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Age-related macular degeneration (Fritsche, 2015)

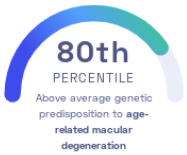
Lars Fritsche, et al.
Nature Genetics

Aging | Eyes

STUDY SUMMARY

Discovery of 16 novel risk loci for age-related macular degeneration.

YOUR RESULT



Above average genetic predisposition to **age-related macular degeneration**

STUDY DESCRIPTION

The retina, located at the back of the eye, contains cells that detect light and generate signals that are sent to the brain enabling us to visualize the world around us. The center part of the retina is known as the macula. It allows us to see in high-resolution and perceive colors. Degeneration of the macula is one of the leading causes of vision loss among the elderly, affecting nearly 160 million individuals worldwide. This genome-wide association study attempted to identify genetic variants that correlate with a person's risk of developing this age-related macular degeneration by examining the genomes of over 40,000 individuals of European ancestry. The study found 34 genetic loci, 16 of which are novel, that are associated with age-related macular degeneration. Some of the implicated genes play a role in the formation of extracellular matrix, that embeds the eyes and other organs. Other implicated genes are known to be involved in inflammation control.

DID YOU KNOW?

Smoker's have a significantly increased risk of developing macular degeneration. When the chemicals from cigarette smoke get in the eyes, they irritate the retina and can cause lasting damage over time.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to age-related macular degeneration we summed up the effects of genetic variants that were linked to age-related macular degeneration 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 age-related macular degeneration. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to age-related macular degeneration. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to age-related macular degeneration. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for age-related macular degeneration to be 0.79**. 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 age-related macular degeneration is in the **80th percentile**. This means that it is higher than the polygenic scores 80% of people. We consider this to be an **above average genetic predisposition to age-related macular degeneration**. 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
rs3750846_C	T / C	1.03 (↑)	44%	6.50×10^{-735}
rs10922109_A	C / A	-0.97 (↓)	22%	9.60×10^{-618}
rs116503776_A	/	-0.56 (–)	9%	1.20×10^{-103}
rs2230199_G	G / G	0.36 (↑)	27%	3.80×10^{-69}
rs429358_C	T / T	-0.36 (–)	10%	2.40×10^{-42}
rs5754227_C	T / T	-0.26 (–)	11%	1.10×10^{-24}
rs10033900_T	T / C	0.14 (↑)	51%	5.40×10^{-17}
rs2043085_C	C / C	-0.14 (↓)	35%	4.30×10^{-16}
rs943080_C	C / T	-0.13 (↓)	47%	1.10×10^{-14}
rs62358361_T	NA	0.59 (–)	2%	1.30×10^{-14}
rs62247658_C	C / T	0.13 (↑)	47%	1.80×10^{-14}
rs72802342_A NEW	C / C	-0.24 (–)	7%	5.00×10^{-12}
rs140647181_C	NA	0.46 (–)	2%	1.40×10^{-11}
rs6565597_T NEW	C / T	0.12 (↑)	40%	1.50×10^{-11}
rs79037040_G	/	-0.11 (–)	45%	4.50×10^{-11}
rs8135665_T	C / C	0.13 (–)	22%	5.50×10^{-11}
rs61985136_C	C / T	-0.11 (↓)	36%	1.60×10^{-10}
rs142450006_T NEW	TTTCTTTCTTTCT / TTTCTTTCTTTCT	-0.16 (–)	12%	2.40×10^{-10}
rs9564692_T	C / T	-0.12 (↓)	28%	3.30×10^{-10}
rs1626340_A	G / G	-0.13 (–)	19%	3.80×10^{-10}
rs61941274_A NEW	NA	0.41 (–)	2%	1.10×10^{-9}
rs1142_T NEW	C / T	0.10 (↑)	37%	1.40×10^{-9}
rs10781182_T NEW	G / G	0.10 (–)	33%	2.60×10^{-9}
rs3138141_A NEW	C / A	0.15 (↑)	22%	4.30×10^{-9}
rs7803454_T NEW	C / C	0.12 (–)	21%	4.80×10^{-9}
rs11080055_A NEW	C / C	-0.09 (–)	46%	1.00×10^{-8}
rs2740488_C NEW	A / C	-0.11 (↓)	26%	1.20×10^{-8}
rs114092250_A NEW	NA	-0.36 (–)	2%	2.10×10^{-8}
rs67538026_T NEW	C / C	-0.11 (–)	46%	2.60×10^{-8}
rs11884770_T NEW	T / C	-0.11 (↓)	26%	2.90×10^{-8}
rs71507014_G NEW	G / GC	0.10 (↑)	43%	3.00×10^{-8}
rs12357257_A NEW	G / A	0.10 (↑)	24%	4.40×10^{-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.