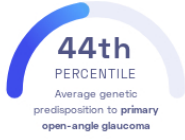


STUDY SUMMARY

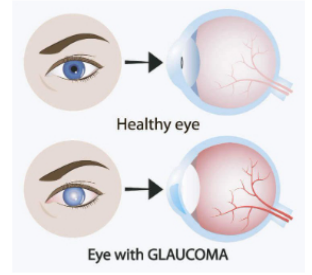
Identification of 127 genetic variants associated with primary open-angle glaucoma, a leading cause of blindness.

YOUR RESULT



STUDY DESCRIPTION

Visual information is relayed from the eyes to the brain through a communication line known as the optic nerve. Glaucoma describes a group of eye conditions where the optic nerve becomes damaged, which leads to progressive vision loss and eventually complete blindness. This study sought to discover genetic variants associated with primary open-angle glaucoma (POAG), a highly heritable glaucoma subtype. The study analyzed genomic data of nearly 384,000 individuals of European, African, and Asian ancestries and identified 127 variants associated with POAG. Of these 127 variants, 44 were novel. Among genes linked to POAG were HLA-G/HLA-H and GJA1. The HLA genes play a central role in the immune system, while GJA1 encodes for a communication channel between cells.



Glaucoma is typically caused by a blockage of a drainage canal in the eye which leads to fluid build up, increase in eye pressure and damage to the optic nerve.

























DID YOU KNOW?

A 2002 study found that blindness is one of the most common health-related fears that people exhibit. It ranks third after fear of cancer and heart disease.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to primary open-angle glaucoma we summed up the effects of genetic variants that were linked to primary open-angle glaucoma 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 primary open-angle glaucoma. The variants highlighted in blue have **negative effect sizes** and decrease your genetic predisposition to primary open-angle glaucoma. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to primary open-angle glaucoma. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for primary open-angle glaucoma to be -3.19**. 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 primary open-angle glaucoma is in the **44th percentile**. This means that it is higher than the polygenic scores 44% of people. We consider this to be an **average genetic predisposition to primary open-angle glaucoma**. 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 [Ⓞ]
rs944801_C	G / G	0.24 (-)	59%	3.71×10^{-123}
rs2790053_C	G / G	0.30 (-)	15%	2.48×10^{-100}
rs2472494_T	T / C	0.18 (↑)	43%	9.38×10^{-83}
rs33912345_A	C / A	-0.16 (↓)	61%	2.18×10^{-62}
rs74315329_A	NA	1.70 (-)	< 1%	3.76×10^{-52}
rs938604_A	A / A	-0.14 (↓)	62%	7.88×10^{-51}
rs9913911_A	A / A	0.14 (↑)	65%	7.26×10^{-49}
rs3829849_T	C / C	-0.13 (-)	37%	1.39×10^{-38}
rs2745572_A	G / G	0.11 (-)	65%	1.29×10^{-28}
rs72904286_T	C / C	-0.16 (-)	11%	4.58×10^{-27}
rs10257125_A	A / T	-0.12 (↓)	76%	7.15×10^{-26}
rs2514879_T	G / G	-0.13 (-)	12%	1.20×10^{-22}
rs1658436_A	G / A	-0.09 (↓)	51%	6.03×10^{-22}
rs4612174_T	T / G	0.09 (↑)	64%	7.29×10^{-22}
rs56233426_A	A / A	0.09 (↑)	47%	2.20×10^{-21}
rs13101042_C	G / C	-0.10 (↓)	62%	5.08×10^{-21}
rs2326788_A	G / G	-0.09 (-)	39%	1.78×10^{-20}
rs1893261_A	G / A	0.09 (↑)	38%	4.34×10^{-20}
rs2627761_T	C / T	-0.10 (↓)	82%	1.37×10^{-18}
rs1579050_A	A / G	-0.09 (↓)	41%	1.59×10^{-18}
rs8141610_T	C / C	-0.13 (-)	15%	3.10×10^{-18}
rs62283813_A	A / A	-0.14 (↓)	95%	1.03×10^{-17}
rs7344_T	T / C	-0.09 (↓)	75%	5.72×10^{-17}
rs5750494_T	G / G	0.08 (-)	43%	2.42×10^{-16}
rs4414666_T	T / T	-0.08 (↓)	66%	2.80×10^{-16}
rs12699251_A	A / G	-0.08 (↓)	59%	4.19×10^{-16}
rs10792871_T	C / C	0.11 (-)	8%	6.49×10^{-16}
rs112142644_A	G / G	0.13 (-)	8%	1.02×10^{-16}
rs4769194_A	A / G	0.09 (↑)	18%	1.24×10^{-16}
rs66819623_T	C / C	0.08 (-)	27%	1.83×10^{-16}
rs1550437_T	C / C	-0.09 (-)	21%	4.62×10^{-16}
rs324788_T	T / C	-0.07 (↓)	42%	8.24×10^{-16}
rs7924522_A	A / A	0.08 (↑)	66%	2.13×10^{-14}
rs6787621_T	G / G	-0.08 (-)	30%	2.95×10^{-14}
rs57866767_T	T / C	-0.07 (↓)	57%	3.77×10^{-14}
rs7111873_A	A / G	0.07 (↑)	29%	4.36×10^{-14}
rs10230941_C	C / G	-0.08 (↓)	64%	4.49×10^{-14}
rs12623251_T	C / T	-0.07 (↓)	41%	1.43×10^{-13}

rs3753841_A	G / A	-0.07 (↓)	61%	1.65 × 10 ⁻¹³
rs5762776_A	A / G	-0.08 (↓)	32%	2.66 × 10 ⁻¹³
rs1346_A	A / A	0.09 (↑)	82%	5.05 × 10 ⁻¹³
rs17146835_T 	T / T	-0.07 (↓)	80%	6.70 × 10 ⁻¹³
rs12566440_A 	A / A	-0.07 (↓)	64%	9.34 × 10 ⁻¹³
rs200621439_A	A / A	-0.14 (↓)	32%	1.21 × 10 ⁻¹²
rs1286770_T	C / C	-0.09 (-)	12%	1.82 × 10 ⁻¹²
rs12013166_A 	A / A	-0.07 (↓)	28%	2.02 × 10 ⁻¹²
rs407238_C 	G / G	-0.07 (-)	38%	2.71 × 10 ⁻¹²
rs10882167_T	C / T	0.07 (↑)	40%	5.17 × 10 ⁻¹²
rs2431023_A	T / T	0.07 (-)	42%	1.07 × 10 ⁻¹¹
rs9544024_A	G / G	-0.07 (-)	48%	1.11 × 10 ⁻¹¹
rs7805468_T 	T / G	0.06 (↑)	41%	1.21 × 10 ⁻¹¹
rs11868441_A	G / G	0.08 (-)	21%	1.49 × 10 ⁻¹¹
rs59101260_G	G / GA	0.09 (↑)	62%	1.77 × 10 ⁻¹¹
rs4652902_A	C / A	0.08 (↑)	74%	1.80 × 10 ⁻¹¹
rs941125_T 	G / T	0.06 (↑)	35%	2.01 × 10 ⁻¹¹
rs4141194_A	C / C	0.06 (-)	28%	4.53 × 10 ⁻¹¹
rs31916_A	A / A	0.07 (↑)	71%	5.45 × 10 ⁻¹¹
rs8176749_T	C / C	0.10 (-)	8%	5.52 × 10 ⁻¹¹
rs2935057_A	A / G	0.08 (↑)	11%	5.76 × 10 ⁻¹¹
rs6667536_A	G / G	-0.14 (-)	6%	6.12 × 10 ⁻¹¹
rs62521288_A	A / A	-0.13 (↓)	94%	1.05 × 10 ⁻¹⁰
rs2439386_T 	T / T	-0.07 (↓)	72%	1.09 × 10 ⁻¹⁰
rs9308689_T	C / C	-0.06 (-)	39%	1.53 × 10 ⁻¹⁰
rs17527016_T 	C / C	-0.08 (-)	23%	1.60 × 10 ⁻¹⁰
rs10517281_A 	A / G	0.07 (↑)	82%	1.70 × 10 ⁻¹⁰
rs13194719_A	A / A	0.06 (↑)	45%	2.21 × 10 ⁻¹⁰
rs10430737_T	C / C	0.06 (-)	41%	2.32 × 10 ⁻¹⁰
rs12613800_A 	T / A	0.06 (↑)	46%	2.55 × 10 ⁻¹⁰
rs216197_A	G / G	-0.06 (-)	37%	3.08 × 10 ⁻¹⁰
rs754458_A	G / A	0.06 (↑)	51%	3.08 × 10 ⁻¹⁰
rs12433661_A	A / A	0.06 (↑)	58%	3.21 × 10 ⁻¹⁰
rs7137828_T	C / T	0.07 (↑)	53%	3.32 × 10 ⁻¹⁰
rs2191828_C 	G / G	0.08 (-)	7%	3.74 × 10 ⁻¹⁰
rs1192415_A	A / A	-0.07 (↓)	82%	5.01 × 10 ⁻¹⁰
rs2875238_T	T / C	-0.07 (↓)	33%	5.02 × 10 ⁻¹⁰
rs4361323_A	A / G	-0.06 (↓)	57%	5.28 × 10 ⁻¹⁰
rs1106287_T	T / T	-0.06 (↓)	76%	5.66 × 10 ⁻¹⁰
rs12438742_C	C / C	-0.06 (↓)	40%	6.72 × 10 ⁻¹⁰
rs6101546_T	T / C	0.06 (↑)	46%	6.93 × 10 ⁻¹⁰
rs10248136_T	C / T	0.06 (↑)	52%	7.74 × 10 ⁻¹⁰
rs242569_A 	C / C	-0.07 (-)	76%	8.80 × 10 ⁻¹⁰
rs687914_T	G / T	-0.07 (↓)	22%	8.89 × 10 ⁻¹⁰
rs4903352_C 	C / C	0.07 (↑)	84%	9.60 × 10 ⁻¹⁰
rs13050568_A 	G / A	0.07 (↑)	22%	1.27 × 10 ⁻⁹
rs6713914_T 	C / C	0.05 (-)	58%	1.42 × 10 ⁻⁹
rs4542196_A 	G / A	-0.06 (↓)	77%	1.80 × 10 ⁻⁹
rs9530143_A	G / G	-0.06 (-)	38%	2.01 × 10 ⁻⁹
rs1500708_T 	C / C	-0.06 (-)	21%	2.13 × 10 ⁻⁹
rs13049669_A 	T / T	-0.09 (-)	6%	2.23 × 10 ⁻⁹
rs114367221_T	C / C	0.11 (-)	6%	2.24 × 10 ⁻⁹
rs2811688_C 	C / C	-0.06 (↓)	51%	2.56 × 10 ⁻⁹
rs11710845_T	C / C	-0.07 (-)	21%	2.60 × 10 ⁻⁹
rs12846405_T 	T / T	0.05 (↑)	37%	2.63 × 10 ⁻⁹
rs35740987_CA 	C / C	0.07 (-)	57%	2.98 × 10 ⁻⁹
rs190157577_A 	A / A	-0.94 (↓)	> 99%	3.23 × 10 ⁻⁹
rs6602453_A 	C / C	0.06 (-)	32%	3.38 × 10 ⁻⁹
rs6552711_T 	G / T	0.05 (↑)	52%	3.88 × 10 ⁻⁹
rs10237321_A	C / C	-0.06 (-)	36%	3.92 × 10 ⁻⁹

rs12447201_T	C / C	-0.06 (-)	32%	4.22×10^{-9}
rs2515437_C	C / G	-0.12 (↓)	95%	5.61×10^{-9}
rs12667675_A	A / A	0.07 (↑)	84%	6.62×10^{-9}
rs62478245_T	NA	0.16 (-)	4%	8.95×10^{-9}
rs1348518145_A	/	-0.06 (-)	53%	9.24×10^{-9}
rs3777688_A	G / G	0.06 (-)	64%	9.36×10^{-9}
rs72837408_A	A / A	-0.06 (↓)	69%	9.38×10^{-9}
rs12139208_T	T / C	0.05 (↑)	33%	9.55×10^{-9}
rs1037013_A	A / G	0.05 (↑)	57%	9.82×10^{-9}
rs280875_T	T / T	-0.10 (↓)	92%	1.01×10^{-8}
rs61751937_C	NA	0.17 (-)	4%	1.05×10^{-8}
rs7972874_A	T / A	-0.06 (↓)	24%	1.12×10^{-8}
rs2218471_C	C / G	-0.06 (↓)	50%	1.39×10^{-8}
rs7120067_A	C / C	0.05 (-)	35%	1.60×10^{-8}
rs17006576_A	G / G	0.06 (-)	29%	1.67×10^{-8}
rs172531_A	G / G	-0.06 (-)	67%	1.93×10^{-8}
rs10444329_A	G / A	-0.06 (↓)	81%	2.36×10^{-8}
rs4076000_A	C / C	0.05 (-)	39%	2.44×10^{-8}
rs6124885_A	A / A	-0.08 (↓)	95%	2.78×10^{-8}
rs8038628_A	A / G	-0.07 (↓)	86%	2.91×10^{-8}
rs6732795_A	C / C	-0.05 (-)	62%	3.47×10^{-8}
rs6957752_T	C / T	0.05 (↑)	63%	3.63×10^{-8}
rs6437682_A	C / C	-0.05 (-)	38%	3.76×10^{-8}
rs57400569_A	G / A	-0.06 (↓)	24%	4.22×10^{-8}
rs72692789_T	T / T	0.09 (↑)	88%	4.25×10^{-8}
rs2637250_T	T / T	0.05 (↑)	55%	4.66×10^{-8}
rs7760346_A	A / A	-0.05 (↓)	63%	4.71×10^{-8}
rs9852634_A	A / C	0.05 (↑)	50%	4.82×10^{-8}
rs10908363_T	C / T	0.05 (↑)	55%	4.89×10^{-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.