

Kathryn P Burdon

List of Publications by Year in descending order

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Version: 2024-02-01

188
papers

10,409
citations

38742

50
h-index

45317

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. <i>Nature Genetics</i> , 2016, 48, 134-143.	21.4	1,167
2	Genome-wide meta-analyses of multi-ancestry cohorts identify multiple new susceptibility loci for refractive error and myopia. <i>Nature Genetics</i> , 2013, 45, 314-318.	21.4	398
3	Genome-wide association study identifies susceptibility loci for open angle glaucoma at TMCO1 and CDKN2B-AS1. <i>Nature Genetics</i> , 2011, 43, 574-578.	21.4	381
4	Common variants near CAV1 and CAV2 are associated with primary open-angle glaucoma. <i>Nature Genetics</i> , 2010, 42, 906-909.	21.4	357
5	Genome-wide association analyses identify multiple loci associated with central corneal thickness and keratoconus. <i>Nature Genetics</i> , 2013, 45, 155-163.	21.4	269
6	Genome-wide meta-analysis identifies five new susceptibility loci for cutaneous malignant melanoma. <i>Nature Genetics</i> , 2015, 47, 987-995.	21.4	218
7	Genome-wide analysis of multi-ancestry cohorts identifies new loci influencing intraocular pressure and susceptibility to glaucoma. <i>Nature Genetics</i> , 2014, 46, 1126-1130.	21.4	212
8	Genome-wide association analysis identifies TXNRD2, ATXN2 and FOXC1 as susceptibility loci for primary open-angle glaucoma. <i>Nature Genetics</i> , 2016, 48, 189-194.	21.4	211
9	Multitrait analysis of glaucoma identifies new risk loci and enables polygenic prediction of disease susceptibility and progression. <i>Nature Genetics</i> , 2020, 52, 160-166.	21.4	192
10	Common variants near ABCA1, AFAP1 and GMDS confer risk of primary open-angle glaucoma. <i>Nature Genetics</i> , 2014, 46, 1120-1125.	21.4	186
11	A Systematic Meta-Analysis of Genetic Association Studies for Diabetic Retinopathy. <i>Diabetes</i> , 2009, 58, 2137-2147.	0.6	180
12	Angiopoietin receptor TEK mutations underlie primary congenital glaucoma with variable expressivity. <i>Journal of Clinical Investigation</i> , 2016, 126, 2575-2587.	8.2	175
13	Ancestral LOXL1 variants are associated with pseudoexfoliation in Caucasian Australians but with markedly lower penetrance than in Nordic people. <i>Human Molecular Genetics</i> , 2007, 17, 710-716.	2.9	152
14	Genome-wide association study of intraocular pressure uncovers new pathways to glaucoma. <i>Nature Genetics</i> , 2018, 50, 1067-1071.	21.4	152
15	Genome-wide association study identifies five new susceptibility loci for primary angle closure glaucoma. <i>Nature Genetics</i> , 2016, 48, 556-562.	21.4	147
16	Nine Loci for Ocular Axial Length Identified through Genome-wide Association Studies, Including Shared Loci with Refractive Error. <i>American Journal of Human Genetics</i> , 2013, 93, 264-277.	6.2	139
17	Genome-wide association meta-analyses combining multiple risk phenotypes provide insights into the genetic architecture of cutaneous melanoma susceptibility. <i>Nature Genetics</i> , 2020, 52, 494-504.	21.4	138
18	Common Genetic Variants near the Brittle Cornea Syndrome Locus ZNF469 Influence the Blinding Disease Risk Factor Central Corneal Thickness. <i>PLoS Genetics</i> , 2010, 6, e1000947.	3.5	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.	2.9	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.	21.4	114
22	Genome-wide association study identifies seven novel susceptibility loci for primary open-angle glaucoma. Human Molecular Genetics, 2018, 27, 1486-1496.	2.9	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.	6.2	107
24	A common variant near TGFBR3 is associated with primary open angle glaucoma. Human Molecular Genetics, 2015, 24, 3880-3892.	2.9	105
25	Meta-analysis of gene-environment-wide association scans accounting for education level identifies additional loci for refractive error. Nature Communications, 2016, 7, 11008.	12.8	104
26	Genome-wide association study identifies three novel loci in Fuchs endothelial corneal dystrophy. Nature Communications, 2017, 8, 14898.	12.8	101
27	Insights into keratoconus from a genetic perspective. Australasian journal of optometry, The, 2013, 96, 146-154.	1.3	97
28	A common variant mapping to CACNA1A is associated with susceptibility to exfoliation syndrome. Nature Genetics, 2015, 47, 387-392.	21.4	97
29	Mutations of theEPHA2receptor tyrosine kinase gene cause autosomal dominant congenital cataract. Human Mutation, 2009, 30, E603-E611.	2.5	96
30	The genetics of central corneal thickness. British Journal of Ophthalmology, 2010, 94, 971-976.	3.9	96
31	Meta-analysis of genome-wide association studies identifies novel loci that influence cupping and the glaucomatous process. Nature Communications, 2014, 5, 4883.	12.8	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.	2.9	79
34	Mutations in the <i>NDP</i> gene: contribution to Norrie disease, familial exudative vitreoretinopathy and retinopathy of prematurity. Clinical and Experimental Ophthalmology, 2006, 34, 682-688.	2.6	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.	5.2	74
36	Mutational Analysis of<i>MIR184</i>in 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. <i>Diabetologia</i> , 2015, 58, 2288-2297.	6.3	73
38	Identification of LOXL1 protein and Apolipoprotein E as components of surgically isolated pseudoexfoliation material by direct mass spectrometry. <i>Experimental Eye Research</i> , 2009, 89, 479-485.	2.6	72
39	Meta-analysis of Genome-Wide Association Studies Identifies Novel Loci Associated With Optic Disc Morphology. <i>Genetic Epidemiology</i> , 2015, 39, 207-216.	1.3	72
40	Apparent autosomal dominant keratoconus in a large Australian pedigree accounted for by digenic inheritance of two novel loci. <i>Human Genetics</i> , 2008, 124, 379-386.	3.8	70
41	Homozygous Mutations in PXDN Cause Congenital Cataract, Corneal Opacity, and Developmental Glaucoma. <i>American Journal of Human Genetics</i> , 2011, 89, 464-473.	6.2	68
42	Copy Number Variations of TBK1 in Australian Patients With Primary Open-Angle Glaucoma. <i>American Journal of Ophthalmology</i> , 2015, 159, 124-130.e1.	3.3	68
43	Measurement of Systemic Mitochondrial Function in Advanced Primary Open-Angle Glaucoma and Leber Hereditary Optic Neuropathy. <i>PLoS ONE</i> , 2015, 10, e0140919.	2.5	66
44	Common Sequence Variation in the VEGFA Gene Predicts Risk of Diabetic Retinopathy. , 2009, 50, 5552.		64
45	Australian and New Zealand Registry of Advanced Glaucoma: methodology and recruitment. <i>Clinical and Experimental Ophthalmology</i> , 2012, 40, 569-575.	2.6	64
46	Cross-ancestry genome-wide association analysis of corneal thickness strengthens link between complex and Mendelian eye diseases. <i>Nature Communications</i> , 2018, 9, 1864.	12.8	63
47	Association of TCF4 and CLU polymorphisms with Fuchs's endothelial dystrophy and implication of CLU and TGFBI proteins in the disease process. <i>European Journal of Human Genetics</i> , 2012, 20, 632-638.	2.8	61
48	Serum selenium status in Graves' disease with and without orbitopathy: a case-control study. <i>Clinical Endocrinology</i> , 2014, 80, 905-910.	2.4	58
49	WNT10A exonic variant increases the risk of keratoconus by decreasing corneal thickness. <i>Human Molecular Genetics</i> , 2015, 24, 5060-5068.	2.9	58
50	Genetic analysis of the soluble epoxide hydrolase gene, EPHX2, in subclinical cardiovascular disease in the Diabetes Heart Study. <i>Diabetes and Vascular Disease Research</i> , 2008, 5, 128-134.	2.0	57
51	Sequence Variation in DDAH1 and DDAH2 Genes Is Strongly and Additively Associated with Serum ADMA Concentrations in Individuals with Type 2 Diabetes. <i>PLoS ONE</i> , 2010, 5, e9462.	2.5	54
52	Mutation in TMEM98 in a Large White Kindred With Autosomal Dominant Nanophthalmos Linked to 17p12-q12. <i>JAMA Ophthalmology</i> , 2014, 132, 970.	2.5	54
53	Multiethnic Genome-Wide Association Study of Diabetic Retinopathy Using Liability Threshold Modeling of Duration of Diabetes and Glycemic Control. <i>Diabetes</i> , 2019, 68, 441-456.	0.6	54
54	A novel mutation in the Connexin 46 gene causes autosomal dominant congenital cataract with incomplete penetrance. <i>Journal of Medical Genetics</i> , 2004, 41, e106-e106.	3.2	53

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55	Diabetic Retinopathy Is Associated With Elevated Serum Asymmetric and Symmetric Dimethylarginines. <i>Diabetes Care</i> , 2009, 32, 2084-2086.	8.6	53
56	A single-nucleotide polymorphism in the MicroRNA-146a gene is associated with diabetic nephropathy and sight-threatening diabetic retinopathy in Caucasian patients. <i>Acta Diabetologica</i> , 2016, 53, 643-650.	2.5	53
57	A genome-wide association study suggests new evidence for an association of the <i>NADPH Oxidase 4 (NOX4)</i> gene with severe diabetic retinopathy in type 2 diabetes. <i>Acta Ophthalmologica</i> , 2018, 96, e811-e819.	1.1	52
58	Association Between Erythropoietin Gene Polymorphisms and Diabetic Retinopathy. <i>JAMA Ophthalmology</i> , 2010, 128, 102.	2.4	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. <i>BMC Medical Genetics</i> , 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. <i>Ophthalmology</i> , 2013, 120, 1135-1143.	5.2	48
62	Genetic and Environmental Risk Factors for Keratoconus. <i>Annual Review of Vision Science</i> , 2020, 6, 25-46.	4.4	46
63	T-786C Polymorphism of the Endothelial Nitric Oxide Synthase Gene Is Associated with Albuminuria in the Diabetes Heart Study. <i>Journal of the American Society of Nephrology: JASN</i> , 2005, 16, 1085-1090.	6.1	45
64	Rapid inexpensive genome-wide association using pooled whole blood. <i>Genome Research</i> , 2009, 19, 2075-2080.	5.5	45
65	Glaucoma spectrum and age-related prevalence of individuals with FOXC1 and PITX2 variants. <i>European Journal of Human Genetics</i> , 2017, 25, 839-847.	2.8	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. <i>PLoS ONE</i> , 2013, 8, e67903.	2.5	42
68	Association of Î±2-Heremans-Schmid Glycoprotein Polymorphisms with Subclinical Atherosclerosis. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2007, 92, 345-352.	3.6	40
69	Aldose Reductase Gene Polymorphisms and Diabetic Retinopathy Susceptibility. <i>Diabetes Care</i> , 2010, 33, 1834-1836.	8.6	39
70	Association of Genetic Variants in the <i>TMCO1</i> Gene 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. <i>Journal of Proteome Research</i> , 2011, 10, 3522-3529.	3.7	37
72	Replication and Meta-Analysis of Candidate Loci Identified Variation at <i>RAB3GAP1</i> Associated 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. <i>Molecular Vision</i> , 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. <i>Human Genetics</i> , 2010, 127, 33-44.	3.8	36
75	A multi-ethnic genome-wide association study implicates collagen matrix integrity and cell differentiation pathways in keratoconus. <i>Communications Biology</i> , 2021, 4, 266.	4.4	36
76	Variability of Serum Soluble Intercellular Adhesion Molecule-1 Measurements Attributable to a Common Polymorphism. <i>Clinical Chemistry</i> , 2004, 50, 2185-2187.	3.2	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). <i>American Heart Journal</i> , 2006, 151, 706-711.	2.7	35
78	Prenatal detection of congenital bilateral cataract leading to the diagnosis of Nance-Horan syndrome in the extended family. <i>Prenatal Diagnosis</i> , 2007, 27, 662-664.	2.3	35
79	Mutations in the EPHA2 Gene Are a Major Contributor to Inherited Cataracts in South-Eastern Australia. <i>PLoS ONE</i> , 2013, 8, e72518.	2.5	35
80	Chromosome 9p21 primary open-angle glaucoma susceptibility locus: a review. <i>Clinical and Experimental Ophthalmology</i> , 2014, 42, 25-32.	2.6	35
81	Association of Genetic Variation With Keratoconus. <i>JAMA Ophthalmology</i> , 2020, 138, 174.	2.5	34
82	Heritability and Expression of C-Reactive Protein in Type 2 Diabetes in the Diabetes Heart Study. <i>Annals of Human Genetics</i> , 2006, 70, 717-725.	0.8	33
83	Analysis combining correlated glaucoma traits identifies five new risk loci for open-angle glaucoma. <i>Scientific Reports</i> , 2018, 8, 3124.	3.3	33
84	Prevalence of <i>FOXC1</i> Variants in Individuals With a Suspected Diagnosis of Primary Congenital Glaucoma. <i>JAMA Ophthalmology</i> , 2019, 137, 348.	2.5	33
85	Biallelic CPAMD8 Variants Are a Frequent Cause of Childhood and Juvenile Open-Angle Glaucoma. <i>Ophthalmology</i> , 2020, 127, 758-766.	5.2	33
86	The PITX3 gene in posterior polar congenital cataract in Australia. <i>Molecular Vision</i> , 2006, 12, 367-71.	1.1	33
87	Identification of podocin (NPHS2) gene mutations in African Americans with nondiabetic end-stage renal disease. <i>Kidney International</i> , 2005, 68, 256-262.	5.2	32
88	Human Lipoxygenase Pathway Gene Variation and Association with Markers of Subclinical Atherosclerosis in the Diabetes Heart Study. <i>Mediators of Inflammation</i> , 2010, 2010, 1-9.	3.0	32
89	Macular Ganglion Cell Inner Plexiform Layer Loss Precedes Peripapillary Retinal Nerve Fiber Layer Loss in Glaucoma with Lower Intraocular Pressure. <i>Ophthalmology</i> , 2019, 126, 1119-1130.	5.2	32
90	Myocilin Gene Gln368Ter Variant Penetrance and Association With Glaucoma in Population-Based and Registry-Based Studies. <i>JAMA Ophthalmology</i> , 2019, 137, 28.	2.5	32

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91	Association of <i>eNOS</i> Polymorphisms 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.	3.3	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.	2.5	29
94	Identification of a Novel Oligomerization Disrupting Mutation in <i>CRY1A</i> Associated with Congenital Cataract in a South Australian Family. Human Mutation, 2013, 34, 435-438.	2.5	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.	3.3	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 the <i>FTL</i> gene causes hereditary hyperferritinemia cataract syndrome (HHCS) by alteration of the transcription start site. Human Mutation, 2007, 28, 742-742.	2.5	25
99	Biological effect of <i>LOXL1</i> coding variants associated with pseudoexfoliation syndrome. Experimental Eye Research, 2016, 146, 212-223.	2.6	25
100	Myocilin Predictive Genetic Testing for Primary Open-Angle Glaucoma Leads to Early Identification of At-Risk Individuals. Ophthalmology, 2017, 124, 303-309.	5.2	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.	4.8	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.	2.2	24
104	TGC repeat expansion in the <i>TCF4</i> gene increases the risk of Fuchs' endothelial corneal dystrophy in Australian cases. PLoS ONE, 2017, 12, e0183719.	2.5	24
105	The association of hepatocyte growth factor (<i>HGF</i>) 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 <i>OR52B4</i> gene with anti-VEGF treatment response in age-related macular degeneration. Scientific Reports, 2016, 6, 37924.	3.3	23
107	Review of the prevalence of diabetic retinopathy in Indigenous Australians. Clinical and Experimental Ophthalmology, 2014, 42, 875-882.	2.6	22
108	Evaluation of Shared Genetic Susceptibility to High and Low Myopia and Hyperopia. JAMA Ophthalmology, 2021, 139, 601.	2.5	22

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109	Severe prekallikrein deficiency associated with homozygosity for an Arg94Stop nonsense mutation. <i>British Journal of Haematology</i> , 2004, 127, 220-223.	2.5	21
110	Occurrence of <i>CYP1B1</i> Mutations in Juvenile Open-Angle Glaucoma With Advanced Visual Field Loss. <i>JAMA Ophthalmology</i> , 2015, 133, 826.	2.5	21
111	Assessment of polygenic effects links primary open-angle glaucoma and age-related macular degeneration. <i>Scientific Reports</i> , 2016, 6, 26885.	3.3	21
112	Novel missense mutation in the bZIP transcription factor, MAF, associated with congenital cataract, developmental delay, seizures and hearing loss (Aym�-Gripp syndrome). <i>BMC Medical Genetics</i> , 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. <i>PLoS ONE</i> , 2018, 13, e0199178.	2.5	21
114	Association of Protein Tyrosine Phosphatase-N1 Polymorphisms With Coronary Calcified Plaque in the Diabetes Heart Study. <i>Diabetes</i> , 2006, 55, 651-658.	0.6	20
115	Genome-wide association studies in the hunt for genes causing primary open-angle glaucoma: a review. <i>Clinical and Experimental Ophthalmology</i> , 2012, 40, 358-363.	2.6	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. <i>Ophthalmology</i> , 2015, 122, 1828-1836.	5.2	20
118	High-Throughput Genetic Screening of 51 Pediatric Cataract Genes Identifies Causative Mutations in Inherited Pediatric Cataract in South Eastern Australia. <i>G3: Genes, Genomes, Genetics</i> , 2017, 7, 3257-3268.	1.8	20
119	The telomere of human chromosome 1p contains at least two independent autosomal dominant congenital cataract genes. <i>British Journal of Ophthalmology</i> , 2005, 89, 831-834.	3.9	19
120	Association of genes of lipid metabolism with measures of subclinical cardiovascular disease in the Diabetes Heart Study. <i>Journal of Medical Genetics</i> , 2005, 42, 720-724.	3.2	19
121	Ethnic and Mouse Strain Differences in Central Corneal Thickness and Association with Pigmentation Phenotype. <i>PLoS ONE</i> , 2011, 6, e22103.	2.5	19
122	Association of Arachidonate 12-Lipoxygenase Genotype Variation and Glycemic Control With Albuminuria in Type 2 Diabetes. <i>American Journal of Kidney Diseases</i> , 2008, 52, 242-250.	1.9	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. <i>Diabetes</i> , 2017, 66, 3130-3141.	0.6	17
125	Key challenges in bringing CRISPR-mediated somatic cell therapy into the clinic. <i>Genome Medicine</i> , 2017, 9, 85.	8.2	17
126	The genetic and clinical landscape of nanophthalmos and posterior microphthalmos in an Australian cohort. <i>Clinical Genetics</i> , 2020, 97, 764-769.	2.0	17

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127	EPHA2 MUTATIONS CONTRIBUTE TO CONGENITAL CATARACT THROUGH DIVERSE MECHANISMS. <i>Molecular Vision</i> , 2016, 22, 18-30.	1.1	16
128	Recurrent mutation in the crystallin alpha A gene associated with inherited paediatric cataract. <i>BMC Research Notes</i> , 2016, 9, 83.	1.4	15
129	Diabetic macular oedema: clinical risk factors and emerging genetic influences. <i>Australasian journal of optometry, The</i> , 2017, 100, 569-576.	1.3	15
130	Identification of novel mutations causing pediatric cataract in Bhutan, Cambodia, and Sri Lanka. <i>Molecular Genetics & Genomic Medicine</i> , 2018, 6, 555-564.	1.2	15
131	Genetic study of diabetic retinopathy: recruitment methodology and analysis of baseline characteristics. <i>Clinical and Experimental Ophthalmology</i> , 2014, 42, 486-493.	2.6	14
132	Predictive genetic testing in minors for Myocilin juvenile onset open angle glaucoma. <i>Clinical Genetics</i> , 2015, 88, 584-588.	2.0	14
133	Rare variants in optic disc area gene <i>CARD10</i> enriched in primary open-angle glaucoma. <i>Molecular Genetics & Genomic Medicine</i> , 2016, 4, 624-633.	1.2	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 <i>ZNF469</i> 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. <i>Ophthalmic Genetics</i> , 2018, 39, 221-227.	1.2	13
138	Ocular Expression and Distribution of Products of the POAG-Associated Chromosome 9p21 Gene Region. <i>PLoS ONE</i> , 2013, 8, e75067.	2.5	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. <i>BMC Medical Genetics</i> , 2016, 17, 30.	2.1	12
141	Partial duplication of the CRYBB1-CRYBA4 locus is associated with autosomal dominant congenital cataract. <i>European Journal of Human Genetics</i> , 2017, 25, 711-718.	2.8	12
142	Candidate gene study to investigate the genetic determinants of normal variation in central corneal thickness. <i>Molecular Vision</i> , 2010, 16, 562-9.	1.1	12
143	Relationship between DDAH gene variants and serum ADMA level in individuals with type 1 diabetes. <i>Journal of Diabetes and Its Complications</i> , 2012, 26, 195-198.	2.3	11
144	Predictive genetic testing experience for myocilin primary open-angle glaucoma using the Australian and New Zealand Registry of Advanced Glaucoma. <i>Genetics in Medicine</i> , 2014, 16, 558-563.	2.4	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 <sc><i>COL8A2</i></sc> gene in an <sc>A</sc>ustralian family with earlyâ€onset <sc>F</sc>uchsâ€™ endothelial corneal dystrophy. Clinical and Experimental Ophthalmology, 2014, 42, 198-200.	2.6	10
147	Pooled genome wide association detects association upstream of FCRL3 with Gravesâ€™ disease. BMC Genomics, 2016, 17, 939.	2.8	10
148	Epha2 genotype influences ultraviolet radiation induced cataract in mice. Experimental Eye Research, 2019, 188, 107806.	2.6	10
149	Common variants in SOX-2 and congenital cataract genes contribute to age-related nuclear cataract. Communications Biology, 2020, 3, 755.	4.4	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.	5.2	9
154	Ethical Considerations for the Return of Incidental Findings in Ophthalmic Genomic Research. Translational Vision Science and Technology, 2016, 5, 3.	2.2	9
155	Rapid and efficient cataract gene evaluation in FO zebrafish using CRISPR-Cas9 ribonucleoprotein complexes. Methods, 2021, 194, 37-47.	3.8	9
156	Identifying Genetic Risk Factors for Diabetic Macular Edema and the Response to Treatment. Journal of Diabetes Research, 2020, 2020, 1-12.	2.3	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.	2.5	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.	2.0	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.	2.6	7
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