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Cerebral small vessel disease (Sargurupremraj, 2020)

Muralidharan Sargurupremraj, et al.
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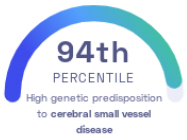
STUDY SUMMARY

Discovery of 27 genomic regions associated with cerebral small vessel disease.

YOUR RESULT



STUDY DESCRIPTION

Like all other organs and tissues across the body, the brain needs a constant supply of blood to receive oxygen and nutrients. Cerebral small vessel disease is a condition that results from damage to the small blood vessels in the brain. The disease can affect the brain function, eventually leading to cognitive decline and problems with muscle coordination. This study examined the genetic predisposition to cerebral small vessel disease by analyzing genetic data from over 50,000 individuals of European and African-American ancestry. A total of 27 unique regions of the genome were linked to cerebral small vessel disease, 18 of which have not been previously identified. Interestingly, over half of the identified variants have previosly been associated with predisposition to high blood pressure.

DID YOU KNOW?

Cerebral small vessel disease has been shown to contribute to the risk of stroke. Roughly 25% of all strokes may be attributable to the condition. It is also believed to slow recovery in individuals that suffered a stroke.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to cerebral small vessel disease we summed up the effects of genetic variants that were linked to cerebral small vessel 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 cerebral small vessel disease. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to cerebral small vessel disease. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to cerebral small vessel disease. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for cerebral small vessel disease to be 1.53**. 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 cerebral small vessel disease is in the **94th percentile**. This means that it is higher than the polygenic scores 94% of people. We consider this to be a **high genetic predisposition to cerebral small vessel 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	EFFECT SIZE	VARIANT FREQUENCY	SIGNIFICANCE
rs34974290_A	G / G	0.10 (–)	19%	2.59×10^{-48}
rs7596872_A	C / A	0.10 (↑)	10%	3.81×10^{-24}
rs6503417_C NEW	C / C	0.05 (↑)	63%	3.43×10^{-19}
rs12921170_A NEW	A / A	0.05 (↑)	58%	9.82×10^{-18}
rs6940540_G	T / T	0.04 (–)	41%	7.17×10^{-16}
rs73923006_G	G / G	0.06 (↑)	81%	2.00×10^{-14}
rs6797002_C NEW	C / C	0.05 (↑)	73%	8.18×10^{-14}
rs4630220_G	G / G	0.05 (↑)	71%	1.46×10^{-13}
rs7603972_A	A / G	0.07 (↑)	87%	2.23×10^{-13}
rs10786772_G	G / A	0.04 (↑)	64%	1.56×10^{-12}
rs17205972_T NEW	G / G	0.05 (–)	20%	4.76×10^{-12}
rs62172472_G NEW	G / G	0.05 (↑)	79%	3.67×10^{-11}
rs2303655_T NEW	T / T	0.05 (↑)	78%	4.03×10^{-11}
rs1948948_C NEW	C / T	0.04 (↑)	56%	1.17×10^{-10}
rs1285847_T NEW	T / T	0.04 (↑)	55%	1.24×10^{-10}
rs73184312_G NEW	G / G	0.04 (↑)	72%	2.18×10^{-9}
rs71471298_T NEW	C / T	0.05 (↑)	11%	2.72×10^{-9}
rs55940034_G	A / A	0.04 (–)	29%	4.49×10^{-9}
rs6540873_A NEW	A / C	0.03 (↑)	61%	1.37×10^{-8}
rs7157599_C	C / C	0.04 (↑)	29%	1.49×10^{-8}
rs11257311_G NEW	T / G	0.05 (↑)	70%	2.01×10^{-8}
rs5762197_C NEW	C / C	0.04 (↑)	71%	2.67×10^{-8}
rs12443113_G NEW	G / G	0.03 (↑)	55%	3.42×10^{-8}
rs11249945_A NEW	G / G	0.03 (–)	35%	3.60×10^{-8}
rs72680374_T NEW	T / A	0.03 (↑)	63%	5.45×10^{-8}
rs7004825_T NEW	T / T	0.03 (↑)	47%	9.18×10^{-8}
rs786921_A NEW	G / G	0.03 (–)	60%	2.52×10^{-7}