

Health Explorer

DEMO REPORT

Visual

The visual system comprises the sensory organ (the eye) and parts of the central nervous system (the retina containing photoreceptor cells, the optic nerve, the optic tract and the visual cortex) which gives organisms the sense of sight (the ability to detect and process visible light) as well as enabling the formation of several non-image photo response functions. It detects and interprets information from the optical spectrum perceptible to that species to "build a representation" of the surrounding environment.

Disclaimer This report is intended as educational information. It is not intended to provide medical advice or be used solely by the customer in the diagnosis, cure, mitigation, treatment or prevention of disease. If you have any serious medical condition(s), including but not limited to, being over or under weight, or having diabetes or heart disease, you should not make any changes to your diet or exercise without consulting your doctor. Under no circumstances, should you make changes to your medication or other medical care without consulting your physician

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8 entries

Full View

Sort by risk

Sort by name

Age-related macular degeneration

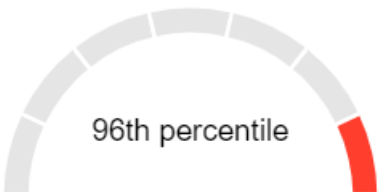
This result is based on **19 genetic variants** associated with "Age-related macular degeneration" analyzed in the scientific paper [\(2013 Apr - Fritsche LG\)](#).



Visual

Your results

Very high genetic predisposition



Your genetic predisposition is higher than the 96% average person from your genetic population

Study description

Age-related macular degeneration (AMD) is a common cause of blindness in older individuals. To accelerate the understanding of AMD biology and help design new therapies, we executed a collaborative genome-wide association study, including >17,100 advanced AMD cases and >60,000 controls of European and Asian ancestry. We identified 19 loci associated at $P < 5 \times 10^{-8}$. These loci show enrichment for genes involved in the regulation of complement activity, lipid metabolism, extracellular matrix remodeling and angiogenesis. Our results include seven loci with associations reaching $P < 5 \times 10^{-8}$ for the first time, near the genes COL8A1-FILIP1L, IER3-DDR1, SLC16A8, TGFBR1, RAD51B, ADAMTS9 and B3GALT1. A genetic risk score combining SNP genotypes from all loci showed similar ability to distinguish cases and controls in all samples examined. Our findings provide new directions for biological, genetic and therapeutic studies of AMD.

Learn more



Amblyopia

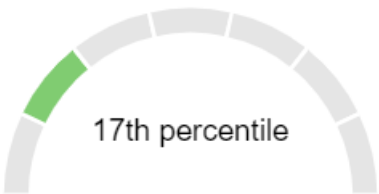
This result is based on **15 genetic variants** associated with "Amblyopia" analyzed in the scientific paper [\(08/01/2018 - Shaaban S\)](#).



Visual

Your results

Low genetic predisposition



Your genetic predisposition is lower than the 83% average person from your genetic population

Study description

To identify genetic variants conferring susceptibility to esotropia. Esotropia is the most common form of comitant strabismus, has its highest incidence in European ancestry populations, and is believed to be inherited as a complex trait.

Learn more



Central corneal thickness

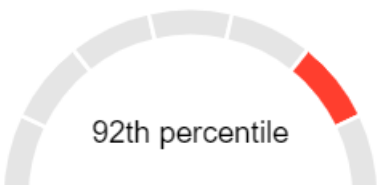
This result is based on **34 genetic variants** associated with "Central corneal thickness" analyzed in the scientific paper [\(11/27/2019 - Bonnemajjer PWM\)](#).



Visual

Your results

High genetic predisposition



Your genetic predisposition is higher than the 92% average person from your genetic population

Study description

A new avenue of mining published genome-wide association studies includes the joint analysis of related traits. The power of this approach depends on the genetic correlation of traits, which reflects the number of pleiotropic loci, i.e. genetic loci influencing multiple traits. Here, we applied new meta-analyses of optic nerve head (ONH) related traits implicated in primary open-angle glaucoma (POAG); intraocular pressure and central corneal thickness using Haplotype reference consortium imputations. We performed a multi-trait analysis of ONH parameters cup area, disc area and vertical cup-disc ratio. We uncover new variants; rs11158547 in PPP1R36-PLEKHG3 and rs1028727 near SERPINE3 at genome-wide significance that replicate in independent Asian cohorts imputed to 1000 Genomes. At this point, validation of these variants in POAG cohorts is hampered by the high degree of heterogeneity. Our results show that multi-trait analysis is a valid approach to identify novel pleiotropic variants for ONH.

Learn more



Corneal astigmatism

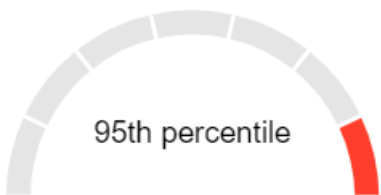
This result is based on 39 genetic variants associated with "Corneal astigmatism" analyzed in the scientific paper [\(2018 Dec - Shah RL\)](#).



Visual

Your results

Very high genetic predisposition



Your genetic predisposition is higher than the 95% average person from your genetic population

Study description

Previous studies have suggested that naturally occurring genetic variation contributes to the risk of astigmatism. The purpose of this investigation was to identify genetic markers associated with corneal and refractive astigmatism in a large-scale European ancestry cohort (UK Biobank) who underwent keratometry and autorefraction at an assessment centre. Genome-wide association studies for corneal and refractive astigmatism were performed in individuals of European ancestry (N = 86,335 and 88,005 respectively), with the mean corneal astigmatism or refractive astigmatism in fellow eyes analysed as a quantitative trait (dependent variable). Genetic correlation between the two traits was calculated using LD Score regression. Gene-based and gene-set tests were carried out using MAGMA. Single marker-based association tests for corneal astigmatism identified four genome-wide significant loci ($P < 5 \times 10^{-8}$) near the genes ZC3H11B (1q41), LINC00340 (6p22.3), HERC2/OCA2 (15q13.1) and NPLOC4/TSPAN10 (17q25.3). Three of these loci also demonstrated genome-wide significant association with refractive astigmatism: LINC00340, HERC2/OCA2 and NPLOC4/TSPAN10. The genetic correlation between corneal and refractive astigmatism was 0.85 (standard error = 0.068, $P = 1.37 \times 10^{-35}$). Here, we have undertaken the largest genome-wide association studies for corneal and refractive astigmatism to date and identified four novel loci for corneal astigmatism, two of which were also novel loci for refractive astigmatism. These loci have previously demonstrated association with axial length (ZC3H11B), myopia (NPLOC4), spherical equivalent refractive error (LINC00340) and eye colour (HERC2). The shared role of these novel candidate genes for astigmatism lends further support to the shared genetic susceptibility of myopia and astigmatism.

Learn more



Glaucoma

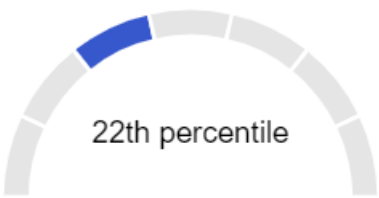
This result is based on **39 genetic variants** associated with "Glaucoma" analyzed in the scientific paper [\(2020 Feb - Craig, JE\)](#).



Visual

Your results

Below average genetic predisposition



Your genetic predisposition is lower than the 78% average person from your genetic population

Study description

Glaucoma, a disease characterized by progressive optic nerve degeneration, can be prevented through timely diagnosis and treatment. We characterize optic nerve photographs of 67,040 UK Biobank participants and use a multitrait genetic model to identify risk loci for glaucoma. A glaucoma polygenic risk score (PRS) enables effective risk stratification in unselected glaucoma cases and modifies penetrance of the MYOC variant encoding p.Gln368Ter, the most common glaucoma-associated myocilin variant. In the unselected glaucoma population, individuals in the top PRS decile reach an absolute risk for glaucoma 10 years earlier than the bottom decile and are at 15-fold increased risk of developing advanced glaucoma (top 10% versus remaining 90%, odds ratio = 4.20). The PRS predicts glaucoma progression in prospectively monitored, early manifest glaucoma cases ($P = 0.004$) and surgical intervention in advanced disease ($P = 3.6 \times 10^{-6}$). This glaucoma PRS will facilitate the development of a personalized approach for earlier treatment of high-risk individuals, with less intensive monitoring and treatment being possible for lower-risk groups.

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High myopia

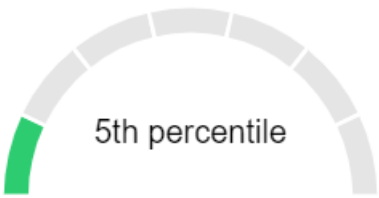
This result is based on **17 genetic variants** associated with "High myopia"



Visual

Your results

Very low genetic predisposition



Your genetic predisposition is lower than the 95% average person from your genetic population



Refractive astigmatism

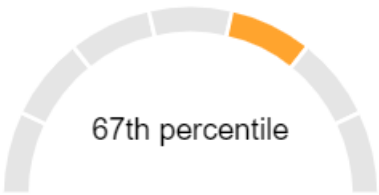
This result is based on **42 genetic variants** associated with "Refractive astigmatism" analyzed in the scientific paper [\(2018 Dec - Shah RL\)](#).



Visual

Your results

Above average genetic predisposition



Your genetic predisposition is higher than the 67% average person from your genetic population

Study description

Previous studies have suggested that naturally occurring genetic variation contributes to the risk of astigmatism. The purpose of this investigation was to identify genetic markers associated with corneal and refractive astigmatism in a large-scale European ancestry cohort (UK Biobank) who underwent keratometry and autorefraction at an assessment centre. Genome-wide association studies for corneal and refractive astigmatism were performed in individuals of European ancestry (N = 86,335 and 88,005 respectively), with the mean corneal astigmatism or refractive astigmatism in fellow eyes analysed as a quantitative trait (dependent variable). Genetic correlation between the two traits was calculated using LD Score regression. Gene-based and gene-set tests were carried out using MAGMA. Single marker-based association tests for corneal astigmatism identified four genome-wide significant loci ($P < 5 \times 10^{-8}$) near the genes ZC3H11B (1q41), LINC00340 (6p22.3), HERC2/OCA2 (15q13.1) and NPLOC4/TSPAN10 (17q25.3). Three of these loci also demonstrated genome-wide significant association with refractive astigmatism: LINC00340, HERC2/OCA2 and NPLOC4/TSPAN10. The genetic correlation between corneal and refractive astigmatism was 0.85 (standard error = 0.068, $P = 1.37 \times 10^{-35}$). Here, we have undertaken the largest genome-wide association studies for corneal and refractive astigmatism to date and identified four novel loci for corneal astigmatism, two of which were also novel loci for refractive astigmatism. These loci have previously demonstrated association with axial length (ZC3H11B), myopia (NPLOC4), spherical equivalent refractive error (LINC00340) and eye colour (HERC2). The shared role of these novel candidate genes for astigmatism lends further support to the shared genetic susceptibility of myopia and astigmatism.

Learn more



Strabismus

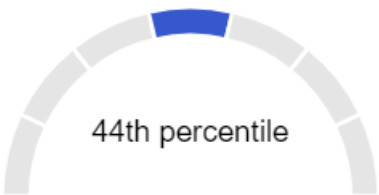
This result is based on **22 genetic variants** associated with "Strabismus" analyzed in the scientific paper [\(2019 Jul - Plotnikov D\)](#).



Visual

Your results

Average genetic predisposition



Your genetic predisposition is lower than the 56% average person from your genetic population

Study description

Strabismus refers to an abnormal alignment of the eyes leading to the loss of central binocular vision. Concomitant strabismus occurs when the angle of deviation is constant in all positions of gaze and often manifests in early childhood when it is considered to be a neurodevelopmental disorder of the visual system. As such, it is inherited as a complex genetic trait, affecting 2-4% of the population. A genome-wide association study (GWAS) for self-reported strabismus (1345 cases and 65,349 controls from UK Biobank) revealed a single genome-wide significant locus on chromosome 17q25. Approximately 20 variants across the NPLOC4-TSPAN10-PDE6G gene cluster and in almost perfect linkage disequilibrium (LD) were most strongly associated (lead variant: rs75078292, OR = 1.26, $p = 2.24 \times 10^{-8}$). A recessive model provided a better fit to the data than an additive model. Association with strabismus was independent of refractive error, and the degree of association with strabismus was minimally attenuated after adjustment for amblyopia. The association with strabismus was replicated in an independent cohort of clinician-diagnosed children aged 7 years old (116 cases and 5084 controls; OR = 1.85, $p = 0.009$). The associated variants included 2 strong candidate causal variants predicted to have functional effects: rs6420484, which substitutes tyrosine for a conserved cysteine (C177Y) in the TSPAN10 gene, and a 4-bp deletion variant, rs397693108, predicted to cause a frameshift in TSPAN10. The population-attributable risk for the locus was approximately 8.4%, indicating an important role in conferring susceptibility to strabismus.

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