, regarding the diagnosis and rehabilitation of DM1 in clinical practice, it should be remembered that the evolution of the disease potentiates the complications of oropharyngeal swallowing. Thus, screening and referral for diagnosis of swallowing disorders should accompany the diagnosis of the disease because of the need for intervention in order to minimize the negative effects of the disorder on hydration, nutrition, feeding pleasure, and pulmonary aspects.

FINAL COMMENTS

The progression of DM1 impairs oropharyngeal swallowing. During the longitudinal monitoring of oropharyngeal swallowing in the present DM1 case, the objective findings of oropharyngeal swallowing and the report of weight loss indicated impairment of swallowing efficiency much more than impairment of swallowing safety. At the beginning of the disease, the signs and symptoms of dysphagia compromise the efficiency of swallowing due to the impairment of the oral phase, of oral food transport and of pharyngeal cleaning as a consequence of muscle weakness, with a possible limitation of the food supply for the patient. This question was present since the beginning of the evaluations, with constant posterior oral spillage after the intake of all food consistencies. Thus, a multidisciplinary approach is suggested in order to guarantee the efficiency and safety of oropharyngeal swallowing, as well as the general well-being of the patient. With the advancement of the disease, in addition to the difficulties of swallowing efficiency, there is the beginning of impaired safety of this function, with an increased risk of pulmonary complications due to the greater severity of pharyngeal residues and to the presence of laryngotracheal aspiration. Thus, an integrated approach by physicians, speech therapists, nutritionists and physiotherapists may support the quality of life of patients with DM1.

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