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★ Resistance to transmissible Creutzfeldt-Jakob disease (Mead, 2009)

Simon Mead, et al.
The Lancet Neurology

Brain

STUDY SUMMARY

Identification of a genetic variant that confers resistance to transmissible Creutzfeldt-Jakob disease.

STUDY DESCRIPTION

Prion diseases are brain disorders caused by misfolded proteins that form aggregates leading to progressive, fatal dementia. Some prion diseases are transmissible. For example, consuming meat infected with misfolded prion proteins can induce the body's own prion proteins to misfold and aggregate. This study examined the genetic information of over 5,000 individuals from the United Kingdom and Papua New Guinea to understand the susceptibility to a transmissible Creutzfeldt-Jakob disease. The study identified one variant allele that confers resistance to infection with the CJD prion protein. This variant allele is located in the human prion gene and prevents induced misfolding and aggregation of the human prion proteins.

DID YOU KNOW?

While prion diseases are exceedingly rare, there is currently no cure. The spread of transmissible prion disease must be carefully controlled through the destruction of tainted meat and proper sterilization of medical equipment.

YOUR DETAILED RESULTS

The variants highlighted in green have **positive effect sizes** and increase your genetic predisposition to resistance to transmissible CJD. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to resistance to transmissible CJD. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to resistance to transmissible CJD. 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_G 	A / A	Resistance to transmissible Creutzfeldt-Jakob disease	999.00 [-]	30%	2.00×10^{-27}