*B-ENT*, 2015, **11**, 249-256

# Swallowing dysfunction in myotonic dystrophy: a retrospective study of symptomatology and radiographic findings

*A. Willaert*[1], *M. Jorissen*[1] *and A. Goeleven*[2]

[1]Department of ENT – Head and Neck Surgery, University Hospitals Leuven, Leuven, Belgium; [2]Department of Speech Language pathology, Swallowing Clinic, University Hospitals Leuven, Leuven, Belgium

**Key-words.** Myotonic dystrophy; oropharyngeal dysphagia; fluoroscopy

**Abstract.** *Swallowing dysfunction in myotonic dystrophy: a retrospective study of symptomatology and radiographic findings. Background*: Swallowing dysfunction is a common symptom of myotonic dystrophy, but it is poorly documented in large patient series. This retrospective study was designed to investigate the presence of swallowing symptoms in a large study population and to describe a specific pattern of clinical and radiographic abnormalities.
*Methods*: A retrospective analysis was made of 169 files of patients with confirmed MD. Neuromuscular assessment was made by means of a standardised neurological examination; clinical swallowing symptoms were listed, and video-fluoroscopic images were analysed.
*Results*: More than half the patients reported swallowing complaints. The major symptoms were frequent choking, difficult pharyngeal transport and piecemeal deglutition. The pharyngeal phase of swallowing was most frequently compromised. This was shown radiographicaly in reduced pharyngeal peristalsis, hypopharyngeal stasis and fragmented swallowing. Aspiration was seen in half of the patients, mostly during swallowing. A typical 'hung position' of the hyoid was also seen.
Different onset types of MD seem to be accompanied by comparable subjective complaints and radiographic symptoms.
*Conclusions*: Pharyngeal transport was most affected in this patient population. Muscular weakness seems to be the major contributor to swallowing impairment in MD. Swallowing abnormalities may be present even if patients report only a few symptoms and even if the severity of the disease is not pronounced.

**Abbreviations**

MD: Myotonic dystrophy.
DMPK: dystrophia myotonica protein kinase.
NMRC: Neuromuscular Reference Centre.
PEG: percutaneous endoscopic gastrostomy.
UES: Upper oesophageal sphincter.

## Introduction

Myotonic dystrophy (MD), which is also known as dystrophia myotonica or Steinert's disease, is the most common inherited neuromuscular disease affecting adults and children, with a prevalence of 2.4 to 12/100.000.[1,2] It is an autosomal dominant multisystem disorder caused by an unstable trinucleotide repeat expansion in the dystrophia myotonica protein kinase gene (DMPK). The amplification of this repeat is correlated to the severity of the disease and to the age of onset, with more severe symptoms and younger onset in more extensive repeats.[3,4] In clinical practice, patients are therefore allocated to three disease types based on age at onset: congenital onset (from birth), childhood onset (before the age of 18) and adult onset (from young adulthood onwards). The course of MD is one of slow progression and, to date, there is no effective cure.[5]

Unique among the muscular dystrophies, MD is characterised by myotonia in combination with progressive muscular dystrophy and multiple systemic complications such as cataracts, cardiac arrhythmia, restrictive pulmonary dysfunction, mental retardation, dysphagia and hyper-somnolence.[1,6-10] It is rarely a difficult diagnosis to make in patients presenting with the typical triad of myotonia, muscle weakness and wasting, provided that the clinician is aware of the disease. Despite

Presented by Dr. Annelore Willaert at the annual meeting of the Royal Belgian Society for Ear, Nose, Throat, Head and Neck Surgery, March 23, 2013, where she received the 2013 RBS Lecturer Award for Laryngology, Head and Neck Pathology.

this, the correct diagnosis is often delayed. In part, this reflects the uncomplaining nature of most patients[7,11] but another possible explanation is that many patients do not complain of neuromuscular symptoms but of systemic disorders.[1,5,6,12,13] One of the most frequent of these complaints is dysphagia. Symptoms of swallowing dysfunction may even dominate the clinical picture or they may be manifest long before musculoskeletal disturbances become apparent.[2,8,9,10,11] In many cases, these symptoms may also be accompanied by dysarthria and nasal speech. Patients with MD therefore often consult an otolaryngologist initially. ENT clinicians concentrating on these immediate problems may easily overlook or misinterpret a degree of myotonia and muscle weakness that would have been obvious if sought for specifically.

Nearly all MD patients have swallowing disorders during the course of the disease. However, the onset and progression of these swallowing difficulties vary greatly between individuals. Secondary medical and social complications of dysphagia such as dehydration, inadequate energy intake, aspiration pneumonia or social isolation and embarrassment may have a major impact on quality of life.[4,6,14,15] As a matter of fact, aspiration pneumonia is the most common cause of death in patients with MD.[16]

Oropharyngoesophageal motor involvement in MD has been evaluated in some studies, using radiology, radio-isotopes, fibroscopy, electro-physiology and manometry.[2,10,12-14,16-24] All agree that the alteration of the upper gastrointestinal tract musculature is a common finding in patients with MD, even after disregarding dysphagia.[13,23] However, patient series studied in the past have always been quite small, and several matters remain unclear. The aim of this retrospective study was therefore, firstly, to describe the frequency and nature of swallowing symptoms in these patients in a large study population and, secondly, to determine a specific pattern of radiographic abnormalities associated with disease onset.

## Materials and methods

This study reviewed 169 files of consecutive patients with a genetically proven diagnosis of MD. All patients had been referred to the Neuromuscular Reference Centre (NMRC) of the University Hospitals Leuven, Belgium, for annual follow-up between October 1997 and October 2010. In that period, 82 men and 87 women were seen with a mean age of 38 years (range: 19 to 74 years): 112 patients with adult onset, 46 with childhood onset and 11 with congenital onset. Neuromuscular involvement was assessed in a standard neurological clinical examination by a neurologist (neurological symptoms displayed in Table 1). All patients were questioned systematically and evaluated clinically by a speech-language pathologist for the presence of swallowing symptoms (the swallowing complaints covered by the questions are listed in Table 2). The severity of reported dysphagia was rated retrospectively using a 7-point swallowing scale (SS) (see Table 3 for description of degree of severity). In nearly half the patient population (n = 47/101), the subsequent radiographic evaluation of swallowing was performed with videofluoroscopy. In order to allow for the accurate comparison of radiographic findings, only the investigation with a liquid bolus was used in this study since this consistency was tested in all patients and semi-liquids and solids were not evaluated in every patient. The dynamic radiographic assessments were performed using a standard protocol. Each patient was seated in a swallowing chair (VessH) and was viewed laterally. A standard amount of liquid contrast material (10cc MicropaqueH or UltravistH) was given and the patient was asked to swallow. Subsequently, radiological images were analysed for swallowing abnormalities using a standard dichotomic protocol, including the features displayed in Table 4. In addition, reduced elevation of the hyoid, delayed reposition of the hyoid, disturbed or reduced oesophageal peristalsis and incomplete or excessively brief opening of the upper oesophageal sphincter (UES) were evaluated in some patients.

## Results

Of the total study population of 169 patients with MD, 101/169 patients (60%) reported swallowing complaints during their clinical follow-up. Of this group, 47/101 were referred for the additional radiographic evaluation of the safety and efficiency of swallowing. It should be noted that 19% of the patients had already complained about dysphagia before the diagnosis of MD was made. As for the different types of MD, 62% of the patients with the

*Table 1*

Neuromuscular symptoms in patients with swallowing complaints in total study population (n = 100*)

| Neuromuscular symptom in MD | Population (n = 100) |
|---|---|
| Distal muscle weakness | 100% |
| Bifacial weakness | 97% |
| Myotonia | 87% |
| Dysarthric speech | 58% |
| Restrictive pulmonary | 57% |
| Myopathic facies | 54% |
| Mental retardation | 42% |
| Hypersomnolence | 41% |
| Global areflexia | 40% |
| Cardiac arrhythmia | 38% |
| Bilateral catarct | 28% |
| Proximal muscle weakness | 23% |
| Nasal speech | 23% |
| Confined to wheelchair | 9% |

* One patient had no documented neurological examination at the time of the onset of swallowing problems.

*Table 2*

Frequency of subjective swallowing complaints in total population (n = 101) in descending order of frequency

| Swallowing complaint | Population (n = 101) |
|---|---|
| Frequent choking | 72% |
| Difficult pharyngeal transport | 69% |
| Piecemeal deglutition | 45% |
| Smaller bite size | 42% |
| Need for adaption of mealtime duration | 26% |
| Difficult to chew | 22% |
| Difficult oral transport | 19% |
| Swallowing complaints before diagnosis of MD | 18% |
| Regurgitation | 17% |
| Dietary consitency changes to soft diet | 14% |
| Nasal backflow | 4% |
| Need for additional nutrition | 4% |
| Dietary consistency changes to liquified diet | 0 |
| No oral feeding | 0 |

adult type had swallowing complaints, 54% of the childhood type, and 64% of the congenital type.

Most of the patients had a significant degree of neuromuscular functional incapacity. Table 1 shows the frequency of the assessed neuromuscular symptoms. These symptoms were evaluated when swallowing difficulties were first reported. At that time, distal muscle weakness was present in all patients and bifacial weakness in 97%. Slightly more than half the patients (54%) presented with the recognisable myopathic facies. In a considerable number - 87 % - myotonia could be provoked with ease clinically in the hands and/or in the tongue.

All 169 patients were on complete oral feeding when referred for the first time to the NMRC. By the end of the study period, oral feeding was considered unsafe in two patients, who needed a percutaneous endoscopic gastrostomy (PEG). The most frequently reported swallowing symptoms were: frequent choking, impression of difficult pharyngeal transport, piecemeal deglutition, need for smaller bite size, need for adaption of mealtime duration and difficulty with chewing (Table 2). Table 3 shows the distribution of different swallowing scale scores in the total population, and for the MD disease type. Most patients reported minor swallowing abnormalities and were therefore given a score of 2.

*Objective swallowing dysfunction*

The swallowing function of 47 MD patients (28% of the total population of 169) was checked by means of videofluoroscopy. All the patients, and even three patients who subjectively reported no swallowing complaints, had swallowing abnormalities to some extent (Table 4). The pharyngeal phase of swallowing was most compromised, as demonstrated radiographically in reduced pharyngeal peristalsis, delayed pharyngeal initiation, vallecular and piriform stasis and fragmented swallowing. Aspiration was seen in 49% of the patients and it occurred mostly during swallowing.

In the oral phase, the most frequently observed signs were lack of bolus control (38%) and difficult initiation of swallowing (19%).

Remarkably, a typical 'hung position' of the hyoid after swallowing was seen in 42% of the patients evaluated for this feature (n = 24). The following (as described previously by Bosma and Brodie[20]) seemed to happen: during the first swallow, the hyoid rose normally upward and forward from its resting position. It then remained briefly in that elevated position before returning only partly to its resting position. During the next swallow, the hyoid bone rose again from this

*Table 3*

Scores on Swallowing Scale (SS) in total population and according to disease type

| Score | Description of the subclass | total population | adult | childhood | congenital |
|---|---|---|---|---|---|
| | | n=101 | n=69 | n=25 | n=7 |
| 1 | Asymptomatic swallowing dysfunction: no subjective complaints. | 0% | 0% | 0% | 0% |
| 2 | Minor abnormalities: sporadic formation of residue, choking and/or coughing, complete oral intake. | 65% | 75% | 56% | 0% |
| 3 | Complete oral intake, need to adapt mealtime duration or amount of food intake during the meal, no need for additional nutrition. | 16% | 15% | 12% | 43% |
| 4 | Complete oral intake, but need for restriction of food consistency. | 15% | 7% | 24% | 57% |
| 5 | Complete oral intake, but with additional nutrition by mouth. | 4% | 3% | 8% | 0% |
| 6 | Need for additional non-oral nutrition. | 0% | 0% | 0% | 0% |
| 7 | Complete non-oral nutrition. | 0% | 0% | 0% | 0% |

abnormal 'hung' position. As a consequence, the tongue was displaced ventrally and therefore failed to move dorsally in the normal rolling sequence to close the oropharynx around the bolus. Disturbed pharyngeal initiation was the result.[20]

Generally speaking, the same trends were found in all onset types. Regurgitation was the only complaint that was clearly more prevalent in the adult type. Patients with the childhood type needed to switch to a soft diet more often, and they reported more recurrent pulmonary infections. Comparison of the videofluoroscopic data showed no differences between the different onset types. No clear link could be found between the reported swallowing problems and the objective oropharyngeal alterations. It is worth noting that we found the same constellation of radiographic abnormalities in the three patients who reported no swallowing complaints. Aspiration was even observed in two of them.

## Discussion

Until now, the effect of myotonic dystrophy on the swallowing function has not been investigated in large patient series. Since MD is a relatively rare disease, most of the published papers suffer from bias associated with the low number of patients studied.[4]

### *Swallowing symptoms*

Different studies have recounted a prevalence of dysphagia in MD ranging from 25% to 95%.[4,13,4,15,24] In our study population, 60% of the patients with MD reported swallowing difficulties. The complaints were in a descending order of frequency: frequent choking, impression of difficult pharyngeal transport, piecemeal deglutition, need for prolongation of mealtime duration, difficulty with chewing and regurgitation. This concurs with recent literature. Willig *et al*. also reported that choking was listed by two thirds of 110 MD respondents questioned about alimentation, making it the most frequent complaint.[24] In our population, a considerable percentage – 19% – already had swallowing difficulties before the diagnosis of MD was made. This area has been investigated in the past by Ronnblom *et al*. only, who found that 28% of their study group of 40 patients with MD had gastro-intestinal problems preceding the actual diagnosis of MD, with dysphagia as the second most frequent complaint.[15] Given the early involvement of the muscles of the head and neck in this disease, this finding is not very surprising.

### *Radiographic findings: frame-by-frame evaluation revealed a recognisable pattern*

#### Oral Phase

In accordance with previous studies, the most striking difficulties were lack of bolus control and difficult oral initiation. Both lead to a risk of aspiration before the actual swallowing movement. Weakness and diminished coordination of the tongue could be the origin of these abnormalities, and these difficulties are probably exacerbated by

*Table 4*
Radiographic oropharyngeal symptoms in total population and according to disease type

| Fluid | Total group (n=47) | Adult (n=29) | Childhood (n=13) | Congenital (n=5) |
|---|---|---|---|---|
| Oral phase | | | | |
| Oral backflow | 0% | 0% | 0% | 0% |
| Oral stasis | 2% | 3% | 0% | 0% |
| Disturbed oral movements | 2% | 3% | 0% | 0% |
| Difficult initiation | 19% | 21% | 8% | 40% |
| Difficult oral transport | 11% | 10% | 15% | 0% |
| Lack of bolus control | 38% | 33% | 38% | 60% |
| Pharyngeal phase | | | | |
| Nasal backflow | 0% | 0% | 0% | 0% |
| Delayed initiation | 47% | 55% | 23% | 60% |
| Stasis in the valleculae | 47% | 48% | 61% | 0% |
| Stasis in the piriform sinusses | 85% | 77% | 92% | 100% |
| Reduced pharyngeal peristalsis | 87% | 83% | 92% | 100% |
| Penetration[a] | 38% | 37% | 54% | 0% |
| Aspiration[b] before swallowing | 6% | 3% | 15% | 0% |
| Aspiration[b] during swallowing | 30% | 31% | 23% | 40% |
| Aspiration[b] after swallowing | 13% | 14% | 15% | 0% |
| Fragmented swallowing[c] | 64% | 72% | 38% | 80% |
| Oesophageal phase | | | | |
| Regurgitation | 2% | 3% | 0% | 0% |

[a]: penetration: entrance of contrast material in the laryngeal vestibule above the level of the vocal cords.
[b]: aspiration: entrance of contrast material in the laryngeal vestibule below the level of the vocal cords.
[c]: fragmented swallowing: piecemeal deglutition or not being able to swallow entire bolus at one time.

the bifacial muscle weakness and wasting that characterises MD.

In addition, various studies report myotonia of the tongue, a typical sign of MD during neurological examination, as a cause of a lack of lingual coordination and the disturbance of the oropharyngeal passage.[11,24] Some patients complain spontaneously of sudden cramps in the tongue during eating or speaking. Myotonia is commonly most marked in patients with relatively minor muscle weakness and wasting, and may be difficult to elicit in advanced cases with severe wasting[1].

## Pharyngeal phase

The main feature we observed was reduced pharyngeal peristalsis in 87% of the patients examined, with subsequent vallecular and pyriform stasis. This has also been the most consistent finding in previous studies, and it leads to prolonged pharyngeal transit time with a consequential risk of tracheal aspiration and nasal regurgitation.[2,11,13,24] More drastically, Swick *et al*.[21] and Pierce *et al*.[18] found pharyngeal hypocontractility in all their study subjects. A fluoroscopic study by Pruzansky and Profis showed actual ballooning of the pyriform sinuses and valleculae after swallowing, in conjunction with a greatly prolonged clearance time, up to 90 minutes after deglutition.[10]

When bolus material passes the ramus mandibulae before the pharyngeal phase is initiated, it is called delayed swallowing initiation. This second important abnormality occurred in 47% of our study group and it reflects a delay in the triggering of the swallowing reflex. Ertekin *et al*.[14] recently suggested a combination of reasons for this delay in MD: it may be the result of poor tongue and sub-mental muscle control due to dystrophy and subsequent weakness, sometimes complicated by myotonia of the tongue and laryngeal elevator muscles, in combination with the dysfunctional involvement of the corticobulbar pyramidal fibres, which are normally responsible for initiating

voluntarily induced swallows. It should be emphasised that none of these possible mechanisms can be solely responsible for the witnessed delayed swallowing initiation.[14]

Thirdly, fragmented swallowing was demonstrated in 64% of patients. Pharyngeal hypocontractility in combination with a reduced lingual driving force would seem to be a possible cause. This is partly compatible with Ertekin *et al*., who found abnormal piecemeal deglutition and repetitive swallows in 67% of their patients.[14] Nevertheless, they considered it exclusively as an oral phase abnormality, arguing that bolus control and formation is mainly provided by the tongue, masticatory and buccal muscles. If these muscles are weak, the bolus will be divided into pieces and swallowed successively.[14] We believe that reduced pharyngeal peristalsis also contributes to the fragmented swallowing.

Finally, as a fourth radiographic feature during the pharyngeal phase, aspiration occurred in half of the patients in this study, mostly during swallowing. Laryngeal hypocontractility or delayed laryngeal contraction appears to be the origin. The risk of aspiration also increases when the oral phase problems are aggravated by pharyngeal hypocontractility. As hypopharyngeal stasis proved to be the most frequent finding in our study group, one could consider most patients in the current study to be at risk of aspiration after swallowing. Leonard *et al*. reached the same conclusions in their study population and proposed advising conservative reflux precautions routinely for MD patients with objective significant pharyngeal residue.[24] These findings are of considerable importance since aspiration in conjunction with weak respiratory muscles and restrictive pulmonary changes is frequently the cause of death in MD.[1,6,16,18,23,24]

## Myotonia

In this retrospective radiographic analysis, we found a single straightforward item of evidence that myotonia plays a part in swallowing difficulties: the delayed reposition of the hyoid bone after the swallowing movement was observed in 42% of our patients. This phenomenon was first described by Bosma and Brodie, who stated that it was typical of the early stages of myotonic dystrophy, referring to it as a 'hung position'.[8,20] This myotonic disturbance could be useful in detecting MD in patients in whom dysphagia is an early symptom. On the other hand, Bosma and Brodie also warned that it could easily be mistaken for a disorder affecting the coordination of swallowing.[20]

Other evidence supporting oropharyngeal prolonged contraction showed myotonia in the musculus tensor veli palatini and sustained downward displacement of the soft palate during swallowing and speech.[20] This picture was supported by Pruzansky and Profis,[10] who also described frequent lingual myotonia in their study population. Recently, Leonard *et al*. showed that relaxation of the epiglottis was significantly delayed in patients with MD by comparison with healthy controls.[24]

In conclusion, the constellation of radiographic findings in patients with MD in our study as well as in others is not, to our knowledge, compatible with any other disease.[2,18] Given the role of videofluoroscopy in the early identification of oropharyngeal abnormalities with possible clinical implications, we suggest that the videofluoroscopic study of swallowing should be included in the evaluation of MD patients.

### *Relation between symptoms, radiographic findings and degree of myopathy?*

Evidence relating to the consistent relationship between the occurrence of swallowing symptoms and severity of the myopathic disorder has been conflicting. Some authors have reported a statistically significant correlation of swallowing dysfunction with the degree of the muscular involvement.[10,19] Others tend to describe only a positive correlation with the duration of the skeletal muscle disease[4]. Some researchers have found no correlation at all.[13,22] Leonard *et al*. concluded that the severity of swallow impairment in MD subjects differed in ways that were not predictable on the basis of subject age or time since diagnosis.[24] We agree since, on the basis of our review of records and images in this larger study, it was also difficult to establish a clear difference between the three types of MD in swallowing complaints, as well as in radiographic alterations. It is of further interest that researchers generally agree that there is no definite association between the occurrence and severity of dysphagia symptoms, and the presence of oropharyngo-oesophageal abnormalities. Patients without symptoms of dysphagia also often prove to have abnormal swallowing when evaluated

radiographically.[10,11,13,17,19,21-24] Bellini *et al*.[4] suggested the concept of *motor reserve* in the swallowing mechanism as a possible explanation. Even when fairly weakened, as in MD, the tongue and pharyngeal muscles can contract sufficiently to push a bolus through the UES into the oesophagus, after which the force of gravity is enough to complete the swallowing process. Moreover, because of the slow evolution of the disease, MD patients may develop compensatory mechanisms and therefore gradually adapt to the impairment of their swallowing function.[4,13]

However, this compensation for the slow deterioration of the bulbar function means that swallowing problems are frequently underestimated by both the patient and the physician. Furthermore, patients with MD often have personality disorders and some impairment of intellectual functions, including a lack of interest in their disease in particular.[4,14,21,23,24] This could play a significant part in their lack of awareness of their swallowing dysfunction. Patients should therefore be closely monitored and questioned to make appropriate management decisions.[14]

*Weaknesses of the present study*

A retrospective study will always have certain limitations since it draws on existing data that have been recorded for reasons other than research, and that are unblended and uncontrolled. We therefore depended on the availability and accuracy of the medical records of each patient in the NMRC. We also regret the absence of an additional functional endoscopic examination of swallowing during follow-up for these patients.

Furthermore, radiographic data for all patients were available for fluid consistencies only. We can presume that more solid contrast material may have resulted in more findings of oropharyngeal stasis and aspiration after swallowing.

Finally, our data are descriptive only and they cannot be compared statistically with a control group.

## Conclusion

The findings of this retrospective study confirm and extend previous reports about the presence and nature of swallowing dysfunction in a large number of patients with MD. The pharyngeal phase of swallowing is most compromised, with common complaints such as frequent choking, formation of residue and fragmented swallowing. Radiographic evaluation revealed a unique and recognisable pattern of oropharyngeal motility disturbance, mainly characterised by reduced pharyngeal peristalsis, hypopharyngeal stasis, delayed swallowing initiation and fragmented swallowing. These abnormalities may be present even if symptoms stated by the patient or the severity of the disease are not remarkable. There is a significant risk of aspiration. Muscular weakness seems to be the major contributor to swallowing impairment in MD. Nevertheless, myotonia can be seen in early disease in the tongue, palatal muscles and the muscles suspending the hyoid bone. The relationship between the degree of muscular dystrophy, oropharyngeal motility and clinical symptoms is still debated and requires investigation in greater depth. Dysphagia can be the only symptom of MD and it may precede the muscular signs by many years. We therefore suggest that examination of the swallowing function may be a valuable diagnostic tool in certain early or atypical cases. The potential benefit should not to be underestimated since a diagnosis of MD allows for appropriate precautions to be taken prior to anaesthesia and surgery (for example with respect to the restrictive pulmonary function) and therefore be lifesaving, even in asymptomatic patients.

## References

1. Harper PS. *Myotonic Dystrophy – Major problems in neurology*. Series vol 9. W.B. Saunders, Philadelphia: 1979.
2. Hillarp B, Ekberg O, Jacobsson S, Nylander G, Aberg M. Myotonic dystrophy revealed at videoradiography of deglutition and speech in adult patients with velopharyngeal insufficiency: Presentation of four cases. *Cleft Palate Craniofac J*. 1994;31(2):125-133.
3. Omim Database: *Dystrophia Myotonica*. Available at: http://www.omim.org/entry/160900. Accessed November 5, 2015.
4. Bellini M, Biagi S, Stasi C, Costa F, Mumolo MG, Ricchiuti A, Marchi S. Gastrointestinal manifestations in myotonic muscular dystrophy. *World J Gastroenterol*. 2006;12(12):1821-1828.
5. de Die-Smulders C E M. *Long-term clinical and genetic studies in myotonic dystrophy*. Proefschrift Universiteit van Maastricht. Universitaire Pers Maastricht, Maastricht: 2000.
6. Howeler CJ. Nieuwe inzichten bij dystrophia myotonica. *Ned Tijdschr Geneeskd*. 1988;132(23):1076-1078.

7. Gagnon C, Chouinard MC, Laberge L, Veillette S, Bégin P, Breton R, Jean S, Brisson D, Gaudet D, Mathieu J. Health supervision and anticipatory guidance in adult myotonic dystrophy type 1. *Neuromuscul disord*. 2010;20(12):847-851.
8. Kilman WJ, Goyal RK. Disorders of pharyngeal and upper esophageal sphincter motor function. *Arch Intern Med*. 1976;136(5):592-601.
9. Linoli G, Ceccatelli P, Malentacchi GM. Pharyngo-esophageal motility disorders in Steinert's myotonic dystrophy. Description of a case presenting with dysphagia. *Pathologica*. 1992;84(1092):523-530.
10. Pruzanski W, Profis A. Dysfunction of the alimentary tract in myotonic dystrophy. *Israel J Med Sci*. 1966;2(1):59-64.
11. Nowak TV, Ionasescu V, Anuras S. Gastrointestinal manifestations of the muscular dystrophies. *Gastroenterology*. 1982;82(4):800-810.
12. Casey EB, Aminoff MJ. Dystrophia Myotonica presenting with dysphagia. *Br Med J*. 1971;2(5759):443.
13. Modolell I, Mearin F, Baudet J-S, Gamez J, Cervera C, Malagelada J-R. Pharyngo-esophageal motility disturbances in patients with myotonic dystrophy. *Scand J Gastroenterol*.1999;34(9):878-882.
14. Ertekin C, Yüceyar N, Aydoğdu I., Karasoy H. Electrophysiological evaluation of oropharyngeal swallowing in myotonic dystrophy. *J Neurol Neurosurg Psychiatry*. 2001;70(3),363-371.
15. Ronnblom A, Forsberg H, Danielsson A. Gastrointestinal symptoms in myotonic dystrophy. *Scand J Gastroenterol*. 1996;31(7):654-657.
16. Garrett JM, DuBose TD Jr, Jackson JE, Norman JR. Esophageal and pulmonary Disturbances in Myotonic Dystrophica. *Arch Intern Med*. 1969;123(1):26-32.
17. Hughes DT, Swann JC, Gleeson JA, Lee FI. Abnormalities in swallowing associated with dystrophia myotonica. *Brain*. 1965;88(5):1037-1042.
18. Pierce JW, Creamer B, Macdermot V. Pharynx and oesophagus in dystrophia myotonica. *Gut*. 1965;6(4):392.
19. Siegel CI, Hendrix TR, Harvey JC. The swallowing disorder in myotonica dystrophia. *Gastroenterology*. 1966;50(4):541-550.
20. Bosma JF, Brodie DR. Cineradiographic demonstration of pharyngeal area myotonia in myotonic patients. *Radiology*. 1969;92(1):104-109.
21. Swick HM, Werlin SL, Dodds WJ, Hogan WJ. Pharyngeal motor function in patients with myotonic dystrophy. *Ann Neurol*. 1981;10(5):454-457.
22. Eckardt VF, Nix W, Kraus W, Bohl J. Esophageal motor dysfunction in patients with muscular dystrophy. *Gastroenterology*. 1986;90(3):628-35.
23. Marcon M, Briani C, Ermani M, Menegazzo E, Iurilli V, Feltrin GP, Novelli G, Gennarelli M, Angelini C. Positive correlation of CTG expansion and pharyngoesophageal alterations in myotonic dystrophy patients. *Ital J Neurol Sci*. 1998;19(2):75-80.
24. Leonard RJ, Kendall KA, Johnson R, Mckenzie S. Swallowing in myotonic muscular dystrophy: a videofluoroscopic study. *Arch Phys Med Rehab*. 2001;82(7):979-985.
25. Cappabianca S, Reginelli A, Monaco L, Del Vecchio L, Di Martino N, Grassi R. Combined videofluoroscopiy and manometry in the diagnosis of oropharyngeal dysphagia: examination technique and preliminary experience. *Radiol Med*. 2008;113(6):923-940.

Ann Goeleven
UZ Leuven – Campus Sint Rafael Dienst NKO-GH-MUCLA
Kapucijnenvoer 33
3000 Leuven, Belgium
E-mail: ann.goeleven@uzleuven.be