

GENETICS RELATED ARTICLE FROM POPULAR PRESS

In the recent article titled 'Mutation Helps Even Carriers of Alzheimer's Gene Avoid Alzheimer's,' published on US News, the author discusses a study shedding light on a rare genetic mutation's protective effects against Alzheimer's disease. The study reveals that individuals carrying the APOE4 gene variant, a known risk factor for Alzheimer's, may still avoid developing the disease due to the presence of this mutation. The news article explores the role of genetics, particularly a rare mutation, in influencing Alzheimer's disease (AD) risk. It discusses how this mutation provides protection against AD, even for carriers of the APOE4 gene variant, which is a known risk factor for the disease. By highlighting the significance of genetic factors in determining susceptibility to AD and the potential for certain mutations to mitigate its effects, the article underscores the importance of understanding genetics in both the development and prevention of neurodegenerative diseases like AD.

The article discusses a study revealing a rare genetic mutation that protects carriers of the APOE4 gene variant from developing Alzheimer's disease. While the APOE4 gene is a known risk factor for Alzheimer's, researchers found that individuals with both the APOE4 gene and the protective mutation were significantly less likely to develop the disease. This discovery challenges previous assumptions about the inevitability of Alzheimer's for APOE4 carriers and opens new possibilities for understanding the disease's mechanisms and potential treatments. The study's findings offer hope for personalized approaches to Alzheimer's prevention and highlight the importance of further research into genetic factors influencing disease risk.

The scientific study, “The APOE-R136S mutation protects against APOE4-driven Tau pathology, neurodegeneration and neuroinflammation”, investigates the protective effects of a rare APOE variant, APOE3-R136S, against Alzheimer's disease (AD) pathologies driven by APOE4, the strongest genetic risk factor for late-onset AD. Using mouse models and human induced pluripotent stem cell-derived neurons, the researchers demonstrate that the homozygous R136S mutation rescues APOE4-driven Tau pathology, neurodegeneration, and neuroinflammation, while the heterozygous mutation partially protects against these effects. Single-nucleus RNA sequencing reveals that the R136S mutation increases disease-protective and decreases disease-associated cell populations in a gene dose-dependent manner. These findings suggest that the APOE-R136S mutation could serve as a target for therapeutic development against APOE4-related AD.

Both articles discussed in this summary focus on identifying protective factors against Alzheimer's disease (AD). While the previous news article highlights a rare mutation (APOE3-R136S) that protects carriers from developing early-onset AD, the scientific study delves deeper into investigating whether this mutation can also protect against AD pathologies driven by the APOE4 gene variant, which is a major risk factor for late-onset AD. The study's findings demonstrate that the APOE3-R136S mutation indeed confers protection against APOE4-driven AD pathologies, providing potential targets for therapeutic development against AD. Therefore, both articles align in their exploration of genetic factors that influence AD risk and potential avenues for intervention.

Based on the findings in the scientific review article, it is evident that the APOE-R136S mutation indeed confers protection against APOE4-driven Tau pathology, neurodegeneration, and neuroinflammation. The study's results validate the accuracy of the news article's claims regarding the protective role of the R136S mutation against AD, further emphasizing the significance of genetic factors in influencing AD risk and potential therapeutic targets.

CITATION

Thompson, D. (2024, March 29). Mutation Helps Even Carriers of Alzheimer's Gene Avoid Alzheimer's. U.S. News & World Report. <https://www.usnews.com/news/health-news/articles/2024-03-29/mutation-helps-even-carriers-of-alzheimers-gene-avoid-alzheimers>

Nelson, M.R. et al. The APOE-R136S mutation protects against APOE4-driven Tau pathology, neurodegeneration and neuroinflammation. Nature Neuroscience; <https://doi.org/10.1038/s41593-023-01480-8> (2023).