

Researchers ID gene behind primary cervical dystonia, a neck-twisting disorder

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Researchers have identified a gene that causes adult-onset primary cervical dystonia, an often-painful condition in which patients' necks twist involuntarily. The discovery by a team from the Jacksonville, Fla., campus of Mayo Clinic and the University of Tennessee Health Sciences Center sheds light on a movement disorder that physicians previously could seldom explain. Their research appears in the [*Annals of Neurology*](#).

In 1990, a man with a crooked neck came to see Ryan Uitti, M.D., a [neurologist](#) then at Mayo Clinic in Rochester, Minn. Dr. Uitti knew about adult-onset primary cervical dystonia, which results in involuntary twisting of the neck to the left or right, backward or forward. Most people who have it suffer from [muscle pain](#) and abnormalities in head position. Some don't think it is all that unusual and may not seek medical help, Dr. Uitti says.

"They think they slept wrong at some point, or, because the twisting might straighten out with another maneuver, such as walking backwards, they might actually be accused of being a little crazy," Dr. Uitti says.

Dr. Uitti had been taught that there is usually no explanation for the disorder, when it shows up in adulthood. But working with a team of neurologists who have found the [genetic causes](#) of other rare conditions, Dr. Uitti began to investigate.

His patient first said no one in his family had the same problem. Dr.

Uitti soon found out that his patient had an identical twin whose head was also twisted, but in the opposite direction. And when Dr. Uitti went to visit their sister, she had the same kind of dystonia. Eventually, seven people in the extended family were diagnosed with this mysterious condition. "I heard a lot of explanations by the family for it, such as that one member got hit by lightning," he says.

In 1994, when Dr. Uitti relocated to Mayo Clinic's campus in Florida, he continued his research into the [genetic basis](#) of this [neurological disorder](#), which is also known as spasmodic torticollis. He used the research infrastructure provided by the Morris K. Udall Center of Excellence for Parkinson's Disease Research, funded by the National Institutes of Health, and collaborated with a Mayo team that included Parkinson's gene hunter Zbigniew Wszolek, M.D.

They and researchers from the University of Tennessee Health Sciences Center, with Mark LeDoux, M.D., Ph.D., as the lead neurogeneticist, are reporting the first gene that causes primary cervical dystonia. Their finding is based on genetic material donated by this family — the first extended "cohort" identified — and others with the condition.

The researchers found a mutation in the CIZ1 gene that makes a protein expressed in certain nerve cells in the brain and which seems to be involved in cell cycle activities. The actual mechanism has not yet been identified, Dr. Uitti says: "It is interesting because the brain tissue of folks with this disorder looks absolutely normal."

Textbooks say that adult-onset primary cervical dystonia affects about 30 of every 100,000 people, but Dr. Uitti believes it is not that rare. "Cervical dystonia is the most common focal, fixed, adult-onset dystonia. But I suspect most people don't seek medical attention for a little bit of neck twisting or tilting," he says.

There are several treatments. Most common is the use of botulinum toxin injections to incapacitate the nerve in the affected muscle, eliminating chronic pain and muscle pulling/contraction.

Dr. Uitti believes CIZ1 is one genetic cause of this disorder, and that other genes will be found. But he is elated that at least one explanation for it has been found. "While it took over 20 years, at least it took place in my lifetime," he says. "This discovery reflects the first genetic cause for this condition ever identified."

Provided by Mayo Clinic

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