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Pharyngeal Dystonia Mimicking Spasmodic Dysphonia

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Summary

Objective—The aim of this study was to describe the presentation of pharyngeal dystonia (PD), which can occur as a focal or segmental dystonia with a primarily pharyngeal involvement for the discussion of treatment methods for controlling consequent symptoms. PD is specific to speech-related tasks.

Methods—A retrospective medical record review of four patients with PD was performed.

Results—All patients were initially misdiagnosed with adductor spasmodic dysphonia and failed standard treatment with botulinum toxin type A (BTX). On laryngoscopy, the patients were discovered to have segmental or focal dystonia primarily affecting the pharyngeal musculature contributing to their vocal manifestations. A novel treatment regimen was designed, which involved directing BTX injections into the muscles involved in spasmodic valving at the oropharyngeal level. After titrating to an optimal dose, all patients showed improvement in their voice and speech with only mild dysphagia. These patients have maintained favorable results with repeat injections at 6- to 12-week intervals.

Conclusions—PD, or dystonia with predominant pharyngeal involvement, is a rare entity with vocal manifestations that are not well described. It can be easily mistaken for spasmodic dysphonia. PD is specific to speech-related tasks. A novel method of BTX injections into the involved muscles results in a significant improvement in voice without significant dysphagia.

Keywords

Dystonia; Dysphonia; Segmental dystonia; Speech-language disorder; Spasmodic dysphonia

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SUPPLEMENTARY DATA

Supplementary data related to this article can be found online at doi:10.1016/j.jvoice.2017.05.004.
INTRODUCTION

Adult-onset focal and segmental dystonias affect a limited region of the body and can be task specific.¹ ² Spasmodic dysphonia (SD) is a focal dystonia characterized by laryngeal muscle spasms during phonation that is commonly the source of voice breaks.³ Adductor SD is the most common type of SD and is caused by overactivity of the thyroarytenoid muscles. Consequent hyperadduction of the vocal folds results in a strained-strangled voice quality that tends to occur with vowel-laden phrases. On the other hand, abductor SD is much less common and is caused by an overactivity of the posterior cricoarytenoid muscles, resulting in an excessive opening of the vocal cords and breathy voice breaks. Mixed SD can sometimes occur when laryngeal adductor and abductor muscles are simultaneously involved.⁴

In this paper, we describe a rare, poorly described form of dystonia that most significantly and predominately affects the pharyngeal musculature, known as pharyngeal dystonia (PD). This distinct entity can exist as either an isolated focal or segmental dystonia and could easily be mistaken for more common dystonias of this anatomic region. In this report, we describe four patients who presented with prominent voice breaks mimicking SD because of effortful speech and a constricted or strained-strangled voice. However, flexible laryngoscopy readily revealed spasms primarily of the pharyngeal rather than the laryngeal muscles leading to their symptoms. These patients exhibit oropharyngeal spasms during connected speech resulting in a distinctive-sounding dysphonia characterized by dystonic breaks principally with vowels and nasals that are best characterized by the voice of the cartoon character “Donald Duck.” All patients were evaluated by a board-certified neurologist to rule out systematic neurologic disease as a potential etiology. Finally, an effective treatment regimen is also described in our report, which involves botulinum toxin type A (BTX) injections directly into the muscles involved in spasmodic valving at the oropharyngeal level. This method allows for chemodenervation of the affected muscles with only mild dysphagic side effects.

METHODS

A retrospective medical record review of four patients with PD was performed over an 11-year period. Clinical notes and laryngeal videostroboscopies were reviewed. Patients were evaluated based only on available data in the patient files, which included the Voice Handicap Index-10 (VHI-10) score, the Voice-Related Quality of Life (V-RQOL) score, and the Consensus Auditory Perceptual Evaluation of Voice (CAPE-V) score. The anchors of the V-RQOL are 0 (lowest quality of life) and 100 (highest quality of life), and the anchors of the CAPE-V are 0 (normal voice) and 100 (most impaired).

CASE REPORTS

Case 1

A 56-year-old man presented to our clinic in August 2001, 2 months after developing acute-onset dysphonia following an upper respiratory infection. Laryngeal symptoms were the presenting and sole complaint of the disorder. Symptoms were progressive and had no clear
etiology. The physical examination revealed a strained-strangled speech quality, worse with vowels and nasals, but noted across all speech-related tasks. Flexible laryngoscopy revealed a hyperadduction of the supraglottis and vocal folds. Given these findings, a preliminary diagnosis of SD was rendered. After an initial course of voice therapy failed, the patient underwent electromyogram-guided injections of 1.25 units of Botox (BTX) into the thyroarytenoid-lateral cricoarytenoid (TA/LCA) complex. Following injection, the patient experienced an expected breathy voice. Unfortunately, there was limited improvement in the voice. Flexible laryngoscopy after BTX injections revealed a reduction in supraglottic spasms, vocal fold bowing from chemodenervation, and persistent spasms involving the oropharyngeal musculature with medial contraction of the pharyngeal walls and posterior displacement of the base of the tongue that was most prominent with vowels and nasals (Table 1).

The diagnosis was revised to adductor SD with segmental dystonia involving the tongue base. BTX injections of the false vocal folds (FVF) and strap muscle complex were administered in an effort to reduce breathiness, which resulted in slight improvement and reduced breathiness, but the patient remained dissatisfied with his voice results. The patient was prescribed 0.5 mg clonazepam three times daily by mouth. The patient also underwent injection of 8 units of BTX unilaterally into the TA/LCA complex and 25 units in the submental region, but three consecutive treatments yielded no improvement.

Given the limited improvement with the above interventions and the ongoing pharyngeal and tongue base muscle spasm with speech, a diagnosis of PD was entertained. BTX was injected superficially at the pharyngoepiglottic fold (PEF) in an effort to reduce posterior tongue base movement and pharyngeal contraction (see Figures 1 and 2A–C). Anatomically, this site approximates the location of the styloglossus, a tongue base retractor that is not an intrinsic muscle of the tongue. The dose was started at 5 units bilaterally and up-titrated in 2.5-unit increments. Increasing the dosage resulted in a significant improvement in the patient’s voice with decreasing breathiness. At 17.5 units injected bilaterally, however, the patient reported a dysphagia for solids, and the dosage was reduced to 15 units superficially in each PEF. This regimen maximized vocal benefits with only mild dysphagia. Our patient continued these treatments every 2–3 months for the past 7 years with consistently good results.

**Case 2**

A 67-year-old woman presented with an 8-year history of dysphonia characterized by pitch and voice breaks, reduced projection, increased vocal effort and strain, and vocal fatigue. Laryngeal symptoms, which were the presenting and sole complaint, were progressively worsening over time. There was no clear associated etiology. The patient was previously diagnosed with adductor SD based on clinical and examination findings but failed to benefit from voice therapy or BTX injections into the TA/LCA muscles. Flexible laryngoscopy (see Video S1) revealed spasms involving the oropharyngeal musculature with a medial contraction of the pharyngeal walls and a posterior displacement of the base of the tongue that were most prominent with vowels and nasals. Glottic and supraglottic compression were present to a lesser degree and there was no tremor. BTX injections into the posterior
cricoarytenoid muscles were attempted with a successful reduction in vocal fold abduction but no improvement in voice, indicating that the oropharyngeal spasms were not due to a compensatory abductor SD.

A diagnosis of PD was suspected. The patient received a peroral injection of 10 units of BTX superficially in each PEF and 5 units in each FVF, which resulted in some improvement in the patient’s voice. The dose of the PEF injections was serially increased to 20 units bilaterally while withholding the FVF injections, and continued vocal improvement was appreciated. Dosage continued to be titrated to maximize effectiveness. However, dysphagia developed at a dose of 23 units bilaterally, at which point the injection was reduced to 20 units bilaterally. Over a 29-month period, our patient received a total of 17 20-unit injections to each PEF with continued vocal improvement. Concomitant FVF injections have been used in half of the patient’s treatments, with some added benefit. The patient’s VHI-10 score on presentation was 40, which improved to 10 after each successful treatment. The patient’s best voice typically occurred within 5 days of injection and was a self-rated 75% of normal. Improvement was maintained for 5 weeks and the patient returned at 6-week intervals for reinjection. The patient continued to have dysphagia for solids after injection but these were well tolerated with appropriate dietary changes.

**Case 3**

A 76-year-old woman presented with a 3-year history of voice impairment. During the first year, the onset was insidious and progressive, but then subsequently stabilized. The patient’s vocal impairment was her presenting and sole complaint, and she characterized her voice as constricted with a Donald Duck quality. There was no clear etiology for the patient’s symptoms. On examination, running speech was marked by articulatory imprecision on lingual alveolar, lingual dental, palatal, and velar sounds. The manner of voicing did not appear to impact production. The voice was hyponasal, most likely because of posterior tongue carriage. Sustained vowels were produced without tremor, although there were irregular interruptions of sound. On presentation, our patient had a V-RQOL score of 23 with a global voice rating of 3 out of 7 (0 = no useable voice, 7 = perfect voice). The CAPE-V, a 100-mm visual analog rating scale, was used to assess the overall severity of voice quality and to quantify aberrant perceptual features in the voice. The patient received an overall score of 58 out of 100, indicating moderate-severe voice quality deficits.

Flexible laryngoscopy demonstrated a posterior retrusion of the tongue base and a pharyngeal constriction at the PEF during all voicing tasks, which was unchanged by phonetic loading. These findings were suggestive of PD. Initial trials of trihexyphenidyl and ethopropazine proved to be ineffective. Based on the patient’s clinical and examination findings, a BTX injection of 5 units to the inferior right palatopharyngeus and PEF regions was given, and the patient reported a marked improvement of her voice with no consequent airway or dysphagia symptoms. Two weeks after the patient’s initial injection, her V-RQOL score progressed to 70, her global voice rating increased to 7/7, and her CAPE-V score improved to 35 out of 100. Residual symptoms remained after the initial injection, and 10 units of BTX were injected to the left PEF region. This procedure resulted in 2 months of sustained voice improvement, but, by the third month, our patient reported a gradual vocal
deterioration. Repeat injection using 10 units of BTX was performed to both PEFs, which yielded excellent voice results without any dysphagia side effects.

Case 4

A 51-year-old man with a 3-year history of cervical dystonia presented to our clinic for difficulty in speaking, chewing, and swallowing, and with an abnormal rightward deviation of his lower face. The patient’s cervical dystonia initially presented with torticollis and neck muscle overactivity; the onset of the patient’s vocal complaints and PD occurred about 2 years after dystonic symptoms began in other locations. These symptoms were gradual in onset and progressive. The patient takes 2 mg clonazepam by mouth three times daily and has received regular BTX injections to posterolateral neck muscles with only a mild relief of his dystonia and no improvement to his voice. On examination, lip and jaw movements were normal, but the patient’s voice “cuts out” on occasion. Mild right torticollis was observed, with an episodic backward jerking of the head and downward tucking movements of the chin triggered by speaking. There was a visible overactivity of multiple neck muscles including the submental muscles, platysma, sternocleidomastoids, scalene complex, and levator scapulae. The patient’s running speech had a significant hyponasal quality with fluctuation in volume and intermittent disruptions of voicing suggestive of abductor SD. The patient reported a V-RQOL score of 12.5 out of 100 on initial presentation, representing a significant impact on quality of life.

On laryngeal videostroboscopy, voice breaks were found to be due to dystonic contractions of the pharyngeal, suprahypoid, and tongue musculatures resulting in an involuntary elevation of the larynx. Based on these clinical findings, trials of BTX injections were initially targeted to the suprahypoid, thyrohyoid, and cricothyroid muscles bilaterally, but they yielded limited vocal improvement. Subsequently, BTX injections to the PEF and the base of tongue were attempted, which achieved more significant improvement in our patient’s voice. The dosage was serially increased to 15 units bilaterally with no development of dysphagia. Over a course of 7 months, the patient returned five times to the clinic for BTX injections, reporting an ultimate V-RQOL score of 37.5 out of 100.

DISCUSSION

This case series describes a rare and infrequently discussed entity known as PD, which is defined as dystonia affecting primarily the pharyngeal musculature that can occur as an isolated focal or segmental disease. PD is characterized by pharyngeal involvement that affects the muscles at the oropharyngeal level. PD can easily be mistaken on presentation for SD because of effortful speech and constricted or strained-strangled voice. The mechanism for voice breaks is likely to involve constriction of the pharynx, with brief superimposed spasms leading to a transient block in the airflow. In the clinic, patients with PD have a distinctive clinical presentation with a voice quality similar to that of the cartoon character Donald Duck. These vocal manifestations are especially apparent on flexible laryngoscopy, which reveals spasms or constriction of the oropharyngeal musculature, especially during voice-related tasks and often coinciding with voice breaks. The voice breaks are clinically difficult to distinguish from those in SD because of spasms of the laryngeal muscles but,
unlike laryngeal dystonia, can be readily seen by flexible laryngoscopy. These observations are important because they imply that voice breaks are not specific to SD.

Because of the anatomic proximity between the pharyngeal, laryngeal, and oromandibular musculatures, it is difficult to separate dystonias involving these closely related areas by clinical examination alone. Consequently, PDs have only been previously described when accompanied by laryngeal or oromandibular involvement, resulting in terms such as “laryngopharyngeal” or “oropharyngeal” dystonia, and its unique vocal symptomatology has not been well documented. Furthermore, dystonia affecting primarily the pharyngeal musculature can be an early feature of more complex dystonias, which later progress to involve the larynx or oromandibular regions, such as idiopathic adultonset focal dystonia, neuroleptic-associated tardive syndrome, or systemic neurologic disorders such as Machado-Joseph disease. For example, in a case series of patients with lower cranial dystonia, pharyngeal involvement was identified as a potential presentation. As a result, it is difficult to isolate this disease process. In the few reports where PD was described as a unique phenomenon, the focus was primarily on the consequent dysphagia and did not adequately describe the vocal constriction or breaks also caused by the dystonia.

In addition, we also describe a novel treatment regimen for patients diagnosed with PD. We have found that targeting BTX injections specifically into muscles involved in spasmodic valving at the oropharyngeal level has resulted in a significant voice improvement. There are very few descriptions in the literature for BTX injections in the oropharynx, principally because of the risk of significant dysphagia. Our report demonstrates that, with an adequate refinement of the injection dosage, oropharyngeal spasmodic activity can be reduced while still maintaining an acceptable swallowing function. In our experience, the optimal dosage varied from patient to patient, but ranged from 15 to 20 units of BTX injected into the PEF via endoscopic guidance. Although two of our four patients experienced some dysphagia for solids with treatment, the benefits of the voice improvement outweighed the side effects, which our patients were able to manage through dietary modifications.

CONCLUSIONS

Given the similar clinical presentations of laryngeal SD and PD, it is difficult to differentiate between the common adductor SD and the rare variant PD based on clinical findings alone. Although the treatment modalities for these two entities are similar in that both involve BTX injections, the target sites are very different. An injection to the laryngeal musculature will have a significant impact on a laryngeal SD but will not improve voice symptoms in PD and vice versa. As such, otolaryngologists and speech pathologists must remain cognizant of these two variants of spasmodic dystonia to more accurately direct future treatment and management.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.
Acknowledgments

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Author roles: L.L. Shi: manuscript writing and editing. C.B. Simpson: research project conception, organization, and execution; manuscript writing and review. E.R. Hapner: research project conception, organization, and execution; manuscript writing and review. H.A. Jinnah: research project conception, organization, and execution; manuscript writing and review. M.M. Johns III: research project conception, organization, and execution; manuscript writing and review.

Conflicts of interest: H.A. Jinnah has received research grant support from the National Institutes of Health (NIH), the Atlanta Clinical & Translational Science Institute, the Emory University Research Council, the Lesch-Nyhan Syndrome Children’s Research Foundation, the Dystonia Medical Research Foundation, the Bachmann-Strauss Dystonia & Parkinson’s Foundation, and the Benign Essential Blepharospasm Research Foundation. He also is the principal investigator for the Dystonia Coalition, which receives the majority of its support through NIH grant NS065701 from the Office of Rare Diseases Research in the National Center for Advancing Translational Sciences and National Institute of Neurological Disorders and Stroke. The Dystonia Coalition receives additional material or administrative support from industry sponsors (Allergan Inc., Ipsen Biopharm, Medtronics Inc, and Merz Pharmaceuticals), as well as private foundations (The American Dystonia Society, The Bachmann-Strauss Dystonia and Parkinson Foundation, BeatDystonia, The Benign Essential Blepharospasm Foundation, Dystonia Europe, Dystonia Ireland, The Dystonia Medical Research Foundation, The Dystonia Society, The Foundation for Dystonia Research, The National Spasmodic Dysphonia Association, and The National Spasmodic Torticollis Association). Dr. Jinnah serves on the Scientific Advisory Boards for Cure Dystonia Now, the Dystonia Medical Research foundation, Tyler’s Hope for a Dystonia Cure, the Lesch-Nyhan Syndrome Children’s Research Foundation, and Lesch-Nyhan Action France.

References

FIGURE 1.
Diagram illustrating the site of injection at the pharyngoepiglottic fold.
FIGURE 2.
A. Flexible laryngoscopy showing a curved needle approaching the pharyngoepiglottic fold. 
B. Flexible laryngoscopy showing a curved needle entering the pharyngoepiglottic fold. 
C. Flexible laryngoscopy showing curved needle placement during injection.
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<td>1</td>
<td>Strained-strangled speech, worse with vowels and nasals</td>
<td></td>
<td>Hyperadduction of the supraglottis and vocal folds, oropharyngeal spasms with pharyngeal wall contraction and posterior displacement of the tongue</td>
<td>15 units of BTX superficially to PEF bilaterally</td>
<td>Significant improvement.</td>
<td>Mild dysphagia</td>
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<td>Pitch and voice breaks, reduced projection, increased vocal effort and strain, vocal fatigue</td>
<td>VHI-10</td>
<td>Oropharyngeal spasms with pharyngeal wall contraction and posterior displacement of the base of the tongue; mild glottic and supraglottic compression</td>
<td>20 units of BTX to PEF bilaterally</td>
<td>VHI-10\40</td>
<td>Dysphagia for solids managed with diet</td>
</tr>
<tr>
<td>3</td>
<td>Articulatory imprecision on lingual alveolar, lingual dental, palatal, and velar sound; hyponasal voice, irregular interruptions of sound</td>
<td>V-RQOL\23, CAPE-V\58, Global Voice Rating\3/7</td>
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<td>V-RQOL\7/0, CAPE-V\35, Global Voice Rating\7/7</td>
<td>No dysphagia</td>
</tr>
<tr>
<td>4</td>
<td>Significant hyponasal quality, fluctuation in volume, and intermittent vocal disruptions</td>
<td>V-RQOL\12.5</td>
<td>Dystonic contractions of the pharyngeal, suprahypoid, and tongue musculature</td>
<td>15 units of BTX to PEF bilaterally</td>
<td>V-RQOL\37.5</td>
<td>No dysphagia</td>
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Abbreviations: BTX, botulinum toxin type A; CAPE-V, Consensus Auditory Perceptual Evaluation of Voice; PEF, pharyngoepiglottic fold; VHI-10, Voice Handicap Index-10; V-RQOL, Voice-Related Quality of Life.