Brief Reports

Clinical, Etiological, and Therapeutic Features of Jaw-opening and Jaw-closing Oromandibular Dystonias: A Decade of Experience at a Single Treatment Center

Pedro Gonzalez-Alegre[1*], Robert L. Schneider[2] & Henry Hoffman[3]

[1] Department of Neurology, Carver College of Medicine, The University of Iowa, Iowa City, Iowa, United States of America, [2] Department of Oral & Maxillofacial Surgery, College of Dentistry, The University of Iowa, Iowa City, Iowa, United States of America, [3] Department of Otolaryngology, Carver College of Medicine, The University of Iowa, Iowa City, Iowa, United States of America

Abstract

Background: Dystonia is a heterogeneous hyperkinetic disorder. The anatomical location of the dystonia helps clinicians guide their evaluation and treatment plan. When dystonia involves masticatory, lingual, and pharyngeal muscles, it is referred to as oromandibular dystonia (OMD).

Methods: We identified patients diagnosed with OMD in a Movement Disorders Clinic and Laryngeal Movement Disorders Clinic from a single institution. Demographic, etiological, clinical, and therapeutic information was retrospectively reviewed for patients with jaw-opening (O-OMD) and jaw-closing (C-OMD) OMD.

Results: Twenty-seven patients were included. Their average age of onset was in the sixth decade of life and there was a female predominance. Etiological factors linked in this study to OMD included a family history of dystonia or essential tremor, occupation, cerebellar disease, a dental disorder, and tardive syndrome. Clinically, patients with C-OMD presented with more prominent feeding difficulties, but seemed to respond better to therapy than those with O-OMD. In addition to the known benefits of botulinum toxin therapy, patients who described sensory tricks obtained benefit from the use of customized dental prosthesis.

Discussion: This works provides useful information on potential etiological factors for OMD and its response to therapy, and highlights the potential benefit of dental prosthesis for the treatment of OMD.

Keywords: Oromandibular dystonia, botulinum toxin, sensory trick, etiology

Citation: Gonzalez-Alegre P, Schneider RL, Hoffman H. Clinical, etiological, and therapeutic features of jaw-opening and jaw-closing oromandibular dystonias: a decade of experience at a single treatment center. Tremor Other Hyperkinet Mov. 2014; 4. doi: 10.7916/D8TH8JSM

Introduction

Dystonia is a movement disorder characterized by sustained or intermittent muscle contractions that cause abnormal, often repetitive, movements, postures, or both.[1] Dystonia is a very heterogeneous phenomenon, and its clinical variability, together with the low frequency of some of its forms, presents diagnostic and therapeutic challenges.[2,3]

A recent consensus update proposed classifying patients with dystonia according to clinical features and etiology.[1] The classification along the clinical axis aims to identify clinically defined forms of dystonia to aid in their diagnosis and treatment. Dystonia can present as a wide range of clinical syndromes that share the presence of this hyperkinetic movement disorder in different body parts. Due to this variability, there is no uniform clinical approach that is applicable to all.[3] For instance, adult-onset focal leg dystonia requires a diagnostic investigation, as it is likely to be acquired.[4,5] On the other hand, adult-onset cervical dystonia is usually idiopathic and rarely requires diagnostic testing beyond a good clinical examination.[6]

While some forms of dystonia are relatively common, such as adult-onset cervical dystonia,[6] others are less frequent and some clinicians don't have enough clinical experience to guide their practice. Therefore, it is important to share the experience of centers that

evaluate uncommon dystonia syndromes. This will help us to gain a better understanding of features that are specifically observed in these forms of dystonia, aiding clinicians and researchers in their quest to improve the quality of life of those suffering from this hyperkinetic disorder.

In this work, we retrospectively analyzed the etiological, clinical, and therapeutic features of a series of consecutive patients diagnosed over a decade with jaw-opening or jaw-closing oromandibular dystonia (O-OMD and C-OMD, respectively).

Methods

We performed a retrospective chart review of patients evaluated for a diagnosis of O-OMD and C-OMD at the University of Iowa Hospitals and Clinics in the period between 2004 and 2013. We first identified patients who had been evaluated in both the Movement Disorders Clinic (Department of Neurology) and the Laryngeal Movement Disorders Clinic (Department of Otolaryngology) through our electronic medical records. Of those, we searched their diagnostic coding information for the term dystonia. We used these criteria because in our institution, botulinum toxin treatment for patients with OMD is performed at the Laryngeal Movement Disorders Clinic. However, most of these patients are also evaluated in the Movement Disorders Clinic for the possibility of presence of dystonia outside of the oromandibular area and for consideration of additional diagnostic testing or therapeutic interventions.

Once identified, their medical records were reviewed, and only those diagnosed with C-OMD and O-OMD by one of our movement disorders experts and an otolaryngologist with expertise on oroman-dibular movement disorders (H.H.) were included in the study. We collected demographic, clinical, and therapeutic information including age at onset and at diagnosis, clinical diagnosis, presumed etiology, factors potentially linked to the development of dystonia as indicated by the patient or clinician (such as occupation, injury, etc.), family history of dystonia or other movement disorders, presence of a *geste-antagoniste* or sensory trick,[2] reported dysphagia, weight loss and body mass index (BMI), medical treatments attempted and the results, therapeutic use of dental prosthesis, and application of botulinum toxin treatments (including dose and muscles selected for the treatment of O-OMD and C-OMD). This protocol was reviewed and approved by the Institutional Review Board of our institution and, as a retrospective record review, the requirement to obtain informed consent from the patients was waived.

Results

A total of 67 patients who had been evaluated in the Movement Disorders and Laryngeal Movement Disorders Clinics during the defined period and had the term "dystonia" included in their diagnostic coding information. A review of their chart confirmed the diagnosis of O-OMD or C-OMD in 27 of them. The 40 excluded patients had a diagnosis of laryngeal dystonia (12), voice tremor (eight), classical tardive dyskinesias (eight), dystonia outside the oromandibular and laryngeal areas (six), or other unrelated diagnoses (six).

The demographic, clinical and etiological characteristics of the 27 patients are shown in Table 1. Four (15%) patients had a family history of dystonia or ET. Among potential etiological factors, three patients with idiopathic O-OMD related this disorder to their frequent use of loud speech due to their occupation (a teacher, an auctioneer, and a preacher). Furthermore, two patients developed their dystonia as a presumed consequence of cerebellar disease (a primary cerebellar degenerative disease and a cerebellar stroke). From a clinical point of view, we found that self-reported weight loss (though not objectively documented) was commonly seen in our patients with OMD, more prominently for the C-OMD group.

As shown in Table 2, most patients received botulinum toxin injections as a treatment for their OMD. Of the 11 with O-OMD who received at least one treatment, only eight had repeated injections, but the eight patients with C-OMD who initiated this treatment at our institution had repeated injections. For O-OMD, we targeted the anterior belly of the digastric (ABD) with or without additional injections into the lateral pterygoid muscles bilaterally, and for C-OMD the masseters and/or temporalis. Patients with additional anatomical involvement received injections in other muscles (not shown). The average dose in their more recent treatment was higher for all targeted muscles than in the initial application, although, for instance, the lateral pterygoid was abandoned as a target in all but two patients with O-OMD.

At our institution, patients who present sensory tricks that relieve their OMD symptoms are offered an evaluation at the Maxillofacial Prosthodontics Clinic for fitting with prosthetic dental prosthesis. Four out of eight patients evaluated in that clinic reported significant benefit and, in two cases, botulinum toxin treatments were not pursued at all due to the efficacy of these devices (Figure 1).

Discussion

In this study, we report a series of consecutive patients with C-OMD and O-OMD seen at a single institution over a decade. In addition to adding important information to the literature on the clinical features of these relatively rare forms of dystonia, this work highlights important features of OMD. First, consistent with previous reports, some of our OMD patients reported family members diagnosed with dystonia suggesting a genetic predisposition. Second, cerebellar disease and occupational public speaking could play a role in the pathogenesis of OMD. Third, while the small sample size precluded us from performing a reliable statistical comparison between C-OMD and O-OMD, our data suggested potential differences. Self-reports of weight loss seem to be more frequent in C-OMD, although patients with C-OMD responded better than those with O-OMD to botulinum toxin treatment. Finally, a sizable proportion of patients with OMD exhibit sensory tricks that can be used for the development of therapeutic prosthetic devices.

The retrospective nature of this study adds different sources of potential bias, such as clinician preference for specific treatment approaches or self-reported symptoms by patients that could not be confirmed (i.e., weight loss). In addition, the description of the

Table 1. Demographic and Clinical Characteristics of Patients with OMD.

			O-OMD	C-OMD	Total
Demographics					
	N		16 (59%)	11 (41%)	27
	Women		56%	64%	59%
	Caucasian		94%	82%	89%
Etiology					
	Idiopathic/primary		14 (88%)	6 (55%)	20 (74%)
	Secondary		2 (12%)	5 (45%)	7 (26%)
		Tardive	0	2	2
		Peripheral[1]	1	1	2
		Degenerative	1[2]	0	1
		Other	0	2[3]	2
Family history					
	Dystonia		1 (6%)	1 (9%)	2 (7%)
	PD		0	0	0
	Essential tremor		2 (12%)	0	2 (7%)
Clinical					
	Age onset (SD)		56 (14.8)	52.6 (16.4)	54.8 (15)
	DD (≥1 year:<1 year)		5:8	3:5	8:13
	Phenomenology				
		Sensory trick	10 (63%)	2 (18%)	12 (44%)
		Task-specific	1 (6%)	1 (9%)	2 (7%)
		Focal	11 (69%)	4 (36%)	15 (56%)
		+Cervical	0	3 (27%)	3 (11%)
		+Cranial	3 (19%)	1 (9%)	4 (15%)
		+Craniocervical	2 (12%)	2 (18%)	4 (15%)
		+Other	0	1 (9%)	1 (4%)
	Weight loss		2 (12%)	6 (55%)	8 (30%)
	BMI (SD)[4]		27.5 (7.4)	22.5 (7.8)	26.4 (6.3)

BMI, Body Mass Index; C-, Jaw Closing; DD, Diagnostic Delay; O-, Jaw Opening; OMD, Oromandibular Dystonia; PD, Parkinson's Disease; SD, Standard Deviation.

[1]Onset after dental procedures.

[2]Progressive cerebellar degeneration of unknown etiology.

[3]Cerebellar stroke and cerebral palsy.

[4]Taken from most recent visit (non-significant trend).

Table 2. Response to Treatment of Patients with OMD.

|		O-OMD	C-OMD
Botulinum toxin[1]			
	N	11	8
	Muscles (N/mean initial dose)[2]	ABD (11/6.1 U)	Temporalis (5/9.5 U)
		Lat. pterygoid (5/7 U)	Masseter (7/12.9 U)
	Muscles (N/mean final dose)[2]	ABD (8/9.4 U)	Temporalis (5/14 U)
		Lat. pterygoid (2/8.8 U)	Masseter (7/34.3 U)
Other medical therapies (total/benefit[3])			
	Anticholinergics	4/2	2/0
	Dopaminergic	3/0	2/0
	Benzodiazepines	4/1	1/0
	Baclofen	1/0	2/1
	Tizanidine	0/0	1/1
	Reserpine	1/1	0/0
	Amantadine	1/1	0/0

ABD, anterior belly of digastric; C-, Jaw Closing; lat., lateral; O-, Jaw Opening; OMD, Oromandibular Dystonia; U, units.

[1]All patients received initially botulinum toxin type A (Botox). One patient in the O-OMD group changed to botulinum toxin type B (Myobloc, dose not included in the analysis).

[2]Dose injected unilaterally. However, all injections were bilateral, with the same dose in both sides for all patients.

[3]Benefit was always reported as mild or transient. No significant and sustained benefit was described with medical treatment.

phenomenology is limited. In most patients, the more prominent component of the OMD was either jaw-opening or jaw-closing. However, a component of lateral deviation or tongue involvement could also have been present. We did not include those in this description because their presence or documentation was more variable. None of the patients with OMD had comorbid laryngeal dystonia. As a result, the data reported here have to be interpreted with caution and should be followed by prospective studies.

The demographic and clinical characteristics of our patients, such as female predominance, are comparable to and consistent with the few previously reported series of patients with OMD.[7–11] Similar to ours, Singer and Papapetropoulos[7] included only patients with C-OMD and O-OMD and compared their clinical features, whereas most other articles report all forms of OMD in aggregate. Therefore, we will briefly compare our findings to theirs. In contrast to our study, they found more males in their C-OMD group, and the reported mean age at symptom onset is higher in our group of patients than theirs (56 vs. 48 years). The retrospective nature of both studies and potential recall bias could explain these differences. Taking all reports in aggregate, and consistent with other forms of primary dystonia,[12,13] it is reasonable to conclude that OMD is more prevalent in women.

Interestingly, both studies describe several consistent findings. A family history of dystonia was found in ∼10% of subjects in both studies, a more frequent cervical involvement in subjects with O-OMD (41% and 27% in Singer and Papapetropoulos and this report, respectively) than C-OMD (18% and 0%), and a more frequent presence of sensory tricks in O-OMD (33% and 63%) than in C-OMD (0% and 18%). Furthermore, Singer and Papapetropoulos reported a moderate or marked improvement to botulinum toxin in 90% of patients with C-OMD but only in 43% of those with O-OMD. In our series, this intervention was more frequently abandoned in patients with O-OMD. Overall, these findings suggest that O-OMD and C-OMD have different clinical and therapeutic features, perhaps reflecting differences in the underlying pathophysiology. Replication of these findings in future prospective studies would be important for dystonia researchers. For instance, if the clinical and therapeutic differences between both forms of OMD are confirmed, patients should not be clustered together in clinical trials under the umbrella of OMD, but stratified as O-OMD or C-OMD.

We were interested in the identification of potential etiological factors in patients with OMD. Dystonia is thought to result from a combination of genetic and environmental factors. Our findings are
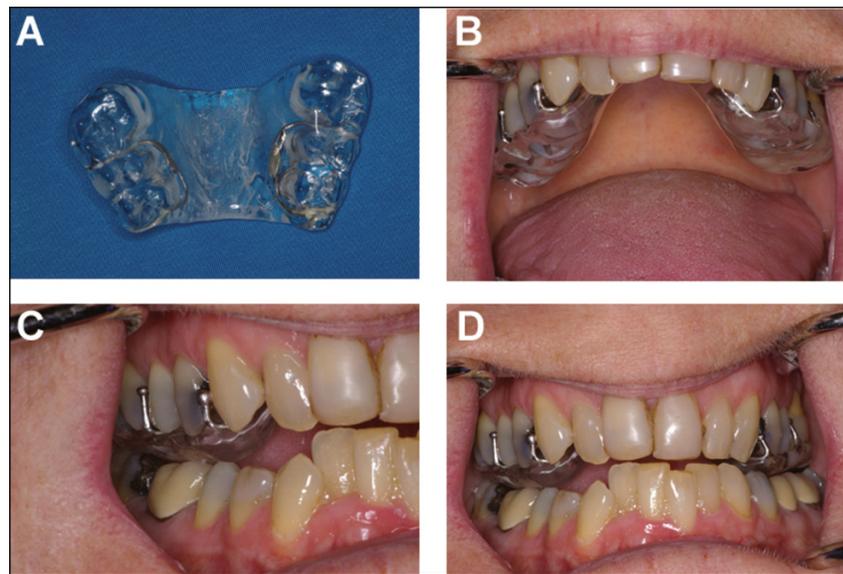
Figure 1. Dental Prosthesis Used in the Treatment of Oromandibular Dystonia. (A) Occlusal view of PMMA (polymethyl methacrylate) prosthesis with built in right side "pivot" point. (B) Prosthesis seated on the maxillary teeth. (C) Patient closing on the right side "pivot" point. (D) Frontal view of prosthesis in place with right side touching and left side just out of closing contact.

consistent with a potential genetic predisposition and a role of specific environmental factors in the development of OMD. When analyzing potential genetic susceptibility, two of our patients reported having at least another family member diagnosed with dystonia. This lends some support to the role of genetic factors in the pathogenesis of different forms of dystonia. Multiple studies have identified dystonia in some family members of subjects with different forms of presumably sporadic dystonia, including musician's dystonia.[14] This led to the hypothesis that "sporadic" dystonia is a dominantly inherited disorder with very low penetrance.[15] Environmental factors could be important determinants of penetrance in genetically susceptible individuals. In addition to two patients that developed OMD following an invasive dental procedure (diagnosed as "acquired"), we identified three patients with presumed idiopathic OMD that linked their disease to their occupation (school teacher, auctioneer, and preacher). OMD has been previously reported in an auctioneer,[16] a bingo caller,[17] and a telephone operator,[18] suggesting a real association. Future prospective studies should study the potential role of loud and frequent voicing as an environmental risk factor for OMD.

The neuroanatomical substrate underlying dystonia remains unclear. Traditionally, the basal ganglia have been considered to be the primary dysfunctional site in dystonia. Recent studies, however, have highlighted a potential role of the cerebellum.[19] Interestingly, two of our patients were diagnosed with acquired forms of OMD as a consequence of an idiopathic cerebellar degenerative process and a cerebellar stroke. This association, previously reported in another OMD patient,[20] lends additional support to the proposed role of the cerebellum in the pathogenesis of this movement disorder. Furthermore, two of our subjects had family members diagnosed with essential tremor, another hyperkinetic movement disorder that involves the cerebellum and is frequently comorbid with dystonia.[21]

In terms of treatment, all C-OMD, but not O-OMD, were followed up for repeated injections. It is unclear whether this is a consequence of lack of benefit or other reasons, as this was a retrospective study. Nevertheless, it is in agreement with a larger study of patients with OMD, where those with C-OMD had better functional improvement after botulinum toxin treatments than those with O-OMD.[8,11]

An interesting finding in our sample is the common presence of sensory tricks, more often in patients with O-OMD than C-OMD. This is particularly intriguing because, as indicated above, O-OMD seems to be more resistant to botulinum toxin therapy, and other therapeutic options are often needed. The use of dental prosthetic devices for the treatment of OMD has been described previously.[22,23] The protocol followed in our institution for the evaluation of this intervention has been particularly helpful[22] and will be described in some detail. Patients who find relief of their dystonia by a sensory trick are evaluated by a prosthodontics expert (R.L.S.). A prosthesis is fabricated for the maxillary arch, as it is less obtrusive for most patients because it does not interfere with their tongue (Figure 1). Dental impressions are made of both the maxilla (upper) and the mandibular (lower) dental arches. We routinely fabricate two clear acrylic resin prostheses with a "pivot" center for posterior tooth contact. The pivot center is made on opposite sides of each prosthesis, one on the right and one on the left, for the patient to use alternately for several days. The prosthesis only covers the biting surface of the back teeth so as to interfere minimally with the tongue. This allows the patient to have their mouth slightly open for speech with back tooth contact open approximately 3–5 mm. The prosthesis pivot point contact somehow permits the patient significant reduction/elimination of their dystonia. Previously, with the

fabrication of one prosthesis with a pivot point, it was noted the dystonia returned after several months of wear. The positive effect of the prosthesis is extended with alternating the pivot point periodically as previously described.[22] We propose that this type of intervention should be pursued in patients with OMD who benefit from a sensory trick, and certainly in those for whom botulinum toxin does to provide enough benefit.

In summary, we describe a series of patients with O-OMD and C-OMD seen in a single institution over a decade. Collectively with previous reports, this study helps us better understand important features of OMD. Furthermore, we identified several interesting etiological, clinical, and therapeutic aspects of C-OMD and O-OMD that should be prospectively evaluated in future, larger studies.

References

1. Albanese A, Bhatia K, Bressman SB, et al. Phenomenology and classification of dystonia: A consensus update. *Mov Disord* 2013;28:863–873, doi: http://dx.doi.org/10.1002/mds.25475.

2. Frucht SJ. The definition of dystonia: Current concepts and controversies. *Mov Disord* 2013;28:884–888, doi: http://dx.doi.org/10.1002/mds.25529.

3. Fung VS, Jinnah HA, Bhatia K, Vidailhet M. Assessment of patients with isolated or combined dystonia: An update on dystonia syndromes. *Mov Disord* 2013;28:889–898, doi: http://dx.doi.org/10.1002/mds.25549.

4. Pont-Sunyer C, Marti MJ, Tolosa E. Focal limb dystonia. *Eur J Neurol* 2010;17(Suppl 1):22–27, doi: http://dx.doi.org/10.1111/j.1468-1331.2010.03046.x.

5. Strader S, Rodnitzky RL, Gonzalez-Alegre P. Secondary dystonia in a botulinum toxin clinic: Clinical characteristics, neuroanatomical substrate and comparison with idiopathic dystonia. *Parkinsonism Relat Disord* 2011;17:749–752, doi: http://dx.doi.org/10.1016/j.parkreldis.2011.07.013.

6. Dauer WT, Burke RE, Greene P, Fahn S. Current concepts on the clinical features, aetiology and management of idiopathic cervical dystonia. *Brain* 1998;121(Pt 4):547–560, doi: http://dx.doi.org/10.1093/brain/121.4.547.

7. Singer C, Papapetropoulos S. A comparison of jaw-closing and jaw-opening idiopathic oromandibular dystonia. *Parkinsonism Relat Disord* 2006;12:115–118, doi: http://dx.doi.org/10.1016/j.parkreldis.2005.07.007.

8. Tan EK, Jankovic J. Botulinum toxin A in patients with oromandibular dystonia: Long-term follow-up. *Neurology* 1999;53:2102–2107, doi: http://dx.doi.org/10.1212/WNL.53.9.2102.

9. Bakke M, Larsen BM, Dalager T, Moller E. Oromandibular dystonia--functional and clinical characteristics: A report on 21 cases. *Oral Surg Oral Med Oral Pathol Oral Radiol* 2013;115:e21–26, doi: http://dx.doi.org/10.1016/j.oooo.2012.04.023.

10. Merz RI, Deakin J, Hawthorne MR. Oromandibular dystonia questionnaire (OMDQ-25): A valid and reliable instrument for measuring health-related quality of life. *Clin Otolaryngol* 2010;35:390–396, doi: http://dx.doi.org/10.1111/j.1749-4486.2010.02194.x.

11. Sinclair CF, Gurey LE, Blitzer A. Oromandibular dystonia: Long-term management with botulinum toxin. *Laryngoscope* 2013 Dec;123(12):3078–83.

12. Defazio G. The epidemiology of primary dystonia: Current evidence and perspectives. *Eur J Neurol* 2010;17(Suppl 1):9–14, doi: http://dx.doi.org/10.1111/j.1468-1331.2010.03053.x.

13. Steeves TD, Day L, Dykeman J, Jette N, Pringsheim T. The prevalence of primary dystonia: A systematic review and meta-analysis. *Mov Disord* 2012;27:1789–1796, doi: http://dx.doi.org/10.1002/mds.25244.

14. Schmidt A, Jabusch HC, Altenmuller E, et al. Etiology of musician's dystonia: Familial or environmental? *Neurology* 2009;72:1248–1254, doi: http://dx.doi.org/10.1212/01.wnl.0000345670.63363.d1.

15. Lohmann K, Klein C. Genetics of dystonia: What's known? What's new? What's next? *Mov Disord* 2013;28:899–905, doi: http://dx.doi.org/10.1002/mds.25536.

16. Scolding NJ, Smith SM, Sturman S, Brookes GB, Lees AJ. Auctioneer's jaw: A case of occupational oromandibular hemidystonia. *Mov Disord* 1995;10:508–509, doi: http://dx.doi.org/10.1002/mds.870100418.

17. Diaz-Sanchez M, Martinez-Castrillo JC. Botulinum toxin in a task-specific oromandibular dystonia in a bingo caller. *J Neurol* 2008;255:942–943, doi: http://dx.doi.org/10.1007/s00415-008-0716-y.

18. Kang SY, Kim H, Ma HI, et al. Highly task-specific oromandibular dystonia in a telephone operator. *Eur J Neurol* 2011;18:e136, doi: http://dx.doi.org/10.1111/j.1468-1331.2011.03468.x.

19. Jinnah HA, Hess EJ. A new twist on the anatomy of dystonia: The basal ganglia and the cerebellum? *Neurology* 2006;67:1740–1741, doi: http://dx.doi.org/10.1212/01.wnl.0000246112.19504.61.

20. Waln O, LeDoux MS. Delayed-onset oromandibular dystonia after a cerebellar hemorrhagic stroke. *Parkinsonism Relat Disord* 2010;16:623–625, doi: http://dx.doi.org/10.1016/j.parkreldis.2010.07.010.

21. Hedera P, Phibbs FT, Fang JY, Cooper MK, Charles PD, Davis TL. Clustering of dystonia in some pedigrees with autosomal dominant essential tremor suggests the existence of a distinct subtype of essential tremor. *BMC Neurol* 2010;10:66, doi: http://dx.doi.org/10.1186/1471-2377-10-66.

22. Schneider R, Hoffman HT. Oromandibular dystonia: A clinical report. *J Prosthet Dent* 2011;106:355–358, doi: http://dx.doi.org/10.1016/S0022-3913(11)60145-5.

23. Maestre-Ferrin L, Burguera JA, Penarrocha-Diago M. Oromandibular dystonia: A dental approach. *Med Oral Patol Oral Cir Bucal* 2010;15:e25–27.