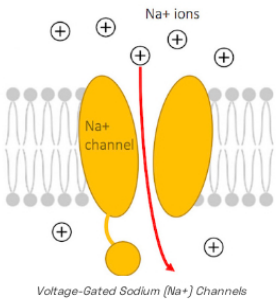
09/2014

Epilepsy (International League Against Epilepsy Consortium on Complex Epilepsies, 2014)

International League Against Epilepsy Consortium on Complex Epilepsies
The Lancet Neurology

Mind Brain

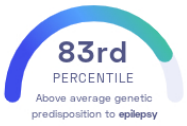


Voltage-Gated Sodium (Na+) Channels

STUDY SUMMARY

Genetic variants in the SCN1A and PCDH7 genes may influence the risk of epilepsy.

YOUR RESULT



STUDY DESCRIPTION

Epilepsy is a condition that causes unpredictable seizures and other health problems. This study looked at patients with general epilepsy as well as specific types of epilepsy (focal and unclassified) to determine which genetic variants are linked to the disease. After examining 34,853 individuals of European, Asian, or African ancestry, the most significant variants were identified as being located in the SCN1A and PCDH7 genes. SCN1A encodes for a subunit of voltage-dependent *sodium channel*s. These membrane proteins allow sodium to flow into the cell, which is essential for the activity of muscle cells and neurons. The PCDH7 gene encodes a protein that is thought to play a role in cell-cell recognition and adhesion.

DID YOU KNOW?

To reduce your risk of developing epilepsy, try to prevent traumatic brain injuries, get vaccinated, and lower your chances of stroke and heart disease by eating healthily and exercising regularly.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to epilepsy we summed up the effects of genetic variants that were linked to epilepsy 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 epilepsy. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to epilepsy. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to epilepsy. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for epilepsy to be 0.23**. 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 epilepsy is in the **83rd percentile**. This means that it is higher than the polygenic scores 83% of people. We consider this to be an **above average genetic predisposition to epilepsy**. 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	EFFECT SIZE	VARIANT FREQUENCY	SIGNIFICANCE
rs6732655_A	T / T	-0.12 (-)	22%	8.71×10^{-10}
rs28498976_G NEW	A / A	-0.11 (-)	54%	5.44×10^{-9}
rs2947349_C	A / A	0.21 (-)	26%	9.99×10^{-9}
rs1939012_T	T / T	0.11 (↑)	40%	2.37×10^{-8}