

Case Reports

Sudden Onset of Oromandibular Dystonia after Cerebellar Stroke

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Abstract

Background: We present the case of a 65-year-old female with sudden-onset involuntary mouth opening, deviation of the jaw, facial grimacing, and tongue movements that started 6 months prior to her admission.

Case Report: She was diagnosed with oromandibular dystonia. Differential diagnosis of oromandibular dystonia and various etiologies were investigated. Neuroimaging studies revealed a left cerebellar infarction.

Discussion: To our knowledge, this case is the first oromandibular dystonia presenting with cerebellar ischemic stroke. Possible roles of the cerebellum for the pathophysiology of oromandibular dystonia are discussed.

Keywords: Dystonia, oromandibular dystonia, cerebrovascular disease

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Introduction

Oromandibular dystonia (OMD) is a cranial segmental dystonia of the lips, jaw, and tongue, causing involuntary mouth closure or opening, deviation of the jaw, tongue movements, or any combination of these due to repetitive or sustained spasms of masticatory, facial, or lingual muscles. OMD is the most frequent cranial dystonia after blepharospasm¹ and can occur in isolation or appear together with other forms of craniocervical dystonia.² Although drug-induced, post-anoxic, neurodegenerative disorder-associated, and head injury-associated etiologies have been reported, the cause of OMD is unknown in the majority of patients. Other types of dystonia associated with cerebellar disease are known,^{3–5} but reports of OMD related to cerebellar stroke are rare.⁶ We present a case of OMD following a cerebellar ischemic stroke.

Case Report

A 65-year-old female was admitted because of involuntary mouth and tongue movements. She reported a sudden onset of gait problems accompanying these abnormal movements beginning 6 months prior

to her referral. Her medical history revealed hypertension and diabetes mellitus. Her medication history was unremarkable for dystonia. She had no history of consanguineous marriage and she had seven first-degree relatives who had never experienced any neurological disease. On neurological examination she exhibited deviation of the jaw to the left, right, or front, or a combination of these, together with lingual and perioral dystonia (Video 1). These movements were more pronounced during speech and chewing. The dystonic movements diminished with oral sensory feedback, such as holding a toothpick in her mouth. Her left upper and lower extremities were dysmetric and dysdiadochokinetic. She had slightly hyperactive deep tendon reflexes on the left side and her gait was ataxic. Her psychiatric examination was normal. Her complete blood count and biochemical tests were normal, except for high blood sugar levels (250 mg/dL) and hyperlipidemia (low-density lipoprotein: 176 mg/dL). Tests for ferritin, folate, and vitamin B12 levels were normal. She had high blood pressure (175/90 mm/Hg). Magnetic resonance imaging (MRI) showed chronic anterior inferior cerebellar artery infarction (Figure 1). Wilson's disease was excluded as the Kayser–Fleischer ring was not seen on ophthalmologic



Video 1. Patient with Oromandibular Dystonia. The video shows repetitive contractions of jaw, tongue, and perioral muscles in our patient.

examination and the ceruplasmin value was normal (24.3 mg/dL). Her peripheral blood smear was normal. She was treated with electromyographically guided botulinum toxin-A injections (Botox®), with 10 units to both masseter muscles, 5 to both digastric muscles, 15 to both lateral pterygoidei muscles, and 5 units to the platysma muscles, which provided moderate benefit.

Discussion

For decades, dystonia was thought to be a disorder of the basal ganglia.⁷ However, along with the basal ganglia, the cerebellum has been shown to contribute to the pathophysiology of dystonia.⁸ This contribution has been demonstrated in several imaging and molecular studies, most of which were focused on primary dystonia, shedding light on the mechanism of the dystonic phenomenon.

The problem underlying the dystonic phenomenon is now understood at the circuit level, namely the cerebello-thalamo-cortical network.⁹ In macaques, the link between the dentate nucleus and the striatum was shown by histological tract tracing.¹⁰ Another study showed a return pathway from the basal ganglia to the cerebellum.¹¹ Aberrant cerebellar activities in dystonic animals and decrease in

dystonia with the removal of the cerebellum in rats have been reported.¹²

In humans, the modulation of cortical excitability by cerebellar outputs via the basal ganglia-thalamo-cortical network has been shown.¹³ The GABAergic inhibition in the primary motor cortex by the cerebello-thalamo-cortical network may be deficient in dystonia. It has been proposed that cerebellar outputs alter basal ganglia activity, leading to dystonic movements.¹⁴ Moreover, changes of striatal dopamine levels with cerebellar stimulation⁸ and loss of cerebellar Purkinje cells in adult-onset primary focal dystonia have been shown.¹⁵ Furthermore, reduction in the connection between the cerebellum and the thalamus and microstructural abnormalities in the vicinity of the superior cerebellar peduncle have been reported in DYT mutation carriers.¹⁶ All of these reports favor the connection of the cerebello-thalamo-cortical and basal ganglia-thalamo-cortical networks in both animals and humans.

The cerebellum is thought to be responsible for processing proprioceptive information and changing the somatosensory thresholds in the cortex.¹⁷ In this sense, dystonia may be the result of the abnormal inhibition and plasticity of sensorimotor pathways.⁸ Indeed, lack of inhibition, sensory dysfunction, and abnormal plasticity may all be major causes of the dystonic phenomena.¹⁸ Detection of increased metabolism in the cerebellum and dorsal midbrain using fluorodeoxyglucose positron emission tomography in dopa-responsive dystonia supports this notion.¹⁹ A study with voxel-based morphometry also found increased gray matter volume in the cortex, basal ganglia, and cerebellum in idiopathic cervical dystonia.²⁰

Studies showing the relationship between cerebellar disease and OMD have been reported.^{3,4} Although the cause is not known in the majority of cases, there are some cases induced by drugs, anoxia, cerebellopontine angle meningioma, or cerebrotendinous xanthomatosis.^{21,22} On the other hand, OMD related to a cerebellar stroke has only been reported once: a case of late-onset OMD following a cerebellar hemorrhage.⁶ Such late-onset dystonia following an event has been attributed to maladaptive plasticity. Waln and LeDoux⁶ noted the slow development of reorganization leading to supersensitivity and abnormal rewiring within the cerebellar nuclei. These reports are in accordance with the concept about abnormal plasticity.

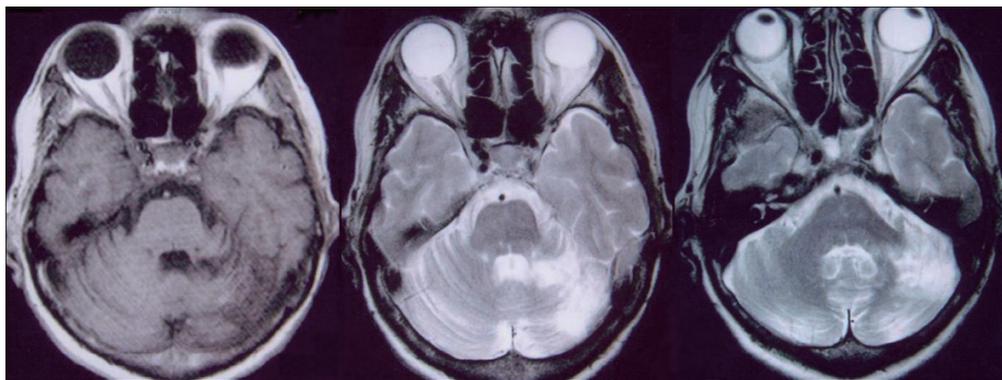


Figure 1. T1 and T2 Images. The images show chronic anterior inferior cerebellar artery infarction.

We herein report a case of OMD due to ischemia of the cerebellum. To our knowledge, this case is the first report of OMD following a cerebellar ischemic stroke. We argue that acute ischemia might have caused disruption of the output from Purkinje cells, resulting in the loss of GABAergic inhibition of the motor cortex via the cerebello-thalamo-cortical network.

By the time our patient was admitted, it was already 6 months since the beginning of the dystonic movements. She asserted that her dystonia and ataxia started simultaneously. As we had not followed the patient from the beginning, the possibility of other etiologies also needed to be discussed; in our clinical and laboratory work-up, we could find no evidence in favor of neurodegeneration. A review of the prescription, her family history, and other neuropsychiatric features, together with MRI and blood tests, enabled us to exclude drug-induced, hereditary degenerative dystonia and dystonia-plus syndromes. The main shortcoming was that we could not perform a genetic analysis in this case. However, we thought that a diagnosis of primary dystonia was unlikely in our patient, considering her overall radiologic and clinical picture. She had a sudden-onset focal dystonia without a tendency for generalization. Her age was even higher than that for late-onset primary dystonia-like DYT 7 or 13.²³ Moreover, apparent evidence for brain injury and additional cerebellar symptoms strongly suggested secondary dystonia as a diagnosis for our case.

A remarkably unusual point in our case is the instant onset of dystonia following the stroke. It raises a possibility that the lesion and the dystonia may be unrelated. In that case, our patient would fall into a category of late-onset primary focal dystonia, which we believe unlikely for the reasons we have discussed above (particularly with sudden and simultaneous onset with other cerebellar stroke signs). Generally, one would expect dystonia to appear later on, due to the process of maladaptive plasticity, ranging from months to years.²⁴ However, although rare, it may appear in a much shorter time, such as in days.^{25,26} Moreover, Scott and Jankovic²⁴ reported that the older the patient, the shorter the latent period between the cause and the dystonia.

Nevertheless, we were cautious about believing our patient's assertion, since it had been 6 months from the beginning of the symptoms. If we assumed a latent period for dystonia in our patient, it still had to be a very short period. We lost contact with the patient after her discharge, so we are unable to confirm this statement or re-examine her. We believe that lack of latency for dystonia in our patient indicates that slow processes such as maladaptive plasticity or reorganization are unlikely as causes of OMD.

Cerebellar diseases related with dystonia vary broadly in etiology but less in the clinical outcome. Rumbach et al.³ reported a case with hemidystonia due to infarction of the posterior and superior cerebellar arteries. This case had a similar but larger infarct than our case. One might argue that our case could have a more generalized dystonia, if she had had a larger lesion. Also, Alarcón et al.⁴ reported a central mass of the cerebellum with upper limb dystonia. In our patient, however, the lesion was more lateral. Our patient's dystonia was in the

mouth and tongue, which were anatomically in accordance with the cerebellar homunculus.²⁷

The cellular pathophysiology of ataxia and dystonia seems to be different. Lehéricy et al.²⁸ proposed that dystonia may be caused by dysfunction of the cerebellum, whereas ataxia is the result of destruction. Furthermore, as the cerebellum has structurally different functional areas, one region may be affected more severely than the other by means of "selective vulnerability," resulting in the destruction of one area and distortion of the other area, yielding different clinical pictures.¹⁵ In our patient, the ischemia was partial; she had an ataxia along with a focal dystonia, but we are unable to comment on whether she would still have dystonia if she had a different lesion. Destruction in the cerebellar tissue as the cause of ataxia is more pronounced than dystonia in patients with ischemia. As studies about the cerebello-thalamo-cortical network accumulate and more cases with cerebellar stroke and OMD are reported, the exact role of the cerebellum in OMD and other focal dystonia may be elucidated.

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