# Pirro G Hysi

## List of Publications by Year in Descending Order



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| 178 papers | 10,831 citations | 54 h-index | 101 g-index |
| 208 ext. papers | 13,837 ext. citations | 10.8 avg, IF | 5.41 L-index |

| # | Paper | IF | Citations |
|---|---|---|---|
| 178 | Temporal trends in frequency, type and severity of myopia and associations with key environmental risk factors in the UK: Findings from the UK Biobank Study.. *PLoS ONE*, **2022**, 17, e0260993 | 3.7 | 0 |
| 177 | Associations of Alcohol Consumption and Smoking With Disease Risk and Neurodegeneration in Individuals With Multiple Sclerosis in the United Kingdom.. *JAMA Network Open*, **2022**, 5, e220902 | 10.4 | 0 |
| 176 | Rare variant analysis in eczema identifies exonic variants in DUSP1, NOTCH4 and SLC9A4. *Nature Communications*, **2021**, 12, 6618 | 17.4 | 2 |
| 175 | Macular thickness varies with age-related macular degeneration genetic risk variants in the UK Biobank cohort. *Scientific Reports*, **2021**, 11, 23255 | 4.9 | 2 |
| 174 | Machine Learning Algorithms to Detect Subclinical Keratoconus: Systematic Review.. *JMIR Medical Informatics*, **2021**, 9, e27363 | 3.6 | 0 |
| 173 | At What Age Does Age-Related Macular Degeneration Start?. *JAMA Ophthalmology*, **2021**, 139, 1226-1227 | 3.9 | |
| 172 | Genome-wide association study in almost 195,000 individuals identifies 50 previously unidentified genetic loci for eye color. *Science Advances*, **2021**, 7, | 14.3 | 11 |
| 171 | A multi-ethnic genome-wide association study implicates collagen matrix integrity and cell differentiation pathways in keratoconus. *Communications Biology*, **2021**, 4, 266 | 6.7 | 10 |
| 170 | Genetic variation affects morphological retinal phenotypes extracted from UK Biobank optical coherence tomography images. *PLoS Genetics*, **2021**, 17, e1009497 | 6 | 5 |
| 169 | The genomic loci of specific human tRNA genes exhibit ageing-related DNA hypermethylation. *Nature Communications*, **2021**, 12, 2655 | 17.4 | 3 |
| 168 | A large multiethnic GWAS meta-analysis of cataract identifies new risk loci and sex-specific effects. *Nature Communications*, **2021**, 12, 3595 | 17.4 | 7 |
| 167 | New Polygenic Risk Score to Predict High Myopia in Singapore Chinese Children. *Translational Vision Science and Technology*, **2021**, 10, 26 | 3.3 | 1 |
| 166 | Retinal asymmetry in multiple sclerosis. *Brain*, **2021**, 144, 224-235 | 11.2 | 4 |
| 165 | Testing the impact of trait prevalence priors in Bayesian-based genetic prediction modeling of human appearance traits. *Forensic Science International: Genetics*, **2021**, 50, 102412 | 4.3 | 0 |
| 164 | Looking for Sunshine: Genetic Predisposition to Sun Seeking in 265,000 Individuals of European Ancestry. *Journal of Investigative Dermatology*, **2021**, 141, 779-786 | 4.3 | 2 |
| 163 | Genome-wide meta-analysis identifies 127 open-angle glaucoma loci with consistent effect across ancestries. *Nature Communications*, **2021**, 12, 1258 | 17.4 | 47 |
| 162 | A GWAS in Latin Americans identifies novel face shape loci, implicating VPS13B and a Denisovan introgressed region in facial variation. *Science Advances*, **2021**, 7, | 14.3 | 7 |

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| 161 | Association Between Medication-Taking and Refractive Error in a Large General Population-Based Cohort **2021**, 62, 15 | | 1 |
| 160 | Prevalence of electronegative electroretinograms in a healthy adult cohort. *BMJ Open Ophthalmology*, **2021**, 6, e000751 | 3.2 | 0 |
| 159 | Population screening for glaucoma in UK: current recommendations and future directions. *Eye*, **2021**, | 4.4 | 3 |
| 158 | Characteristics of p.Gln368Ter Myocilin Variant and Influence of Polygenic Risk on Glaucoma Penetrance in the UK Biobank. *Ophthalmology*, **2021**, 128, 1300-1311 | 7.3 | 5 |
| 157 | Genetic Determinants of Intraocular Pressure. *Annual Review of Vision Science*, **2021**, 7, 727-746 | 8.2 | 2 |
| 156 | Real-time tracking of self-reported symptoms to predict potential COVID-19. *Nature Medicine*, **2020**, 26, 1037-1040 | 50.5 | 750 |
| 155 | A multiethnic genome-wide analysis of 44,039 individuals identifies 41 new loci associated with central corneal thickness. *Communications Biology*, **2020**, 3, 301 | 6.7 | 14 |
| 154 | Genome-wide association meta-analysis of corneal curvature identifies novel loci and shared genetic influences across axial length and refractive error. *Communications Biology*, **2020**, 3, 133 | 6.7 | 9 |
| 153 | Meta-analysis of 542,934 subjects of European ancestry identifies new genes and mechanisms predisposing to refractive error and myopia. *Nature Genetics*, **2020**, 52, 401-407 | 36.3 | 68 |
| 152 | Multitrait analysis of glaucoma identifies new risk loci and enables polygenic prediction of disease susceptibility and progression. *Nature Genetics*, **2020**, 52, 160-166 | 36.3 | 78 |
| 151 | Rhegmatogenous Retinal Detachment in the Age of Genomic Medicine. *JAMA Ophthalmology*, **2020**, 138, 678-679 | 3.9 | |
| 150 | Variants associated with HHIP expression have sex-differential effects on lung function. *Wellcome Open Research*, **2020**, 5, 111 | 4.8 | 0 |
| 149 | Comparison of Associations with Different Macular Inner Retinal Thickness Parameters in a Large Cohort: The UK Biobank. *Ophthalmology*, **2020**, 127, 62-71 | 7.3 | 20 |
| 148 | Genome-wide association study of corneal biomechanical properties identifies over 200 loci providing insight into the genetic etiology of ocular diseases. *Human Molecular Genetics*, **2020**, 29, 3154-3164 | 5.6 | 13 |
| 147 | The Role of Chromosome X in Intraocular Pressure Variation and Sex-Specific Effects **2020**, 61, 20 | | 2 |
| 146 | Common variants in SOX-2 and congenital cataract genes contribute to age-related nuclear cataract. *Communications Biology*, **2020**, 3, 755 | 6.7 | 3 |
| 145 | Variants associated with expression have sex-differential effects on lung function. *Wellcome Open Research*, **2020**, 5, 111 | 4.8 | 0 |
| 144 | Association of Pharmacogenetic Markers With Atazanavir Exposure in HIV-Infected Women. *Clinical Pharmacology and Therapeutics*, **2020**, 107, 315-318 | 6.1 | 1 |

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| 143 | Genetic Heritability of Pigmentary Glaucoma and Associations With Other Eye Phenotypes. *JAMA Ophthalmology*, **2020**, 138, 294-299 | 3.9 | 4 |
| 142 | Genome-wide association analysis of 95 549 individuals identifies novel loci and genes influencing optic disc morphology. *Human Molecular Genetics*, **2019**, 28, 3680-3690 | 5.6 | 11 |
| 141 | Associations with Corneal Hysteresis in a Population Cohort: Results from 96 010 UK Biobank Participants. *Ophthalmology*, **2019**, 126, 1500-1510 | 7.3 | 14 |
| 140 | Multiple sclerosis genomic map implicates peripheral immune cells and microglia in susceptibility. *Science*, **2019**, 365, | 33.3 | 309 |
| 139 | Quantile regression analysis reveals widespread evidence for gene-environment or gene-gene interactions in myopia development. *Communications Biology*, **2019**, 2, 167 | 6.7 | 18 |
| 138 | Genetic Correlations Between Diabetes and Glaucoma: An Analysis of Continuous and Dichotomous Phenotypes. *American Journal of Ophthalmology*, **2019**, 206, 245-255 | 4.9 | 6 |
| 137 | In-utero epigenetic factors are associated with early-onset myopia in young children. *PLoS ONE*, **2019**, 14, e0214791 | 3.7 | 11 |
| 136 | A commonly occurring genetic variant within the NPLOC4-TSPAN10-PDE6G gene cluster is associated with the risk of strabismus. *Human Genetics*, **2019**, 138, 723-737 | 6.3 | 16 |
| 135 | Genome-Wide Association Studies Identify Multiple Genetic Loci Influencing Eyebrow Color Variation in Europeans. *Journal of Investigative Dermatology*, **2019**, 139, 1601-1605 | 4.3 | 10 |
| 134 | Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. *American Journal of Human Genetics*, **2019**, 104, 948-956 | 11 | 17 |
| 133 | Twin Registries Moving Forward and Meeting the Future: A Review. *Twin Research and Human Genetics*, **2019**, 22, 201-209 | 2.2 | 2 |
| 132 | Genetic variants linked to myopic macular degeneration in persons with high myopia: CREAM Consortium. *PLoS ONE*, **2019**, 14, e0220143 | 3.7 | 5 |
| 131 | Genetic Variants Associated With Corneal Biomechanical Properties and Potentially Conferring Susceptibility to Keratoconus in a Genome-Wide Association Study. *JAMA Ophthalmology*, **2019**, 137, 1005-1012 | 3.9 | 25 |
| 130 | Novel genetic loci affecting facial shape variation in humans. *ELife*, **2019**, 8, | 8.9 | 22 |
| 129 | Multi-trait genome-wide association study identifies new loci associated with optic disc parameters. *Communications Biology*, **2019**, 2, 435 | 6.7 | 10 |
| 128 | Ascorbic acid metabolites are involved in intraocular pressure control in the general population. *Redox Biology*, **2019**, 20, 349-353 | 11.3 | 14 |
| 127 | Early life factors for myopia in the British Twins Early Development Study. *British Journal of Ophthalmology*, **2019**, 103, 1078-1084 | 5.5 | 18 |
| 126 | Artificial Intelligence Applied to Osteoporosis: A Performance Comparison of Machine Learning Algorithms in Predicting Fragility Fractures From MRI Data. *Journal of Magnetic Resonance Imaging*, **2019**, 49, 1029-1038 | 5.6 | 32 |

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| 125 | Antisense Therapy for a Common Corneal Dystrophy Ameliorates TCF4 Repeat Expansion-Mediated Toxicity. *American Journal of Human Genetics*, **2018**, 102, 528-539 | 11 | 33 |
| 124 | A GWAS meta-analysis from 5 population-based cohorts implicates ion channel genes in the pathogenesis of irritable bowel syndrome. *Neurogastroenterology and Motility*, **2018**, 30, e13358 | 4 | 21 |
| 123 | Genome-wide association meta-analysis of individuals of European ancestry identifies new loci explaining a substantial fraction of hair color variation and heritability. *Nature Genetics*, **2018**, 50, 652-656 | 36.3 | 59 |
| 122 | Genome-wide association study identifies nine novel loci for 2D:4D finger ratio, a putative retrospective biomarker of testosterone exposure in utero. *Human Molecular Genetics*, **2018**, 27, 2025-2038 | 5.6 | 27 |
| 121 | Ensemble landmarking of 3D facial surface scans. *Scientific Reports*, **2018**, 8, 12 | 4.9 | 42 |
| 120 | Retinal thickness measurements in sickle cell patients with HbSS and HbSC genotype. *Canadian Journal of Ophthalmology*, **2018**, 53, 420-424 | 1.4 | 7 |
| 119 | A Genome-Wide Association Study Identifies a Candidate Gene Associated With Atazanavir Exposure Measured in Hair. *Clinical Pharmacology and Therapeutics*, **2018**, 104, 949-956 | 6.1 | 1 |
| 118 | Obligatory and facilitative allelic variation in the DNA methylome within common disease-associated loci. *Nature Communications*, **2018**, 9, 8 | 17.4 | 77 |
| 117 | Genetics of the human face: Identification of large-effect single gene variants. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, E676-E685 | 11.5 | 33 |
| 116 | Meta-analysis of genome-wide association studies identifies 8 novel loci involved in shape variation of human head hair. *Human Molecular Genetics*, **2018**, 27, 559-575 | 5.6 | 33 |
| 115 | Genome-wide association study in 176,678 Europeans reveals genetic loci for tanning response to sun exposure. *Nature Communications*, **2018**, 9, 1684 | 17.4 | 51 |
| 114 | A genome-wide association study of corneal astigmatism: The CREAM Consortium. *Molecular Vision*, **2018**, 24, 127-142 | 2.3 | 5 |
| 113 | Twin studies, genome-wide association studies and myopia genetics. *Annals of Eye Science*, **2018**, 2, 69-69 | 0.9 | 4 |
| 112 | Genome-wide association study of primary open-angle glaucoma in continental and admixed African populations. *Human Genetics*, **2018**, 137, 847-862 | 6.3 | 25 |
| 111 | Low-Frequency and Rare-Coding Variation Contributes to Multiple Sclerosis Risk. *Cell*, **2018**, 175, 1679-1687.e7 | 36.2 | 72 |
| 110 | Genome-wide association meta-analysis highlights light-induced signaling as a driver for refractive error. *Nature Genetics*, **2018**, 50, 834-848 | 36.3 | 135 |
| 109 | Genome-wide analyses identify 68 new loci associated with intraocular pressure and improve risk prediction for primary open-angle glaucoma. *Nature Genetics*, **2018**, 50, 778-782 | 36.3 | 122 |
| 108 | Cross-ancestry genome-wide association analysis of corneal thickness strengthens link between complex and Mendelian eye diseases. *Nature Communications*, **2018**, 9, 1864 | 17.4 | 37 |

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| 107 | A Metabolome-Wide Study of Dry Eye Disease Reveals Serum Androgens as Biomarkers. *Ophthalmology*, **2017**, 124, 505-511 | 7.3 | 26 |
| 106 | New insights into the genetics of primary open-angle glaucoma based on meta-analyses of intraocular pressure and optic disc characteristics. *Human Molecular Genetics*, **2017**, 26, 438-453 | 5.6 | 80 |
| 105 | A GWAS meta-analysis suggests roles for xenobiotic metabolism and ion channel activity in the biology of stool frequency. *Gut*, **2017**, 66, 756-758 | 19.2 | 12 |
| 104 | Phenotypic and genotypic correlation between myopia and intelligence. *Scientific Reports*, **2017**, 7, 45977 | 4.9 | 16 |
| 103 | Haplotype reference consortium panel: Practical implications of imputations with large reference panels. *Human Mutation*, **2017**, 38, 1025-1032 | 4.7 | 20 |
| 102 | Genetically low vitamin D concentrations and myopic refractive error: a Mendelian randomization study. *International Journal of Epidemiology*, **2017**, 46, 1882-1890 | 7.8 | 34 |
| 101 | Heritability maps of human face morphology through large-scale automated three-dimensional phenotyping. *Scientific Reports*, **2017**, 7, 45885 | 4.9 | 32 |
| 100 | Evaluation of the Myocilin Mutation Gln368Stop Demonstrates Reduced Penetrance for Glaucoma in European Populations. *Ophthalmology*, **2017**, 124, 547-553 | 7.3 | 17 |
| 99 | Replication of SNP associations with keratoconus in a Czech cohort. *PLoS ONE*, **2017**, 12, e0172365 | 3.7 | 16 |
| 98 | Genetic correlations between intraocular pressure, blood pressure and primary open-angle glaucoma: a multi-cohort analysis. *European Journal of Human Genetics*, **2017**, 25, 1261-1267 | 5.3 | 9 |
| 97 | Systems genetics identifies a role for Cacna2d1 regulation in elevated intraocular pressure and glaucoma susceptibility. *Nature Communications*, **2017**, 8, 1755 | 17.4 | 21 |
| 96 | A meta-analysis of reflux genome-wide association studies in 6750 Northern Europeans from the general population. *Neurogastroenterology and Motility*, **2017**, 29, e12923 | 4 | 15 |
| 95 | Comparison of HapMap and 1000 Genomes Reference Panels in a Large-Scale Genome-Wide Association Study. *PLoS ONE*, **2017**, 12, e0167742 | 3.7 | 21 |
| 94 | A principal component meta-analysis on multiple anthropometric traits identifies novel loci for body shape. *Nature Communications*, **2016**, 7, 13357 | 17.4 | 46 |
| 93 | Meta-analysis of gene-environment-wide association scans accounting for education level identifies additional loci for refractive error. *Nature Communications*, **2016**, 7, 11008 | 17.4 | 79 |
| 92 | Childhood gene-environment interactions and age-dependent effects of genetic variants associated with refractive error and myopia: The CREAM Consortium. *Scientific Reports*, **2016**, 6, 25853 | 4.9 | 57 |
| 91 | New loci for body fat percentage reveal link between adiposity and cardiometabolic disease risk. *Nature Communications*, **2016**, 7, 10495 | 17.4 | 180 |
| 90 | An Automatic 3D Facial Landmarking Algorithm Using 2D Gabor Wavelets. *IEEE Transactions on Image Processing*, **2016**, 25, 580-8 | 8.7 | 12 |

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| 89 | Genome-wide association analysis identifies TXNRD2, ATXN2 and FOXC1 as susceptibility loci for primary open-angle glaucoma. *Nature Genetics*, **2016**, 48, 189-94 | 36.3 | 159 |
| 88 | A meta-analysis of 120 246 individuals identifies 18 new loci for fibrinogen concentration. *Human Molecular Genetics*, **2016**, 25, 358-70 | 5.6 | 54 |
| 87 | Genetic and Dietary Factors Influencing the Progression of Nuclear Cataract. *Ophthalmology*, **2016**, 123, 1237-44 | 7.3 | 20 |
| 86 | Changes in quality of life shortly after routine cataract surgery. *Canadian Journal of Ophthalmology*, **2016**, 51, 282-287 | 1.4 | 10 |
| 85 | Deep Intronic Sequence Variants in COL2A1 Affect the Alternative Splicing Efficiency of Exon 2, and May Confer a Risk for Rhegmatogenous Retinal Detachment. *Human Mutation*, **2016**, 37, 1085-96 | 4.7 | 9 |
| 84 | Novel regional age-associated DNA methylation changes within human common disease-associated loci. *Genome Biology*, **2016**, 17, 193 | 18.3 | 22 |
| 83 | Big data challenges in bone research: genome-wide association studies and next-generation sequencing. *BoneKEy Reports*, **2015**, 4, 635 | | 9 |
| 82 | Validation of rs2956540:G>C and rs3735520:G>A association with keratoconus in a population of European descent. *European Journal of Human Genetics*, **2015**, 23, 1581-3 | 5.3 | 27 |
| 81 | Directional dominance on stature and cognition in diverse human populations. *Nature*, **2015**, 523, 459-462 | 50.4 | 119 |
| 80 | Increasing Prevalence of Myopia in Europe and the Impact of Education. *Ophthalmology*, **2015**, 122, 1489-97 | 7.3 | 220 |
| 79 | Genetic Factors Influencing Coagulation Factor XIII B-Subunit Contribute to Risk of Ischemic Stroke. *Stroke*, **2015**, 46, 2069-74 | 6.7 | 12 |
| 78 | Genome-wide association study for refractive astigmatism reveals genetic co-determination with spherical equivalent refractive error: the CREAM consortium. *Human Genetics*, **2015**, 134, 131-46 | 6.3 | 20 |
| 77 | Prevalence of vitreomacular interface abnormalities on spectral domain optical coherence tomography of patients undergoing macular photocoagulation for centre involving diabetic macular oedema. *British Journal of Ophthalmology*, **2015**, 99, 1078-81 | 5.5 | 16 |
| 76 | Molecular mechanisms underlying variations in lung function: a systems genetics analysis. *Lancet Respiratory Medicine,the*, **2015**, 3, 782-95 | 35.1 | 52 |
| 75 | Integrative pathway genomics of lung function and airflow obstruction. *Human Molecular Genetics*, **2015**, 24, 6836-48 | 5.6 | 20 |
| 74 | Multi-ancestry genome-wide association study of 21,000 cases and 95,000 controls identifies new risk loci for atopic dermatitis. *Nature Genetics*, **2015**, 47, 1449-1456 | 36.3 | 329 |
| 73 | The UK10K project identifies rare variants in health and disease. *Nature*, **2015**, 526, 82-90 | 50.4 | 776 |
| 72 | The Influence of Age and Sex on Genetic Associations with Adult Body Size and Shape: A Large-Scale Genome-Wide Interaction Study. *PLoS Genetics*, **2015**, 11, e1005378 | 6 | 220 |

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| 71 | Frequency and Distribution of Refractive Error in Adult Life: Methodology and Findings of the UK Biobank Study. *PLoS ONE*, **2015**, 10, e0139780 | 3.7 | 28 |
| 70 | Genetics of skin color variation in Europeans: genome-wide association studies with functional follow-up. *Human Genetics*, **2015**, 134, 823-35 | 6.3 | 97 |
| 69 | Sixteen new lung function signals identified through 1000 Genomes Project reference panel imputation. *Nature Communications*, **2015**, 6, 8658 | 17.4 | 79 |
| 68 | Interocular asymmetries in axial length and refractive error in 4 cohorts. *Ophthalmology*, **2015**, 122, 648-9 | 7.3 | 6 |
| 67 | Meta-analysis of Genome-Wide Association Studies Identifies Novel Loci Associated With Optic Disc Morphology. *Genetic Epidemiology*, **2015**, 39, 207-16 | 2.6 | 58 |
| 66 | APLP2 Regulates Refractive Error and Myopia Development in Mice and Humans. *PLoS Genetics*, **2015**, 11, e1005432 | 6 | 59 |
| 65 | Low copy number of the salivary amylase gene predisposes to obesity. *Nature Genetics*, **2014**, 46, 492-7 | 36.3 | 177 |
| 64 | The pattern and distribution of retinal breaks in eyes with rhegmatogenous retinal detachment. *American Journal of Ophthalmology*, **2014**, 157, 221-226.e1 | 4.9 | 19 |
| 63 | Genome-wide association analysis identifies six new loci associated with forced vital capacity. *Nature Genetics*, **2014**, 46, 669-77 | 36.3 | 104 |
| 62 | Clarifying the role of ATOH7 in glaucoma endophenotypes. *British Journal of Ophthalmology*, **2014**, 98, 562-6 | 5.5 | 11 |
| 61 | Genome-wide analysis of multi-ancestry cohorts identifies new loci influencing intraocular pressure and susceptibility to glaucoma. *Nature Genetics*, **2014**, 46, 1126-1130 | 36.3 | 171 |
| 60 | Outcomes of ptosis surgery assessed using a patient-reported outcome measure: an exploration of time effects. *British Journal of Ophthalmology*, **2014**, 98, 387-90 | 5.5 | 9 |
| 59 | Genome-wide association studies of refractive error and myopia, lessons learned, and implications for the future **2014**, 55, 3344-51 | | 54 |
| 58 | Association of CHRDL1 mutations and variants with X-linked megalocornea, Neuhäuser syndrome and central corneal thickness. *PLoS ONE*, **2014**, 9, e104163 | 3.7 | 20 |
| 57 | Genome-wide meta-analysis of myopia and hyperopia provides evidence for replication of 11 loci. *PLoS ONE*, **2014**, 9, e107110 | 3.7 | 36 |
| 56 | The heritability of dry eye disease in a female twin cohort **2014**, 55, 7278-83 | | 26 |
| 55 | Prevalence and risk factors of dry eye disease in a British female cohort. *British Journal of Ophthalmology*, **2014**, 98, 1712-7 | 5.5 | 123 |
| 54 | Meta-analysis of genome-wide association studies identifies novel loci that influence cupping and the glaucomatous process. *Nature Communications*, **2014**, 5, 4883 | 17.4 | 71 |

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| 53 | Common mechanisms underlying refractive error identified in functional analysis of gene lists from genome-wide association study results in 2 European British cohorts. *JAMA Ophthalmology*, **2014**, 132, 50-6 | 3.9 | 19 |
| 52 | A genome-wide association study of intra-ocular pressure suggests a novel association in the gene FAM125B in the TwinsUK cohort. *Human Molecular Genetics*, **2014**, 23, 3343-8 | 5.6 | 32 |
| 51 | Optic disc planimetry, corneal hysteresis, central corneal thickness, and intraocular pressure as risk factors for glaucoma. *American Journal of Ophthalmology*, **2014**, 157, 441-6 | 4.9 | 24 |
| 50 | Relationship between dry eye symptoms and pain sensitivity. *JAMA Ophthalmology*, **2013**, 131, 1304-8 | 3.9 | 67 |
| 49 | First all-in-one diagnostic tool for DNA intelligence: genome-wide inference of biogeographic ancestry, appearance, relatedness, and sex with the Identitas v1 Forensic Chip. *International Journal of Legal Medicine*, **2013**, 127, 559-72 | 3.1 | 38 |
| 48 | Candidate gene study of macular response to supplemental lutein and zeaxanthin. *Experimental Eye Research*, **2013**, 115, 172-7 | 3.7 | 23 |
| 47 | Germline genetic contributions to risk for esophageal adenocarcinoma, Barrett's esophagus, and gastroesophageal reflux. *Journal of the National Cancer Institute*, **2013**, 105, 1711-8 | 9.7 | 75 |
| 46 | Ischemic stroke is associated with the ABO locus: the EuroCLOT study. *Annals of Neurology*, **2013**, 73, 16-31 | 9.4 | 105 |
| 45 | Nine loci for ocular axial length identified through genome-wide association studies, including shared loci with refractive error. *American Journal of Human Genetics*, **2013**, 93, 264-77 | 11 | 116 |
| 44 | Genetic variants associated with idiopathic pulmonary fibrosis susceptibility and mortality: a genome-wide association study. *Lancet Respiratory Medicine,the*, **2013**, 1, 309-317 | 35.1 | 341 |
| 43 | Genome-wide meta-analyses of multiancestry cohorts identify multiple new susceptibility loci for refractive error and myopia. *Nature Genetics*, **2013**, 45, 314-8 | 36.3 | 314 |
| 42 | Focusing in on the complex genetics of myopia. *PLoS Genetics*, **2013**, 9, e1003442 | 6 | 45 |
| 41 | Genome-wide association study of intraocular pressure identifies the GLCCI1/ICA1 region as a glaucoma susceptibility locus. *Human Molecular Genetics*, **2013**, 22, 4653-60 | 5.6 | 24 |
| 40 | Macular displacement following rhegmatogenous retinal detachment repair. *British Journal of Ophthalmology*, **2013**, 97, 1297-302 | 5.5 | 35 |
| 39 | Genome-wide association analyses identify multiple loci associated with central corneal thickness and keratoconus. *Nature Genetics*, **2013**, 45, 155-63 | 36.3 | 222 |
| 38 | Age of myopia onset in a British population-based twin cohort. *Ophthalmic and Physiological Optics*, **2013**, 33, 339-45 | 4.1 | 22 |
| 37 | Association mapping of the high-grade myopia MYP3 locus reveals novel candidates UHRF1BP1L, PTPRR, and PPFIA2 **2013**, 54, 2076-86 | | 21 |
| 36 | Identification of a candidate gene for astigmatism **2013**, 54, 1260-7 | | 23 |

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| 35 | Copy number variation at chromosome 5q21.2 is associated with intraocular pressure **2013**, 54, 3607-12 | | 10 |
| 34 | Large scale international replication and meta-analysis study confirms association of the 15q14 locus with myopia. The CREAM consortium. *Human Genetics*, **2012**, 131, 1467-80 | 6.3 | 57 |
| 33 | Common genetic determinants of intraocular pressure and primary open-angle glaucoma. *PLoS Genetics*, **2012**, 8, e1002611 | 6 | 131 |
| 32 | Genome-wide joint meta-analysis of SNP and SNP-by-smoking interaction identifies novel loci for pulmonary function. *PLoS Genetics*, **2012**, 8, e1003098 | 6 | 108 |
| 31 | Common polymorphisms in the SERPINI2 gene are associated with refractive error in the 1958 British Birth Cohort **2012**, 53, 440-7 | | 3 |
| 30 | A genome-wide association study identifies five loci influencing facial morphology in Europeans. *PLoS Genetics*, **2012**, 8, e1002932 | 6 | 194 |
| 29 | Validation of printed and computerised crowded Kay picture logMAR tests against gold standard ETDRS acuity test chart measurements in adult and amblyopic paediatric subjects. *Eye*, **2012**, 26, 593-600 | 4.4 | 26 |
| 28 | A genome-wide association study of female sexual dysfunction. *PLoS ONE*, **2012**, 7, e35041 | 3.7 | 15 |
| 27 | Meta-analysis of genome-wide association studies identifies three new risk loci for atopic dermatitis. *Nature Genetics*, **2011**, 44, 187-92 | 36.3 | 244 |
| 26 | Genome-wide association and large-scale follow up identifies 16 new loci influencing lung function. *Nature Genetics*, **2011**, 43, 1082-90 | 36.3 | 313 |
| 25 | A genome-wide association study identifies a susceptibility locus for refractive errors and myopia at 15q14. *Nature Genetics*, **2010**, 42, 897-901 | 36.3 | 181 |
| 24 | A genome-wide association study for myopia and refractive error identifies a susceptibility locus at 15q25. *Nature Genetics*, **2010**, 42, 902-5 | 36.3 | 179 |
| 23 | Common genetic variants near the Brittle Cornea Syndrome locus ZNF469 influence the blinding disease risk factor central corneal thickness. *PLoS Genetics*, **2010**, 6, e1000947 | 6 | 106 |
| 22 | Genome-wide association identifies ATOH7 as a major gene determining human optic disc size. *Human Molecular Genetics*, **2010**, 19, 2716-24 | 5.6 | 118 |
| 21 | A genome-wide association study of optic disc parameters. *PLoS Genetics*, **2010**, 6, e1000978 | 6 | 157 |
| 20 | Genome-wide association study identifies two novel regions at 11p15.5-p13 and 1p31 with major impact on acute-phase serum amyloid A. *PLoS Genetics*, **2010**, 6, e1001213 | 6 | 20 |
| 19 | Digital quantification of human eye color highlights genetic association of three new loci. *PLoS Genetics*, **2010**, 6, e1000934 | 6 | 135 |
| 18 | Evidence for keratoconus susceptibility locus on chromosome 14: a genome-wide linkage screen using single-nucleotide polymorphism markers. *JAMA Ophthalmology*, **2010**, 128, 1191-5 | | 38 |

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| 17 | Common variants in interleukin-1-Beta gene are associated with intracranial hemorrhage and susceptibility to brain arteriovenous malformation. *Cerebrovascular Diseases*, **2009**, 27, 176-82 | 3.2 | 72 |
| 16 | Integrating genomic and clinical medicine: searching for susceptibility genes in complex lung diseases. *Translational Research*, **2008**, 151, 181-93 | 11 | 11 |
| 15 | Sequencing of the CHST6 gene in Czech macular corneal dystrophy patients supports the evidence of a founder mutation. *British Journal of Ophthalmology*, **2008**, 92, 265-7 | 5.5 | 12 |
| 14 | Population stratification in a case-control study of brain arteriovenous malformation in Latinos. *Neuroepidemiology*, **2008**, 31, 224-8 | 5.4 | 14 |
| 13 | Arteriovenous malformation. *Journal of Neurosurgery*, **2007**, 106, 731-2; author reply 732-3 | 3.2 | 8 |
| 12 | The Roles of PAX6 and SOX2 in Myopia: lessons from the 1958 British Birth Cohort. *Investigative Ophthalmology and Visual Science*, **2007**, 48, 4421-5 | | 31 |
| 11 | Molecular analysis of the VSX1 gene in familial keratoconus. *Molecular Vision*, **2007**, 13, 1887-91 | 2.3 | 35 |
| 10 | Study of p.N247S KERA mutation in a British family with cornea plana. *Molecular Vision*, **2007**, 13, 1339-47 | 2.3 | 16 |
| 9 | TUCAN (CARD8) genetic variants and inflammatory bowel disease. *Gastroenterology*, **2006**, 131, 1190-6 | 13.3 | 84 |
| 8 | Association between a complex insertion/deletion polymorphism in NOD1 (CARD4) and susceptibility to inflammatory bowel disease. *Human Molecular Genetics*, **2005**, 14, 1245-50 | 5.6 | 260 |
| 7 | Haplotypes and asthma. *American Journal of Respiratory and Critical Care Medicine*, **2005**, 171, 1066-7 | 10.2 | 1 |
| 6 | NOD1 variation, immunoglobulin E and asthma. *Human Molecular Genetics*, **2005**, 14, 935-41 | 5.6 | 219 |
| 5 | LD mapping of maternally and non-maternally derived alleles and atopy in FcepsilonRI-beta. *Human Molecular Genetics*, **2003**, 12, 2577-85 | 5.6 | 40 |
| 4 | Real-time tracking of self-reported symptoms to predict potential COVID-19 | | 1 |
| 3 | A large cross-ancestry meta-analysis of genome-wide association studies identifies 69 novel risk loci for primary open-angle glaucoma and includes a genetic link with Alzheimer's disease | | 4 |
| 2 | Loss of smell and taste in combination with other symptoms is a strong predictor of COVID-19 infection | | 74 |
| 1 | A multiethnic GWAS meta-analysis of 585,243 individuals identifies new risk loci associated with cataract and reveals sex-specific effects | | 1 |