

New Alzheimer's Gene Identified



By Jenifer Goodwin

HealthDay Reporter

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WEDNESDAY, April 14 (HealthDay News) -- Researchers have pinpointed a gene variant that nearly doubles the risk of developing late-onset Alzheimer's disease, a new study says.

A U.S. research team examined gene variations across the human genome, or full DNA sequence, of 2,269 people with late-onset Alzheimer's and 3,107 people without the disease. This research -- known as a genome-wide association study -- looks throughout the entire genome for small differences, or variants, in long stretches of DNA that are more prevalent in those with a particular disease or condition.

About 9 percent of those with late-onset Alzheimer's had a specific variation in the gene MTHFD1L on chromosome 6, according to the study. Only about 5 percent of those who did not have Alzheimer's had the variant.

Late-onset Alzheimer's, which affects those 60 and up, is the most common form of the brain disorder.

With the number of people with Alzheimer's expected to nearly double from 18 million worldwide to 34 million by 2025, according to the World Health Organization, researchers have been hunting for genes that play a role in Alzheimer's disease. The hope is that understanding the function of the genes could help in developing better treatments, which are sorely lacking.

So far, the primary known genetic contributor to late-onset Alzheimer's is a variant of the gene APOE on chromosome 19. The Alzheimer's-linked APOE variant occurs in about 40 percent of people who develop late-onset Alzheimer's, while about 25 to 30 percent of the general population has it, according to the National Institute on Aging.

The influence of the MTHFD1L variation is not as strong as APOE, and the variation itself is not as common in the population, said principal investigator Margaret Pericak-Vance, director of the University of Miami Miller School of Medicine's John P. Hussman Institute for Human Genomics.

But what makes the current finding so interesting is how it might connect to previous research about MTHFD1L. The gene is involved with the metabolism of folate, which in turn can influence levels of homocysteine.

Elevated homocysteine, which is often tied to folic acid deficiencies in the diet, have been shown to

be a risk factor for coronary artery disease and late-onset Alzheimer's.

Previous genome-wide studies have also implicated another variation in MTHFD1L in coronary artery disease.

Taken together, the research hints at ways in which the gene variant might be associated with changes in blood vessel function in the brain that impact Alzheimer's, Pericak-Vance said.

"The key reason people are excited about this is that it melds the genetics and the biology," Pericak-Vance said. "Maybe we can put the biology together with genetics and come up with some way to either treat it or approach it."

While lots of genetic variants have been singled out as possible contributors to Alzheimer's, the findings often can't be replicated or repeated, leaving researchers unsure if the results are a coincidence or actually important, said Dr. Ron Peterson, director of the Mayo Alzheimer's Disease Research Center in Rochester, Minn.

"The strength of his study is it includes a large number of subjects, they looked at a large number of [DNA sequence variations], and they replicated previously reported findings, which gives you confidence that they are correct," Peterson said.

The study is slated to be presented April 14 at the American Academy of Neurology's meeting in Toronto.

More information

The [National Institute on Aging](#) has more on the genetics of Alzheimer's.