

Team aids discovery of first dystonia gene found in African-Americans

March 7 2013

A pair of studies tells the tale of how a neuroscientist at Mayo Clinic in Florida helped to discover the first African-American family to have inherited the rare movement disorder dystonia, which causes repetitive muscle contractions and twisting, resulting in abnormal posture. The research may improve diagnosis of this neurological condition in a population not known to suffer from it.

In the first study, published in 2011 in the journal *Parkinsonism and Related Disorders*, Mayo Clinic's Zbigniew Wszolek, M.D., and a team of [neuroscientists](#) from other institutions in the U.S. described three generations of an African-American family in Georgia who had [dystonia](#). The team excluded mutations in genes previously associated with dystonia. The study was the first description of an African-American family with late-onset primary dystonia.

In the second study, published online this week in [Human Molecular Genetics](#), Dr. Wszolek was part of an international team of researchers led by Mark LeDoux, M.D., Ph.D., a neurologist and neurogeneticist from the University of Tennessee Health Science Center in Memphis. The investigators identified the specific [genetic abnormality](#) seen in the African-American family and in several other white families. In the African-American family, the mutation produced a protein in which one amino acid was substituted for another.

While this isn't the only gene anomaly linked to dystonia, it is the first found in an African-American family. All other genes found to be linked

to this disorder were discovered in families of other [ethnic origins](#).

The findings may improve diagnosis and treatment of dystonia in African-Americans, says Dr. Wszolek, who has been a driving force behind international research efforts to uncover genes that play a role in neurological disorders.

"This is a very hard disease to diagnose," Dr. Wszolek says. "The patient who came to see me had been to many other physicians who did not help him. His disorder, which resulted in contortions of his upper body, led to the loss of his job, and it meant that he couldn't drive because he was not able to keep his head straight to see in front of him," Dr. Wszolek says.

Through work with the patient, Dr. Wszolek came to know the rest of his family, which allowed him to do the genetic work that was critical to discovery of the mutated gene.

When dystonia was finally diagnosed in the patient, Dr. Wszolek and the Mayo Clinic health care team treated him with botulinum toxin (also known as Botox) to reduce muscle spasms and improve his head position.

"Our study now tells physicians that dystonia can occur in African-American patients, and that a blood sample that reveals a genetic abnormality can help diagnose the disease," Dr. Wszolek says. "There is no cure, but treatment can really help. Now the patient has less pain, he can drive and he works part time."

Provided by Mayo Clinic

Citation: Team aids discovery of first dystonia gene found in African-Americans (2013, March 7) retrieved 7 May 2024 from

<https://medicalxpress.com/news/2013-03-team-aids-discovery-dystonia-gene.html>

This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.