

New gene associated with increased risk of Alzheimer's disease

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Researchers have identified a gene that appears to increase a person's risk of developing late-onset Alzheimer's disease, the most common type of Alzheimer's disease. The research will be presented as part of the late-breaking science program at the American Academy of Neurology's 62nd Annual Meeting in Toronto, April 10 - 17, 2010. The gene, abbreviated MTHFD1L, is located on chromosome six.

"Only recently have common variants in [genes](#) other than APOE been convincingly shown to be associated with a person's risk of developing late-onset [Alzheimer's disease](#)," said senior author Margaret Pericak-Vance, PhD, the principal investigator of the study and Director of the University of Miami Miller School of Medicine's John P. Hussman Institute for Human Genomics in Miami, Florida.

Researchers looked at [gene variation](#) throughout the human genomes of 2,269 people with late-onset Alzheimer's disease and 3,107 people without the disease through what's known as a genome-wide association study. Such studies involve looking at long stretches of DNA to identify small differences in the [genetic sequence](#) between people with and without Alzheimer's disease.

The study found that individuals with a particular variation in the gene MTHFD1L may be almost twice as likely to develop Alzheimer's disease as those people without the variation. "We are hopeful our identification of MTHFD1L as a risk gene for Alzheimer's disease will help us to better understand how this disease develops and potentially serve as a

marker for people who may be at increased risk," said Adam Naj, PhD, with the University of Miami Miller School of Medicine's John P. Hussman Institute for Human Genomics in Miami and the first author of the abstract reporting the discovery.

"Identifying this gene is important because the gene is known to be involved in influencing the body's levels of [homocysteine](#), and high levels of homocysteine are strong risk factor for late-onset Alzheimer's disease," said Pericak-Vance. "In addition, variations of the MTHFD1L gene have been reported to possibly increase the risk of coronary artery disease. Since the function of blood vessels in the brain may affect Alzheimer's disease, this finding may also help us understand how homocysteine levels and blood vessel function in the brain affect Alzheimer's disease."

The World Health Organization estimates that there are currently 18 million people worldwide with Alzheimer's disease, and this figure is projected to nearly double to 34 million by 2025. There are currently at least five million Americans living with Alzheimer's disease today.

Provided by American Academy of Neurology

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