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Melanoma (Landi, 2020)

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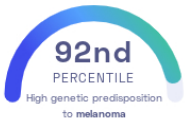
Skin Cancer

STUDY SUMMARY

Identification of 54 genomic regions associated with melanoma risk.

YOUR RESULT



STUDY DESCRIPTION

Melanoma is the most serious type of skin cancer. This study compared ~36,000 melanoma patients with ~375,000 healthy individuals of European descent and identified 68 genetic variants in 54 different genomic regions. The study also found associations between melanoma risk and lighter skin color as well as a larger number of moles on the body. One of the regions associated with both melanoma and moles is near the PARP1 gene, a DNA repair gene that helps control the melanoma causing gene MITF. Another variant was associated with the *HLA* region, which suggests a role for immunity in the development of skin cancer. Other genes were involved in *telomere* maintenance. The genomic regions discovered in this study collectively explain ~8.5% of melanoma risk heritability.

DID YOU KNOW?

There are a lot of warning signs to determine if a spot is just a mole or is actually melanoma. The easiest check is called the "Ugly Duckling" test -- if a mole looks different from the other moles on your body like the ugly duckling looked different from its siblings, it's worth getting checked out! Melanomas are often asymmetric, have unusual borders, have different colors, are large in diameter, or change over time.

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 -4.85**. 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 **92nd percentile**. This means that it is higher than the polygenic scores 92% of people. We consider this to be a **high 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
rs1805007_C	C / T	-0.56 (↓)	94%	5.86×10^{-52}
rs6059655_A	A / G	0.37 (↑)	6%	2.52×10^{-42}
rs1126809_G	G / G	-0.19 (↓)	76%	4.78×10^{-37}
rs408825_C	C / T	-0.12 (↓)	41%	1.03×10^{-32}
rs16891982_C	G / G	-0.67 (–)	12%	1.96×10^{-28}
rs7902587_C	C / T	-0.15 (↓)	90%	2.68×10^{-23}
rs871024_C	A / A	0.17 (–)	48%	2.72×10^{-23}
rs132941_T	T / T	0.10 (↑)	55%	8.80×10^{-23}
rs1801516_G	G / A	0.13 (↑)	86%	2.22×10^{-21}
rs117132860_G	G / G	-0.34 (↓)	98%	3.83×10^{-21}
rs4364713_A	G / G	0.10 (–)	36%	8.50×10^{-21}
rs1339759_C NEW	C / C	0.10 (↑)	67%	5.61×10^{-19}
rs6914598_T	T / T	-0.09 (↓)	68%	1.18×10^{-18}
rs10739220_C	T / T	0.10 (–)	26%	1.34×10^{-18}
rs2695237_T	T / T	0.10 (↑)	63%	1.53×10^{-18}
rs13178866_C	C / C	-0.14 (↓)	55%	2.59×10^{-18}
rs32578_G NEW	G / A	0.09 (↑)	66%	6.58×10^{-17}
rs4731207_G NEW	G / G	-0.07 (↓)	54%	2.22×10^{-15}
rs76798800_G NEW	G / G	-0.08 (↓)	75%	3.86×10^{-15}
rs1800440_T	C / C	0.10 (–)	82%	6.97×10^{-15}
rs8444_G	G / A	0.08 (↑)	65%	3.89×10^{-14}
rs4420522_A NEW	A / G	-0.07 (↓)	69%	8.34×10^{-14}
rs149617956_G NEW	G / G	-0.94 (↓)	> 99%	9.00×10^{-14}
rs143190905_G NEW	G / T	0.14 (↑)	91%	6.54×10^{-13}
rs72834823_T NEW	T / T	0.10 (↑)	82%	1.04×10^{-12}
rs10960710_G NEW	G / T	-0.07 (↓)	39%	3.08×10^{-12}
rs12913832_A	G / G	-0.13 (–)	34%	4.85×10^{-12}
rs1278768_G NEW	C / C	-0.06 (–)	49%	6.33×10^{-12}
rs117648907_C NEW	C / C	-0.22 (↓)	98%	7.29×10^{-12}
rs3950296_C NEW	C / C	0.08 (↑)	75%	4.47×10^{-11}
rs10859996_C NEW	C / T	0.07 (↑)	64%	2.09×10^{-10}
rs78378222_T NEW	T / G	-0.27 (↓)	99%	3.33×10^{-10}
rs12984831_G NEW	G / G	-0.43 (↓)	98%	3.86×10^{-10}
rs113469387_G NEW	G / G	-0.09 (↓)	91%	8.76×10^{-10}
rs4237963_T NEW	A / A	-0.07 (–)	21%	1.27×10^{-9}
rs7941496_G NEW	G / G	0.06 (↑)	52%	1.40×10^{-9}
rs5766565_A NEW	G / G	0.06 (–)	65%	1.44×10^{-9}
rs2967383_G NEW	T / T	0.06 (–)	27%	2.24×10^{-9}
rs1056927_A NEW	A / G	-0.07 (↓)	56%	2.74×10^{-9}

rs6908626_G	G / T	0.09 (↑)	84%	3.92×10^{-9}
rs6994183_A	A / A	-0.08 (↓)	87%	4.84×10^{-9}
rs12473635_T	T / C	-0.07 (↓)	78%	5.17×10^{-9}
rs12215602_G	G / G	-0.06 (↓)	72%	7.91×10^{-9}
rs79966207_T	T / T	-0.08 (↓)	85%	8.68×10^{-9}
rs7778378_C	C / T	-0.07 (↓)	25%	8.93×10^{-9}
rs670318_T	NA	-0.15 (–)	5%	1.21×10^{-8}
rs2369633_T	C / C	0.10 (–)	8%	1.24×10^{-8}
rs28986343_C	C / C	0.14 (↑)	95%	1.61×10^{-8}
rs12539524_C	C / T	-0.07 (↓)	85%	1.65×10^{-8}
rs3780269_G	G / A	-0.06 (↓)	69%	1.92×10^{-8}
rs10931936_T	T / T	0.08 (↑)	28%	2.17×10^{-8}
rs12290699_T	T / C	-0.06 (↓)	75%	2.20×10^{-8}
rs13263376_G	G / A	-0.07 (↓)	36%	2.28×10^{-8}
rs12523094_T	T / T	0.07 (↑)	57%	1.74×10^{-6}

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.