Spasmodic Dysphonia

**Diagnosis**

Spasmodic dysphonia is a neurological disorder caused by a dystonia (abnormal sustained muscle contraction) of the vocal cords. Depending on the vocal cord muscles that are affected, there are three types: adductor (involuntary vocal cord closing), abductor (involuntary vocal cord opening), or mixed. Adductor SD is more common and presents with a strained or strangled voice with irregular voice breaks. The severity of spasmodic dysphonia varies throughout the day and from day to day. Abductor dysphonia presents as excessive breathiness, pauses in the middle of the word, and difficulty in generating adequate volume of speech. Mixed dysphonia patients often have a chaotic combination of inappropriate adduction and abduction, resulting in halting speech and variable breathiness and tightness. Singing, shouting, and laughter may change the characteristics of all three types of dysphonia. In addition to their phonation difficulties, many patients with SD have an associated tremor of the voice, hands and head. This condition may be very debilitating especially for professionals such as teachers, trial attorneys or performers.

Little is currently known about the incidence and prevalence of SD. There may be between 10,000 to 30,000 affected individuals in the United States as reported by the National Spasmodic Dysphonia Association, although this may grossly underestimate the true prevalence. Spasmodic dysphonia, like most focal dystonias, tends to emerge gradually in young adulthood or midlife (peak time of onset age 30-50 years) and then reaches a plateau, when the severity of symptoms remains constant. The disorder predominantly affects women (2.5:1). Spontaneous
remission occurs in a small percentage of patients, but it is usually transient and the symptoms may recur after a few weeks or months. Repetitive vocal cord strain and laryngeal trauma may precipitate SD.

The diagnosis of SD is challenging. Individuals suffering from this disorder are often initially misdiagnosed as psychogenic (so-called muscle tension dysphonia). An accurate diagnosis is critical as misdiagnosis delays the time to initiation of effective therapy. Most commonly, SD is diagnosed based on the clinical evaluation (see characteristics of the different types of SD above) combined with direct visualization of the vocal cords (videolaryngoscopy).

Cause
The cause of SD remains unknown, but the disorder is classified as a form of focal dystonia (syndrome of abnormal sustained muscle contractions) involving the vocal muscles. This neurologic disorder is probably caused by impairment in the normal motor control mechanisms within the basal ganglia (deep brain structures which help regulate movement). A few cases with single mutations in THAP1, a gene involved in transcription regulation, suggest that there may be a weak genetic predisposition. SD may occur isolated or as part of a segmental dystonia, when it is associated with involuntary spasms of the face, jaw, and neck muscles (cranial or craniocervical dystonia). SD may also occur in patients with more generalized dystonia, also involving the trunk and limbs. This form of dystonia is often of genetic origin with several genes or gene markers already identified (see related information on dystonia).

Treatment
Minor adductor overactivity may improve with speech therapy, relaxation techniques or alteration of vocal pitch. Oral medications (such as trihexyphenidyl or benzodiazepines) may be effective in mild cases, but botulinum toxin injections are clearly the treatment of choice for most patients. Techniques of injecting botulinum toxin into the vocal cords include percutaneously (through the skin) unilaterally or
bilaterally, or through a transnasal fiberoptic approach. The effect may last up to three months and most patients are able to sustain a benefit with 2-4 injections per year. Voice therapy and voice rest following the injections of botulinum toxin may prolong the benefit of botulinum toxin. Treatment with botulinum toxin injections is proven to be effective in treating SD and it has markedly changed the prognosis of the disease. Finally, surgical treatments, such as neuromodulation (direct electrical stimulation of the muscle) and muscle surgery, has been attempted with mixed results.

Selected References
Jankovic J, Schwartz K, Donovan DT. Botulinum toxin treatment of cranial-cervical

Support Organizations

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