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Venous thromboembolism (Klarin, 2019)

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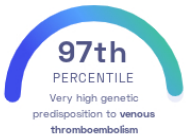
Nature Genetics

Vasculature

STUDY SUMMARY

Identification of 22 new genetic variants associated with venous thromboembolism.

YOUR RESULT



STUDY DESCRIPTION

Venous thromboembolism is characterized by the formation of blood clots, typically in veins of the legs. Such blood clots can block the blood flow, resulting in painful swelling of the limbs. The blood clots can also travel to the lungs, resulting in pulmonary embolism, a dangerous condition associated with high mortality. To better understand genetic factors contributing to the risk of venous thromboembolism, this genome-wide association study examined over 650,000 individuals of European, African, and Hispanic ancestry and discovered 22 new genetic variants. Some of the implicated genes play a role in the regulation of LDL cholesterol (our "bad cholesterol") in the blood.

DID YOU KNOW?

Pulmonary embolism often goes underdiagnosed. The symptoms can range from chest pains and shortness of breath to pain or tenderness in the limbs.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to venous thromboembolism we summed up the effects of genetic variants that were linked to venous thromboembolism 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 venous thromboembolism. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to venous thromboembolism. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to venous thromboembolism. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for venous thromboembolism to be 3.40**. 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 venous thromboembolism is in the **97th percentile**. This means that it is higher than the polygenic scores 97% of people. We consider this to be a **very high genetic predisposition to venous thromboembolism**. 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
rs6025_T	NA	0.93 (-)	2%	1.00×10^{-300}
rs9411377_A	A / C	0.30 (↑)	31%	1.12×10^{-224}
rs4253417_C	T / T	0.20 (-)	39%	7.63×10^{-96}
rs2066865_A	G / A	0.20 (↑)	25%	2.17×10^{-88}
rs1799963_A	NA	0.63 (-)	1%	1.06×10^{-69}
rs78707713_T	T / T	0.24 (↑)	88%	1.65×10^{-55}
rs10747514_A	G / A	0.12 (↑)	32%	3.99×10^{-32}
rs2288904_G	G / G	0.12 (↑)	78%	4.40×10^{-24}
rs12445050_T NEW	C / C	0.12 (-)	14%	2.10×10^{-21}
rs4734879_A	A / A	0.09 (↑)	71%	4.21×10^{-19}
rs10886430_G NEW	A / A	0.13 (-)	12%	1.94×10^{-18}
rs6048_A NEW	A / A	0.07 (↑)	72%	3.12×10^{-17}
rs216296_G NEW	G / G	0.13 (↑)	91%	6.27×10^{-16}
rs174536_A NEW	A / A	0.08 (↑)	66%	9.34×10^{-15}
rs7585314_T NEW	C / C	0.07 (-)	34%	9.10×10^{-14}
rs3002417_T NEW	C / C	0.06 (-)	53%	2.05×10^{-13}
rs3211752_G NEW	A / A	0.06 (-)	48%	2.44×10^{-12}
rs7051718_T	T / T	0.07 (↑)	75%	3.55×10^{-12}
rs9607928_A NEW	C / C	0.09 (-)	13%	6.05×10^{-12}
rs6083037_A	A / A	0.09 (↑)	83%	7.25×10^{-12}
rs7739314_C NEW	C / C	0.06 (↑)	53%	1.38×10^{-11}
rs1654425_C NEW	C / C	0.08 (↑)	84%	8.31×10^{-11}
rs1867312_C NEW	C / A	0.06 (↑)	44%	1.35×10^{-10}
rs6795524_G NEW	A / A	0.14 (-)	7%	1.38×10^{-10}
rs1048483_T NEW	T / T	0.06 (↑)	47%	1.79×10^{-10}
rs16867574_C NEW	C / T	0.06 (↑)	32%	3.42×10^{-10}
rs57328376_G NEW	A / A	0.06 (-)	30%	8.83×10^{-10}
rs12824685_G NEW	G / G	0.07 (↑)	80%	1.15×10^{-9}
rs2842700_A NEW	C / C	0.10 (-)	10%	4.49×10^{-9}
rs13084580_T NEW	C / C	0.08 (-)	11%	4.60×10^{-9}
rs10087301_A NEW	A / A	0.07 (↑)	81%	0.00×10^{0}
rs2074492_T NEW	T / T	0.05 (↑)	62%	0.00×10^{0}
rs731839_A NEW	A / A	0.05 (↑)	66%	4.79×10^{-8}

N/A indicates variants that could not be imputed using the 1000 genomes project datasets and variants that have a frequency of < 5%. Your genome was sequenced at 30x/100x coverage and is not imputed. However, to calculate percentiles, we need to compare your data with other users imputed data. To make the data comparable, we need to exclude some of the variants from your data.