Task-specific focal dystonia

Task-specific focal dystonia is a movement disorder that interferes with the performance of particular tasks, such as writing, playing a musical instrument, or participating in a sport. Dystonias are a group of movement problems characterized by involuntary, sustained muscle contractions, tremors, and other uncontrolled movements. The term "focal" refers to a type of dystonia that affects a single part of the body, such as the hand or jaw.

Researchers have described several forms of task-specific focal dystonia. The most common is writer's cramp, in which muscle cramps or spasms in the hand, wrist, or forearm interfere with holding a pen or pencil. Writer's cramp begins in the hand used for writing (the dominant hand) and is usually limited to that task, but with time it can spread to the other hand and affect other fine-motor activities such as shaving or typing.

Musician's dystonia is a form of task-specific focal dystonia characterized by muscle cramps and spasms that occur while playing a musical instrument. This condition can affect amateur or professional musicians, and the location of the dystonia depends on the instrument. Some musicians (such as piano, guitar, and violin players) develop focal hand dystonia, which causes loss of fine-motor control in the hand and wrist muscles. This condition reduces finger coordination, speed, and endurance while playing. Musicians who play woodwind or brass instruments can develop what is known as embouchure dystonia. This condition causes muscle cramps or spasms involving the lips, tongue, or jaw, which prevents normal positioning of the mouth around the instrument's mouthpiece. Musician's dystonia often occurs only when playing a particular instrument. However, over time focal hand dystonia may impair other activities, and embouchure dystonia can worsen to affect eating and speech.

Task-specific focal dystonia can affect people who play sports and engage in other occupations involving repetitive, highly practiced movements. For example, some golfers experience involuntary jerking of the wrists during putting, a condition known informally as "the yips." Cramps and spasms of the hand and arm muscles can also affect tennis players, billiard players, dart throwers, and other athletes. Additionally, task-specific dystonia has been reported in tailors, shoemakers, hair stylists, and people who frequently type or use a computer mouse.

The abnormal movements associated with task-specific focal dystonia are usually painless, although they can cause anxiety when they interfere with musical performance and other activities. Severe cases can cause professional disability.
Frequency

Task-specific focal dystonia affects an estimated 7 to 69 per million people in the general population. Musician's dystonia that is severe enough to impact performance occurs in about 1 percent of musicians.

Causes

The causes of task-specific focal dystonia are unknown, although the disorder likely results from a combination of genetic and environmental factors. Certain genetic changes probably increase the likelihood of developing this condition, and environmental factors may trigger the onset of symptoms in people who are at risk. It is possible that the different forms of task-specific focal dystonia have different underlying causes.

Having a family history of dystonia, particularly focal dystonia, is one of the only established risk factors for task-specific focal dystonia. Studies suggest that previous injury, changes in practice routine, and exposure to anti-psychotic drugs (which can cause other types of dystonia) are not major risk factors. Nor does the condition appear to be a form of performance anxiety. Task-specific focal dystonia may be associated with dysfunction in areas of the brain that regulate movement. In particular, researchers have found that at least some cases of the condition are related to malfunction of the basal ganglia, which are structures deep within the brain that help start and control movement.

Although genetic factors are almost certainly involved in task-specific focal dystonia, no genes have been clearly associated with the condition. Researchers have looked for mutations in several genes known to be involved in other forms of dystonia, but these genetic changes do not appear to be a major cause of task-specific focal dystonia. Researchers are working to determine which genetic factors are related to this disorder.

Inheritance Pattern

Most cases of task-specific focal dystonia are sporadic, which means they occur in people with no history of the condition in their family. However, at least 10 percent of affected individuals have a family history of focal dystonia. (For example, writer's cramp and musician's dystonia have been reported to occur in the same family.) The dystonia often appears to have an autosomal dominant pattern of inheritance, based on the observation that some affected people have a parent with the condition.

Other Names for This Condition

- focal hand dystonia
- focal task-specific dystonia
- FTSD
- occupational cramp
• occupational dystonia
• task-specific dystonia

Diagnosis & Management

Research Studies from ClinicalTrials.gov
• ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22task-specific+focal+dystonia%22+OR+%22embouchure+dystonia%22+OR+%22focal+hand+dystonia%22+OR+%22musician%22+OR+%22task-specific+dystonia%22

Other Diagnosis and Management Resources
• Dystonia Medical Research Foundation: Symptoms and Classifications
  https://dystonia-foundation.org/what-is-dystonia/symptoms-classifications/
• Dystonia Medical Research Foundation: Treatment
  https://dystonia-foundation.org/living-dystonia/treatment/
• GeneReview: Hereditary Dystonia Overview
  https://www.ncbi.nlm.nih.gov/books/NBK1155

Additional Information & Resources

Health Information from MedlinePlus
• Health Topic: Dystonia
  https://medlineplus.gov/dystonia.html

Genetic and Rare Diseases Information Center
• Focal dystonia
  https://rarediseases.info.nih.gov/diseases/6458/focal-dystonia

Additional NIH Resources
• National Institute of Neurological Disorders and Stroke: Dystonias
  https://www.ninds.nih.gov/Disorders/All-Disorders/Dystonias-Information-Page

Educational Resources
• Dystonia Medical Research Foundation: Hand Dystonia
  https://dystonia-foundation.org/what-is-dystonia/types-dystonia/hand-dystonia/
• Dystonia Medical Research Foundation: Musician's Dystonias
  https://dystonia-foundation.org/what-is-dystonia/types-dystonia/musicians/
• MalaCards: focal hand dystonia
  https://www.malacards.org/card/focal_hand_dystonia
• Merck Manual Consumer Version: Dystonia
• Orphanet: Focal, segmental or multifocal dystonia
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=1866

Patient Support and Advocacy Resources
• National Organization for Rare Disorders (NORD): Dystonia
  https://rarediseases.org/rare-diseases/dystonia/
• Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/dystonia.html

Clinical Information from GeneReviews
• Hereditary Dystonia Overview
  https://www.ncbi.nlm.nih.gov/books/NBK1155

Scientific Articles on PubMed
• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Dystonic+Disorders%5BMAJR%5D%29+AND+%28%28task-specific+focal+dystonia%5BTIAB%5D%29+OR+%28embouchure+dystonia%5BTIAB%5D%29+OR+%28focal+hand+dystonia%5BTIAB%5D%29+OR+%28musician's+dystonia%5BTIAB%5D%29+OR+%28writer's+cramp%5BTIAB%5D%29+OR+%28yips%5BTIAB%5D%29+OR+%28task-specific+dystonia%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmhm%5D+AND+%22last+1080+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM
• DYSTONIA, FOCAL, TASK-SPECIFIC
  http://omim.org/entry/611284

Sources for This Summary
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16859794

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18785647

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16924027

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19349605
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2677486/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22569836

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18990127
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2652841/

Reprinted from Genetics Home Reference:

Reviewed: December 2012
Published: March 3, 2020

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services