10/2020

Sporadic Creutzfeldt-Jakob disease (Jones, 2020)

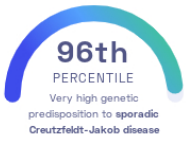
Emma Jones, et al.
The Lancet Neurology

Brain Dementia

STUDY SUMMARY

Discovery of 4 genomic regions associated with the sporadic Creutzfeldt-Jakob disease, a form of neurodegenerative prion disease.

YOUR RESULT



STUDY DESCRIPTION

Prion diseases occur in the brain when certain proteins fold into the wrong configuration, causing them to aggregate together. Neurodegeneration begins to occur as a result of this clumping leading to a progressive and fatal form of *dementia*. Prion diseases can be inherited (familial) or can occur through the consumption of infected meat (variant), but the vast majority of cases are known to occur spontaneously (sporadic). Little is known about what may make an individual more susceptible to acquiring a form of prion disease known as sporadic Creutzfeldt-Jakob disease (sCJD). This genome-wide association study examined over 5,000 individuals of European ancestry affected with sCJD to better understand the influence of genetics on the condition. Four regions of the genome were found to be strongly associated with the risk of the disease. In addition to a variant in the prion protein itself, the study found other variants near genes known as GAL3ST1, STX6, and BMERB1.

DID YOU KNOW?

Mad cow disease is one example of variant Creutzfeldt-Jakob disease. Though extremely rare, the condition results from eating nerve tissue (such as the brain) of an infected animal. Amazingly, infectious prion proteins can handle extreme heat and cannot simply be neutralized through cooking like pathogens such as bacteria. Even sterilization processes used by hospitals aren't effective at getting rid of the infectious proteins.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to sporadic Creutzfeldt-Jakob disease we summed up the effects of genetic variants that were linked to sporadic Creutzfeldt-Jakob disease in the study that this report is based on. These variants can be found in the table below. The variants highlighted in green have **positive effect sizes** and increase your genetic predisposition to sporadic Creutzfeldt-Jakob disease. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to sporadic Creutzfeldt-Jakob disease. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to sporadic Creutzfeldt-Jakob disease. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for sporadic Creutzfeldt-Jakob disease to be 1.12**. To determine whether your score is high or low, we compared it to the scores of 5,000 other Nebula Genomics users. We found that your polygenic score for sporadic Creutzfeldt-Jakob disease is in the **96th percentile**. This means that it is higher than the polygenic scores 96% of people. We consider this to be a **very high genetic predisposition to sporadic Creutzfeldt-Jakob disease**. However, please note that genetic predispositions do not account for important non-genetic factors like lifestyle. Furthermore, the genetics of most traits has not been fully understood yet and many associations between traits and genetic variants remain unknown. For additional explanations, click on the column titles in the table below and visit our Nebula Library tutorial.

VARIANT	YOUR GENOTYPE	COMMENTS	EFFECT SIZE	VARIANT FREQUENCY	SIGNIFICANCE
rs1799990_A	A / A	Near PRNP gene	0.21 (↑)	69%	9.61×10^{-17}
rs3747957_A NEW	A / A	Near STX6 gene	0.15 (↑)	43%	1.23×10^{-10}
rs2267161_C NEW	C / T	Near GAL3ST1 gene	0.17 (↑)	70%	1.97×10^{-10}
rs6498552_T NEW	T / G	Near BMERB1 gene	0.24 (↑)	18%	6.45×10^{-8}