

Vascular disorder in the brain often misdiagnosed as multiple sclerosis

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A devastating vascular disorder of the brain called CADASIL, which strikes young adults and leads to early dementia, often is misdiagnosed as multiple sclerosis, Loyola University Health System researchers report.

CADASIL occurs when a thickening of [blood vessel walls](#) blocks [blood flow](#) in the brain. Migraine headaches are an early symptom of this condition, which progresses to strokes and mini-strokes, depression, apathy, motor disability and executive dysfunction (an inability to plan and organize everyday activities.) The final symptom is [dementia](#).

CADASIL is caused by mutations of a single gene called NOTCH 3. If an individual carries the mutated gene, he or she inevitably will develop the disease, and there's a 50 percent chance that each of the individual's children will inherit the mutation and the disease.

Researchers conducted an exhaustive series of genetic, physical and psychological tests and exams on 11 CADASIL patients. "We found a delay in the detection of this pathology and previous diagnostic errors in some patients and their relatives," researchers wrote. "[Multiple sclerosis](#) was the most frequent misdiagnosis."

The study is published in *Revista de Neurologia* ([Journal of Neurology](#)) in Spain.

The study was a subset of a larger study to determine whether the

Alzheimer's disease drug donepezil (trade name, Aricept®) can help in CADASIL patients. This larger study found there generally was no benefit to the drug.

CADASIL stands for cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy. It was realistically portrayed by one of the main characters in the critically acclaimed 2004 movie "The Sea Inside."

"It is a terrible disease that runs in families, and, unfortunately, we as yet don't have effective treatments," said Dr. José Biller, senior author of the study and chairman of the Department of Neurology of Loyola University Chicago Stritch School of Medicine.

There are several reasons why CADASIL is misdiagnosed as MS. Both diseases tend to strike young adults. There are similarities in brain MRIs, and both diseases can cause focal neurologic signs and symptoms.

While there currently are no effective treatments, researchers are making significant progress in better understanding CADASIL, Biller said. "The field is exploding, and there is hope down the road that there will be new treatments for these patients," Biller said.

Provided by Loyola University

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