Stuart MacGregor, et al. Nature Genetics



STUDY SUMMARY

Identification of over 24 genetic variants that correlate with the development of glaucoma.

YOUR RESULT

STUDY DESCRIPTION



Glaucoma is one of the leading causes of blindness in older adults, though it can occur at any age. It develops when the optic nerve that connects the eye to the brain becomes damaged, often as a result of increased pressure within the eye. One method of halting glaucoma progression is to decrease that pressure. This genome-wide association study of over 130,000 participants of European descent identified 24 genetic variants associated with increased <u>intraocular pressure</u> and the risk of developing glaucoma. Two of the variants are near genes that play a role in development and function of the optic nerve (CDKN/2B and SIX6).

DID YOU KNOW

While damage from glaucoma can't be reversed, early detection can help slow the progression of vision loss. Regular eye exams can detect increased intraocular pressure before the vision becomes impaired.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to glaucoma we summed up the effects of genetic variants that were linked to 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 glaucoma. The variants highlighted in blue have negative effects sizes and decrease your genetic predisposition to glaucoma. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to glaucoma. By adding up the effect sizes of the highlighted variants we calculated your polygenic score for glaucoma to be -1.31. 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 glaucoma is in the 62nd percentile. This means that it is higher than the polygenic scores 62% of people. We consider this to be an above average genetic predisposition to 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 [®]
rs7518099_T	T/T	-0.31 (↓)	89%	2.35 x 10 ⁻⁵²
rs944801_C	G / G	0.20 (-)	70%	8.00 x 10 ⁻³⁶
rs2472493_A	G / A	-0.17 (↓)	60%	4.30 x 10 ⁻³⁰
rs2093210_T	C/T	-0.15 (↓)	47%	6.29 x 10 ⁻²²
rs9913911_A	A / A	0.15 (1)	68%	2.13 x 10 ⁻²¹
rs28795989_A	G / G	0.14 (-)	57%	1.90 x 10 ⁻²⁰
rs945686_C	G / C	-0.15 (↓)	99%	2.58 x 10 ⁻¹⁷
rs2745572_A	G / G	0.12 (-)	69%	1.35×10^{-13}
rs9284802_A	G / A	-0.11 (↓)	43%	1.56 x 10 ⁻¹²
rs58073046_A	A / A	-0.16 (↓)	90%	1.99 x 10 ⁻¹²
rs12699251_A 🖐	A / G	-0.11 (↓)	68%	4.16 x 10 ⁻¹²
rs10505100_A 💮	C/C	-0.17 (-)	13%	4.86×10^{-12}
rs2024211_A	A / C	-0.11 (↓)	76%	9.48 x 10 ⁻¹²
rs9853115_A	T/T	-0.11 (-)	50%	4.35 × 10 ⁻¹¹
rs61394862_T 🐡	C/C	-0.11 (-)	28%	4.13×10^{-10}
rs2935057_A 💮	A / G	0.14 (1)	26%	8.02 × 10 ⁻¹⁰
rs2073006_T 💮	C/C	0.13 (-)	12%	1.20 × 10 ⁻⁹
rs61861119_A	A / G	-0.09 (↓)	56%	2.56 x 10 ⁻⁹
rs8141433_A	A / A	0.14 (↑)	74%	3.04 x 10 ⁻⁹
rs4141671_T 🐡	T/C	-0.09 (↓)	50%	8.67 x 10 ⁻⁹
rs73174345_T 💮	T/T	-0.17 (↓)	89%	1.53 x 10 ⁻⁸
rs1013278_C 🙌	G / C	0.09 (1)	40%	2.99 x 10 ⁻⁸
rs7924522_A 🖐	A / A	0.09 (1)	70%	3.33 x 10 ⁻⁸
rs11710139_A 💮	G / G	-0.11 (-)	20%	5.00 x 10 ⁻⁸

