Kathryn P Burdon

List of Publications by Citations

186 papers	7,783 citations	47 h-index	82 g-index
222 ext. papers	9,252 ext. citations	7.5 avg, IF	5.15 L-index

#	Paper	IF	Citations
186	A large genome-wide association study of age-related macular degeneration highlights contributions of rare and common variants. *Nature Genetics*, **2016**, 48, 134-43	36.3	769
185	Genome-wide association study identifies susceptibility loci for open angle glaucoma at TMCO1 and CDKN2B-AS1. *Nature Genetics*, **2011**, 43, 574-8	36.3	329
184	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
183	Common variants near CAV1 and CAV2 are associated with primary open-angle glaucoma. *Nature Genetics*, **2010**, 42, 906-9	36.3	303
182	Genome-wide association analyses identify multiple loci associated with central corneal thickness and keratoconus. *Nature Genetics*, **2013**, 45, 155-63	36.3	222
181	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
180	Genome-wide meta-analysis identifies five new susceptibility loci for cutaneous malignant melanoma. *Nature Genetics*, **2015**, 47, 987-995	36.3	162
179	A systematic meta-analysis of genetic association studies for diabetic retinopathy. *Diabetes*, **2009**, 58, 2137-47	0.9	160
178	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
177	Common variants near ABCA1, AFAP1 and GMDS confer risk of primary open-angle glaucoma. *Nature Genetics*, **2014**, 46, 1120-1125	36.3	141
176	Ancestral LOXL1 variants are associated with pseudoexfoliation in Caucasian Australians but with markedly lower penetrance than in Nordic people. *Human Molecular Genetics*, **2008**, 17, 710-6	5.6	129
175	Angiopoietin receptor TEK mutations underlie primary congenital glaucoma with variable expressivity. *Journal of Clinical Investigation*, **2016**, 126, 2575-87	15.9	117
174	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
173	Genome-wide association study identifies five new susceptibility loci for primary angle closure glaucoma. *Nature Genetics*, **2016**, 48, 556-62	36.3	109
172	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
171	Association of polymorphisms in the hepatocyte growth factor gene promoter with keratoconus **2011**, 52, 8514-9		101
170	Mutations in a novel gene, NHS, cause the pleiotropic effects of Nance-Horan syndrome, including severe congenital cataract, dental anomalies, and mental retardation. *American Journal of Human Genetics*, **2003**, 73, 1120-30	11	92

169	Mutations of the EPHA2 receptor tyrosine kinase gene cause autosomal dominant congenital cataract. *Human Mutation*, **2009**, 30, E603-11	4.7	87
168	Genome-wide association study of intraocular pressure uncovers new pathways to glaucoma. *Nature Genetics*, **2018**, 50, 1067-1071	36.3	86
167	A common variant near TGFBR3 is associated with primary open angle glaucoma. *Human Molecular Genetics*, **2015**, 24, 3880-92	5.6	84
166	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
165	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
164	Insights into keratoconus from a genetic perspective. *Australasian journal of optometry, The*, **2013**, 96, 146-54	2.7	79
163	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
162	The genetics of central corneal thickness. *British Journal of Ophthalmology*, **2010**, 94, 971-6	5.5	76
161	Genetic association study of exfoliation syndrome identifies a protective rare variant at LOXL1 and five new susceptibility loci. *Nature Genetics*, **2017**, 49, 993-1004	36.3	72
160	Genome-wide association study identifies seven novel susceptibility loci for primary open-angle glaucoma. *Human Molecular Genetics*, **2018**, 27, 1486-1496	5.6	72
159	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
158	A common variant mapping to CACNA1A is associated with susceptibility to exfoliation syndrome. *Nature Genetics*, **2015**, 47, 387-92	36.3	70
157	Functional and structural implications of the complement factor H Y402H polymorphism associated with age-related macular degeneration. *Investigative Ophthalmology and Visual Science*, **2008**, 49, 1763-70		70
156	Mutations in the NDP gene: contribution to Norrie disease, familial exudative vitreoretinopathy and retinopathy of prematurity. *Clinical and Experimental Ophthalmology*, **2006**, 34, 682-8	2.4	67
155	Genome-wide association study identifies three novel loci in Fuchs endothelial corneal dystrophy. *Nature Communications*, **2017**, 8, 14898	17.4	66
154	Identification of LOXL1 protein and Apolipoprotein E as components of surgically isolated pseudoexfoliation material by direct mass spectrometry. *Experimental Eye Research*, **2009**, 89, 479-85	3.7	65
153	Glaucoma risk alleles at CDKN2B-AS1 are associated with lower intraocular pressure, normal-tension glaucoma, and advanced glaucoma. *Ophthalmology*, **2012**, 119, 1539-45	7.3	64
152	Apparent autosomal dominant keratoconus in a large Australian pedigree accounted for by digenic inheritance of two novel loci. *Human Genetics*, **2008**, 124, 379-86	6.3	63

151	ARHGEF12 influences the risk of glaucoma by increasing intraocular pressure. *Human Molecular Genetics*, **2015**, 24, 2689-99	5.6	62
150	Mutational analysis of MIR184 in sporadic keratoconus and myopia **2013**, 54, 5266-72		61
149	Genome-wide association study for sight-threatening diabetic retinopathy reveals association with genetic variation near the GRB2 gene. *Diabetologia*, **2015**, 58, 2288-97	10.3	60
148	Meta-analysis of Genome-Wide Association Studies Identifies Novel Loci Associated With Optic Disc Morphology. *Genetic Epidemiology*, **2015**, 39, 207-16	2.6	58
147	Common sequence variation in the VEGFA gene predicts risk of diabetic retinopathy **2009**, 50, 5552-8		55
146	Copy number variations of TBK1 in Australian patients with primary open-angle glaucoma. *American Journal of Ophthalmology*, **2015**, 159, 124-30.e1	4.9	54
145	Homozygous mutations in PXDN cause congenital cataract, corneal opacity, and developmental glaucoma. *American Journal of Human Genetics*, **2011**, 89, 464-73	11	54
144	Association of TCF4 and CLU polymorphisms with FuchsTendothelial dystrophy and implication of CLU and TGFBI proteins in the disease process. *European Journal of Human Genetics*, **2012**, 20, 632-8	5.3	51
143	Genetic analysis of the soluble epoxide hydrolase gene, EPHX2, in subclinical cardiovascular disease in the Diabetes Heart Study. *Diabetes and Vascular Disease Research*, **2008**, 5, 128-34	3.3	51
142	WNT10A exonic variant increases the risk of keratoconus by decreasing corneal thickness. *Human Molecular Genetics*, **2015**, 24, 5060-8	5.6	50
141	A single-nucleotide polymorphism in the MicroRNA-146a gene is associated with diabetic nephropathy and sight-threatening diabetic retinopathy in Caucasian patients. *Acta Diabetologica*, **2016**, 53, 643-50	3.9	49
140	Measurement of Systemic Mitochondrial Function in Advanced Primary Open-Angle Glaucoma and Leber Hereditary Optic Neuropathy. *PLoS ONE*, **2015**, 10, e0140919	3.7	47
139	Sequence variation in DDAH1 and DDAH2 genes is strongly and additively associated with serum ADMA concentrations in individuals with type 2 diabetes. *PLoS ONE*, **2010**, 5, e9462	3.7	47
138	Australian and New Zealand Registry of Advanced Glaucoma: methodology and recruitment. *Clinical and Experimental Ophthalmology*, **2012**, 40, 569-75	2.4	45
137	Association between erythropoietin gene polymorphisms and diabetic retinopathy. *JAMA Ophthalmology*, **2010**, 128, 102-6		45
136	A novel mutation in the Connexin 46 gene causes autosomal dominant congenital cataract with incomplete penetrance. *Journal of Medical Genetics*, **2004**, 41, e106	5.8	45
135	Diabetic retinopathy is associated with elevated serum asymmetric and symmetric dimethylarginines. *Diabetes Care*, **2009**, 32, 2084-6	14.6	42
134	T-786C polymorphism of the endothelial nitric oxide synthase gene is associated with albuminuria in the diabetes heart study. *Journal of the American Society of Nephrology: JASN*, **2005**, 16, 1085-90	12.7	42

133	Mutation in TMEM98 in a large white kindred with autosomal dominant nanophthalmos linked to 17p12-q12. *JAMA Ophthalmology*, **2014**, 132, 970-7	3.9	40
132	Heritability of central corneal thickness in nuclear families **2009**, 50, 4087-90		40
131	Genome-wide association meta-analyses combining multiple risk phenotypes provide insights into the genetic architecture of cutaneous melanoma susceptibility. *Nature Genetics*, **2020**, 52, 494-504	36.3	39
130	Association of alpha2-Heremans-Schmid glycoprotein polymorphisms with subclinical atherosclerosis. *Journal of Clinical Endocrinology and Metabolism*, **2007**, 92, 345-52	5.6	39
129	Serum selenium status in GravesTdisease with and without orbitopathy: a case-control study. *Clinical Endocrinology*, **2014**, 80, 905-10	3.4	38
128	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
127	Higher prevalence of myocilin mutations in advanced glaucoma in comparison with less advanced disease in an Australasian disease registry. *Ophthalmology*, **2013**, 120, 1135-43	7.3	36
126	A genome-wide association study suggests new evidence for an association of the NADPH Oxidase 4 (NOX4) gene with severe diabetic retinopathy in type 2 diabetes. *Acta Ophthalmologica*, **2018**, 96, e811-e819	3.7	36
125	Rapid inexpensive genome-wide association using pooled whole blood. *Genome Research*, **2009**, 19, 2075-80	9.7	35
124	Replication and meta-analysis of candidate loci identified variation at RAB3GAP1 associated with keratoconus **2013**, 54, 5132-5		34
123	Matrix metalloproteinase-9 genetic variation and primary angle closure glaucoma in a Caucasian population. *Molecular Vision*, **2011**, 17, 1420-4	2.3	34
122	Association of genetic variants in the TMCO1 gene with clinical parameters related to glaucoma and characterization of the protein in the eye **2012**, 53, 4917-25		33
121	Elevation of serum asymmetrical and symmetrical dimethylarginine in patients with advanced glaucoma **2012**, 53, 1923-7		33
120	Variants of the CD40 gene but not of the CD40L gene are associated with coronary artery calcification in the Diabetes Heart Study (DHS). *American Heart Journal*, **2006**, 151, 706-11	4.9	33
119	Variability of serum soluble intercellular adhesion molecule-1 measurements attributable to a common polymorphism. *Clinical Chemistry*, **2004**, 50, 2185-7	5.5	33
118	Genome-wide association studies for diabetic macular edema and proliferative diabetic retinopathy. *BMC Medical Genetics*, **2018**, 19, 71	2.1	32
117	Mutations in the EPHA2 gene are a major contributor to inherited cataracts in South-Eastern Australia. *PLoS ONE*, **2013**, 8, e72518	3.7	31
116	Heritability and expression of C-reactive protein in type 2 diabetes in the Diabetes Heart Study. *Annals of Human Genetics*, **2006**, 70, 717-25	2.2	31

115	Multiethnic Genome-Wide Association Study of Diabetic Retinopathy Using Liability Threshold Modeling of Duration of Diabetes and Glycemic Control. *Diabetes*, **2019**, 68, 441-456	0.9	31
114	Aldose reductase gene polymorphisms and diabetic retinopathy susceptibility. *Diabetes Care*, **2010**, 33, 1834-6	14.6	30
113	MALDI-MS-imaging of whole human lens capsule. *Journal of Proteome Research*, **2011**, 10, 3522-9	5.6	30
112	Novel quantitative trait loci for central corneal thickness identified by candidate gene analysis of osteogenesis imperfecta genes. *Human Genetics*, **2010**, 127, 33-44	6.3	30
111	Identification of podocin (NPHS2) gene mutations in African Americans with nondiabetic end-stage renal disease. *Kidney International*, **2005**, 68, 256-62	9.9	30
110	Chromosome 9p21 primary open-angle glaucoma susceptibility locus: a review. *Clinical and Experimental Ophthalmology*, **2014**, 42, 25-32	2.4	29
109	Association of genetic variants with primary angle closure glaucoma in two different populations. *PLoS ONE*, **2013**, 8, e67903	3.7	29
108	Human lipoxygenase pathway gene variation and association with markers of subclinical atherosclerosis in the diabetes heart study. *Mediators of Inflammation*, **2010**, 2010, 170153	4.3	29
107	The PITX3 gene in posterior polar congenital cataract in Australia. *Molecular Vision*, **2006**, 12, 367-71	2.3	29
106	Prenatal detection of congenital bilateral cataract leading to the diagnosis of Nance-Horan syndrome in the extended family. *Prenatal Diagnosis*, **2007**, 27, 662-4	3.2	28
105	Glaucoma spectrum and age-related prevalence of individuals with FOXC1 and PITX2 variants. *European Journal of Human Genetics*, **2017**, 25, 839-847	5.3	25
104	Association of open-angle glaucoma loci with incident glaucoma in the Blue Mountains Eye Study. *American Journal of Ophthalmology*, **2015**, 159, 31-6.e1	4.9	25
103	Analysis combining correlated glaucoma traits identifies five new risk loci for open-angle glaucoma. *Scientific Reports*, **2018**, 8, 3124	4.9	25
102	Genetic investigation into the endophenotypic status of central corneal thickness and optic disc parameters in relation to open-angle glaucoma. *American Journal of Ophthalmology*, **2012**, 154, 833-842.e2	4.9	25
101	Genetic analysis of the clusterin gene in pseudoexfoliation syndrome. *Molecular Vision*, **2008**, 14, 1727-36	2.3	25
100	Association of eNOS polymorphisms with primary angle-closure glaucoma **2013**, 54, 2108-14		24
99	Novel causative mutations in patients with Nance-Horan syndrome and altered localization of the mutant NHS-A protein isoform. *Molecular Vision*, **2008**, 14, 1856-64	2.3	24
98	Association of Genetic Variation With Keratoconus. *JAMA Ophthalmology*, **2020**, 138, 174-181	3.9	24

97	A novel deletion in the FTL gene causes hereditary hyperferritinemia cataract syndrome (HHCS) by alteration of the transcription start site. *Human Mutation*, **2007**, 28, 742	4.7	23
96	The association of hepatocyte growth factor (HGF) gene with primary angle closure glaucoma in the Nepalese population. *Molecular Vision*, **2011**, 17, 2248-54	2.3	23
95	Genetic Association at the 9p21 Glaucoma Locus Contributes to Sex Bias in Normal-Tension Glaucoma **2016**, 57, 3416-21		23
94	Macular Ganglion Cell-Inner Plexiform Layer Loss Precedes Peripapillary Retinal Nerve Fiber Layer Loss in Glaucoma with Lower Intraocular Pressure. *Ophthalmology*, **2019**, 126, 1119-1130	7.3	21
93	Identification of a novel oligomerization disrupting mutation in CRYAA associated with congenital cataract in a South Australian family. *Human Mutation*, **2013**, 34, 435-8	4.7	21
92	Common Sequence Variation in the VEGFC Gene Is Associated with Diabetic Retinopathy and Diabetic Macular Edema. *Ophthalmology*, **2015**, 122, 1828-36	7.3	18
91	Biallelic CPAMD8 Variants Are a Frequent Cause of Childhood and Juvenile Open-Angle Glaucoma. *Ophthalmology*, **2020**, 127, 758-766	7.3	18
90	GWAS study using DNA pooling strategy identifies association of variant rs4910623 in OR52B4 gene with anti-VEGF treatment response in age-related macular degeneration. *Scientific Reports*, **2016**, 6, 37924	4.9	18
89	Myocilin Predictive Genetic Testing for Primary Open-Angle Glaucoma Leads to Early Identification of At-Risk Individuals. *Ophthalmology*, **2017**, 124, 303-309	7.3	17
88	Assessment of polygenic effects links primary open-angle glaucoma and age-related macular degeneration. *Scientific Reports*, **2016**, 6, 26885	4.9	17
87	Severe prekallikrein deficiency associated with homozygosity for an Arg94Stop nonsense mutation. *British Journal of Haematology*, **2004**, 127, 220-3	4.5	17
86	The telomere of human chromosome 1p contains at least two independent autosomal dominant congenital cataract genes. *British Journal of Ophthalmology*, **2005**, 89, 831-4	5.5	17
85	Prevalence of FOXC1 Variants in Individuals With a Suspected Diagnosis of Primary Congenital Glaucoma. *JAMA Ophthalmology*, **2019**, 137, 348-355	3.9	16
84	Association of protein tyrosine phosphatase-N1 polymorphisms with coronary calcified plaque in the Diabetes Heart Study. *Diabetes*, **2006**, 55, 651-8	0.9	16
83	Biological effect of LOXL1 coding variants associated with pseudoexfoliation syndrome. *Experimental Eye Research*, **2016**, 146, 212-223	3.7	16
82	Genetic and Environmental Risk Factors for Keratoconus. *Annual Review of Vision Science*, **2020**, 6, 25-46	8.2	16
81	The Association Between Vitamin D and Multiple Sclerosis Risk: 1,25(OH)D Induces Super-Enhancers Bound by VDR. *Frontiers in Immunology*, **2019**, 10, 488	8.4	15
80	Occurrence of CYP1B1 Mutations in Juvenile Open-Angle Glaucoma With Advanced Visual Field Loss. *JAMA Ophthalmology*, **2015**, 133, 826-33	3.9	15

79	TGC repeat expansion in the TCF4 gene increases the risk of FuchsTendothelial corneal dystrophy in Australian cases. *PLoS ONE*, **2017**, 12, e0183719	3.7	15
78	Genome-wide association studies in the hunt for genes causing primary open-angle glaucoma: a review. *Clinical and Experimental Ophthalmology*, **2012**, 40, 358-63	2.4	15
77	Accurate Imputation-Based Screening of Gln368Ter Myocilin Variant in Primary Open-Angle Glaucoma **2015**, 56, 5087-93		15
76	Review of the prevalence of diabetic retinopathy in Indigenous Australians. *Clinical and Experimental Ophthalmology*, **2014**, 42, 875-82	2.4	15
75	Association of arachidonate 12-lipoxygenase genotype variation and glycemic control with albuminuria in type 2 diabetes. *American Journal of Kidney Diseases*, **2008**, 52, 242-50	7.4	15
74	Association of genes of lipid metabolism with measures of subclinical cardiovascular disease in the Diabetes Heart Study. *Journal of Medical Genetics*, **2005**, 42, 720-4	5.8	15
73	EPHA2 MUTATIONS CONTRIBUTE TO CONGENITAL CATARACT THROUGH DIVERSE MECHANISMS. *Molecular Vision*, **2016**, 22, 18-30	2.3	15
72	Rare, potentially pathogenic variants in 21 keratoconus candidate genes are not enriched in cases in a large Australian cohort of European descent. *PLoS ONE*, **2018**, 13, e0199178	3.7	14
71	A Turkish family with Nance-Horan Syndrome due to a novel mutation. *Gene*, **2013**, 525, 141-5	3.8	14
70	Differential Gene Expression Profiling of Orbital Adipose Tissue in Thyroid Orbitopathy **2015**, 56, 6438-47		14
69	Ethnic and mouse strain differences in central corneal thickness and association with pigmentation phenotype. *PLoS ONE*, **2011**, 6, e22103	3.7	14
68	Myocilin Gene Gln368Ter Variant Penetrance and Association With Glaucoma in Population-Based and Registry-Based Studies. *JAMA Ophthalmology*, **2019**, 137, 28-35	3.9	14
67	Genetically Determined Plasma Lipid Levels and Risk of Diabetic Retinopathy: A Mendelian Randomization Study. *Diabetes*, **2017**, 66, 3130-3141	0.9	13
66	High-Throughput Genetic Screening of 51 Pediatric Cataract Genes Identifies Causative Mutations in Inherited Pediatric Cataract in South Eastern Australia. *G3: Genes, Genomes, Genetics*, **2017**, 7, 3257-3268	2.2	13
65	Novel missense mutation in the bZIP transcription factor, MAF, associated with congenital cataract, developmental delay, seizures and hearing loss (Aym□Gripp syndrome). *BMC Medical Genetics*, **2017**, 18, 52	2.1	13
64	Rare, Potentially Pathogenic Variants in ZNF469 Are Not Enriched in Keratoconus in a Large Australian Cohort of European Descent **2017**, 58, 6248-6256		12
63	Candidate gene study to investigate the genetic determinants of normal variation in central corneal thickness. *Molecular Vision*, **2010**, 16, 562-9	2.3	12
62	Predictive genetic testing experience for myocilin primary open-angle glaucoma using the Australian and New Zealand Registry of Advanced Glaucoma. *Genetics in Medicine*, **2014**, 16, 558-63	8.1	11

61	Genetic study of diabetic retinopathy: recruitment methodology and analysis of baseline characteristics. *Clinical and Experimental Ophthalmology*, **2014**, 42, 486-93	2.4	11
60	Contribution of Mutations in Known Mendelian Glaucoma Genes to Advanced Early-Onset Primary Open-Angle Glaucoma **2017**, 58, 1537-1544		11
59	Recurrent mutation in the crystallin alpha A gene associated with inherited paediatric cataract. *BMC Research Notes*, **2016**, 9, 83	2.3	11
58	Partial duplication of the CRYBB1-CRYBA4 locus is associated with autosomal dominant congenital cataract. *European Journal of Human Genetics*, **2017**, 25, 711-718	5.3	10
57	Key challenges in bringing CRISPR-mediated somatic cell therapy into the clinic. *Genome Medicine*, **2017**, 9, 85	14.4	10
56	DNA methylation at the 9p21 glaucoma susceptibility locus is associated with normal-tension glaucoma. *Ophthalmic Genetics*, **2018**, 39, 221-227	1.2	10
55	Ocular expression and distribution of products of the POAG-associated chromosome 9p21 gene region. *PLoS ONE*, **2013**, 8, e75067	3.7	10
54	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
53	Identification of novel mutations causing pediatric cataract in Bhutan, Cambodia, and Sri Lanka. *Molecular Genetics & Genomic Medicine*, **2018**, 6, 555	2.3	10
52	Diabetic macular oedema: clinical risk factors and emerging genetic influences. *Australasian journal of optometry, The*, **2017**, 100, 569-576	2.7	9
51	MicroRNA-Related Genetic Variants Are Associated With Diabetic Retinopathy in Type 1 Diabetes Mellitus **2019**, 60, 3937-3942		9
50	Testosterone Pathway Genetic Polymorphisms in Relation to Primary Open-Angle Glaucoma: An Analysis in Two Large Datasets **2018**, 59, 629-636		9
49	Relationship between DDAH gene variants and serum ADMA level in individuals with type 1 diabetes. *Journal of Diabetes and Its Complications*, **2012**, 26, 195-8	3.2	9
48	Predictive genetic testing in minors for Myocilin juvenile onset open angle glaucoma. *Clinical Genetics*, **2015**, 88, 584-8	4	9
47	A novel locus for X-linked congenital cataract on Xq24. *Molecular Vision*, **2008**, 14, 721-6	2.3	9
46	A novel de novo Myocilin variant in a patient with sporadic juvenile open angle glaucoma. *BMC Medical Genetics*, **2016**, 17, 30	2.1	8
45	P-selectin gene haplotype associations with albuminuria in the Diabetes Heart Study. *Kidney International*, **2005**, 68, 741-6	9.9	8
44	Whole exome sequencing implicates eye development, the unfolded protein response and plasma membrane homeostasis in primary open-angle glaucoma. *PLoS ONE*, **2017**, 12, e0172427	3.7	8

43	Tag SNPs detect association of the CYP1B1 gene with primary open angle glaucoma. *Molecular Vision*, **2010**, 16, 2286-93	2.3	8
42	Ethical Considerations for the Return of Incidental Findings in Ophthalmic Genomic Research. *Translational Vision Science and Technology*, **2016**, 5, 3	3.3	8
41	The genetic and clinical landscape of nanophthalmos and posterior microphthalmos in an Australian cohort. *Clinical Genetics*, **2020**, 97, 764-769	4	7
40	Pooled genome wide association detects association upstream of FCRL3 with GravesTdisease. *BMC Genomics*, **2016**, 17, 939	4.5	7
39	The role of toll-like receptor variants in acute anterior uveitis. *Molecular Vision*, **2011**, 17, 2970-7	2.3	7
38	Compound heterozygote myocilin mutations in a pedigree with high prevalence of primary open-angle glaucoma. *Molecular Vision*, **2012**, 18, 3064-9	2.3	7
37	Epha2 genotype influences ultraviolet radiation induced cataract in mice. *Experimental Eye Research*, **2019**, 188, 107806	3.7	6
36	A functional polymorphism in the lymphotoxin-alpha gene is associated with carotid artery wall thickness: the Diabetes Heart Study. *European Journal of Cardiovascular Prevention and Rehabilitation*, **2006**, 13, 655-7		6
35	molecular analysis and genotype-phenotype correlations in families with aniridia from Australasia and Southeast Asia. *Molecular Vision*, **2018**, 24, 261-273	2.3	6
34	Association of Polymorphisms in MACRO Domain Containing 2 With Thyroid-Associated Orbitopathy **2016**, 57, 3129-37		6
33	Long-term survival rates of patients undergoing vitrectomy for diabetic retinopathy in an Australian population: a population-based audit. *Clinical and Experimental Ophthalmology*, **2019**, 47, 598-604	2.4	5
32	Screening of the COL8A2 gene in an Australian family with early-onset FuchsTendothelial corneal dystrophy. *Clinical and Experimental Ophthalmology*, **2014**, 42, 198-200	2.4	5
31	Rare variants in optic disc area gene enriched in primary open-angle glaucoma. *Molecular Genetics & Genomic Medicine*, **2016**, 4, 624-633	2.3	5
30	Reduced expression of apolipoprotein E and immunoglobulin heavy constant gamma 1 proteins in Fuchs endothelial corneal dystrophy. *Clinical and Experimental Ophthalmology*, **2019**, 47, 1028-1042	2.4	4
29	Visual outcomes following vitrectomy for diabetic retinopathy amongst Indigenous and non-Indigenous Australians in South Australia and the Northern Territory. *Clinical and Experimental Ophthalmology*, **2018**, 46, 417-423	2.4	4
28	Identification of a novel MYOC mutation, p.(Trp373), in a family with open angle glaucoma. *Gene*, **2014**, 545, 271-5	3.8	4
27	Investigation of eight candidate genes on chromosome 1p36 for autosomal dominant total congenital cataract. *Molecular Vision*, **2008**, 14, 1799-804	2.3	4
26	CYP1B1 copy number variation is not a major contributor to primary congenital glaucoma. *Molecular Vision*, **2015**, 21, 160-4	2.3	4

25	Evaluation of Shared Genetic Susceptibility to High and Low Myopia and Hyperopia. *JAMA Ophthalmology*, **2021**, 139, 601-609	3.9	4
24	Promoter polymorphism at the tumour necrosis factor/lymphotoxin-alpha locus is associated with type of diabetes but not with susceptibility to sight-threatening diabetic retinopathy. *Diabetes and Vascular Disease Research*, **2016**, 13, 164-7	3.3	3
23	A novel syndrome of paediatric cataract, dysmorphism, ectodermal features, and developmental delay in Australian Aboriginal family maps to 1p35.3-p36.32. *BMC Medical Genetics*, **2010**, 11, 165	2.1	3
22	Common variants in SOX-2 and congenital cataract genes contribute to age-related nuclear cataract. *Communications Biology*, **2020**, 3, 755	6.7	3
21	Ferritin light chain gene mutation in a large Australian family with hereditary hyperferritinemia-cataract syndrome. *Ophthalmic Genetics*, **2017**, 38, 171-174	1.2	2
20	A novel genetic syndrome characterized by pediatric cataract, dysmorphism, ectodermal features, and developmental delay in an indigenous Australian family. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 633-9	2.5	2
19	Association of Smoking, Alcohol Consumption, Blood Pressure, Body Mass Index, and Glycemic Risk Factors With Age-Related Macular Degeneration: A Mendelian Randomization Study. *JAMA Ophthalmology*, **2021**,	3.9	2
18	Generation of MNZTASi001-A, a human pluripotent stem cell line from a person with primary progressive multiple sclerosis. *Stem Cell Research*, **2021**, 57, 102568	1.6	2
17	Identifying Genetic Risk Factors for Diabetic Macular Edema and the Response to Treatment. *Journal of Diabetes Research*, **2020**, 2020, 5016916	3.9	2
16	Innate and Adaptive Gene Single Nucleotide Polymorphisms Associated With Susceptibility of Severe Inflammatory Complications in Acanthamoeba Keratitis **2021**, 62, 33		2
15	The utility of genomic testing in the ophthalmology clinic: A review. *Clinical and Experimental Ophthalmology*, **2021**, 49, 615-625	2.4	2
14	Rapid and efficient cataract gene evaluation in F0 zebrafish using CRISPR-Cas9 ribonucleoprotein complexes. *Methods*, **2021**, 194, 37-47	4.6	2
13	Does the association between TMEM98 and nanophthalmos require further confirmation?-Reply. *JAMA Ophthalmology*, **2015**, 133, 359-60	3.9	1
12	Utilising multi-large omics data to elucidate biological mechanisms within multiple sclerosis genetic susceptibility loci. *Multiple Sclerosis Journal*, **2021**, 27, 2141-2149	5	1
11	A 127 kb truncating deletion of PGRMC1 is a novel cause of X-linked isolated paediatric cataract. *European Journal of Human Genetics*, **2021**, 29, 1206-1215	5.3	1
10	Comparing vision and macular thickness in neovascular age-related macular degeneration, diabetic macular oedema and retinal vein occlusion patients treated with intravitreal antivascular endothelial growth factor injections in clinical practice. *BMJ Open Ophthalmology*, **2021**, 6, e000749	3.2	1
9	Differential gene expression analysis of corneal endothelium indicates involvement of phagocytic activity in FuchsTendothelial corneal dystrophy. *Experimental Eye Research*, **2021**, 210, 108692	3.7	1
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