HealthHub

Genetic Culprit Identified in Progression of Alzheimer's

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Does genetics play a role in the varying rates of cognitive decline among Alzheimer's patients?

Thanks to some intercontinental teamwork, researchers have identified a gene that may help explain why certain Alzheimer's disease patients experience a more rapid decline in cognitive (thinking) abilities.

Brigham and Women's Hospital (BWH) medical geneticist Dr. Robert C. Green first recognized that there may be a genetic explanation for why cognitive decline rates vary widely among Alzheimer's disease patients after analyzing data from a large treatment trial. Even after screening out individuals with vascular disease and other medical conditions known to influence cognition, Dr. Green found that there was still significant variability in the rate of decline among remaining participants.

In an effort to find the gene responsible for this disparity, researchers scanned millions of markers (identifiable genetic traits) across complete sets of DNA in a group of Alzheimer's patients experiencing both slow and fast rates of cognitive decline. This required DNA samples from a large sample of Alzheimer's patients and periodic assessments of each individual's cognitive abilities as the disease progressed. The culprit was eventually determined to be the spondin 1 gene (*SPON1*).

"The discovery of *SPON1* as a gene that influences rate of progression in Alzheimer's disease could go a long way toward improving our understanding of why people decline rapidly in Alzheimer's, but the work is far from complete," says Dr. Green. "As with all genome-wide association studies, replication of the initial findings with larger numbers is very important. We are continuing to add investigators and datasets with the goal of having more than 10,000 individuals to analyze over the coming year."

The collaboration of a vast number of genetic researchers from throughout the US and Europe makes such significant, large-scale studies possible. In this case, Dr. Green benefits from access to numerous datasets from the Genetic Architecture of Rate of Alzheimer's Decline Consortium (GENAROAD), an international partnership of investigators that collects, analyzes, and shares data in an effort to "accelerate the discovery of biological pathways involved in Alzheimer's disease."

"The future of genetics research will require collaboration with large interdisciplinary teams and the GENAROAD Consortium is an excellent example of that," says Dr. Green, senior author on these studies and a leader of the Consortium. "We are leveraging and combining large amounts of data that have already been collected from many studies to make new discoveries that we hope will identify previously unsuspected targets for prevention and treatment."

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