

## ☆ Keratoconus (McComish, 2019)

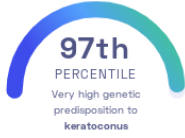
Bennet McComish, et al.  
JAMA Ophthalmology

Eyes

### STUDY SUMMARY

Discovery of 3 genetic regions associated with keratoconus, an eye condition characterized by thinning of the cornea.

### YOUR RESULT



### STUDY DESCRIPTION

The cornea is a thin, clear layer of tissue that covers the front of the eye. Like a car's windshield, the cornea enables you to see clearly while protecting the inner parts of the eye. Keratoconus is a disease characterized by thinning and deformation of the cornea which can lead to blurry vision and nearsightedness. This genome-wide association study sought to discover variants associated with an increased risk of developing keratoconus. By examining the genetic information of nearly 6,000 individuals, researchers identified 3 genetic regions associated with keratoconus.

### DID YOU KNOW?

Long term use of contact lenses can lead to a reduced thickness of the cornea. Limiting the use of contact lenses to a maximum of 14-16 hours per day can help reduce this and other harmful effects.

### 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 0.46**. 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 **97th percentile**. This means that it is higher than the polygenic scores 97% of people. We consider this to be a **very high 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 <sup>Ⓞ</sup>	YOUR GENOTYPE <sup>Ⓞ</sup>	EFFECT SIZE <sup>Ⓞ</sup>	VARIANT FREQUENCY <sup>Ⓞ</sup>	SIGNIFICANCE <sup>Ⓞ</sup>
rs138378_A	G / G	-0.70 (-)	61%	$1.77 \times 10^{-11}$
rs61876744_T	C / C	-0.53 (-)	45%	$7.46 \times 10^{-9}$
rs10831500_G	T / G	0.46 (↑)	33%	$3.91 \times 10^{-7}$