5/2021

Osmotic hemolysis (Page, 2021)

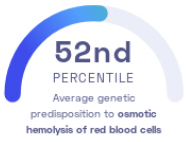
Grier Page, et al.
Journal of Clinical Investigation

Blood

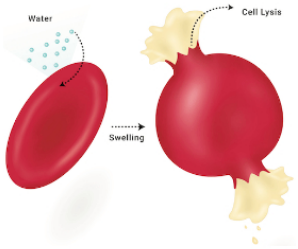
STUDY SUMMARY

This report is based on a study that discovered 21 genetic variants associated with osmotic hemolysis, which causes the destruction of red blood cells.

YOUR RESULT



STUDY DESCRIPTION



Osmotic hemolysis describes rupturing of red blood cells due to water inflow.

Blood plays an extremely important role in providing oxygen and nutrients to all parts of the body. Whether due to trauma or disease, occasionally blood needs to be transferred from one individual to another. This procedure, known as a transfusion. Often, the donated blood must spend time in storage before being transfused, and the transfusion process itself may take in excess many hours. As a result, the red blood cells can get damaged during storage or transfusion. To identify genetic factors that may contribute to damage of red blood cells, this study examined genetic data of over 12,000 individuals of European, African, Asian, and Hispanic ancestries. The researchers identified 21 genes associated with the destruction of red blood cells through osmotic hemolysis, which describes a bursting due to the inflow of water. A number of genes were linked to osmotic hemolysis, including ANK1 and SPTA1, which work to maintain the structural integrity of red blood cells.

DID YOU KNOW?

Someone in the United States needs a blood transfusion every 2 seconds. Since blood cannot be manufactured, hospitals rely on volunteer donors to meet this need.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to osmotic hemolysis of red blood cells we summed up the effects of genetic variants that were linked to osmotic hemolysis of red blood cells 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 osmotic hemolysis of red blood cells. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to osmotic hemolysis of red blood cells. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to osmotic hemolysis of red blood cells. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for osmotic hemolysis of red blood cells to be -39.80**. 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 osmotic hemolysis of red blood cells is in the **52nd percentile**. This means that it is higher than the polygenic scores 52% of people. We consider this to be an **average genetic predisposition to osmotic hemolysis of red blood cells**. 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	GENE	EFFECT SIZE	VARIANT FREQUENCY	SIGNIFICANCE
rs4737010_A NEW	G / G	ANK1	-1.90 (–)	24%	5.85×10^{-28}
rs2022003_A NEW	A / A	SPTA1	1.70 (↑)	30%	1.01×10^{-22}
rs857725_G NEW	T / T	SPTA1	-1.60 (–)	28%	8.75×10^{-21}
rs35365035_T NEW	T / C	MY09B	1.20 (↑)	41%	9.88×10^{-16}
rs551118_C NEW	C / G	PIEZO1	-1.30 (↓)	59%	4.04×10^{-14}
rs55707417_G NEW	G / G	MFSD2B	-1.80 (↓)	14%	1.07×10^{-13}
rs77684561_C NEW	C / T	BRAP, ALDH2, MAKKAPK5	1.50 (↑)	17%	2.24×10^{-13}
rs72805692_A NEW	A / A	HK1	1.80 (↑)	11%	4.90×10^{-11}
rs360153_T NEW	T / C	SWAP70	-1.00 (↓)	60%	1.28×10^{-10}
rs334_A NEW	NA	HBB	-8.20 (–)	< 1%	3.66×10^{-10}
rs5883264_GC NEW	CCC / CCC	AQP1	-1.30 (–)	16%	4.23×10^{-10}
rs78484557_C NEW	NA	EYS	-25.70 (–)	3%	3.20×10^{-9}
rs717662_C NEW	C / C	ARHGAP42	-1.50 (↓)	11%	4.00×10^{-9}
rs118149920_T NEW	NA	NA	-12.50 (–)	1%	9.74×10^{-9}
rs148642995_T NEW	NA	NA	-11.20 (–)	< 1%	1.09×10^{-8}
rs6976036_T NEW	T / T	IKZF1	0.90 (↑)	50%	1.86×10^{-8}
rs7222349_G NEW	A / A	UBTF, SLC4A1	-1.00 (–)	32%	3.62×10^{-8}
rs35558093_A NEW	A / A	TFB2M	-21.20 (↓)	29%	3.87×10^{-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.