Kathryn P Burdon

List of Publications by Year
in descending order

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Version: 2024-02-01

188 papers	10,409 citations	38720 50 h-index	45285 90 g-index
222 all docs	222 docs citations	222 times ranked	11444 citing authors

#	Article	IF	Citations
1	A large genome-wide association study of age-related macular degeneration highlights contributions of rare and common variants. Nature Genetics, 2016, 48, 134-143.	9.4	1,167
2	Genome-wide meta-analyses of multiancestry cohorts identify multiple new susceptibility loci for refractive error and myopia. Nature Genetics, 2013, 45, 314-318.	9.4	398
3	Genome-wide association study identifies susceptibility loci for open angle glaucoma at TMCO1 and CDKN2B-AS1. Nature Genetics, 2011, 43, 574-578.	9.4	381
4	Common variants near CAV1 and CAV2 are associated with primary open-angle glaucoma. Nature Genetics, 2010, 42, 906-909.	9.4	357
5	Genome-wide association analyses identify multiple loci associated with central corneal thickness and keratoconus. Nature Genetics, 2013, 45, 155-163.	9.4	269
6	Genome-wide meta-analysis identifies five new susceptibility loci for cutaneous malignant melanoma. Nature Genetics, 2015, 47, 987-995.	9.4	218
7	Genome-wide analysis of multi-ancestry cohorts identifies new loci influencing intraocular pressure and susceptibility to glaucoma. Nature Genetics, 2014, 46, 1126-1130.	9.4	212
8	Genome-wide association analysis identifies TXNRD2, ATXN2 and FOXC1 as susceptibility loci for primary open-angle glaucoma. Nature Genetics, 2016, 48, 189-194.	9.4	211
9	Multitrait analysis of glaucoma identifies new risk loci and enables polygenic prediction of disease susceptibility and progression. Nature Genetics, 2020, 52, 160-166.	9.4	192
10	Common variants near ABCA1, AFAP1 and GMDS confer risk of primary open-angle glaucoma. Nature Genetics, 2014, 46, 1120-1125.	9.4	186
11	A Systematic Meta-Analysis of Genetic Association Studies for Diabetic Retinopathy. Diabetes, 2009, 58, 2137-2147.	0.3	180
12	Angiopoietin receptor TEK mutations underlie primary congenital glaucoma with variable expressivity. Journal of Clinical Investigation, 2016, 126, 2575-2587.	3.9	175
13	Ancestral LOXL1 variants are associated with pseudoexfoliation in Caucasian Australians but with markedly lower penetrance than in Nordic people. Human Molecular Genetics, 2007, 17, 710-716.	1.4	152
14	Genome-wide association study of intraocular pressure uncovers new pathways to glaucoma. Nature Genetics, 2018, 50, 1067-1071.	9.4	152
15	Genome-wide association study identifies five new susceptibility loci for primary angle closure glaucoma. Nature Genetics, 2016, 48, 556-562.	9.4	147
16	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, 264-277.	2.6	139
17	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.	9.4	138
18	Common Genetic Variants near the Brittle Cornea Syndrome Locus ZNF469 Influence the Blinding Disease Risk Factor Central Corneal Thickness. PLoS Genetics, 2010, 6, e1000947.	1.5	130

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19	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, ddw399.	1.4	120
20	Association of Polymorphisms in the Hepatocyte Growth Factor Gene Promoter with Keratoconus. , 2011, 52, 8514.		114
21	Genetic association study of exfoliation syndrome identifies a protective rare variant at LOXL1 and five new susceptibility loci. Nature Genetics, 2017, 49, 993-1004.	9.4	114
22	Genome-wide association study identifies seven novel susceptibility loci for primary open-angle glaucoma. Human Molecular Genetics, 2018, 27, 1486-1496.	1.4	111
23	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, 1120-1130.	2.6	107
24	A common variant near TGFBR3 is associated with primary open angle glaucoma. Human Molecular Genetics, 2015, 24, 3880-3892.	1.4	105
25	Meta-analysis of geneâ€“environment-wide association scans accounting for education level identifies additional loci for refractive error. Nature Communications, 2016, 7, 11008.	5.8	104
26	Genome-wide association study identifies three novel loci in Fuchs endothelial corneal dystrophy. Nature Communications, 2017, 8, 14898.	5.8	101
27	Insights into keratoconus from a genetic perspective. Australasian journal of optometry, The, 2013, 96, 146-154.	0.6	97
28	A common variant mapping to CACNA1A is associated with susceptibility to exfoliation syndrome. Nature Genetics, 2015, 47, 387-392.	9.4	97
29	Mutations of theEPHA2receptor tyrosine kinase gene cause autosomal dominant congenital cataract. Human Mutation, 2009, 30, E603-E611.	1.1	96
30	The genetics of central corneal thickness. British Journal of Ophthalmology, 2010, 94, 971-976.	2.1	96
31	Meta-analysis of genome-wide association studies identifies novel loci that influence cupping and the glaucomatous process. Nature Communications, 2014, 5, 4883.	5.8	89
32	Functional and Structural Implications of the Complement Factor H Y402H Polymorphism Associated with Age-Related Macular Degeneration. , 2008, 49, 1763.		85
33	ARHGEF12 influences the risk of glaucoma by increasing intraocular pressure. Human Molecular Genetics, 2015, 24, 2689-2699.	1.4	79
34	Mutations in the NDP gene: contribution to Norrie disease, familial exudative vitreoretinopathy and retinopathy of prematurity. Clinical and Experimental Ophthalmology, 2006, 34, 682-688.	1.3	76
35	Glaucoma Risk Alleles at CDKN2B-AS1 Are Associated with Lower Intraocular Pressure, Normal-Tension Glaucoma, and Advanced Glaucoma. Ophthalmology, 2012, 119, 1539-1545.	2.5	74
36	Mutational Analysis ofMIR184in Sporadic Keratoconus and Myopia. , 2013, 54, 5266.		73

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37	Genome-wide association study for sight-threatening diabetic retinopathy reveals association with genetic variation near the GRB2 gene. Diabetologia, 2015, 58, 2288-2297.	2.9	73
38	Identification of LOXL1 protein and Apolipoprotein E as components of surgically isolated pseudoexfoliation material by direct mass spectrometry. Experimental Eye Research, 2009, 89, 479-485.	1.2	72
39	Metaâ€analysis of Genomeâ€Wide Association Studies Identifies Novel Loci Associated With Optic Disc Morphology. Genetic Epidemiology, 2015, 39, 207-216.	0.6	72
40	Apparent autosomal dominant keratoconus in a large Australian pedigree accounted for by digenic inheritance of two novel loci. Human Genetics, 2008, 124, 379-386.	1.8	70
41	Homozygous Mutations in PXDN Cause Congenital Cataract, Corneal Opacity, and Developmental Glaucoma. American Journal of Human Genetics, 2011, 89, 464-473.	2.6	68
42	Copy Number Variations of TBK1 in Australian Patients With Primary Open-Angle Glaucoma. American Journal of Ophthalmology, 2015, 159, 124-130.e1.	1.7	68
43	Measurement of Systemic Mitochondrial Function in Advanced Primary Open-Angle Glaucoma and Leber Hereditary Optic Neuropathy. PLoS ONE, 2015, 10, e0140919.	1.1	66
44	Common Sequence Variation in theVEGFAGene Predicts Risk of Diabetic Retinopathy. , 2009, 50, 5552.		64
45	Australian and New Zealand Registry of Advanced Glaucoma: methodology and recruitment. Clinical and Experimental Ophthalmology, 2012, 40, 569-575.	1.3	64
46	Cross-ancestry genome-wide association analysis of corneal thickness strengthens link between complex and Mendelian eye diseases. Nature Communications, 2018, 9, 1864.	5.8	63
47	Association of TCF4 and CLU polymorphisms with Fuchsâ€™ endothelial dystrophy and implication of CLU and TGFBI proteins in the disease process. European Journal of Human Genetics, 2012, 20, 632-638.	1.4	61
48	Serum selenium status in <scp>G</scp>ravesâ€™ disease with and without orbitopathy: a caseâ€“control study. Clinical Endocrinology, 2014, 80, 905-910.	1.2	58
49	WNT10A exonic variant increases the risk of keratoconus by decreasing corneal thickness. Human Molecular Genetics, 2015, 24, 5060-5068.	1.4	58
50	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, 128-134.	0.9	57
51	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.	1.1	54
52	Mutation inTMEM98in a Large White Kindred With Autosomal Dominant Nanophthalmos Linked to 17p12-q12. JAMA Ophthalmology, 2014, 132, 970.	1.4	54
53	Multiethnic Genome-Wide Association Study of Diabetic Retinopathy Using Liability Threshold Modeling of Duration of Diabetes and Glycemic Control. Diabetes, 2019, 68, 441-456.	0.3	54
54	A novel mutation in the Connexin 46 gene causes autosomal dominant congenital cataract with incomplete penetrance. Journal of Medical Genetics, 2004, 41, e106-e106.	1.5	53

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55	Diabetic Retinopathy Is Associated With Elevated Serum Asymmetric and Symmetric Dimethylarginines. Diabetes Care, 2009, 32, 2084-2086.	4.3	53
56	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, 643-650.	1.2	53
57	A genomeâ€wide association study suggests new evidence for an association of the <scp>NADPH</scp> Oxidase 4 (<scp>NOX</scp>4) gene with severe diabetic retinopathy in type 2 diabetes. Acta Ophthalmologica, 2018, 96, e811-e819.	0.6	52
58	Association Between Erythropoietin Gene Polymorphisms and Diabetic Retinopathy. JAMA Ophthalmology, 2010, 128, 102.	2.6	51
59	Heritability of Central Corneal Thickness in Nuclear Families. , 2009, 50, 4087.		49
60	Genome-wide association studies for diabetic macular edema and proliferative diabetic retinopathy. BMC Medical Genetics, 2018, 19, 71.	2.1	49
61	Higher Prevalence of Myocilin Mutations in Advanced Glaucoma in Comparison with Less Advanced Disease in an Australasian Disease Registry. Ophthalmology, 2013, 120, 1135-1143.	2.5	48
62	Genetic and Environmental Risk Factors for Keratoconus. Annual Review of Vision Science, 2020, 6, 25-46.	2.3	46
63	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, 1085-1090.	3.0	45
64	Rapid inexpensive genome-wide association using pooled whole blood. Genome Research, 2009, 19, 2075-2080.	2.4	45
65	Glaucoma spectrum and age-related prevalence of individuals with FOXC1 and PITX2 variants. European Journal of Human Genetics, 2017, 25, 839-847.	1.4	43
66	Elevation of Serum Asymmetrical and Symmetrical Dimethylarginine in Patients with Advanced Glaucoma. , 2012, 53, 1923.		42
67	Association of Genetic Variants with Primary Angle Closure Glaucoma in Two Different Populations. PLoS ONE, 2013, 8, e67903.	1.1	42
68	Association of Î±2-Heremans-Schmid Glycoprotein Polymorphisms with Subclinical Atherosclerosis. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 345-352.	1.8	40
69	Aldose Reductase Gene Polymorphisms and Diabetic Retinopathy Susceptibility. Diabetes Care, 2010, 33, 1834-1836.	4.3	39
70	Association of Genetic Variants in theTMCO1Gene with Clinical Parameters Related to Glaucoma and Characterization of the Protein in the Eye. , 2012, 53, 4917.		38
71	MALDI-MS-Imaging of Whole Human Lens Capsule. Journal of Proteome Research, 2011, 10, 3522-3529.	1.8	37
72	Replication and Meta-Analysis of Candidate Loci Identified Variation atRAB3GAP1Associated With Keratoconus. , 2013, 54, 5132.		37

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73	Matrix metalloproteinase-9 genetic variation and primary angle closure glaucoma in a Caucasian population. Molecular Vision, 2011, 17, 1420-4.	1.1	37
74	Novel quantitative trait loci for central corneal thickness identified by candidate gene analysis of osteogenesis imperfecta genes. Human Genetics, 2010, 127, 33-44.	1.8	36
75	A multi-ethnic genome-wide association study implicates collagen matrix integrity and cell differentiation pathways in keratoconus. Communications Biology, 2021, 4, 266.	2.0	36
76	Variability of Serum Soluble Intercellular Adhesion Molecule-1 Measurements Attributable to a Common Polymorphism. Clinical Chemistry, 2004, 50, 2185-2187.	1.5	35
77	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, 706-711.	1.2	35
78	Prenatal detection of congenital bilateral cataract leading to the diagnosis of Nance-Horan syndrome in the extended family. Prenatal Diagnosis, 2007, 27, 662-664.	1.1	35
79	Mutations in the EPHA2 Gene Are a Major Contributor to Inherited Cataracts in South-Eastern Australia. PLoS ONE, 2013, 8, e72518.	1.1	35
80	Chromosome 9p21 primary openâ€angle glaucoma susceptibility locus: a review. Clinical and Experimental Ophthalmology, 2014, 42, 25-32.	1.3	35
81	Association of Genetic Variation With Keratoconus. JAMA Ophthalmology, 2020, 138, 174.	1.4	34
82	Heritability and Expression of C-Reactive Protein in Type 2 Diabetes in the Diabetes Heart Study. Annals of Human Genetics, 2006, 70, 717-725.	0.3	33
83	Analysis combining correlated glaucoma traits identifies five new risk loci for open-angle glaucoma. Scientific Reports, 2018, 8, 3124.	1.6	33
84	Prevalence ofFOXC1Variants in Individuals With a Suspected Diagnosis of Primary Congenital Glaucoma. JAMA Ophthalmology, 2019, 137, 348.	1.4	33
85	Biallelic CPAMD8 Variants Are a Frequent Cause of Childhood and Juvenile Open-Angle Glaucoma. Ophthalmology, 2020, 127, 758-766.	2.5	33
86	The PITX3 gene in posterior polar congenital cataract in Australia. Molecular Vision, 2006, 12, 367-71.	1.1	33
87	Identification of podocin (NPHS2) gene mutations in African Americans with nondiabetic end-stage renal disease. Kidney International, 2005, 68, 256-262.	2.6	32
88	Human Lipoxygenase Pathway Gene Variation and Association with Markers of Subclinical Atherosclerosis in the Diabetes Heart Study. Mediators of Inflammation, 2010, 2010, 1-9.	1.4	32
89	Macular Ganglion Cellâ€“Inner Plexiform Layer Loss Precedes Peripapillary Retinal Nerve Fiber Layer Loss in Glaucoma with Lower Intraocular Pressure. Ophthalmology, 2019, 126, 1119-1130.	2.5	32
90	Myocilin Gene Gln368Ter Variant Penetrance and Association With Glaucoma in Population-Based and Registry-Based Studies. JAMA Ophthalmology, 2019, 137, 28.	1.4	32

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91	Association ofeNOSPolymorphisms with Primary Angle-Closure Glaucoma. , 2013, 54, 2108.		30
92	Association of Open-Angle Glaucoma Loci With Incident Glaucoma in the Blue Mountains Eye Study. American Journal of Ophthalmology, 2015, 159, 31-36.e1.	1.7	30
93	Association of Smoking, Alcohol Consumption, Blood Pressure, Body Mass Index, and Glycemic Risk Factors With Age-Related Macular Degeneration. JAMA Ophthalmology, 2021, 139, 1299.	1.4	29
94	Identification of a Novel Oligomerization Disrupting Mutation inCRYÎ'AAssociated with Congenital Cataract in a South Australian Family. Human Mutation, 2013, 34, 435-438.	1.1	28
95	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.	1.7	27
96	Genetic Association at the 9p21 Glaucoma Locus Contributes to Sex Bias in Normal-Tension Glaucoma. , 2016, 57, 3416.		26
97	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.	1.1	26
98	A novel deletion in theFTL gene causes hereditary hyperferritinemia cataract syndrome (HHCS) by alteration of the transcription start site. Human Mutation, 2007, 28, 742-742.	1.1	25
99	Biological effect of LOXL1 coding variants associated with pseudoexfoliation syndrome. Experimental Eye Research, 2016, 146, 212-223.	1.2	25
100	Myocilin Predictive Genetic Testing for Primary Open-Angle Glaucoma Leads to Early Identification of At-Risk Individuals. Ophthalmology, 2017, 124, 303-309.	2.5	25
101	The Association Between Vitamin D and Multiple Sclerosis Risk: 1,25(OH)2D3 Induces Super-Enhancers Bound by VDR. Frontiers in Immunology, 2019, 10, 488.	2.2	25
102	Genetic analysis of the clusterin gene in pseudoexfoliation syndrome. Molecular Vision, 2008, 14, 1727-36.	1.1	25
103	A Turkish family with Nance-Horan syndrome due to a novel mutation. Gene, 2013, 525, 141-145.	1.0	24
104	TGC repeat expansion in the TCF4 gene increases the risk of Fuchsâ€™ endothelial corneal dystrophy in Australian cases. PLoS ONE, 2017, 12, e0183719.	1.1	24
105	The association of hepatocyte growth factor (HGF) gene with primary angle closure glaucoma in the Nepalese population. Molecular Vision, 2011, 17, 2248-54.	1.1	24
106	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.	1.6	23
107	Review of the prevalence of diabetic retinopathy in Indigenous <scp>A</scp>ustralians. Clinical and Experimental Ophthalmology, 2014, 42, 875-882.	1.3	22
108	Evaluation of Shared Genetic Susceptibility to High and Low Myopia and Hyperopia. JAMA Ophthalmology, 2021, 139, 601.	1.4	22

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109	Severe prekallikrein deficiency associated with homozygosity for an Arg94Stop nonsense mutation. British Journal of Haematology, 2004, 127, 220-223.	1.2	21
110	Occurrence ofCYP1B1Mutations in Juvenile Open-Angle Glaucoma With Advanced Visual Field Loss. JAMA Ophthalmology, 2015, 133, 826.	1.4	21
111	Assessment of polygenic effects links primary open-angle glaucoma and age-related macular degeneration. Scientific Reports, 2016, 6, 26885.	1.6	21
112	Novel missense mutation in the bZIP transcription factor, MAF, associated with congenital cataract, developmental delay, seizures and hearing loss (AymÃ©-Gripp syndrome). BMC Medical Genetics, 2017, 18, 52.	2.1	21
113	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.	1.1	21
114	Association of Protein Tyrosine Phosphatase-N1 Polymorphisms With Coronary Calcified Plaque in the Diabetes Heart Study. Diabetes, 2006, 55, 651-658.	0.3	20
115	Genomeâ€wide association studies in the hunt for genes causing primary openâ€angle glaucoma: a review. Clinical and Experimental Ophthalmology, 2012, 40, 358-363.	1.3	20
116	Differential Gene Expression Profiling of Orbital Adipose Tissue in Thyroid Orbitopathy. , 2015, 56, 6438.		20
117	Common Sequence Variation in the VEGFC Gene Is Associated with Diabetic Retinopathy and Diabetic Macular Edema. Ophthalmology, 2015, 122, 1828-1836.	2.5	20
118	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.	0.8	20
119	The telomere of human chromosome 1p contains at least two independent autosomal dominant congenital cataract genes. British Journal of Ophthalmology, 2005, 89, 831-834.	2.1	19
120	Association of genes of lipid metabolism with measures of subclinical cardiovascular disease in the Diabetes Heart Study. Journal of Medical Genetics, 2005, 42, 720-724.	1.5	19
121	Ethnic and Mouse Strain Differences in Central Corneal Thickness and Association with Pigmentation Phenotype. PLoS ONE, 2011, 6, e22103.	1.1	19
122	Association of Arachidonate 12-Lipoxygenase Genotype Variation and Glycemic Control With Albuminuria in Type 2 Diabetes. American Journal of Kidney Diseases, 2008, 52, 242-250.	2.1	18
123	Accurate Imputation-Based Screening of Gln368Ter Myocilin Variant in Primary Open-Angle Glaucoma. , 2015, 56, 5087.		17
124	Genetically Determined Plasma Lipid Levels and Risk of Diabetic Retinopathy: A Mendelian Randomization Study. Diabetes, 2017, 66, 3130-3141.	0.3	17
125	Key challenges in bringing CRISPR-mediated somatic cell therapy into the clinic. Genome Medicine, 2017, 9, 85.	3.6	17
126	The genetic and clinical landscape of nanophthalmos and posterior microphthalmos in an Australian cohort. Clinical Genetics, 2020, 97, 764-769.	1.0	17

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127	EPHA2 MUTATIONS CONTRIBUTE TO CONGENITAL CATARACT THROUGH DIVERSE MECHANISMS. Molecular Vision, 2016, 22, 18-30.	1.1	16
128	Recurrent mutation in the crystallin alpha A gene associated with inherited paediatric cataract. BMC Research Notes, 2016, 9, 83.	0.6	15
129	Diabetic macular oedema: clinical risk factors and emerging genetic influences. Australasian journal of optometry, The, 2017, 100, 569-576.	0.6	15
130	Identification of novel mutations causing pediatric cataract in Bhutan, Cambodia, and Sri Lanka. Molecular Genetics & Genomic Medicine, 2018, 6, 555-564.	0.6	15
131	Genetic study of diabetic retinopathy: recruitment methodology and analysis of baseline characteristics. Clinical and Experimental Ophthalmology, 2014, 42, 486-493.	1.3	14
132	Predictive genetic testing in minors for Myocilin juvenile onset open angle glaucoma. Clinical Genetics, 2015, 88, 584-588.	1.0	14
133	Rare variants in optic disc area gene <scp>CARD</scp> 10 enriched in primary openâ€angle glaucoma. Molecular Genetics & Genomic Medicine, 2016, 4, 624-633.	0.6	14
134	Testosterone Pathway Genetic Polymorphisms in Relation to Primary Open-Angle Glaucoma: An Analysis in Two Large Datasets. , 2018, 59, 629.		14
135	Contribution of Mutations in Known Mendelian Glaucoma Genes to Advanced Early-Onset Primary Open-Angle Glaucoma. , 2017, 58, 1537.		13
136	Rare, Potentially Pathogenic Variants in ZNF469 Are Not Enriched in Keratoconus in a Large Australian Cohort of European Descent. , 2017, 58, 6248.		13
137	DNA methylation at the 9p21 glaucoma susceptibility locus is associated with normal-tension glaucoma. Ophthalmic Genetics, 2018, 39, 221-227.	0.5	13
138	Ocular Expression and Distribution of Products of the POAG-Associated Chromosome 9p21 Gene Region. PLoS ONE, 2013, 8, e75067.	1.1	13
139	Association of Polymorphisms in MACRO Domain Containing 2 With Thyroid-Associated Orbitopathy. , 2016, 57, 3129.		12
140	A novel de novo Myocilin variant in a patient with sporadic juvenile open angle glaucoma. BMC Medical Genetics, 2016, 17, 30.	2.1	12
141	Partial duplication of the CRYBB1-CRYBA4 locus is associated with autosomal dominant congenital cataract. European Journal of Human Genetics, 2017, 25, 711-718.	1.4	12
142	Candidate gene study to investigate the genetic determinants of normal variation in central corneal thickness. Molecular Vision, 2010, 16, 562-9.	1.1	12
143	Relationship between DDAH gene variants and serum ADMA level in individuals with type 1 diabetes. Journal of Diabetes and Its Complications, 2012, 26, 195-198.	1.2	11
144	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, 558-563.	1.1	11

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145	MicroRNA-Related Genetic Variants Are Associated With Diabetic Retinopathy in Type 1 Diabetes Mellitus. , 2019, 60, 3937.		11
146	Screening of the <scp>COL8A2</scp> gene in an <scp>A</scp>ustralian family with earlyâ€onset <scp>F</scp>uchsâ€™ endothelial corneal dystrophy. Clinical and Experimental Ophthalmology, 2014, 42, 198-200.	1.3	10
147	Pooled genome wide association detects association upstream of FCRL3 with Gravesâ€™ disease. BMC Genomics, 2016, 17, 939.	1.2	10
148	Epha2 genotype influences ultraviolet radiation induced cataract in mice. Experimental Eye Research, 2019, 188, 107806.	1.2	10
149	Common variants in SOX-2 and congenital cataract genes contribute to age-related nuclear cataract. Communications Biology, 2020, 3, 755.	2.0	10
150	A novel locus for X-linked congenital cataract on Xq24. Molecular Vision, 2008, 14, 721-6.	1.1	10
151	Tag SNPs detect association of the CYP1B1 gene with primary open angle glaucoma. Molecular Vision, 2010, 16, 2286-93.	1.1	10
152	molecular analysis and genotype-phenotype correlations in families with aniridia from Australasia and Southeast Asia. Molecular Vision, 2018, 24, 261-273.	1.1	10
153	P-selectin gene haplotype associations with albuminuria in the Diabetes Heart Study. Kidney International, 2005, 68, 741-746.	2.6	9
154	Ethical Considerations for the Return of Incidental Findings in Ophthalmic Genomic Research. Translational Vision Science and Technology, 2016, 5, 3.	1.1	9
155	Rapid and efficient cataract gene evaluation in F0 zebrafish using CRISPR-Cas9 ribonucleoprotein complexes. Methods, 2021, 194, 37-47.	1.9	9
156	Identifying Genetic Risk Factors for Diabetic Macular Edema and the Response to Treatment. Journal of Diabetes Research, 2020, 2020, 1-12.	1.0	8
157	Whole exome sequencing implicates eye development, the unfolded protein response and plasma membrane homeostasis in primary open-angle glaucoma. PLoS ONE, 2017, 12, e0172427.	1.1	8
158	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, 164-167.	0.9	7
159	Longâ€term survival rates of patients undergoing vitrectomy for diabetic retinopathy in an Australian population: a populationâ€based audit. Clinical and Experimental Ophthalmology, 2019, 47, 598-604.	1.3	7
160	The utility of genomic testing in the ophthalmology clinic: A review. Clinical and Experimental Ophthalmology, 2021, 49, 615-625.	1.3	7
161	The role of toll-like receptor variants in acute anterior uveitis. Molecular Vision, 2011, 17, 2970-7.	1.1	7
162	Compound heterozygote myocilin mutations in a pedigree with high prevalence of primary open-angle glaucoma. Molecular Vision, 2012, 18, 3064-9.	1.1	7

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163	A functional polymorphism in the lymphotoxin-Î± gene is associated with carotid artery wall thickness: The Diabetes Heart Study. European Journal of Cardiovascular Prevention and Rehabilitation, 2006, 13, 655-657.	3.1	6
164	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.	1.3	6
165	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.	1.3	6
166	Identifying Genetic Biomarkers Predicting Response to Anti-Vascular Endothelial Growth Factor Injections in Diabetic Macular Edema. International Journal of Molecular Sciences, 2022, 23, 4042.	1.8	5
167	Identification of a novel MYOC mutation, p.(Trp373*), in a family with open angle glaucoma. Gene, 2014, 545, 271-275.	1.0	4
168	Innate and Adaptive Gene Single Nucleotide Polymorphisms Associated With Susceptibility of Severe Inflammatory Complications in Acanthamoeba Keratitis. , 2021, 62, 33.		4
169	A 127â€‰kb truncating deletion of PGRMC1 is a novel cause of X-linked isolated paediatric cataract. European Journal of Human Genetics, 2021, 29, 1206-1215.	1.4	4
170	Genotype, Age, Genetic Background, and Sex Influence Epha2-Related Cataract Development in Mice. , 2021, 62, 3.		4
171	Generation of MNZTASi001-A, a human pluripotent stem cell line from a person with primary progressive multiple sclerosis. Stem Cell Research, 2021, 57, 102568.	0.3	4
172	Investigation of eight candidate genes on chromosome 1p36 for autosomal dominant total congenital cataract. Molecular Vision, 2008, 14, 1799-804.	1.1	4
173	CYP1B1 copy number variation is not a major contributor to primary congenital glaucoma. Molecular Vision, 2015, 21, 160-4.	1.1	4
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