Electrophysiological evaluation of oropharyngeal swallowing in myotonic dystrophy

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Abstract

Objective—Oropharyngeal dysphagia is a common feature of patients with myotonic dystrophy and is not usually perceived due to their emotional deficits and lack of interest. The aim was to show the existence and frequency of subclinical electrophysiological abnormalities in oropharyngeal swallowing and to clarify the mechanisms of dysphagia in myotonic dystrophy.

Methods—Eighteen patients with myotonic dystrophy were examined for oropharyngeal phase of swallowing by clinical and electrophysiological methods. Ten patients had dysphagia whereas 11 patients had signs and symptoms reflecting CNS involvement. Four patients with myotonia congenita and 30 healthy volunteers served as controls. Laryngeal movements were detected by means of a piezoelectric sensor. EMG activities of the submental muscle (SM-EMG) and needle EMG of the cricopharyngeal muscle of the upper oesophageal sphincter (CP-EMG) were also recorded during swallowing.

Results—In about 70% of the patients with myotonic dystrophy, the existence of oropharyngeal dysphagia was indicated objectively by means of the technique of “dysphagia limit” and by clinical evaluation. Duration of the swallowing reflex as defined by the laryngeal relocation time (0–2 time interval) and submental muscle excitation as a part of the swallowing reflex (A–C interval) were significantly prolonged in patients with myotonic dystrophy, especially in dysphagic patients. Triggering time of the swallowing reflex (A–0 interval) also showed significant prolongation, especially in the patients having both dysphagia and CNS involvement. During swallowing, CP muscle activity was abnormal in 40% of the patients with myotonic dystrophy.

Conclusion—Both myopathic weakness and myotonia encountered in oropharyngeal muscles play an important part in the oral and the pharyngeal phases of swallowing dysfunction in myotonic dystrophy. It was also suggested that CNS involvement might contribute to the delay of the triggering of the swallowing reflex and some abnormal EMG findings in the CP sphincter, resulting in oropharyngeal dysphagia in myotonic dystrophy.

(J Neurol Neurosurg Psychiatry 2001;70:363–371)

Keywords: myotonic dystrophy; oropharyngeal dysphagia; central nervous system; electrophysiological evaluation

Myotonic dystrophy (MyD) is the most common adult form of muscular dystrophy and pneumonia was reported to be the most common cause of death in these patients.¹ Pneumonia in MyD results from a multiplicity of problems. Oropharyngeal dysphagia and oesophageal motility disorders were found to be the most important reasons causing aspiration pneumonia.^{2–6} Dysphagia may be even a more prominent problem when the swallowing disorder seems to be present early in the course of disease but is not usually subjectively perceived until the advanced stages of disease are reached.⁶

Patients with MyD often exhibit disorders of personality and impairment of intellectual and cognitive functions, especially lack of interest in their disease.⁷ This might play a part in the unawareness of the swallowing dysfunction in these patients. Therefore, it is necessary to evaluate the oropharyngeal deglutition and oesophageal motility in every patient with MyD.

Alterations in pharyngeal and oesophageal functions have been reported in MyD on the basis of both manometric and radiographic studies.^{2,3,6,8–11} These methods are important; however they are time consuming, expensive, and sometimes difficult to apply, especially when patients will not cooperate. Furthermore, the physiological basis of swallowing dysfunction is not clearly understood, as some conflicting results exist.^{9–11} The oropharyngeal swallowing function in MyD has not been studied by electrophysiological techniques. Therefore we had two aims in this study; the first was to show the existence and frequency of subclinical physiological abnormalities in oropharyngeal swallowing, the second was to consider the mechanisms leading to oropharyngeal dysphagia in MyD.

Patients and methods

Eighteen patients with myotonic muscular dystrophy were studied. Diagnosis of MyD was based on clinical and electrophysiological findings and the history of the patients' pedigrees. The severity of disease was classified from 1 to 5 using the muscular disability rating scale described by Mathieu *et al.*¹² The mean age of the patients was 38.8 (range 19–66 years). Eight were women. The mean duration of the disease was 14.3 years (range 1–35 years). Clinical features of all patients are summarised in table 1. Clinical diagnosis of dysphagia was defined when there were persistent symptoms (nasal regurgitation, frequent necessity to clear the throat, feeling of food in the nasopharynx) obtained from their questionnaire and abnor-

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Received 2 July 2000 and in revised form
2 November 2000
Accepted 6 November 2000

Table 1 Summary of the clinical features of patients with myotonic dystrophy

Patient	Age/sex	Duration of MyD (y)	MDR scale	Dysphagic symptoms	Other GI symptoms	CNS Symptoms
1	23, F	7	3	+	+	AP, MRI
2	40, F	3	3	+	+	AP, EDS, MRI
3	23, M	16	2	-	+	AP, BIQ, EDS
4	20, F	5	2	-	-	AP, BIQ
5	42, M	27	3	+	+	AP, EDS, MRI
6	33, M	23	4	-	-	AP, BIQ, EDS
7	19, M	12	2	+	-	AP, BIQ, EDS
8	50, M	35	4	+	+	AP, BIQ, EDS, MRI
9	54, F	2	3	-	-	AP, BIQ, EDS
10	46, M	26	3	+	+	AP, BIQ, EDS, MRI
11	39, M	14	3	+	-	AP, BIQ, EDS
12	49, M	5	3	+	-	-
13	66, F	23	4	-	-	-
14	42, F	1	1	-	-	-
15	41, M	14	2	-	-	-
16	31, F	9	3	+	+	-
17	45, M	30	3	+	-	-
18	35, F	5	4	-	+	-

MDR=Muscular disability rating scale¹²; +=present; -=absent; AP=apathy; EDS=excessive daytime sleepiness; BIQ=borderline IQ or mental retardation; MRI=abnormal findings by cranial MRI¹²⁻¹⁴; GI=gastrointestinal.

mal signs closely related to swallowing disorder (cough during swallowing, accumulation of saliva in the mouth, "wet" voice after swallowing water, nasal voice, palatal paresis, abnormal head posture during swallowing). The patients having uncertain symptoms but lacking any objective signs that could be related to swallowing were not considered to be dysphagic. Therefore 10 patients with dysphagia and eight patients without dysphagia were classified according to the clinical evaluation for dysphagia. Clinical and laboratory findings in patients were documented in detail in our neuromuscular unit and gastrointestinal symptoms and signs were especially evaluated by the clinic of gastroenterology.

Patients with MyD were also reclassified for their symptoms reflecting CNS involvement such as excessive daytime sleepiness, apathy, and mental retardation.^{13, 14} Their MRI findings are shown in table 1. Eleven patients had symptoms and signs suggesting CNS involvement (patient 1 to patient 11 in table 1).

Four patients with myotonia congenita were studied to clarify the pure effect of myotonia on the electrophysiological findings. Patients with congenital myotonia had generalised myotonia without any weakness determined by both clinical and EMG methods. They had clinically no dysphagia. Their mean age was 33 (range 17–40 years, all men).

The mean values and individual results of swallowing variables obtained from patients were compared with the mean values and ranges of 30 normal control subjects. (mean age 38.5; range 20–65 years, 15 women). The cricopharyngeal (CP) muscle of the upper oesophageal sphincter was investigated in 13 patients with MyD and the results were compared with 20 normal control subjects with a mean age of 39.3 (range 22–61 years, 13 women). Informed consent was obtained from each patient. The investigation was approved by the ethics committee of our university hospital.

The methods used for the evaluation of dysphagia have been described previously.¹⁵ During the examination, the seated patient was instructed to hold his or her head in a neutral upright position. Swallows were initiated vol-

untarily with the bolus (tap water) positioned on the tongue and the tip of the tongue touching the upper incisors. Swallow signals were recorded after the delivery of 3 ml water through a graduated syringe. Mechanical upward and downward laryngeal movements during swallowing were detected by means of a piezoelectric sensor designed in our laboratory. This was a simple piezoelectric wafer with a 4x2.5 mm rubber bulge fixed into its centre. The rubber bulge was placed on the coniotomy region between the cricoid and thyroid cartilages at midline. The sensor was taped onto the neck and its output signal was filtered (band pass 0.01–20 Hz) and fed into one of the channels of the EMG apparatus (Medelec Mystro, MS-20, Surrey, UK). The submental muscle EMG (SM-EMG) was recorded by bipolar silver chloride EEG electrodes taped under the chin over the submental muscle complex (mylohyoid, geniohyoid, and anterior digastric muscles). Signals were filtered (band pass 100 Hz to 10 kHz), amplified, rectified, and integrated.

Cricopharyngeal muscle activity of the upper oesophageal sphincter was recorded with a sterile needle electrode (Medelec disposable needle electrode DMC-37) inserted through the skin at the level of the cricoid cartilage about 1.5 cm lateral to its palpable lateral border in the posterior median direction. The CP sphincter EMG (CP-EMG) was recorded together with the laryngeal movement signals under the same recording conditions as for the SM-EMG.

The laryngeal sensor gave two deflections with generally opposing polarity during each swallow. The first deflection of the laryngeal sensor represented the upward movement of the larynx and the second deflection its downward movement. The mid-region of the first deflection was stabilised on the oscilloscopic screen by using the delay line technique so that the deflections would appear at the same location of each sweep (about 800 ms after the onset of the sweep) throughout successive recordings. In this way, all electrophysiological events were displayed synchronously. The onsets of two deflections in the laryngeal sensor signal recordings were denoted as 0 and 2.

Table 2 Symptoms and signs of patients with myotonic dystrophy with regard to dysphagia

	With dysphagia*	Without dysphagia*
Difficulties in bolus formation and control	8/10	1/8
Feeling of food in the nasopharynx	10/10	0/8
Need to clear throat	8/10	0/8
Abnormal posture during swallowing	8/10	0/8
Nasal regurgitation	6/10	0/8
Cough during swallowing	9/10	0/8
Loss of weight	3/10	0/8
History of pneumonia	2/10	0/8
Slow laryngeal elevation	6/10	0/8
Weak involuntary cough	8/10	0/8
Dysphonia	9/10	2/8
Saliva accumulation in mouth	5/10	1/8
Palatal paresis	6/10	0/8
Facial weakness	10/10	8/8
Weakness in chewing muscles	6/10	0/8
Tongue weakness	5/10	3/8
Tongue myotonia	7/10	5/8

* Number of patients having the sign or symptom/total number of dysphagic or non-dysphagic patients.

When the first deflection was stabilised on the EMG screen, the onset of the second deflection showed variability for the same subject during swallowing boluses of the same volume. This variability of the laryngeal downward movement was measured from the first peak of the superimposed traces of the second deflection to the last peak of them. This was called "swallowing jitter".¹⁶ As SM-EMG was activated by the laryngeal elevator muscles, their activity was considered to give information about the onset and duration of swallowing,¹⁷⁻¹⁹ and was denoted as the A-C interval. We also measured another important index called the A-O interval that was related to the triggering of the reflex swallow for the voluntarily initiated swallowing. This variable was the interval between the onset of the SM-EMG and the first deflection of the laryngeal sensor signal reflecting the upward movement of the larynx, which was one of the first events of the pharyngeal reflex swallowing.¹⁷⁻²⁰⁻²⁴ The preanalysis time was set at 800 ms and the total sweep time was set at 2 or 5 seconds. At least five successive "swallowing signals" were recorded. The signals were examined either as a single trace or as the mean of several superimposed traces.

Piecemeal deglutition and dysphagia limit were also measured electrophysiologically.²⁵ This test is based on determining piecemeal deglutition which is a physiological phenomenon that refers to the division of the bolus into two or three successive swallows. All patients were instructed to swallow doses of water gradually increasing in quantity from 1 to 20 ml. The bolus volume causing the recurrence of the EMG and laryngeal sensor signals related to swallowing within 8 seconds was accepted as the sign of dysphagia limit.

Mean (SD) of all measured quantities were calculated. Student's *t* test for independent samples was then applied for the comparison between patient groups and normal controls. Subgroups within the patient groups were compared both with normal controls and with each other by means of Duncan's multiple range variance analysis test. Spearman's correlation analysis test was performed for correlation studies. Significance was tested at the 5% level.

Results

Ten patients with MyD had symptoms and signs closely related to dysphagia and pharyngolaryngeal muscle involvement (table 2). They all exhibited weakness and wasting in facial muscles. Weakness in jaw and tongue muscles was noted in six and five patients respectively and percussion myotonia in the tongue was elicited in seven out of 10 patients. All patients except one had a nasal voice and anteflexion posture was noticed during each swallow in eight patients. Their dysphagia was not severe enough to necessitate non-oral feeding despite their complaint of swallowing and the clinical signs of dysphagia such as cough during a swallow, wet voice after water drinking, and nasal regurgitation. The remaining eight patients with MyD had no complaint that could be attributed to dysphagia and the clinical examination of the swallowing function of these patients was almost always normal as shown in table 2.

DYSPHAGIA LIMIT

Facial and tongue weakness and tongue myotonia occurred equally in both dysphagic and non-dysphagic patients with MyD (table 2). Therefore it is important to determine the oropharyngeal swallowing objectively in these patients. This was done by the technique of "dysphagia limit". The existence of dysphagia in MyD was indicated objectively by this electrophysiological evaluation. The normal control subjects could swallow up to 20 ml of water without dividing the liquid material. Four patients with myotonia congenita without dysphagia also had normal limits of 20 ml water. In nine dysphagic patients with MyD, the bolus was divided into two or more aliquots while swallowing the lesser volumes of water (fig 1).

In three of eight patients with MyD without clinical dysphagia, the dysphagia limit was lower than 20 ml water. In one patient (patient 10) with clinical signs and symptoms suggesting dysphagia, the dysphagia limit was within normal limits. This patient had a clear anteflexion posture of his head for each volume of swallows. It was shown that the anteflexion posture or chin tuck posture of the head and neck facilitated or normalised swallowing.²⁶⁻²⁸ Consequently 12 out of 18 patients with MyD were accepted as having difficulty in their swallowing function from an electrophysiological point of view. Therefore about 70% of patients with MyD had problems during oropharyngeal swallowing determined both by clinical and electrophysiological evaluation.

LARYNGEAL MOVEMENTS AND SM-EMG DURING OROPHARYNGEAL SWALLOWING

The laryngeal elevation and relocation of the larynx (swallowing reflex) was significantly prolonged as shown by the 0-2 time interval ($p < 0.05$, table 3, fig 2) and this variable was also individually prolonged in nine patients (seven dysphagic, two non-dysphagic) when compared with the age and sex matched normal subjects. Similarly the SM-EMG duration during swallowing (A-C interval) was significantly prolonged ($p < 0.001$, fig 2). Although

Figure 1 Laryngeal sensor signals (top traces in each pair) and integrated SM-EMG activities (lower traces in each pair) obtained from (A) a normal subject and (B) a patient with MyD with dysphagia during swallowing of different amounts of water (3–20 ml). The volume swallowed in a single attempt was up to 20 ml in the normal subject whereas the patient began to divide the bolus even at 3 ml water. The dysphagia limit was therefore 3 ml for this patient. The bolus was divided more as the volume was increasing. Arrows indicate the second and subsequent swallows. Amplitude calibration 50 μV for SM-EMG, time calibration 1000 ms in all traces. (The amplitude of the laryngeal sensor signal is irrelevant for this and subsequent figures.)

the longest duration of SM-EMG or 0–2 time interval was found in eight clinically dysphagic patients, similar and abnormal prolongation of these variables were also encountered in three patients without clinical dysphagia. When dysphagic and non-dysphagic patients were compared, the clinically dysphagic group had a significantly longer time interval only in A-C variables ($p < 0.05$). However, the 0–2 time interval was significantly prolonged in dysphagic patients when compared with the normal control subjects ($p < 0.05$) whereas no significance was found in non-dysphagic patients.

Three of the four patients with myotonia congenita did not differ from the normal subjects individually in 0–2 and A-C variables. However, the A-C interval of the SM muscle excitation was found to be significantly prolonged in myotonia congenita when the results were compared statistically with the normal subjects ($p < 0.05$).

The A-0 time interval indicating the triggering time of the pharyngeal reflex swallow^{24, 29} was also statistically prolonged in patients with MyD ($p < 0.005$) especially in clinically dysphagic patients ($p < 0.05$, fig 2). The non-dysphagic patients with MyD did not differ from the normal control subjects. Interestingly, in patients with myotonia congenita, the A-0 interval was also significantly prolonged by comparison with normal subjects and in one patient with myotonia congenita the A-0 interval was prolonged.

Swallowing jitter can be regarded as the variability in the swallowing apparatus and it can be an important measure of the safety of deglutition during swallowing boluses of the same volumes.^{15, 16} It was extremely high in five patients with MyD, especially in dysphagic patients and the mean value of all patients was significantly prolonged ($p < 0.005$) in comparison with normal control subjects (table 3, fig 2). Patients with myotonia congenita had

Table 3 Summary of the statistical results of patients with myotonic dystrophy and myotonia congenita compared with normal subjects

	Myotonic dystrophy (n=18)	Myotonic dystrophy with dysphagia (n=10)	Myotonic dystrophy without dysphagia (n=8)	Myotonia congenita (n=4)	Normal subjects (n=30)
0–2 Interval (laryngeal relocation time)	661 (161) $p < 0.05$	704 (186) $p < 0.05$	612 (121) NS	518 (114) NS	564 (100)
A–C Interval (SM-EMG duration)	1144 (306) $p < 0.001$	1250 (285) $p < 0.05$	1025 (301) $p < 0.05$	1124 (618) $p < 0.05$	850 (123)
A–0 Interval	400 (185) $p < 0.005$	437 (164) $p < 0.05$	359 (208) NS	454 (236) $p < 0.05$	263 (119)
Swallowing variability (jitter)	133 (74) $p < 0.005$	150 (79) $p < 0.05$	114 (68) NS	57 (30) NS	71 (40)
SM-EMG amp	85 (37) NS	77 (34) NS	94 (41) NS	58 (31) NS	73 (32)

Statistical comparisons between the patients have not been included in the table (see text).

Statistical variables except SM-EMG amplitude (μV) are in ms and the values are shown as mean (SD).

A–0=the time interval at the onset of SM-EMG and the onset of laryngeal upward movements and reflects the time until triggering of the swallowing reflex.^{15, 22, 24}

Figure 2 Laryngeal sensor signals (upper traces in each pair) and integrated SM-EMG (lower traces in each pair) in superimposed recordings obtained from a patient with MyD without dysphagia and a patient with dysphagia while swallowing 3 ml water. The laryngeal relocation time (0–2 time interval), duration of SM-EMG activity (A–C interval), swallowing jitter, and the triggering time interval of the swallowing reflex (A–0 interval) (oblique arrow) were obviously prolonged in the dysphagic patient. The points indicating the onset of the upward movement of the larynx (0) and downward movement (2) and the points indicating the onset and end of SM-EMG activity (A and C respectively) are labelled according to the averaged recordings.

normal values of swallowing jitter varying between 20 and 90 ms.

High variability of jitter and the increase in the A–0 interval were probably the results of the myotonic changes at the onset and end of contractions of the laryngeal elevation, especially in severely dysphagic patients. This was clear when the successive swallowing traces were examined one by one, as shown in fig 3.

We could not find any correlation between dysphagia or electrophysiological variables and either the duration or the severity of the disease except for the significant correlation between the swallowing jitter and the duration of the disease ($r=0.60$; $p<0.01$). A significant correlation was also elicited between the presence of dysphagia and dysphagia limits, suggesting the high sensitivity of the test ($p<0.05$). Eleven patients (table 1, patients 1–11) who had CNS symptoms and signs were compared with the remaining seven patients (table 1, patients 11–17) who had no obvious symptoms and

signs reflecting CNS involvement. The A–0 interval of the triggering of the swallowing reflex was prolonged both in patients with MyD with CNS signs and symptoms and in the patients without CNS involvement (449 (SD 66) ms; 330 (SD 45) ms, respectively). However this prolongation was significant only in patients with MyD with CNS involvement when compared with the normal subjects ($p<0.05$). Other electrophysiological variables did not differ between the two groups with MyD.

CRICOPHARYNGEAL SPHINCTER EMG

The CP muscle of the upper oesophageal sphincter was investigated in 13 of the 18 patients with MyD. Behaviour of the CP-EMG was normal, as in normal control subjects both at rest and during swallowing in eight patients (two patients with and six patients without dysphagia). The resting tonic activity of CP muscle switched off for 400–500 ms (CP-EMG pause) and during this time, the laryngeal upward and downward movements were coordinated.¹⁵ In the remaining five patients, the CP-EMG was pathological and all of the five patients had clinical dysphagia as well as clinical signs and symptoms reflecting CNS involvement. Figure 4 illustrates the CP-EMG recordings from two patients with MyD with dysphagia. The duration of the CP-EMG pause associated with the duration of the passage of the bolus into the oesophagus from the upper oesophageal sphincter tended to be shortened in three patients. The second abnormality of the CP sphincter was its premature closure. CP-EMG pause ended too early before the onset of the laryngeal downward movements in two patients. Figures 4 and 5 show the unexpected motor unit burst activity which appears within the CP-EMG pause and some remarkable variability of the tonic activity and foreburst just before the CP-EMG pause in a patient with MyD.

In summary, the CP sphincter muscle seemed to be intact in most patients with MyD with or without dysphagia. Although in the minority (about 40%; all had dysphagia) the CP muscle of the upper oesophageal sphincter showed various abnormalities. During insertion of the needle electrode into the CP muscle, we rarely heard “dive bomber” in two patients, who also had other types of CP sphincter abnormalities as mentioned above. During swallowing, foreburst and rebound activities appearing around the CP-EMG pause sometimes tended to be of high amplitude and long duration without giving an impression of the dive bomber.

In the three patients with myotonia congenita evaluated, the CP-EMG activity was normal both in the resting condition and during swallowing.

Discussion

The swallowing impairment in myotonic dystrophy has been known since the disease was described by Steinert in 1909.³⁰ Dysphagia is an often encountered problem and has been reported with a prevalence between 25% and

80%.^{6, 31–34} We have found, using both clinical and electrophysiological criteria, that about 70% of patients with MyD have oropharyngeal swallowing problems.

The significant findings of the study can be gathered under five headings to clarify the mechanisms of oropharyngeal swallowing in myotonic dystrophy and these are discussed below.

(1) PIECEMEAL DEGLUTITION AND REDUCTION IN DYSPHAGIA LIMIT

We have found abnormal piecemeal deglutition and repetitive swallows in 67% of the patients (12 out of 18) investigated, as the first swallowing abnormality. This finding that can be considered as the oral phase of abnormality of dysphagia,^{25, 35} is not so surprising. Because the control of the bolus and its formation is mainly provided by the tongue, masticatory, buccal and submental muscles. If these muscles are weak, as in MyD, the bolus will be divided into pieces and swallowed successively.^{23, 25} Despite this attempt, the residual bolus volume remaining in the space of the pharynx will escape either into the airway or down through the upper oesophageal sphincter, opened for a sec-

ond time in a considerable time interval after the first swallow as we found in patients with MyD.

(2) DELAY IN THE TRIGGERING OF THE PHARYNGEAL SWALLOWING REFLEX

As a second swallowing abnormality, the interval between the onset of the SM-EMG (voluntarily onset) and the signal of first deflection of the laryngeal movement (reflex onset)—the A-0 interval—was prolonged during attempts to swallow in myotonic dystrophy in comparison with the normal subjects. This may reflect a delay in the triggering of the swallowing reflex.²² There might be several reasons for this delay in MyD as follows:

(a) It may be the result of poor tongue and submental muscle control due to the involvement of the striated muscles mentioned above. However we have previously shown in dysphagic patients with myasthenia gravis,³⁵ and with polymyositis,²³ that the triggering of reflex swallowing is not significantly disturbed or may even be normal as judged by the normality of the A-0 interval. Therefore a delay in triggering of the swallowing reflex is not restricted only to

Figure 3 Six successive swallowing traces (laryngeal sensor signals in upper traces and SM-EMG activities in lower traces in each pair) while swallowing 3 ml water in each time. Arrows indicate the myotonic changes in submental muscles either at the onset or at the end of SM-EMG activity (A-C interval) during subsequent swallowing.

the muscle weakness of the tongue and other muscles involved in MyD.

(b) Mucosal and deeper mechanoreceptors in the oral-pharyngeal region and tongue and their sensory nerve fibres are obviously needed for the integrity of the triggering mechanism of swallowing. To our knowledge such an oral perception disorder has not been previously described in patients with MyD.

(c) Myotonia of the tongue and laryngeal elevator muscles such as SM muscles may have a role in delaying of the triggering mechanisms although a contrary view has also been proposed.^{9–11} In patients with MyD the irregularities of the SM-EMG traces were recorded more clearly if successive wet swallows were provided (fig 3). These patients had a significantly ($p < 0.05$) prolonged A-0 interval compared with the normal subjects. Therefore, a myotonic phenomenon might have some role in the delaying of the triggering and even in prolongation of the swallowing reflex. However, this does not mean that oropharyngeal muscle myotonia is the permanent cause of dysphagia because percussion myotonia of the tongue is not present in some patients with MyD with dysphagia (table 2). Furthermore, myotonia could not produce swallowing dysfunction in four patients with myotonia congenita.

(d) The corticobulbar pyramidal fibres seem to be responsible for initiating voluntarily induced swallows. Any type of involvement of corticobulbar fibres seen in suprabulbar palsies as in amyotrophic lateral sclerosis²⁴ or lacunar states³⁶ produced delay in triggering of the swallowing reflex. Once the reflex is triggered

the laryngeal elevator muscles will have a reflex contraction and the laryngeal relocation time (0–2 time interval) will be normal.^{24–36}

It can be speculated that CNS involvement might be expected to trigger the cortical drive to be transmitted into the reflex swallow at an interneuron level of the bulbar swallowing centre. The significant prolongation of the A-0 interval found only in patients with MyD having CNS signs and symptoms might be supportive data for this speculation.

(3) SLOWING IN PHARYNGEAL PHASE OF SWALLOWING

As a third swallowing abnormality, the duration of the swallowing reflex (0–2 time interval of the laryngeal relocation time) was significantly longer in MyD than in normal controls. The finding of prolongation of SM muscle excitation during swallowing (A-C interval) accompanying prolongation in the A-0 interval was the most significant finding. These two findings may indicate that the pharyngeal phase of swallowing is slow for transporting the bolus into the upper oesophageal sphincter. The prolongation in the duration of the swallowing reflex is similar to that found in myasthenia gravis³⁵ and in inflammatory myopathy²³ but is somewhat contrary to that reported in amyotrophic lateral sclerosis with suprabulbar palsy, where the duration of the swallowing reflex is normally preserved.^{24–37} The prolongation activity of the weak laryngeal elevators in MyD to keep the larynx at an anterosuperior relocation position during swallowing must be the cause of the longer duration of A-C interval of SM-EMG and the 0–2 time interval of the laryngeal sensor movements in individual bases. The weakness of the pharyngeal muscles was demonstrated manometrically in MyD^{8–11} and it has been reported that the duration of the pharyngeal contraction is also longer than normal.^{6–8} Our findings are compatible with these results.

(4) INCREASE OF THE SWALLOWING JITTER

The fourth swallowing abnormality in MyD was the significant increase of swallowing jitter. The range of swallowing jitter would be expected to increase in acute problems of oropharyngeal swallowing and it increased markedly in cases of acute muscular disease such as myasthenia gravis^{23–35} and in acute strokes with dysphagia.²³ As MyD is a chronic disease with a slowly progressive course, the increasing range of jitter was unexpected. It may result from weakness of the laryngeal elevators. The significant difference in jitter values between patients with MyD and those with myotonia congenita might be explained by the dystrophic changes in MyD. The significant correlation between the duration of the disease and the increase in jitter values may be related to the increase in severity of the laryngeal elevator muscle weakness due to the dystrophic pattern as the disease reaches an advanced stage. However, the myotonic contractions changing their condition from one swallow to

Figure 4 Laryngeal sensor signals (top traces in each pair) and CP-EMG activities (lower traces in each pair) recorded from two dysphagic patients with MyD (in averaged recordings). In patient A, unexpected motor unit burst activity was seen within the CP-EMG pause (oblique arrow) while swallowing 3 ml water. The CP-EMG pause was shortened and the pause ended prematurely before the larynx had descended from its superior position (oblique arrow) in patient B.

Figure 5 Four successive swallowing traces obtained from a dysphagic patient while swallowing 3 ml water. Note the unexpected motor unit burst activity within CP-EMG pause (oblique arrow) and the variability of the tonic activity of CP-EMG during rest and foreburst just before the CP-EMG pause (vertical arrows).

another cannot be ignored in patients with MyD and could have a role in increasing jitter values.

(5) ABNORMALITIES IN THE CRICOPHARYNGEAL MUSCLE OF THE UPPER OESOPHAGEAL SPHINCTER In 60% of patients with MyD, the CP muscle of the upper oesophageal sphincter was found to be normal electrophysiologically both in resting tonic activity and during swallowing. This is by contrast with the results of manometric studies, in which a marked reduction in resting tone of the upper oesophageal sphincter was found.^{2 8 10 11} Evidence for neural dysfunction manifested by abnormalities in completeness and coordination of sphincter relaxation was not detected in patients with MyD. It was also reported that myotonia could not be seen around the upper oesophageal sphincter.⁹⁻¹¹ On the other hand, Siegel *et al*⁸ found that the duration of relaxation of the upper oesophageal sphincter was prolonged. This controversy among different reports must be due to different technology and methodology. In the minority of patients (five) with MyD, who had also clinical dysphagia, there were some abnormalities akin to hyperreflexive sphincter usually encountered in patients with suprabulbar palsy, due to the involvement of pyramidal-corticobulbar motor fibres.^{24 36} As the fifth

electrophysiological abnormality of swallowing, this finding could not be explained either by the weakness or myotonia of the striated muscle of the CP sphincter. Although we have found some EMG changes that could be attributed to myotonia, they were not remarkable. Because all of them were in patients who showed signs and symptoms suggesting CNS involvement, they might be related to brain stem mechanisms.

WHAT ARE THE MECHANISMS OF DYSPHAGIA IN MYD?

Although five swallowing abnormalities were encountered in this study, we suggest that there are three different factors that may be involved in the problems of oropharyngeal swallowing in MyD.

(1) Myopathic weakness in jaw, buccal, tongue, submental and pharyngeal constrictor muscles may be responsible for the problems of bolus formation in the mouth (oral phase problem) and slow transit of the bolus along the pharynx (pharyngeal phase problem). This conclusion is supported by the frequent findings of piecemeal deglutition and abnormal dysphagia limits. The prolongation of the reflex swallowing, shown by 0–2 laryngeal sensor movements individually and long duration of SM-EMG, which was statistically and individually significant in patients with dysphagia, are the other supportive pieces of evidence.

(2) Myotonia encountered in oropharyngeal muscles in MyD is a probable contributing factor to the triggering of the swallowing reflex in some patients and in some swallows due to its variability and changeable nature in time. It may also contribute negatively to the safety of deglutition as it may interfere with the usual contractions of SM muscles during successive swallows. This idea is partly supported by the variability in SM muscles measured from the A-0 interval and from the increasing range of swallowing jitter.

(3) The third factor is speculative, but we provided some data that can only be explained by involving brain stem neural mechanisms. The interneurons in the reticular formation linked with the corticobulbar-pyramidal nerve fibres at the level of the medullary swallowing centre may be involved in MyD. The dysfunction of the interneurons related to deglutition is probably responsible for the triggering difficulties of voluntarily initiated swallows in some cases. Some inhibitory effects of the descending impulses may not be transmitted to the related motor neurons due to the lack of transmission in the interneurons situated in the reticular formation at the medullary level.

The organisation of deglutition at the medullary level is well known through experimental studies.^{20 38-40} Such an approach can also explain the premature closure and shortening of the CP-EMG pause encountered in some dysphagic patients in addition to a delay in triggering of the swallowing reflex. It has recently been shown that in addition to the characteristic muscular symptoms, patients with MyD often show CNS symptoms including apathy, hypersomnia and mild mental

impairment^{13 14 30 41} and abnormal cranial MRI findings.⁴²⁻⁴⁴

In MyD, neuronal loss has been described in the dorsal raphe nucleus, the superior central nucleus, and the medullary reticular formation^{45 46} in which the autonomic respiratory centre is thought to be located. The swallowing centre is also considered to be located at the same medullary region.^{20 39} The nature of swallowing dysfunction often encountered in MyD awaits further studies especially into the role of brain stem mechanisms.

This study is supported by a grant from the Turkish Scientific and Technological Research Council (TUBITAK) (project SBAG-1739). We thank Assistant Professor Nese Çelebisoy for her invaluable help in preparing this manuscript.

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