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Melanoma (Barrett, 2011)

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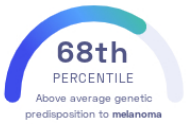
Nature Genetics

Skin Cancer

STUDY SUMMARY

Susceptibility to melanoma is associated with variants in the ATM and CASP8 genes.

YOUR RESULT



STUDY DESCRIPTION

Melanoma is a skin cancer that occurs when pigment-producing skin cells mutate and become cancerous. To identify genetic variants associated with the development of melanoma, this study examined 11,389 individuals of European ancestry. The most significant variants were located in the ATM and CASP8 genes. The protein encoded by the ATM gene checks DNA for damage at various stages of the *cell cycle* and activates other genes if the DNA is damaged. The CASP8 gene plays an essential role in *apoptosis*. ATM and CASP8 have both been implicated in other types of cancers as well.

DID YOU KNOW?

In order to prevent melanoma, you should wear sunscreen (at least 30 SPF) and protective clothing while also avoiding the midday sun and tanning beds.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to melanoma we summed up the effects of genetic variants that were linked to melanoma 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 melanoma. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to melanoma. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to melanoma. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for melanoma to be 0.30**. 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 melanoma is in the **68th percentile**. This means that it is higher than the polygenic scores 68% of people. We consider this to be an **above average genetic predisposition to melanoma**. 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
rs258322_A	A / G	0.53 (↑)	11%	2.70×10^{-27}
rs1393350_A	G / G	0.26 (–)	28%	1.77×10^{-13}
rs13016963_A NEW	A / A	0.13 (↑)	37%	8.60×10^{-10}
rs45430_C NEW	C / T	-0.13 (↓)	39%	2.90×10^{-9}
rs1801516_A NEW	G / A	-0.17 (↓)	13%	3.40×10^{-9}
rs7023329_G	A / G	-0.19 (↓)	49%	7.35×10^{-9}
rs401681_T	C / C	0.18 (–)	46%	2.98×10^{-8}