

Decoding the genetics that drive disease

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Some human genes traditionally labeled as 'bad' are not always what they seem.
Credit: Gerd Altmann from Pixabay

From Alzheimer's to obesity, life can change dramatically if you discover you have a genetic risk of disease. Now, a new study from the Australian Centre for Precision Health, University of South Australia is challenging these predispositions, showing that some of the genes traditionally labeled as 'bad' are not always what they seem.

Examining the three main variants ($\epsilon 4$, $\epsilon 3$, and $\epsilon 2$) of the apolipoprotein E (APOE) gene involved in the metabolism of fats in the

body—researchers found that both the typically 'bad' variant, APOE ε4, and the 'good' variant, APOE ε2 can increase as well as decrease the risk of disease.

Overall, the research revealed APOE-associations with 18 different diseases.

Lead researcher and geneticist, UniSA's Dr. Amanda Lumsden says the findings provide valuable insights about how the APOE gene impacts health and disease.

"APOE-ε4 is notoriously the greatest known risk factor for late onset Alzheimer's disease, and is also connected to cardiovascular disease risk," Dr. Lumsden says.

"Yet, despite its reputation, our research shows it also has qualities that can protect against a range of illnesses, including obesity, type 2 diabetes, chronic airway obstruction, and liver disease.

"Conversely, we found that the rarer APOE ε2 gene variant—which is often considered protective and beneficial to health—can increase the risk of several conditions including peripheral vascular disease, stomach ulcers, diseases of the cervix, and bunions."

The study assessed APOE-associated risks across a spectrum of more than 950 diseases, using information from 337,484 participants in the UK Biobank. Brain neuroimaging, blood biochemistry, body measurement and lung function biomarkers for the same population were also used to support the APOE-associations.

World-renowned genetic epidemiologist and Senior Principal Research Fellow at SAHMRI, Professor Elina Hyppönen, says the study gives hope to people who may carry a genotype that has been classified as

'harmful.'

"An awareness of a genetic susceptibility can cause notable distress, especially if there is little that can be done to prevent that disease," Professor Hyppönen says.

"Here, we show that the same variant that causes harm with respect to some diseases, can be beneficial from other aspects of health, so it's not all doom and gloom based on your [genes](#).

"Of course, it can be helpful to understand our genetic vulnerabilities, as this can motivate us to make positive lifestyle changes that can reduce the risk.

"This is true of several diseases. In our earlier work, we showed that by living a healthy lifestyle you can help mitigate the risk of dementia, even when a person has an elevated genetic risk.

"So, despite what your genes say about predispositions to [disease](#), there are other ways to help you stay healthy and negate the risks."

More information: Amanda L. Lumsden et al, Apolipoprotein E (APOE) genotype-associated disease risks: a phenome-wide, registry-based, case-control study utilising the UK Biobank, *EBioMedicine* (2020). [DOI: 10.1016/j.ebiom.2020.102954](https://doi.org/10.1016/j.ebiom.2020.102954)

Provided by University of South Australia

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