Spasmodic Dysphonia Symptoms as Initial Presentation of Amyotrophic Lateral Sclerosis

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Summary: A patient initially diagnosed with adductor-type spasmodic dysphonia was referred for botulinum toxin (Botox) injections, but found on subsequent evaluation to have amyotrophic lateral sclerosis, and therefore Botox was not administered. This unique case underscores the need to delay botulinum toxin treatments in any patient with recent onset symptoms, and to obtain thorough motor speech and voice, otolaryngologic, and neurologic evaluation in all patients prior to consideration for injection. Key Words: Spasmodic dysphonia—Botulinum toxin—Amyotrophic lateral sclerosis.

Spasmodic dysphonia (SD) is an action-induced laryngeal dystonia caused by a disorder of central nervous system motor processing (1). Adductor-type SD results in a choked, strained-strangled vocal quality, pitch breaks, and voice blocks of tension or effort that interrupt the continuity of phonation (2,3). These speech-related symptoms may coexist with apparently undisrupted nonspeech laryngeal maneuvers, including singing, laughing, coughing, throat clearing, and humming (4). Its exact etiology is unknown (1,5,6). SD may represent a subset of neurologic voice disorders (7). One theory supported by comparative examination of symptoms of SD with the dysarthrias and brain-imaging studies suggests a supranuclear lesion locus in the proximity of the basal ganglia as the likely site (8). It has been classified as a neurologic movement disorder with site of lesion presumed to be somewhere in the extrapyramidal system (6). This syndrome or constellation of symptoms and signs is often associated with other neurologic impairments (9).

Amyotrophic lateral sclerosis (ALS) is a degenerative neurologic disorder characterized by progressive dysfunction of both bilateral corticobulbar tracts (upper motor neuron) and lower motor neuron nuclei (4,10–12). The usual onset is around 50 years of age (13). Symptoms of ALS include weakness, fatigue, muscle atrophy, and fasciculations as evidence of lower motor neuron involvement (14). Signs of upper motor neuron damage include cramps, hyperreflexia, and spasticity (10–12). The disease varies from patient to patient and typically affects the limbs first (15). In up to 25% of cases, the earliest symptoms are bulbar in nature (16,17). Spastic paralysis with pseudobulbar dysphonia or flaccid weakness and hypoadduction of the vocal folds may predominate (4).

ALS characteristically produces a mixed spastic–flaccid dysarthria (4,18,19). Features of spastic dysarthria include slow speech rate, strained–strangled voice, and reduced stress and prosody. Features of flaccid dysarthria include hoarse, breathy vocal quality, consonant distortions, and short phrases. Hypernasality may be a feature of both spastic and flaccid dysarthria. Acoustic and perceptual studies have described phonatory instability (18,20,21) and phonetic contrast errors (19) as features of ALS.
Carrow et al. (22) studied 79 patients with ALS and found that 75% had hypernasality, 80% had harsh voice quality, 60% had strained–strangled voice quality, and 63% had voice tremor. Weakness of speech musculature resulting in dysarthria and dysphagia are common in the terminal stages of the disease (16,23). In cases of ALS, bulbar-onset laryngeal involvement may include flaccid weakness and hypoadduction of the vocal folds (4).

Botulinum toxin (Botox) injections are considered standard therapy for SD (24,25), but are contraindicated in patients with motor neuron disease such as ALS (26). Isolated strained-strangled vocal quality as an initial sign of ALS may be confused with symptoms of adductor SD. We present an unusual case of an ALS patient who at onset appeared to have signs and symptoms confined to the larynx, with these perceptual attributes of adductor-type SD.

CASE REPORT

A 72-year-old white woman sought consultation with an otolaryngologist in July 1992, following a 2-month history of intermittent aphonia and a persistent cough (see Table 1 for time course of symptoms and examinations). Her principal complaint was that her voice would suddenly "quit" and that sometimes she couldn't talk at all. She was a non-smoker and denied hoarseness, but complained of a chronic cough and, more recently, shortness of breath. Her past medical history included a hiatal hernia, tracheitis–bronchitis (treated with beclomethasone, and a transient ischemic attack 3 years earlier. Her initial examination disclosed a hyperactive gag reflex. On indirect mirror examination, the epiglottis, supraglottic larynx, intrinsic larynx, and vocal folds appeared grossly normal. Her voice quality was marked by sudden bursts of increased pitch, loudness, and effort in a spasmlike pattern. She never completely lost her voice during the initial exam. She was given a tentative diagnosis of SD and was referred to a speech–language pathologist for a voice evaluation including a videostroboscopy examination.

Speech evaluation conducted the following week employed a 90° rigid endoscope with topical anesthesia to suppress the gag reflex. Observation light and stroboscopic recordings were made as the patient produced sustained vowel /i/ at normal pitch and loudness, increased and decreased pitch and

loudness, and during laryngeal diadochokinesis. Findings from this recording revealed complete glottic closure at midline, with normal appearance of the larynx and vocal fold structures. There was a marked amount of supraglottic mucus pooling in the posterior glottis, with thick mucus strands between the vocal folds.

Movement patterns of the vocal folds were aberrant, due to consistent spasmlike interruptions of phonation. Excessive tension would build, recruiting ventricular folds and, other supraglottic structures until phonation ceased entirely, followed by a break in tension, with apparently normal abduction of the vocal folds for breath. These intermittent spasms precluded adequate opportunity to assess the vocal fold vibratory features during sustained phonation. There was no evidence of vocal fold stiffness or lesion. Perceptually, the patient's voice was low pitched, with increased tension and effort observed during voice onset and occasionally mid-utterance. Most connected speech was characterized by glottal fry, a low-pitched, low-airflow voicing pattern.

During the evaluation, the patient displayed a consistent nonproductive cough. She denied shortness of breath or other upper respiratory complaints. She also denied swallowing difficulty and problems with gastrointestinal reflux, a history of neck or throat trauma, or other problems with speech production. A formal motor speech examination was not conducted.

Trial therapy maneuvers were attempted to determine the variability and manipulability of the patient's current voice pattern. She did not use a diaphragmatic breathing pattern during general conversation. Under specific directions to increase pitch slightly and increase breath support, she was able to eliminate glottal fry and vocal spasm completely for a trial period lasting ~5 min. Based on her positive response to these gestures, and her own satisfaction with her ability to alter the sound of her voice, it was recommended that the patient receive a trial diagnostic course of voice therapy. The tentative diagnosis of early adductor-type SD was made. She was referred to a local university speech clinic in her community. There, she was seen for one session of diagnostic treatment. Due to minimal voice change, the patient was referred for Botox injections.

The patient's initial evaluation for Botox injections was conducted on September 22, 1992, ~4 months after the onset of speech symptoms. This
TABLE 1. *Symptom time course and examinations*

*July 27, 1992  Otolaryngology exam*
Two-month history of voice "quitting," difficulty voicing, and intermittent aphonia. Denies vocal hoarseness, but confirms intermittent shortness of breath and a persistent cough.

*Exam:* Normal appearance of anatomic structures, but significant gag reflex limits view. Speech described as "strained" and "effortful."

*Impression:* Possible spasmodic dysphonia and chronic cough. Recommend referral to speech-language pathologist for voice evaluation and laryngeal stroboscopic exam.

*July 29, 1992  Voice evaluation and laryngeal videostroboscopy*
Voice unchanged from previous exam both perceptually and by patient report. Denied swallowing difficulties. Consistent cough present.

*Exam:* Intermittent strain-strangled voice, persistent glottal fry, very effortful "bursts" of phonation. Stroboscopic exam revealed some mucus pooling in vallecula, medialization of ventricular and true vocal folds during phonatory "bursts," alternating with near-normal phonation. Speech intelligibility: 100%. (Motor speech examination not conducted.)

*Impression:* Possible early adductor-type spasmodic dysphonia. Recommend referral to home community (local university speech clinic) for trial diagnostic therapy.

*August 17, 1992  University speech pathology clinic visit (one session)*

*Exam:* Speech minimally responsive to behavioral intervention.

*Impression:* Adductor-type spasmodic dysphonia. Recommend referral to medical center for Botox treatment.

*September 22 and October 1, 1992  Medical center voice clinic evaluations*
These included oral mechanism and motor speech exams, acoustic and perceptual analyses of voice, head and neck exam, flexible videendoscopy and stroboscopy exams.

*Exam:* Tongue weakness. Hyperactive suck and gag reflexes. Slow, regular alternate and sequential motion rates. Strained, harsh vocal quality and intermittent hypernasality. Connected speech 95-100% intelligible with monopitch, monoloudness, reduced prosody, articulatory distortions, and imprecise consonants. Emotional lability (pseudobulbar affect). Clinical swallow study revealed good oral preparation with possible delays in oral transit and pharyngeal response. Coughing after swallowing solid texture. C/O nasal regurgitation of food. Flexible laryngoscopy and videostroboscopy showed the vocal cords to be mobile and to meet in midline. Increased supraglottic function and mucus pooling compared with exam of July 29.

*Impression:* Moderate spastic dysarthria (pseudobulbar palsy) and probable oral-pharyngeal dysphagia. Referral to neurology for differential diagnosis. Recommend videofluoroscopic modified barium swallow study.

*October 1, 1992  Neurology evaluation*
Deterioration of speech and swallowing (liquids more than solids); denies difficulty chewing.


*Impression:* Disease affecting upper and lower motor neurons (cranial, axial, and extremity musculature); probably degenerative and probable ALS. Recommended hospital admission for further neurologic workup, including electrical testing and swallow study. Removed from consideration for Botox treatment.

*October 2, 1992  Videofluoroscopic modified barium swallow study (VMBSS)*

*Impression:* Moderate oropharyngeal dysphagia without tracheal aspiration. Recommend diet modifications and management strategies for eating, for example, positioning and bolus size.

*October 22, 1992  Neuromuscular clinic visit*
Patient reports slow but steady progression of symptoms, especially speech and swallowing difficulties.

*Exam:* Results of neurologic examination unchanged. MRI of head revealed brain stem and hemispheric subcortical white matter changes suggestive of small vessel ischemic disease.

*Impression:* Motor neuron disease (ALS of bulbar onset).

*November 5, 1992  Neuromuscular clinic visit*
Patient experiencing continued decline.

*Exam:* 3+ reflexes with pathological spread. Results of physical examination unchanged.

*Impression:* ALS.

*December 9, 1992  VMBSS repeated*

*Impression:* Moderate to severe oropharyngeal dysphagia with tracheal aspiration. Speech essentially unintelligible.

*January 8–18, 1993  Alternative and augmentative communication options evaluated*
Family resistant to recommendations due to questions about long-term use.
team evaluation consisted of exams conducted by a speech pathologist, otolaryngologist, and a neurologist. By this time, the patient complained of slurred speech, muscle "twitches," episodes of generalized weakness associated with shortness of breath, and swallowing difficulties.

A thorough oral mechanism and motor speech examination revealed mild to moderate tongue weakness, distorted consonants, and imprecise articulation. Speech rate and diadochokinesis were slow. Speech prosody was reduced with monopitch and monoloudness. Vocal quality was strained-strangled and harsh. Phonation was marked by intermittent arrests, consistent with impressions of laryngeal spasms. Throughout the examination, episodes of spontaneous emotional lability were noted, suggestive of pseudobulbar affect. A hyperactive gag was also present. The patient's speech was intelligible overall. The patient complained of swallowing problems, as well. The speech diagnosis of spastic dysarthria (pseudobulbar palsy) of moderate severity was made. Botox injections were not recommended.

Evaluation of swallowing function included a videofluoroscopic modified barium swallow study (VMBSS). The findings revealed a moderate oropharyngeal dysphagia, characterized by delays in oral transit and pharyngeal responses, pooling of the bolus in the pharyngeal recesses, and laryngeal vestibule penetration. Residue in the oral cavity and pharynx after the swallow was also evident. There was no evidence of tracheal aspiration on this study. In a follow-up VMBSS conducted 2 months later, increased dysphagia severity was evidenced by prolonged oral transit and pharyngeal response times, increased pooling in the pharyngeal recesses, and consistent tracheal aspiration of thin liquids.

Otolaryngologic evaluation demonstrated a mild vocal tremor. A repeat videostroboscopy examination conducted using flexible fiberoptics revealed mobile vocal folds, with good midline adduction. Increased supraglottic hyperfunction was present compared with the previous videendostroboscopy conducted in July. There was increased pooling of secretions in the hypopharynx. Again, Botox injections were not recommended.

Neurologic evaluation detected an exaggerated jaw jerk and gag reflex, brisk appendicular reflexes, scattered fasciculations, and mild neck weakness; results of sensory exam were within normal limits. A Tensilon test for myasthenia gravis was negative. The neurologic impression was early ALS with a bulbar presentation. Other immunologic and toxic disorders were excluded by a battery of blood and urine studies. Cranial magnetic resonance imaging revealed evidence of small vessel ischemic disease. Initial electromyographic (EMG) testing was suggestive of but not conclusive for motor neuron disease, but ALS was ultimately confirmed on a follow-up EMG 2 months later. Following this examination, the patient was determined to no longer be a candidate for Botox injections.

Over the ensuing months, the patient became anarthric and required an augmentative communication system. She experienced progressive neurologic decline and died in April 1993.

**DISCUSSION**

Because SD represents a complex of symptoms rather than a specific diagnosis, determination of this disorder can be troublesome (27). Perceptual attributes of SD may be similar to those of other neurologic or functional voice disorders. The criteria for differential diagnosis of SD remain elusive. Neither the history of onset nor early signs and symptoms provide differential clarification. Both SD and functional or conversion voice disorders may arise following excessive or extreme vocal demands or following an incident of stress, trauma, or emotional upset (27).

Therefore, differential diagnosis of SD, especially from other neurologic disease, requires complete neurologic exam including a careful oral mechanism and motor speech exam. Speech symptoms may be the only outward sign of neurologic disease within the first months of onset (16). Findings on the voice evaluation, with or without videostroboscopy ex-
amination, may not always provide clear differential diagnosis (27–29). Occasionally, diagnostic treatment may provide the critical information leading to differential diagnosis (30). As seen in this case study, however, even an organic voice disorder secondary to progressive degenerative neurologic disease may show temporary improvement in response to behavioral manipulations. Thus, decisions about the presence and management of SD patients require careful examination of the individual’s speech and voice symptoms across time, attending to the consistency, severity, and resistance to change following traditional treatment methods.

This case report provides an example of the possibility for confusion in the diagnosis of neurologic disorders that affect speech and the laryngeal mechanism. This patient with ALS displayed initial signs and symptoms which were suggestive of isolated adductor-type SD. To our knowledge, this has not been reported previously. Since the patient did not undergo motor speech or neurologic evaluation until 4 months into her illness, it is not known whether she possessed signs of more widespread, subclinical neurologic dysfunction at the time of her initial presentation. However, her only symptomatic complaint concerned her voice.

While Botox treatment is considered standard care in the management of patients with SD, it may be contraindicated in patients with motor neuron diseases such as ALS (26). Botox therapy should be reserved for patients in whom a diagnosis has been established with reasonable certainty (28). SD constitutes a cluster of perceptual symptoms and not a definitive diagnosis, and numerous reports have considered the difficulty in isolating consistent diagnostic criteria (27–30). This case report further illustrates the potential for misdiagnosis of neurologic disorders that affect speech and laryngeal mechanisms. Therefore, it becomes crucial that unrelated neurologic signs separate from the focal dystonia are identified and addressed before consideration of Botox therapy.

This clinical example supports the need for accurate and valid neurologic diagnosis by a skilled interdisciplinary team prior to considering use of Botox as recommended by the National Institutes of Health Consensus Conference statement on the use of Botox treatment (31). In this case, the combined efforts of a speech–language pathologist, neurologist, and otolaryngologist contributed to the correct diagnosis and follow-up.

The speech–language pathology assessment included a thorough motor speech examination to assess the strength, rate, accuracy, range of motion, and symmetry of speech movements. Our experience has shown that a voice evaluation lacking the motor speech examination may fail to reveal the presence of a specific motor speech disorder (for example, dysarthria or apraxia of speech), implicating neurologic disease. A swallowing examination was warranted due to complaints of dysphagia, cough and evidence of excessive mucus pooling noted on laryngeal examination.

Following this case experience, we now recommend that patients with recent onset of voice symptoms observe a waiting period to monitor for signs or symptoms of speech or other neurologic disease before Botox injections. During this time, there should be no evidence of any progressive neurologic dysfunction or evolution of new speech symptoms. Finally, asking the patient, “How is the speech problem affecting your life?” may help clarify the range and severity of the disorder’s impact on individual communication abilities. Decisions about Botox injections are not straightforward. Identification of the best candidates for this procedure may be enhanced by recognition of the possibility for diagnostic confusion and ruling out other motor speech disorder or neurologic disease.

CONCLUSION

We present this unusual case of a patient with ALS whose initial signs and symptoms were consistent with adductor-type spasmodic dysphonia. Clearly, the recommendation for Botox treatment was inappropriate. This case illustrates the potential for confusion in differentially diagnosing SD from signs and symptoms of other progressive neurologic disease. Our experience with this patient has generated the development of strategies to ensure correct diagnosis of speech dystonias and proper selection of candidates for Botox treatment.

REFERENCES


