

★ Keratoconus (Hardcastle, 2021)

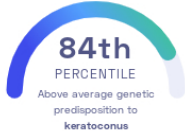
Allison Hardcastle, et al.
Communications Biology

Eyes

STUDY SUMMARY

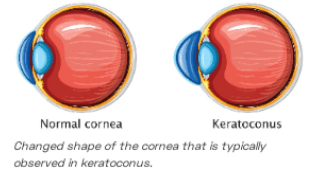
Identification of 36 genomic regions associated with keratoconus, a thinning of the cornea that can cause blindness.

YOUR RESULT



STUDY DESCRIPTION

The cornea acts as the “front window” of the eye, allowing in light but also protecting against dirt and germs that could damage the inner parts of the eye. Keratoconus is a disease that causes the thinning of this protective layer. Over time, the thinning of the cornea causes it to change its shape, which can eventually lead to diminished vision and blindness. While eye trauma is a leading cause of keratoconus, genetics also plays a role in many cases. To identify genetic factors associated with the onset of keratoconus, this study analyzed genomic data of over 120,000 individuals of European, South Asian, and African ancestries. The scientists discovered 36 regions of the genome associated with the condition, many of which were newly identified in this study. In total, these variants may explain about 12% of the keratoconus heritability. Many of the identified variants are near collagen genes (COL6A1, COL1A1, and COL5A1). Collagen is the main structural component of the cornea.



DID YOU KNOW?

The fastest-moving muscles in the body can be found in the eye. There are 6 eye muscles that enable us to perceive changes in the environment very quickly.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to keratoconus we summed up the effects of genetic variants that were linked to keratoconus 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 keratoconus. The variants highlighted in blue have **negative effect sizes** and decrease your genetic predisposition to keratoconus. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to keratoconus. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for keratoconus to be -3.02**. 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 keratoconus is in the **84th percentile**. This means that it is higher than the polygenic scores 84% of people. We consider this to be an **above average genetic predisposition to keratoconus**. 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 [Ⓞ]
rs2721051_T	C / T	0.45 (↑)	8%	5.71 x 10 ⁻³⁵
rs3118518_A	G / G	0.27 (-)	73%	1.83 x 10 ⁻²⁸
rs7117921_T	T / T	0.27 (↑)	47%	1.09 x 10 ⁻²⁶
rs4894414_T	C / C	0.30 (-)	18%	1.21 x 10 ⁻²⁶
rs12912010_T	G / T	-0.31 (↓)	25%	1.99 x 10 ⁻²⁸
rs35523808_A	NA	0.66 (-)	3%	2.90 x 10 ⁻²⁶
rs840464_T	T / T	0.23 (↑)	56%	1.72 x 10 ⁻²⁰
rs11117401_A	NA	0.24 (-)	1%	3.88 x 10 ⁻²⁰
rs1324175_T	T / T	-0.24 (↓)	81%	2.59 x 10 ⁻¹⁹
rs12516400_T	T / T	0.22 (↑)	65%	3.16 x 10 ⁻¹⁸
rs12603055_C	G / G	-0.23 (-)	19%	2.23 x 10 ⁻¹⁴
rs17285550_A	A / A	-0.18 (↓)	71%	2.84 x 10 ⁻¹²
rs2143683_T	T / C	0.18 (↑)	30%	3.51 x 10 ⁻¹²
rs658352_T	T / C	0.18 (↑)	66%	3.71 x 10 ⁻¹²
rs76747345_A	A / A	-0.55 (↓)	99%	6.53 x 10 ⁻¹²
rs4646785_T	C / T	-0.20 (↓)	21%	9.01 x 10 ⁻¹²
rs142493024_A	NA	0.76 (-)	1%	9.07 x 10 ⁻¹²
rs11145948_A	A / A	0.17 (↑)	44%	9.89 x 10 ⁻¹²
rs2417930_T	T / C	0.16 (↑)	32%	2.62 x 10 ⁻¹¹
rs6106210_T	C / C	0.17 (-)	39%	2.85 x 10 ⁻¹¹
rs6904450_A	A / A	-0.19 (↓)	83%	6.44 x 10 ⁻¹¹
rs56161228_A	G / G	-0.21 (-)	18%	2.70 x 10 ⁻¹⁰
rs1453379_T	C / C	0.16 (-)	99%	3.32 x 10 ⁻¹⁰
rs1200108_A	G / A	-0.16 (↓)	65%	4.52 x 10 ⁻¹⁰
rs116792882_T	T / T	-0.69 (↓)	99%	4.82 x 10 ⁻¹⁰
rs3782473_T	T / C	0.17 (↑)	76%	6.60 x 10 ⁻¹⁰
rs11634895_A	A / A	-0.15 (↓)	62%	7.88 x 10 ⁻¹⁰
rs11021221_A	A / A	0.20 (↑)	12%	1.49 x 10 ⁻⁹
rs17340879_T	T / T	-0.49 (↓)	99%	1.77 x 10 ⁻⁹
rs6669560_T	T / C	0.16 (↑)	25%	2.92 x 10 ⁻⁹
rs2075556_C	C / C	-0.19 (↓)	86%	3.35 x 10 ⁻⁹
rs756878_T	T / C	0.16 (↑)	56%	4.01 x 10 ⁻⁹
rs12948086_T	C / C	0.17 (-)	21%	5.33 x 10 ⁻⁹
rs761276_A	A / G	-0.14 (↓)	48%	8.02 x 10 ⁻⁹
rs76194223_T	C / T	0.26 (↑)	6%	1.75 x 10 ⁻⁸
rs117905623_T	T / T	-0.38 (↓)	98%	1.89 x 10 ⁻⁸

