⭐ Age-related macular degeneration (Han, 2020)
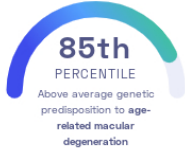
Xikun Han, et al.
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`Eyes`

STUDY SUMMARY

Identification of 12 novel genomic regions associated with age-related macular degeneration.

YOUR RESULT

85th PERCENTILE
Above average genetic predisposition to **age-related macular degeneration**

STUDY DESCRIPTION

The retina is the light-sensitive nerve layer that lines the back of the eye. Age-related macular degeneration describes damage to the macula, the area of the retina that is responsible for central, high-resolution, color vision. Age-related macular degeneration is the leading cause of blindness among elderly individuals, affecting nearly 160 million people worldwide. This genome-wide association study aimed to identify genetic variants associated with age-related macular degeneration by examining the genomes of ~132,000 individuals of mostly European descent. The researchers identified 69 genetic variants associated with age-related macular degeneration, 12 of which are novel. Some of the implicated genes are involved in inflammatory response pathways and eye traits, such as macular thickness.

DID YOU KNOW?

There are two types of macular degeneration: dry and wet. The wet type is characterized by the abnormal growth of blood vessels into the macula. Wet macular degeneration can be treated with drugs that slow down the growth of blood vessels.

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 0.69.** 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 **85th percentile.** This means that it is higher than the polygenic scores 85% 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
rs800292_A	G / A	-0.73 (↓)	23%	3.90×10^{-421}
rs7542235_G	A / A	-0.78 (–)	19%	6.10×10^{-414}
rs2248799_T	T / T	0.61 (↑)	49%	2.70×10^{-412}
rs6428365_A	C / C	-0.69 (–)	14%	6.00×10^{-262}
rs2672589_A	A / A	0.47 (↑)	61%	1.90×10^{-235}
rs7555070_T	T / T	0.46 (↑)	68%	1.10×10^{-209}
rs12116643_C	T / T	-0.51 (–)	16%	2.20×10^{-157}
rs760070_C	T / T	-0.58 (–)	10%	1.20×10^{-133}
rs2672590_C	A / C	-0.37 (↓)	26%	3.50×10^{-119}
rs12134610_G	A / G	-0.53 (↓)	9%	8.80×10^{-97}
rs11200607_T	C / C	-0.34 (–)	25%	2.10×10^{-94}
rs163494_T	C / C	0.31 (–)	23%	6.40×10^{-79}
rs7918867_A	G / A	-0.34 (↓)	17%	4.20×10^{-72}
rs4752699_A	G / G	-0.36 (–)	14%	7.40×10^{-71}
rs4151671_T	NA	-0.56 (–)	4%	3.70×10^{-61}
rs10494757_T	C / C	0.43 (–)	7%	1.80×10^{-51}
rs4420638_G	A / A	-0.26 (–)	19%	5.00×10^{-48}
rs1337169_C	T / C	-0.20 (↓)	68%	8.90×10^{-39}
rs12211410_T	C / C	0.31 (–)	9%	2.70×10^{-34}
rs5754227_C	T / T	-0.25 (–)	13%	3.30×10^{-33}
rs1329423_C	T / T	-0.19 (–)	26%	2.30×10^{-31}
rs247616_T	C / T	0.17 (↑)	33%	6.20×10^{-29}
rs5754206_T	NA	-0.39 (–)	4%	3.10×10^{-27}
rs11076175_G	A / A	-0.20 (–)	18%	1.60×10^{-26}
rs1998345_G	A / A	-0.15 (–)	65%	5.90×10^{-25}
rs943080_T	C / T	0.14 (↑)	50%	2.70×10^{-24}
rs10033900_C	T / C	-0.14 (↓)	52%	1.10×10^{-23}
rs11569520_G	A / A	-0.21 (–)	13%	4.30×10^{-22}
rs2043085_C	C / C	0.14 (↑)	61%	6.10×10^{-21}
rs7266392_G	A / A	-0.27 (–)	6%	5.70×10^{-20}
rs7428936_C	T / C	0.13 (↑)	41%	5.80×10^{-20}
rs8135665_T	C / C	0.15 (–)	20%	6.30×10^{-17}
rs2070895_A	G / A	-0.14 (↓)	22%	5.70×10^{-16}
rs8017304_A	G / A	0.11 (↑)	65%	4.30×10^{-16}
rs13255394_C	T / T	0.11 (–)	54%	5.20×10^{-15}
rs401186_T	C / C	-0.13 (–)	22%	4.20×10^{-14}
rs11771241_A	G / G	0.13 (–)	20%	6.60×10^{-14}
rs13081855_T	G / G	0.18 (–)	9%	1.80×10^{-13}
rs12949956_T	C / T	0.10 (↑)	51%	1.20×10^{-12}

rs2575876_A	G / A	-0.12 (↓)	25%	2.50×10^{-12}
rs9973159_T NEW	C / C	-0.14 (−)	15%	2.90×10^{-12}
rs4943289_A	G / A	-0.11 (↓)	31%	5.10×10^{-12}
rs1142_T	C / T	0.10 (↑)	36%	8.70×10^{-12}
rs2268354_T	NA	-0.31 (−)	2%	1.60×10^{-11}
rs10781180_T	T / T	-0.11 (↓)	69%	1.90×10^{-11}
rs1605677_A	NA	-0.27 (−)	3%	2.20×10^{-11}
rs8056814_A	G / G	-0.16 (−)	8%	9.10×10^{-11}
rs3138142_T	C / T	0.11 (↑)	24%	1.60×10^{-10}
rs6899205_A NEW	G / A	-0.09 (↓)	28%	2.00×10^{-10}
rs11080055_C	C / C	0.09 (↑)	51%	6.80×10^{-10}
rs1064583_G	A / G	-0.08 (↓)	40%	2.00×10^{-9}
rs3760775_T	G / G	-0.19 (−)	6%	2.20×10^{-9}
rs3825991_A NEW	C / A	0.09 (↑)	48%	4.30×10^{-9}
rs1926564_A NEW	A / A	-0.14 (↓)	90%	4.90×10^{-9}
rs11142636_A	A / A	-0.11 (↓)	79%	5.30×10^{-9}
rs11884770_C	T / C	0.10 (↑)	73%	7.40×10^{-9}
rs1378940_A NEW	A / A	0.09 (↑)	68%	8.30×10^{-9}
rs2842343_G	G / G	-0.14 (↓)	91%	8.80×10^{-9}
rs259842_C NEW	T / C	-0.08 (↓)	62%	1.10×10^{-8}
rs11120691_G NEW	T / G	-0.08 (↓)	44%	1.20×10^{-8}
rs2367070_A	C / A	0.10 (↑)	20%	1.30×10^{-8}
rs2011092_C NEW	C / C	0.09 (↑)	35%	1.40×10^{-8}
rs7896471_T NEW	NA	-0.20 (−)	4%	1.90×10^{-8}
rs17356664_T	C / T	-0.08 (↓)	31%	1.90×10^{-8}
rs17421410_G NEW	A / A	0.15 (−)	7%	2.00×10^{-8}
rs16841920_C	NA	0.31 (−)	2%	2.40×10^{-8}
rs1005819_T NEW	T / T	-0.08 (↓)	42%	2.40×10^{-8}
rs2232613_T NEW	C / C	-0.14 (−)	8%	3.00×10^{-8}
rs17576_G	A / G	-0.08 (↓)	35%	3.00×10^{-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.