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Myeloproliferative neoplasms (Bao, 2020)

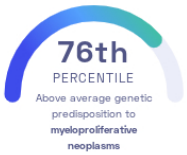
Erik Bao, et al.
Nature

Blood | Cancer

STUDY SUMMARY

Discovery of 17 regions of the genome associated with a risk of myeloproliferative neoplasms, a type of blood cancer.

YOUR RESULT



STUDY DESCRIPTION

Blood cells, including red blood cells, white blood cells, and platelets, are produced in the bone marrow, which is a spongy tissue inside bones. Myeloproliferative neoplasms are a type of blood cancer that occurs when the bone marrow overproduces blood stem cells. Common symptoms of myeloproliferative neoplasms include feelings of weakness, tiredness, headaches, and fever. To identify factors that contribute to a genetic predisposition to this disease, this study examined the genomes of over 1.15 million individuals of European ancestry. The researchers identified 17 variants associated with an increased risk of myeloproliferative neoplasms, including 7 not previously reported. Overall, these variants may explain over 18% of the total genetic risk for developing myeloproliferative neoplasms.

DID YOU KNOW?

There are multiple types of myeloproliferative neoplasms, which are categorized by the cells affected. For example, polycythemia vera is a type of myeloproliferative neoplasm that is characterized by overproduction of red blood cells, while chronic myeloid leukemia is caused by overproduction of white blood cells.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to myeloproliferative neoplasms we summed up the effects of genetic variants that were linked to myeloproliferative neoplasms 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 myeloproliferative neoplasms. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to myeloproliferative neoplasms. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to myeloproliferative neoplasms. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for myeloproliferative neoplasms to be 2.57**. 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 myeloproliferative neoplasms is in the **76th percentile**. This means that it is higher than the polygenic scores 76% of people. We consider this to be an **above average genetic predisposition to myeloproliferative neoplasms**. 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
rs1327494_G	A / G	0.69 (↑)	27%	1.11×10^{-170}
rs7705526_A	C / A	0.46 (↑)	34%	2.42×10^{-64}
rs2853677_G	G / A	0.38 (↑)	42%	4.32×10^{-54}
rs62329718_A	NA	0.75 (-)	4%	2.72×10^{-34}
rs62471615_C	C / A	0.26 (↑)	30%	7.20×10^{-21}
rs7310615_C	C / G	0.24 (↑)	48%	2.46×10^{-18}
rs116466979_C NEW	NA	0.41 (-)	5%	1.86×10^{-12}
rs1633768_T	C / C	0.18 (-)	27%	2.15×10^{-12}
rs1800057_G	NA	0.50 (-)	3%	2.94×10^{-12}
rs74676712_T NEW	C / T	0.26 (↑)	11%	3.64×10^{-11}
rs9847631_T	G / G	0.16 (-)	40%	4.89×10^{-10}
rs8002412_C NEW	T / T	0.18 (-)	18%	5.23×10^{-10}
rs77249081_G NEW	NA	1.31 (-)	1%	5.54×10^{-10}
rs9946154_T NEW	T / T	0.14 (↑)	64%	1.50×10^{-8}
rs9864772_G NEW	A / A	0.14 (-)	61%	2.06×10^{-8}
rs17879961_G	NA	0.80 (-)	2%	3.60×10^{-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.