Velopharyngeal Dystonia: An Unusual Focal Task-specific Dystonia?

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Abstract

Background: Velopharyngeal dysfunction produces a nasal speech pattern because of the inability to close the nasal airway during speech, most often associated with anatomical abnormalities of the palate.

Case Report: We describe two cases of possible velopharyngeal dystonia, a task-specific movement disorder causing a speech pattern similar to velopharyngeal dysfunction. Both patients experienced treatment response with anticholinergic medication.

Discussion: Dystonia affecting speech via involvement of the pharyngeal musculature may be an unrecognized etiology of voice disorders.

Keywords: Velopharyngeal dystonia, functional voice disorder, spasmodic dysphonia

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Introduction

Dystonia can affect speech in myriad ways. Oromandibular dystonia may cause forceful contractions of the mouth, tongue, or jaw to produce abnormal speech patterns. Spasmodic dysphonia, whether by excessive adduction or abduction of the vocal cords, may produce strangled or breathy vocal patterns, respectively. These conditions, whether isolated and idiopathic, or because of a generalized neurological syndrome or injury, are well described phenomenologically. Dystonia affecting the pharyngeal musculature is not as well recognized, and may affect swallowing or breathing as an acute or delayed adverse effect of neuroleptic medication use. Here we describe two unusual cases of possible idiopathic focal task-specific dystonia affecting speech and involving primarily the pharyngeal musculature. Both patients experienced a robust improvement with anticholinergic medication.

Case reports

A 62-year-old male presented with a 2-year history of voice complaints. His symptoms started 6 days after undergoing a dental implant and persisted without progression. He felt that his voice was slurred, especially when pronouncing words with /k/ and /g/ sounds. When he awoke in the morning, his speech felt normal but would decline as the day went on. He had no difficulty singing, and denied dysphagia, odynophagia, difficulty chewing food, or involuntary tongue or jaw movements. His voice did not improve after drinking alcohol.

On examination his voice was hypernasal, particularly when pronouncing words with /k/, /g/, and /qu/ sounds. Whispering and singing were normal. Neither tremor nor breaks in his voice were observed during sustained phonation of /AA/ and /EE/ vowel sounds. Placement of a tongue depressor between his teeth improved his speech noticeably. His tongue thrust was midline and there was no dystonic posturing in the limbs. Muscle tone and strength were normal, and deep tendon reflexes were 2+ and symmetric. One month after starting trihexyphenidyl 2 mg three times a day, he noticed a 50%
subjective improvement in the clarity and nasal quality of his voice. Trihexyphenidyl was increased to 4 mg three times a day and on follow-up 3 months later he reported a 90% subjective improvement. This was reflected in his examination by improvement in hypernasal speech and reduced consonant-specific guttural dysarthria (Video segments 1 and 2).

A 41-year-old female presented with a 16-month history of voice complaints. Her difficulty began suddenly, and she described her voice as having a very “nasal” tone. She noticed that talking in a high-pitched voice improved her speech, and also discovered that putting considerable pressure on her throat and pressing up with her hands while extending her neck also improved her speech. There was no difficulty chewing or swallowing. She had been evaluated by multiple otolaryngologists, the most recent of whom noted velopharyngeal insufficiency in connected speech that resolved with her main sensory geste antagoniste of applying external pressure to the larynx. There was no abnormal laryngeal motion on videostroboscopy during tasks meant to elicit spasms of focal laryngeal dystonia. Neuroimaging was unremarkable. She received speech therapy, a proton pump inhibitor, as well as steroid injections to the vocal cords without benefit. On examination her speaking voice was markedly abnormal, with a simultaneously breathy and nasal quality. She was able to talk quite easily in a high-pitched voice. When she put significant external pressure on her larynx, her voice also improved. Hypernasal speech was triggered by /AA/ and /OO/ vowel sounds. She was treated with trihexyphenidyl 2 mg three times a day. On follow-up 2 months later she reported a 75% subjective improvement in her speech. She was able to speak at work and could be understood on the phone without changing the pitch of her voice or applying pressure to her throat. This was reflected in her examination by better tone production with previously difficult vowel sounds, although the hypernasal quality of her speech remained (Videos segments 3 and 4).

Discussion

These two unusual patients share certain salient features that make velopharyngeal dystonia a possible diagnosis. Symptoms were limited to connected speech, and were notable for hypernasal speech without tremor or prominent voice breaks, suggesting that abnormal movements arose not from the vocal cords but rather the velopharyngeal muscles. Direct examination of the vocal cords also supported this finding. Consistent with the task-specific nature of dystonia, certain phonemes in connected speech were selectively affected. This qualitative observation is similar to the objective findings of acoustic analysis in cases of adductor spasmodic dysphonia, and may be a distinguishing characteristic of dystonia from muscle tension dysphonia (discussed below). Our patients’ nasal speech was mitigated by adjusting pitch or singing, also a common observance in spasmodic dysphonia, or by using a sensory geste, a well-reported feature of oromandibular dystonia.

Videos

Segment 1. Case 1 Initial Evaluation. The patient has a thick, hypernasal voice. Hard consonant sounds are particularly difficult and connected speech is preferentially affected. Sustained vowel phonation is unaffected. Segment 2. Case 1 Post-treatment Follow-up. Three months after treatment with trihexyphenidyl 4 mg three times a day, the patient displayed reduced hypernasality and consonant-specific guttural dysarthria. Segment 3. Case 2 Initial Evaluation. The patient has a simultaneously breathy and hypernasal quality to speech. Changing voice pitch or utilizing a sensory trick improves speech quality transiently. Segment 4. Case 2 Post-treatment Follow-up. Two months after treatment with trihexyphenidyl 2 mg three times a day, the patient had improved tone production, though the hypernasal quality of speech remained.
Finally, both patients responded to anticholinergic medication after previous multimodal therapy failures.

The most likely alternative diagnosis in these cases is of a functional voice disorder. The term “functional” refers to an abnormality of physiology rather than structure, and hints at a psychiatric etiology. However, the accuracy and utility of this term or its synonyms used in clinical practice (e.g., psychogenic, conversion, or muscle misuse) remains a source of contention. The term muscle tension dysphonia (MTD) is sometimes used to delineate the dysregulated or imbalanced laryngeal and paralaryngeal muscle activity seen in functional voice disorders. The term MTD may avoid the possible stigma a patient might associate with the terms "functional" and "psychogenic."6

Indeed, patient 2 carried a diagnosis of functional voice disorder prior to our evaluation, with clinicians citing the acute onset of her speech difficulty as supportive of this diagnosis. She was in fact open to such a diagnosis, and proceeded with recommended speech therapy for many months prior to seeking further consultation. Yet she had no history of emotional trauma, psychiatric illness, or personality disorder. She also continued to work in spite of her voice difficulties and had no evident secondary gain. Her speech difficulty did not appear variable or distractable in the office. While the sudden onset of dystonia is unusual, it has been documented in the case of post-traumatic dystonia. Indeed, it may be more common in cases of oromandibular dystonia, particularly after dental procedures.7 Interestingly, patient 1 had a similar sudden onset of speech difficulty, only a few days after a dental procedure. He too had no history of emotional trauma or psychiatric illness, and also continued to work in spite of his speech difficulties.

The differential diagnosis of velopharyngeal dystonia includes velopharyngeal insufficiency, incompetence, and mislearning. Insufficiency is used to describe the physiologic consequences of anatomical defects (e.g., cleft palate) that impair airflow through the nose and mouth. Velopharyngeal incompetence is synonymous to insufficiency but implies intact anatomy. In either instance, poor movement of the velopharyngeal structures results in incomplete closure of the velopharyngeal valve. Dysarthria, which affects all elements of speech, may have hypernasality from poor velar movement as a prominent characteristic. Patients with speech apraxia will have difficulty in executing the proper sequence of speech events, including that of the velopharyngeal valve. Lower motor neuron damage for cranial nerves IX, X, or XII may lead to weakness of the velum or pharyngeal musculature, thereby causing incomplete closure of the velopharyngeal valve. In these ways, a whole host of congenital and acquired disorders of the central (e.g., stroke, brain tumor, multiple sclerosis, motor neuron disease) and peripheral (e.g., myasthenia gravis, neuropathy) nervous system may cause velopharyngeal incompetence. In each case, associated symptoms of the underlying disorder will suggest a diagnosis.10

There remain limitations in our clinical evaluation of these cases that make a functional voice disorder impossible to exclude. We are unable to visualize the incomplete closure of the velopharyngeal valve we hypothesize to be involved in the abnormal speech production. Diagnoses of dystonia, whether of speech, the face, or limbs, rely on such visualization of the abnormal movement. Also, our measures of speech dysfunction and response to treatment are all subjective. They do not include quantitative assessment with devices such as a nasometer. Indeed, case 2 had no subjective improvement in the nasal quality of her voice, and an argument could be made that her improved speech tone was purely the placebo effect. An evidence-based review of pharmacological treatments for dystonia would not suggest such a robust response to trihexyphenidyl treatment.411 Our clinical experience has also not suggested such a reliable response to anticholinergic medication in other focal dystonias.

Nevertheless, we would call attention to this possibly rare dystonia, in order to suggest a possible treatment path for patients without clear risk factors for psychogenic illness, and thereby spare them ineffective invasive therapies such as botulinum toxin or prolonged courses of speech therapy.

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