Matthew Traylor, et al. The Lancet Neurology

Brain Vasculature

## STUDY SUMMARY

This report is based on a study that discovered 11 novel genetic variants associated with lacunar stroke.

### YOUR RESULT

# STUDY DESCRIPTION



A stroke occurs when the blood flow to the brain is blocked, which results in brain cells being starved of oxygen. A lacunar stroke is a type of stroke, caused by the blockage of arteries deep in the brain. Lacunar strokes represent about 25% of all strokes, making them one of the most common types of stroke. Common symptoms of a lacunar stroke include slurred speech, difficulty moving, confusion, and loss of consciousness. This genome-wide association study aimed to provide a better understanding of the genetic



Lacunar storkes occur when small arteries that are located deep in the brain become occluded.

basis of lacunar strokes. Through the analysis of more than 262,000 individuals of European ancestry, researchers identified 12 regions of the genome associated with an individual's risk of having a lacunar stroke. Of these regions, 11 are novel. One gene connected to lacunar stroke risk in this study is COL4A2. It encodes of a type for collagen that provides structural support to the cells. The COL4A2 gene has previously also been associated with intracerebral hemorrhage and coronary artery disease.

### DID YOU KNOW?

Patients who suffer from lacunar strokes appear to develop dementia 4-12 times more frequently than the average population.

#### VOLID DETAILED DECLITE

To calculate your genetic predisposition to lacunar stroke we summed up the effects of genetic variants that were linked to lacunar stroke 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 lacunar stroke. The variants highlighted in blue have negative effects sizes and decrease your genetic predisposition to lacunar stroke. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to lacunar stroke. By adding up the effect sizes of the highlighted variants we calculated your polygenic score for lacunar stroke to be 1.74. 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 lacunar stroke is in the 84th percentile. This means that it is higher than the polygenic scores 84% of people. We consider this to be an above average genetic predisposition to lacunar stroke. 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 <sup>©</sup>	YOUR GENOTYPE <sup>①</sup>	GENE <sup>⊕</sup>	EFFECT SIZE <sup>①</sup>	VARIANT FREQUENCY <sup>™</sup>	SIGNIFICANCE
rs72934535_T	T/T	ICA1L, WDR12, CARF, NBEAL1	0.20 (1)	89%	5.30 x 10 <sup>-16</sup>
rs61000833_T	T/T	SH3PXD2A	0.07 (1)	60%	6.00 x 10 <sup>-13</sup>
rs2293576_G	G/G	SPI1, SLC39A13, PSMC3, RAPSN	0.13 (1)	67%	6.40 × 10 <sup>-13</sup>
rs11838776_A 🌼	G/G	COL4A2	0.10 (-)	29%	7.90 x 10 <sup>-13</sup>
rs2303655_T	T/T	LOX, ZNF474, LOC100505841	0.11 (1)	81%	1.90 × 10 <sup>-10</sup>
rs2984613_C 💮	C/C	SLC25A44, PMF1, BGLAP	0.09 (1)	64%	8.20 × 10 <sup>-10</sup>
rs79043147_T 🐡	C/C	HTRA1, ARMS2	0.20 (-)	7%	1.60 × 10 <sup>-9</sup>
rs12445022_A	A / A	ZCCHC14	0.11 (1)	34%	3.10 x 10 <sup>-9</sup>
rs7766042_C 💮	Т/Т	FOXF2, FOXQ1	0.17 (-)	11%	5.20 x 10 <sup>-9</sup>
rs4621303_T	T/A	ULK4	0.15 (1)	83%	6.40 x 10 <sup>-9</sup>
rs225744_C 💮	C/C	VTA1, GPR126	0.09 (1)	77%	9.20 x 10 <sup>-9</sup>
rs9958650_G 🐡	A / A	ZBTB14, EPB41L3	0.17 (-)	10%	2.40 × 10 <sup>-8</sup>