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☆ Optic disc size (Hun, 2019)

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Human Molecular Genetics

`Eyes`

STUDY SUMMARY

Identification of 115 genetic variants associated with the size of the optic disc.

YOUR RESULT

63rd
PERCENTILE

Above average genetic predisposition to **larger optic disc**
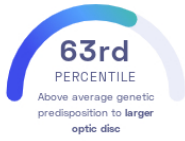
STUDY DESCRIPTION

The optic disc is an oval structure in the back of the eye that represents the exit point of the optic nerve that connects the eye to the brain. A small optic disc size is correlated with vulnerability of optic nerves to various degenerative diseases. To better understand genetic determinants of optic disc size, this study analyzed the genomes of over 95,000 individuals of European ancestry and discovered 115 genetic variants associated with optic disc size of which 90 are novel. Some of the identified variants have been previously associated with eye pressure, glaucoma and nearsightedness.

DID YOU KNOW?

To keep your eyes healthy, protect them from the sun, have annual eye checkups, avoid smoking and alcohol consumption, exercise regularly, and control your blood pressure as well as cholesterol levels.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to larger optic disc we summed up the effects of genetic variants that were linked to larger optic disc 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 larger optic disc. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to larger optic disc. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to larger optic disc. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for larger optic disc to be -0.02.** 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 larger optic disc is in the **63rd percentile.** This means that it is higher than the polygenic scores 63% of people. We consider this to be an **above average genetic predisposition to larger optic disc.** 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 ⓘ
rs7916410_T	C / C	-0.07 (–)	24%	3.30×10^{-246}
rs9988687_A	A / A	0.07 (↑)	77%	3.40×10^{-209}
rs4658101_A	G / G	0.06 (–)	19%	2.80×10^{-160}
rs3835100_A	G / G	-0.08 (–)	6%	1.20×10^{-56}
rs11808696_T	C / C	-0.05 (–)	8%	5.80×10^{-48}
rs5762752_C	C / G	-0.02 (↓)	36%	7.90×10^{-33}
rs6787363_A	A / A	0.02 (↑)	38%	2.30×10^{-31}
rs7196852_A	A / A	-0.02 (↓)	70%	2.30×10^{-31}
rs11079419_T NEW	C / C	0.03 (–)	20%	2.30×10^{-31}
rs10862708_A	A / T	0.02 (↑)	56%	1.80×10^{-29}
rs77587000_T NEW	NA	-0.09 (–)	1%	2.90×10^{-27}
rs12137699_T NEW	C / C	0.02 (–)	24%	4.30×10^{-27}
rs17398137_A	G / G	-0.03 (–)	18%	4.00×10^{-26}
rs7482884_A	T / T	0.02 (–)	23%	4.20×10^{-25}
rs9905786_T NEW	G / T	0.02 (↑)	64%	5.30×10^{-25}
rs56385951_A	G / G	0.03 (–)	11%	2.90×10^{-24}
rs4839470_T NEW	C / T	-0.02 (↓)	24%	3.50×10^{-23}
rs13070614_C	C / C	0.02 (↑)	78%	2.00×10^{-22}
rs10483727_T NEW	T / C	-0.02 (↓)	39%	2.60×10^{-21}
rs12024620_T	C / C	0.04 (–)	6%	7.40×10^{-20}
rs74764079_A NEW	NA	-0.05 (–)	3%	1.70×10^{-19}
rs12436074_A NEW	A / A	-0.02 (↓)	56%	2.30×10^{-19}
rs73173591_A NEW	T / T	0.03 (–)	9%	2.60×10^{-19}
rs72759609_T NEW	T / T	0.03 (↑)	90%	7.50×10^{-19}
rs28514893_T	C / C	0.02 (–)	21%	3.90×10^{-18}
rs76567987_A NEW	A / A	0.02 (↑)	84%	1.70×10^{-17}
rs9534439_T NEW	C / C	0.02 (–)	19%	9.90×10^{-17}
rs3857971_A NEW	G / G	-0.02 (–)	30%	1.90×10^{-16}
rs74056359_A NEW	A / A	-0.02 (↓)	83%	2.50×10^{-16}
rs34607745_T NEW	G / T	-0.02 (↓)	30%	3.40×10^{-16}
rs787540_A	A / A	-0.02 (↓)	66%	7.70×10^{-16}
rs7744813_A NEW	C / A	-0.02 (↓)	59%	1.00×10^{-15}
rs4858678_A	A / G	-0.02 (↓)	72%	1.10×10^{-15}
rs1930941_A NEW	C / A	0.02 (↑)	45%	1.10×10^{-15}
rs8034595_A	A / C	-0.02 (↓)	31%	1.70×10^{-15}
rs742127_C	G / C	0.02 (↑)	31%	3.60×10^{-15}
rs11602582_A NEW	G / G	-0.04 (–)	5%	4.10×10^{-15}
rs74891390_C NEW	G / G	0.03 (–)	6%	4.70×10^{-15}
rs12471698_T NEW	C / C	-0.01 (–)	33%	5.60×10^{-15}

Variant	Genotype	Effect	Frequency	p-value
rs10512176_T [NEW]	T / T	-0.02 (↓)	72%	1.70×10^{-14}
rs61975075_A [NEW]	A / A	0.03 (↑)	94%	1.80×10^{-14}
rs9330799_A [NEW]	A / G	-0.02 (↓)	53%	3.70×10^{-14}
rs11639803_A	A / G	0.02 (↑)	77%	4.80×10^{-14}
rs12547416_T	C / T	-0.01 (↓)	51%	5.20×10^{-14}
rs2092524_A [NEW]	G / G	0.01 (–)	34%	1.30×10^{-13}
rs9967780_T [NEW]	G / T	0.02 (↑)	22%	1.80×10^{-13}
rs906568_T [NEW]	C / T	-0.01 (↓)	36%	2.00×10^{-13}
rs13283933_T	C / C	-0.02 (–)	29%	2.20×10^{-13}
rs1905014_T [NEW]	T / C	-0.01 (↓)	57%	4.40×10^{-13}
rs1550094_A [NEW]	A / A	0.01 (↑)	69%	5.20×10^{-13}
rs9287914_A [NEW]	T / T	0.01 (–)	31%	8.40×10^{-13}
rs10764494_A [NEW]	C / A	0.01 (↑)	68%	8.50×10^{-13}
rs4832012_C [NEW]	C / C	0.01 (↑)	51%	8.90×10^{-13}
rs35285683_A [NEW]	A / A	-0.02 (↓)	85%	2.30×10^{-12}
rs7717697_T [NEW]	T / C	0.01 (↑)	59%	2.40×10^{-12}
rs11564398_T [NEW]	T / C	0.01 (↑)	72%	4.30×10^{-12}
rs7188859_T [NEW]	T / C	0.01 (↑)	63%	8.10×10^{-12}
rs72784719_A [NEW]	A / A	0.06 (↑)	99%	1.00×10^{-11}
rs4285804_A [NEW]	T / A	0.01 (↑)	56%	1.20×10^{-11}
rs77877421_A [NEW]	A / A	-0.03 (↓)	94%	1.30×10^{-11}
rs934237_A	A / A	-0.02 (↓)	79%	1.70×10^{-11}
rs57742792_A [NEW]	A / A	0.01 (↑)	51%	3.50×10^{-11}
rs116072427_C [NEW]	G / G	-0.02 (↓)	7%	4.50×10^{-11}
rs698153_A [NEW]	A / A	-0.03 (↓)	95%	4.70×10^{-11}
rs34536594_A [NEW]	G / G	-0.02 (–)	18%	5.50×10^{-11}
rs71296770_A [NEW]	A / G	-0.02 (↓)	90%	8.20×10^{-11}
rs6673575_A [NEW]	G / G	0.01 (–)	32%	8.80×10^{-11}
rs2033054_T [NEW]	T / T	-0.01 (↓)	65%	1.30×10^{-10}
rs10957177_A [NEW]	A / A	0.01 (↑)	75%	1.30×10^{-10}
rs11584075_A [NEW]	A / A	0.02 (↑)	92%	1.60×10^{-10}
rs10164395_T [NEW]	C / C	-0.01 (–)	33%	1.70×10^{-10}
rs6030_T	T / C	0.01 (↑)	67%	1.80×10^{-10}
rs6999835_T [NEW]	T / T	0.01 (↑)	63%	2.40×10^{-10}
rs17734073_A [NEW]	A / A	-0.02 (↓)	87%	2.70×10^{-10}
rs251526_A [NEW]	A / A	-0.02 (↓)	92%	3.90×10^{-10}
rs2149108_T [NEW]	C / T	-0.01 (↓)	40%	4.00×10^{-10}
rs13417287_T [NEW]	T / C	-0.01 (↓)	77%	5.70×10^{-10}
rs2761882_T [NEW]	C / C	-0.01 (–)	50%	6.40×10^{-10}
rs11114001_C [NEW]	G / C	0.01 (↑)	19%	7.30×10^{-10}
rs12619508_A [NEW]	C / C	-0.01 (–)	47%	7.70×10^{-10}
rs62391624_T [NEW]	C / C	-0.02 (–)	14%	8.70×10^{-10}
rs16909978_A [NEW]	G / A	0.02 (↑)	11%	9.10×10^{-10}
rs9330813_A [NEW]	G / G	-0.01 (–)	31%	1.10×10^{-9}
rs3754442_T [NEW]	C / C	0.01 (–)	59%	1.20×10^{-9}
rs7096311_A [NEW]	C / C	-0.01 (–)	19%	2.70×10^{-9}
rs59199978_A [NEW]	A / A	-0.01 (↓)	82%	3.90×10^{-9}
rs11627052_A [NEW]	G / G	0.01 (–)	22%	4.90×10^{-9}
rs13022913_T [NEW]	C / C	-0.01 (–)	57%	5.50×10^{-9}
rs78977588_A [NEW]	C / C	0.02 (–)	12%	6.30×10^{-9}
rs1126809_A [NEW]	G / G	-0.01 (–)	30%	8.00×10^{-9}
rs340762_T [NEW]	T / T	0.01 (↑)	45%	9.40×10^{-9}
rs10823610_A [NEW]	C / A	0.01 (↑)	56%	9.60×10^{-9}
rs163524_A [NEW]	C / A	-0.01 (↓)	18%	1.00×10^{-8}
rs599892_C [NEW]	C / C	0.01 (↑)	70%	1.10×10^{-8}
rs1852148_A [NEW]	A / A	-0.01 (↓)	47%	1.10×10^{-8}
rs10910_T [NEW]	T / T	0.01 (↑)	69%	1.10×10^{-8}
rs911526_A [NEW]	G / A	-0.01 (↓)	32%	1.10×10^{-8}
rs6860726_C [NEW]	C / C	-0.01 (↓)	49%	1.30×10^{-8}

rs7620608_T [NEW]	T / C	-0.01 (↓)	61%	1.40×10^{-8}
rs7615960_A [NEW]	A / A	-0.02 (↓)	93%	1.50×10^{-8}
rs7610486_A [NEW]	A / A	0.01 (↑)	77%	1.60×10^{-8}
rs2266963_C [NEW]	C / G	-0.01 (↓)	81%	1.90×10^{-8}
rs2220051_T [NEW]	T / C	-0.01 (↓)	34%	2.00×10^{-8}
rs10084346_T	C / C	0.01 (−)	16%	2.20×10^{-8}
rs28840750_T [NEW]	T / T	-0.03 (↓)	96%	2.40×10^{-8}
rs2430356_A [NEW]	A / A	-0.01 (↓)	77%	2.70×10^{-8}
rs11684168_T [NEW]	T / T	0.01 (↑)	83%	2.90×10^{-8}
rs8076249_T [NEW]	C / T	-0.01 (↓)	60%	2.90×10^{-8}
rs80218807_T [NEW]	C / C	0.02 (−)	11%	3.10×10^{-8}
rs9852867_T [NEW]	G / G	-0.01 (−)	41%	3.20×10^{-8}
rs1901440_A [NEW]	C / A	-0.01 (↓)	66%	3.40×10^{-8}
rs30371_T [NEW]	T / C	-0.01 (↓)	63%	3.40×10^{-8}
rs28603236_A [NEW]	A / A	0.01 (↑)	85%	4.30×10^{-8}
rs13264644_A [NEW]	A / G	-0.01 (↓)	57%	4.50×10^{-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.