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Cerebral small vessel disease (Chung, 2019)

Jaeyoon Chung, et al.

Brain

Vasculature | Brain

STUDY SUMMARY

Identification of novel genetic variants linked to cerebral small vessel disease, which can cause a variety of cognitive symptoms.

YOUR RESULT

STUDY DESCRIPTION

Cerebral small vessel disease (CSVD) describes a variety of conditions related to abnormalities or damage to small blood vessels within the brain. Some of the most common conditions are small vessel ischaemic strokes, a result of hardened blood vessels, and intracerebral hemorrhage, which occurs when blood vessels in the brain burst. CSVD generally results in cognitive decline, movement disorders, and can lead to depression. Few genetic variants that correlate with the development of CSVD have been found. This genome-wide association study assesses the genetic data of more than 240,000 people to better determine genetic factors that may contribute to the risk of CSVD. Multiple novel variants were discovered, some that have previously been linked to brain and blood vessel disorders. For example, the PMF1 gene is associated with a breakdown of the blood-brain barrier that protects our brains, and the COL4A2 gene is associated with cells that form a layer on the inner surface of the brain's blood vessels and help regulate blood flow.

DID YOU KNOW?

Lowering your blood pressure may help prevent many types of cardiovascular diseases, including CSVD. Some preventative measures include regular exercise, a healthy diet, and taking prescribed blood pressure medications.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to CSVD we summed up the effects of genetic variants that were linked to CSVD 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 CSVD. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to CSVD. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to CSVD. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for CSVD to be -0.11**. 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 CSVD is in the **42nd percentile**. This means that it is higher than the polygenic scores 42% of people. We consider this to be an **average genetic predisposition to CSVD**. 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
rs9515201_A NEW	**C / C**	0.08 (–)	33%	5.27×10^{-10}
rs72932727_C NEW	**G / C**	-0.11 (↓)	13%	1.65×10^{-8}
rs2758605_C	**G / G**	-0.15 (–)	34%	2.57×10^{-8}