

NIDCD Fact Sheet

Spasmodic Dysphonia

U.S. DEPARTMENT OF HEALTH & HUMAN SERVICES · NATIONAL INSTITUTES OF HEALTH · NATIONAL INSTITUTE ON DEAFNESS AND OTHER COMMUNICATION DISORDERS

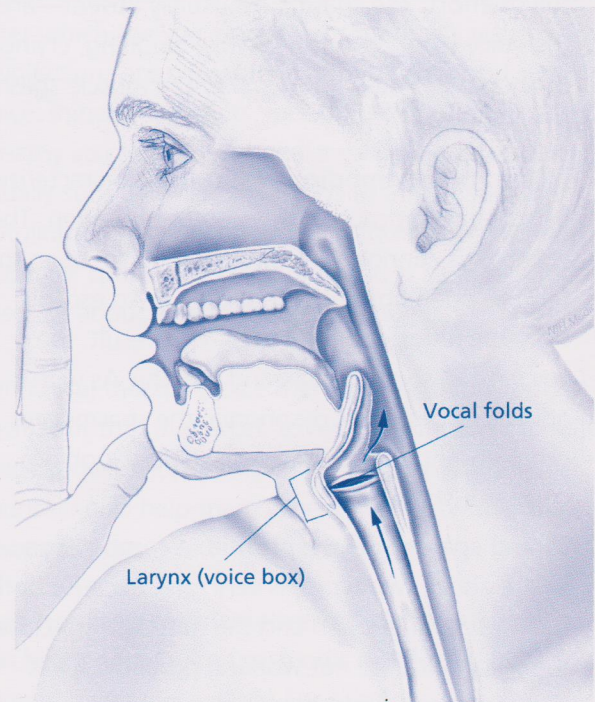
What is spasmodic dysphonia?

Spasmodic dysphonia is a neurological disorder affecting the voice muscles in the larynx, or voice box. When we speak, air from the lungs is pushed between two elastic structures—called vocal folds or vocal cords—with sufficient pressure to cause them to vibrate, producing voice (see figure). In spasmodic dysphonia, the muscles inside the vocal folds experience sudden involuntary movements—called spasms—which interfere with the ability of the folds to vibrate and produce voice.

Spasmodic dysphonia causes voice breaks and can give the voice a tight, strained quality. People with spasmodic dysphonia may have occasional breaks in their voice that occur once every few sentences. Usually, however, the disorder is more severe and spasms may occur on every other word, making a person's speech very difficult for others to understand. At first, symptoms may be mild and occur only occasionally, but they may worsen and become more frequent over time. Spasmodic dysphonia is a chronic condition that continues throughout a person's life.

Spasmodic dysphonia can affect anyone. It is a rare disorder, occurring in roughly one to four people per 100,000 people. The first signs of spasmodic dysphonia are found most often in people between 30 and 50 years of age. It affects women more than men.

Parts of the throat involved in spasmodic dysphonia



What are the types of spasmodic dysphonia?

- **Adductor spasmodic dysphonia** is the most common form of spasmodic dysphonia. It is characterized by spasms that cause the vocal folds to slam together and stiffen. These spasms make it difficult for the vocal folds to vibrate and produce sounds. Words are often cut off or are difficult to start because of muscle spasms. Therefore, speech may be choppy. The voice of



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someone with adductor spasmodic dysphonia is commonly described as strained or strangled and full of effort. The spasms are usually absent—and the voice sounds normal—while laughing, crying, or shouting. Stress often makes the muscle spasms more severe.

- **Abductor spasmodic dysphonia** is characterized by spasms that cause the vocal folds to open. The vocal folds cannot vibrate when they are open too far. The open position also allows air to escape from the lungs during speech. As a result, the voice often sounds weak and breathy. As with adductor spasmodic dysphonia, the spasms are often absent during activities such as laughing, crying, or shouting.
- **Mixed spasmodic dysphonia**, a combination of the above two types, is very rare. Because both the muscles that open and the muscles that close the vocal folds are not working properly, it has features of both adductor and abductor spasmodic dysphonia.

What causes spasmodic dysphonia?

The cause of spasmodic dysphonia is unknown. Because the voice can sound normal or near normal at times, spasmodic dysphonia was once thought to be psychogenic, or originating in a person's mind, rather than from a physical cause. In rare cases, psychogenic forms of spasmodic dysphonia do exist; however, in most instances, the muscle spasms are caused by abnormalities in the central nervous system (the brain).

A disorder that involves involuntary muscle contractions is also called a dystonia; therefore, another name for spasmodic dysphonia is laryngeal dystonia. Spasmodic dysphonia is considered a form

of focal dystonia, a neurological disorder that affects muscle tone in one part of the body. Writer's cramp is another type of focal dystonia. Other dystonias can affect multiple regions of the body or the entire body.

Spasmodic dysphonia may co-occur with other dystonias that cause involuntary and repetitive movement of such muscles as the eyes; face, body, arms, and legs; jaws, lips, and tongue; or neck.

Spasmodic dysphonia is thought to be caused by abnormal functioning in an area of the brain called the basal ganglia. The basal ganglia consist of several clusters of nerve cells deep inside the brain. They help coordinate movements of the muscles throughout the body. Recent research has found abnormalities in other regions of the brain, including the brainstem, the stalk-like part of the brain that connects to the spinal cord.

Symptoms of spasmodic dysphonia generally develop gradually and with no obvious explanation. Some people with spasmodic dysphonia also have vocal tremor, a shaking of the larynx and vocal folds that causes the voice to shake. Although the risk factors for spasmodic dysphonia have not been identified, the voice symptoms can begin following an upper respiratory infection, injury to the larynx, voice overuse, or stress.

In some cases, spasmodic dysphonia may run in families. Although 14 genes have been recently associated with various dystonias, only mutations in one gene, named THAP1, have been associated with forms of whole body dystonia that begin in childhood and that appear with spasmodic dysphonia. This genetic defect does not seem to be associated with the more usual form of focal spasmodic dysphonia that begins in adults, however.

How is spasmodic dysphonia diagnosed?

Diagnosis of spasmodic dysphonia is sometimes difficult because individuals with spasmodic dysphonia often have symptoms similar to other voice disorders. The diagnosis of spasmodic dysphonia usually is made following careful examination by a team that includes an otolaryngologist, a doctor who specializes in diseases of the ear, nose, throat, head, and neck; a speech-language pathologist, a health professional trained to evaluate and treat speech, language, and voice disorders; and a neurologist, a doctor who specializes in nervous system disorders.

The otolaryngologist examines the vocal folds for other possible causes of the voice disorder. A small lighted tube is passed through the nose and into the back of the throat—a procedure called fiberoptic nasolaryngoscopy—allowing the otolaryngologist to evaluate vocal fold structure and movement during speech and other activities. The speech-language pathologist evaluates the types of voice symptoms to see if they are characteristic of spasmodic dysphonia or other voice disorders and voice quality. The neurologist evaluates the patient for signs of other muscle movement disorders.

What treatment is available for spasmodic dysphonia?

There is currently no cure for spasmodic dysphonia; therefore, treatment can only help reduce its symptoms. The most common treatment for spasmodic dysphonia is the injection of very small amounts of botulinum toxin directly into the affected muscles of the larynx. Botulinum toxin is produced by *Clostridium botulinum*, the same bacterium that occurs in improperly canned foods and honey. The toxin

weakens muscles by blocking the nerve impulse to the muscle. Botulinum toxin injections generally improve the voice for a period of three to four months, after which the voice symptoms gradually return. Reinjections are necessary to maintain a good speaking voice. Initial side effects, including a temporary weak, breathy voice and occasional swallowing difficulties, usually subside after a few days to a few weeks. Botulinum toxin will relieve symptoms of most cases of adductor spasmodic dysphonia and is helpful in many cases of abductor spasmodic dysphonia.

Behavioral therapy (voice therapy) is another form of treatment that may work to reduce symptoms in mild cases. Other people may benefit from psychological counseling to help them accept and live with their voice problem.

In some cases, augmentative and alternative devices can help people with spasmodic dysphonia to communicate more easily. For example, some devices can help amplify a person's voice in person or over the phone. Special software can be added to a computer or handheld device such as a personal digital assistant (PDA) or cell phone to translate text into synthetic speech.

When more conventional measures have failed, surgery on the larynx may be performed. Long-term benefits and effects of this procedure are unknown.

What research is being done on spasmodic dysphonia?

Scientists and clinicians are working to understand the causes of spasmodic dysphonia as well as the underlying manner in which the disorder develops. Brain imaging studies enable researchers to better identify possible differences between people with

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spasmodic dysphonia and healthy volunteers as well as to observe how spasmodic dysphonia compares with other dystonias.

Although an animal model is not currently available for the study of spasmodic dysphonia, several have been developed for the study of other forms of dystonia. If a suitable animal model is identified, researchers will have a new tool to explore the underlying cause of spasmodic dysphonia and to test new treatments.

Genetic studies of spasmodic dysphonia are currently ongoing. However, the disorder is rare, and it is difficult for researchers to locate large families with a history of spasmodic dysphonia alone. However, new technologies that rapidly pinpoint small differences in a person's DNA, called single nucleotide polymorphisms, or SNPs, are becoming available that will enable clinician-scientists to study unrelated patients with spasmodic dysphonia. This development will make it easier to determine whether a gene is involved and possible environmental causes of the disorder.

At a 2005 workshop co-sponsored by the National Institute on Deafness and Other Communication Disorders (NIDCD), participants emphasized the need for a standardized three-step procedure in diagnosing spasmodic dysphonia, including: a questionnaire on which a patient reports a possible case of spasmodic dysphonia, a clinical examination to identify a probable case of spasmodic dysphonia, and a fiberoptic nasolaryngoscopy to confirm a case of spasmodic dysphonia. In 2009, a five-year multicenter trial was funded by the National Institutes of Health Office of Rare Diseases Research and the National Institute of Neurological Disorders and Stroke that will determine the accuracy and reliability of this method for diagnosing spasmodic dysphonia.

Where can I get more information?

The NIDCD maintains a directory of organizations that provide information on the normal and disordered processes of hearing, balance, smell, taste, voice, speech, and language. Please see the list of organizations at <http://www.nidcd.nih.gov/directory>.

Use the following keywords to help you search for organizations that can answer questions and provide printed or electronic information on spasmodic dysphonia:

- Spasmodic dysphonia
- Speech-language pathology
- Augmentative and alternative communication

For more information, additional addresses and phone numbers, or a printed list of organizations, contact:

NIDCD Information Clearinghouse

1 Communication Avenue
Bethesda, MD 20892-3456
Toll-free Voice: (800) 241-1044
Toll-free TTY: (800) 241-1055
Fax: (301) 770-8977
E-mail: nidcdinfo@nidcd.nih.gov

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1 Communication Avenue
Bethesda, MD 20892-3456
Toll-free Voice: (800) 241-1044
Toll-free TTY: (800) 241-1055
Fax: (301) 770-8977
E-mail: nidcdinfo@nidcd.nih.gov
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