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ORIGINAL ARTICLE

Oromandibular Dystonia: Demographics and Clinical Data from 240 Patients

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ABSTRACT

Objective To report demographic data from a large cohort of patients with oromandibular dystonia (OMD).

Methods This is a retrospective review of patients with OMD referred to our institution between 1989 and 2015. Demographic (age of onset, gender, and familial history of dystonia) and clinical (type of OMD, associated dystonia, and etiology of dystonia) data were collected from a cohort of 240 individuals.

Results The mean age of onset of OMD was 51.6 years old, with a female predominance (2:1). A family history of dystonia was found in 6 patients (2.5%). One hundred and forty-nine patients (62.1%) had the jaw-opening type of OMD, 48 patients (20.0%) had the jaw-closing type, and 43 patients (17.9%) had a mixed form of OMD. Lingual dystonia was also present in 64 (26.7%) of these patients. Eighty-two patients (34.2%) had a focal dystonia, 131 patients (54.6%) had a segmental dystonia, and 27 patients (11.3%) had a generalized dystonia. One hundred and seventy-one patients (71.3%) had idiopathic OMD.

Conclusion OMD is a chronic and disabling focal dystonia. Our study found a prevalence of female patients, an onset in middle age and a predominantly idiopathic etiology. Unlike other studies, jaw-opening was found to be the most frequent clinical type of OMD.

Key Words Oromandibular dystonia; focal dystonia; movement disorders.

"Dystonia is a movement disorder characterized by sustained or intermittent muscle contractions causing abnormal, often repetitive, movements, postures, or both. Dystonic movements are typically patterned, twisting, and may be tremulous. Dystonia is often initiated or worsened by voluntary action and associated with overflow muscle activation." Dystonia is classified along two axes: clinical characteristics and etiology.[1]

Oromandibular dystonia (OMD) involves the masticatory, lingual, perioral, and platysma muscles. The estimated annual incidence is 3.3 to 6.9 cases per 1 million people.[2,3] The different clinical forms encountered are jaw-opening oromandibular dystonia (JOOD), jaw-closing oromandibular dystonia (JCOD), and mixed OMD (patients with two or more combinations of either JCOD, JOOD, or jaw deviation (JD), and with no clinical predominance of any subtype[4]). OMD is a chronic condition affecting speech and swallowing, leading to an impaired quality of life. OMD can either be a primary disease or secondary to other disorders. Patients are categorized as having focal, segmental, multifocal, or generalized dystonia. OMD can be focal, but more often is part of a segmental or general dystonia.[5] The average age of symptom onset is between 50 and 60 years old, and most studies report a female predominance.[5-8] The

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purpose of this study was to analyze the demographic and clinical data of patients with OMD referred to our institution over a 25-year period and to compare our results to other studies published in the literature.

MATERIALS & METHODS

A retrospective analysis of patients with OMD referred to Fondation Ophtalmologique Adolphe de Rothschild (Paris, France) from 1989 to 2015 was conducted after approval of the affiliated ethical committee (CE_20150630_7_LSM). Clinical evaluation included a fiberoptic transnasal laryngoscopy with a swallowing test. All patients were evaluated by the same otolaryngologist and neurophysiologist. Patients underwent complete neurological testing, including brain imaging. Patients were rated as having JOOD, JCOD, or mixed OMD, with or without lingual involvement. Data concerning gender, age of onset, family history of dystonia, presence of associated anatomic sites with dystonia (blepharospasm, cervical dystonia, spasmodic dysphonia, or limb dystonia) were collected. The etiology of dystonia (idiopathic, tardive, neurodegenerative, post-anoxic, or post-traumatic) was recorded. Dystonia was defined as idiopathic when etiology was not determined.[1] Clinical symptoms such as impairment of speech, mastication or swallowing, pain, and dental effects were also recorded.

Electromyography was used for accurate identification of the muscles involved with the exception of the platysma and perioral muscles.

Statistical analysis was performed using R-software,[9] Fisher's exact test, and the Kruskal-Wallis test. The criterion for statistical significance was set at $p < 0.05$.

RESULTS

Two hundred and forty-four patients with OMD were referred to our institution between 1989 and 2015. Four patients who had initial assessment and treatment performed at another center were excluded. A total of 240 patients were included in our study (Table 1).

The mean age of onset was 51.6 years old (range, 3.0−85.5 years; SD = 18.6 years). The majority of patients were referred to us by neurologists (82%).

Table 1. Demographics and clinical characteristics

Characteristics	OMD (*n* = 240)
Females, *n* (%)	165 (68.8)
Males, *n* (%)	75 (31.2)
Mean age of onset, y (range, ± SD)	51.6 (3.0–85.5, ± 18.6)
OMD type, *n* (%)	
JOOD	149 (62.1)
JCOD	48 (20.0)
Mixed OMD	43 (17.9)
Lingual dystonia, *n* (%)	64 (26.7)
Etiology, *n* (%)	
Idiopathic	171 (71.3)
Tardive	31 (12.9)
Neurodegenerative	18 (7.5)
Post-anoxic	16 (6.7)
Post-traumatic	4 (1.6)
Associated movement disorders, *n* (%)	
Blepharospasm	93 (38.8)
Cervical dystonia	64 (26.7)
Spasmodic dysphonia	49 (20.4)
Limb dystonia	14 (5.8)
Generalized dystonia	27 (11.3)
Symptoms, *n* (%)	
Speech impairment	153 (63.8)
Mastication impairment	118 (49.2)
Swallowing impairment	65 (27.1)
Pain	78 (32.5)
Dental impairment	40 (16.7)
Family history of dystonia, *n* (%)	6 (2.5)

JOOD: jaw-opening oromandibular dystonia, JCOD: jaw-closing oromandibular dystonia, OMD: oromandibular dystonia.

There was a significant female preponderance—165 of 240 (68.8%) patients were women.

A total of 149 out of 240 (62.1%) patients had JOOD, 48 (20.0%) patients had JCOD, and 43 (17.9%) had mixed OMD. There was no significant difference between these groups in gender or age of onset. Lingual dystonia was present in 64 (26.7%) patients and was significantly more associated with JOOD and mixed OMD than with JCOD (32%, 26%, and 10%, respectively).

Eighty-two of 240 (34.2%) patients had a focal dystonia; 131 (54.6%) patients had a segmental dystonia; and 27 (11.3%) patients had a generalized form of OMD. Segmental dystonia categories included blepharospasm as part of Meige syndrome (93/240, 38.8%), cervical dystonia (64/240, 26.7%), spasmodic dysphonia (49/240, 20.4%), and limb dystonia (14/240, 5.8%). There was no correlation between OMD groups and focal versus segmental/generalized presentation.

Dystonic movements impaired speech in 153

(63.8%) patients, mastication in 118 (49.2%) patients, and swallowing in 65 (27.1%) patients. The movements caused pain in 78 (32.5%) patients and dental impairment (wear or early loss of teeth) in 40 (16.7%) patients. Mastication disorders were less present in JCOD than in JOOD and mixed OMD (25%, 56%, and 58%, respectively). Speech impairment was also less frequent in JCOD than in JOOD and mixed OMD (52%, 70%, and 63%, respectively, $p < 0.05$). There was no significant difference in swallowing impairment between these groups. Pain was common in JCOD, less present in mixed OMD and rare in JOOD (71%, 40%, and 18%, respectively, $p < 0.001$). Dental impairment was mostly present in JCOD or mixed OMD and almost absent in JOOD (35%, 37%, and 5%, respectively, $p < 0.001$).

Most patients (171/240, 71.3%) had idiopathic OMD. Thirty-one (12.9%) patients had tardive dystonia, 18 (7.5%) patients suffered from neurodegenerative disease (Parkinson disease $n = 9$, pantothenate kinase-associated neurodegeneration $n = 3$, GM1 gangliosidosis $n = 1$), 16 (6.7%) patients had a post-anoxic form, and 4 (1.6%) patients presented as post-traumatic OMD. Distribution of dystonia varied depending on etiology ($p < 0.001$). Idiopathic forms were either segmental (63%), focal (29%), or generalized (8%). Tardive dystonia patients were either focal (52%) or segmental (48%) but never generalized. Post-anoxic forms were often generalized (56%). A family history of dystonia was present in 6 patients (2.5%). Five of these patients suffered from an idiopathic focal or segmental dystonia, and one suffered from a generalized dystonia due to a familial mitochondriopathy.

DISCUSSION

Two hundred and forty patients diagnosed with OMD were evaluated over a 25-year period at our institution. To our knowledge, this study is the largest reported in the literature. A comparison of our demographic data with other studies is presented in Table 2.

Our results are similar to the data published by others in terms of gender ratio and age of onset.[5-8] The female prevalence has no pathophysiologic explanation.

Focal OMD is rare. OMD is more common as part of a spectrum of cranio-facial segmental or generalized dystonia.[5,9,10] In our study, 34.2% of patients had focal OMD whereas 54.6% had segmental dystonia and 11.3% presented with generalized dystonia. Sinclair et al.[11] reported similar findings with 12 of 59 (34.3%) patients having a focal OMD and 65.7% of patients presenting with a segmental or generalized form. In our study, the segmental form included blepharospasm (Meige syndrome) in 38.8% of patients, cervical dystonia in 26.7%, spasmodic dysphonia in 20.4%, and limb dystonia in 5.8%. Tan and Jankovic[4] also found blepharospasm (50.0%) and cervical dystonia (57.4%) as more frequently associated movement disorders.

In our study, JOOD was the most frequent form of OMD, 62.1% of patients had JOOD, 20.0% had JCOD, and 17.9% had mixed OMD, as opposed to other studies reported in the literature. Sinclair et al.[11] reported 47.4% of patients with JCOD, 35.6% with JOOD, and 16.9% with isolated lateral JD. Tan and Jankovic[4] reported 52.5% of patients with JCOD, 21.6% with JOOD, 1.9% with isolated lateral JD, and 24.0% with mixed OMD. This difference may

Table 2. Comparison of demographics with previously published studies

Clinical data	Sinclair et al.[11]	Tan and Jankovic[4]	Present study
n	59	162	240
Female, *n* (%)	40 (67.8)	111 (68.5)	165 (68.8)
Age of onset, mean (SD)	56.6 (14.0)	57.9 (15.3)	51.6 (18.6)
JOOD, *n* (%)	21 (35.6)	35 (21.6)	149 (62.1)
JCOD, *n* (%)	28 (47.4)	85 (52.5)	48 (20.0)
Mixed OMD, *n* (%)	0	39 (24.0)	43 (17.9)
	JD: 10 (16.9)	JD: 3 (1.9)	
Lingual dystonia, *n* (%)	10 (16.9)	-	64 (26.7)
Idiopathic OMD, *n* (%)	54 (91.5)	102 (63.0)	171 (71.3)
Other movement disorder, *n* (%)	23 (65.7)	-	158 (65.8)
Family history, *n* (%)	-	10 (6.2)	6 (2.5)

JOOD: jaw-opening oromandibular dystonia, JCOD: jaw-closing oromandibular dystonia, OMD: oromandibular dystonia, JD: jaw deviation.

be explained by a bias in the recruitment of our patients whom were mostly referred to us by neurologists (82%) who typically referred JOOD and mixed OMD patients.

Lingual dystonia was present in 26.7% of our cohort and was significantly more associated with JOOD and mixed OMD than with JCOD. There was no correlation between OMD groups and gender, age at diagnosis, or focal versus segmental/generalized presentation. These results are similar to those of Sinclair et al.[11]

Chewing, swallowing, and speech are frequently impaired in OMD, altering quality of life and social relationships.[12] In our cohort, 63.8% of patients had speech impairment, and 49.2% had mastication disorders. Dystonic movements caused pain in 32.5% of patients and dental troubles in 16.7%. Sinclair et al.[11] described similar results with speech and eating impairment in 54.3% of patients, pain in 34.3% and dental effects in 20.0%. Unlike Sinclair et al.,[11] we found a correlation between predominant symptoms and OMD type. Mastication and speech impairments were less present in JCOD than in JOOD or mixed OMD. On the other hand, pain and dental troubles were more common in JCOD and mixed OMD. Dental effects were mostly present in JCOD and mixed OMD and rare in JOOD (35%, 37%, and 5%, respectively). This finding may be due to the hyperactivity of masticatory muscles leading to jaw tension, dental contacts, pain, and other dental effects.

Most patients in our cohort (71.3%) presented as idiopathic OMD. The most frequent secondary form was tardive dystonia (12.9%). Tan and Jankovic[4] observed similar results with 63.0% of patients having idiopathic OMD and 22.8% presenting as tardive type.

Conclusion

OMD is a chronic and disabling focal dystonia. Our study shows a prevalence of female patients, an onset of symptoms between 50 and 60 years of age, and an idiopathic etiology in the majority of patients. JOOD was the most frequent type of OMD, as opposed to other reports.

Conflicts of Interest

The authors have no financial conflicts of interest.

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