David A. Mackey

List of Publications by Year
in descending order

486 papers	25,382 citations	10389 72 h-index	12272 133 g-index
507 all docs	507 docs citations	507 times ranked	21788 citing authors

#	Article	IF	Citations
1	Identification of a Gene That Causes Primary Open Angle Glaucoma. Science, 1997, 275, 668-670.	12.6	1,274
2	A large genome-wide association study of age-related macular degeneration highlights contributions of rare and common variants. Nature Genetics, 2016, 48, 134-143.	21.4	1,167
3	Rare and low-frequency coding variants alter human adult height. Nature, 2017, 542, 186-190.	27.8	544
4	Human TUBB3 Mutations Perturb Microtubule Dynamics, Kinesin Interactions, and Axon Guidance. Cell, 2010, 140, 74-87.	28.9	515
5	Analysis of Myocilin Mutations in 1703 Glaucoma Patients From Five Different Populations. Human Molecular Genetics, 1999, 8, 899-905.	2.9	496
6	A single EFEMP1 mutation associated with both Malattia Leventinese and Doyne honeycomb retinal dystrophy. Nature Genetics, 1999, 22, 199-202.	21.4	453
7	Clinical Features Associated with Mutations in the Chromosome 1 Open-Angle Glaucoma Gene (GLC1A). New England Journal of Medicine, 1998, 338, 1022-1027.	27.0	423
8	Genome-wide meta-analyses of multiancestry cohorts identify multiple new susceptibility loci for refractive error and myopia. Nature Genetics, 2013, 45, 314-318.	21.4	398
9	Genome-wide association study identifies susceptibility loci for open angle glaucoma at TMCO1 and CDKN2B-AS1. Nature Genetics, 2011, 43, 574-578.	21.4	381
10	Common variants near CAV1 and CAV2 are associated with primary open-angle glaucoma. Nature Genetics, 2010, 42, 906-909.	21.4	357
11	Retinal Vascular Caliber: Systemic, Environmental, and Genetic Associations. Survey of Ophthalmology, 2009, 54, 74-95.	4.0	351
12	The Association between Time Spent Outdoors and Myopia in Children and Adolescents. Ophthalmology, 2012, 119, 2141-2151.	5.2	337
13	Mutations in LRP5 or FZD4 Underlie the Common Familial Exudative Vitreoretinopathy Locus on Chromosome 11q. American Journal of Human Genetics, 2004, 74, 721-730.	6.2	333
14	Genome-wide association analyses identify multiple loci associated with central corneal thickness and keratoconus. Nature Genetics, 2013, 45, 155-163.	21.4	269
15	Null Mutations in LTBP2 Cause Primary Congenital Glaucoma. American Journal of Human Genetics, 2009, 84, 664-671.	6.2	255
16	Genetic influences on handedness: Data from 25,732 Australian and Dutch twin families. Neuropsychologia, 2009, 47, 330-337.	1.6	252
17	Mutations in the pre-mRNA splicing factor gene PRPC8 in autosomal dominant retinitis pigmentosa (RP13). Human Molecular Genetics, 2001, 10, 1555-1562.	2.9	251
18	Genome-wide association meta-analysis highlights light-induced signaling as a driver for refractive error. Nature Genetics, 2018, 50, 834-848.	21.4	239

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19	Axial Length Variation Impacts on Superficial Retinal Vessel Density and Foveal Avascular Zone Area Measurements Using Optical Coherence Tomography Angiography. , 2017, 58, 3065.		215
20	The Pedigree Rate of Sequence Divergence in the Human Mitochondrial Genome: There Is a Difference Between Phylogenetic and Pedigree Rates. American Journal of Human Genetics, 2003, 72, 659-670.	6.2	213
21	Genome-wide analysis of multi-ancestry cohorts identifies new loci influencing intraocular pressure and susceptibility to glaucoma. Nature Genetics, 2014, 46, 1126-1130.	21.4	212
22	Genome-wide association analysis identifies TXNRD2, ATXN2 and FOXC1 as susceptibility loci for primary open-angle glaucoma. Nature Genetics, 2016, 48, 189-194.	21.4	211
23	A genome-wide association study for myopia and refractive error identifies a susceptibility locus at 15q25. Nature Genetics, 2010, 42, 902-905.	21.4	204
24	A genome-wide association study identifies a susceptibility locus for refractive errors and myopia at 15q14. Nature Genetics, 2010, 42, 897-901.	21.4	200
25	Genome-wide meta-analysis identifies 127 open-angle glaucoma loci with consistent effect across ancestries. Nature Communications, 2021, 12, 1258.	12.8	196
26	Multitrait analysis of glaucoma identifies new risk loci and enables polygenic prediction of disease susceptibility and progression. Nature Genetics, 2020, 52, 160-166.	21.4	192
27	Common variants near ABCA1, AFAP1 and GMDS confer risk of primary open-angle glaucoma. Nature Genetics, 2014, 46, 1120-1125.	21.4	186
28	A Spectrum of FOXC1 Mutations Suggests Gene Dosage as a Mechanism for Developmental Defects of the Anterior Chamber of the Eye. American Journal of Human Genetics, 2001, 68, 364-372.	6.2	185
29	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.	21.4	180
30	Mutations in TCF8 Cause Posterior Polymorphous Corneal Dystrophy and Ectopic Expression of COL4A3 by Corneal Endothelial Cells. American Journal of Human Genetics, 2005, 77, 694-708.	6.2	177
31	Angiopoietin receptor TEK mutations underlie primary congenital glaucoma with variable expressivity. Journal of Clinical Investigation, 2016, 126, 2575-2587.	8.2	175
32	Directional dominance on stature and cognition inÂdiverse human populations. Nature, 2015, 523, 459-462.	27.8	173
33	Evaluation of optineurin sequence variations in 1,048 patients with open-angle glaucoma. American Journal of Ophthalmology, 2003, 136, 904-910.	3.3	164
34	Common Genetic Determinants of Intraocular Pressure and Primary Open-Angle Glaucoma. PLoS Genetics, 2012, 8, e1002611.	3.5	164
35	Mutations in TSPAN12 Cause Autosomal-Dominant Familial Exudative Vitreoretinopathy. American Journal of Human Genetics, 2010, 86, 248-253.	6.2	161
36	Digital Quantification of Human Eye Color Highlights Genetic Association of Three New Loci. PLoS Genetics, 2010, 6, e1000934.	3.5	161

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37	IMI â€“ Myopia Genetics Report. , 2019, 60, M89.		156
38	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.	2.9	152
39	Genome-wide association study of intraocular pressure uncovers new pathways to glaucoma. Nature Genetics, 2018, 50, 1067-1071.	21.4	152
40	Spectrum, frequency and penetrance of OPA1 mutations in dominant optic atrophy. Human Molecular Genetics, 2001, 10, 1369-1378.	2.9	147
41	The Heritability of Ocular Traits. Survey of Ophthalmology, 2010, 55, 561-583.	4.0	140
42	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.	6.2	139
43	Four Novel Loci (19q13, 6q24, 12q24, and 5q14) Influence the Microcirculation In Vivo. PLoS Genetics, 2010, 6, e1001184.	3.5	134
44	Central Corneal Thickness Is Highly Heritable: The Twin Eye Studies. , 2005, 46, 3718.		133
45	Genome-wide association identifies ATOH7 as a major gene determining human optic disc size. Human Molecular Genetics, 2010, 19, 2716-2724.	2.9	133
46	Common Genetic Variants near the Brittle Cornea Syndrome Locus ZNF469 Influence the Blinding Disease Risk Factor Central Corneal Thickness. PLoS Genetics, 2010, 6, e1000947.	3.5	130
47	Aetiology of congenital and paediatric cataract in an Australian population. British Journal of Ophthalmology, 2002, 86, 782-786.	3.9	129
48	Inhaled corticosteroids, family history, and risk of glaucoma. Ophthalmology, 1999, 106, 2301-2306.	5.2	122
49	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
50	Current state and future prospects of artificial intelligence in ophthalmology: a review. Clinical and Experimental Ophthalmology, 2019, 47, 128-139.	2.6	118
51	Autosomal Dominant Optic Atrophy: Penetrance and Expressivity in Patients With OPA1 Mutations. American Journal of Ophthalmology, 2007, 143, 656-662.e1.	3.3	116
52	Genome-wide association study identifies seven novel susceptibility loci for primary open-angle glaucoma. Human Molecular Genetics, 2018, 27, 1486-1496.	2.9	111
53	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
54	Evidence for genetic heterogeneity within eight glaucoma families, with the GLC1A Gln368STOP mutation being an important phenotypic modifier11None of the authors has a financial interest relating to this article.. Ophthalmology, 2001, 108, 1607-1620.	5.2	106

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55	Sequence Analysis of the Mitochondrial Genomes from Dutch Pedigrees with Leber Hereditary Optic Neuropathy. American Journal of Human Genetics, 2003, 72, 1460-1469.	6.2	106
56	Myocilinallele-specific glaucoma phenotype database. Human Mutation, 2008, 29, 207-211.	2.5	106
57	A common variant near TGFBR3 is associated with primary open angle glaucoma. Human Molecular Genetics, 2015, 24, 3880-3892.	2.9	105
58	Meta-analysis of geneâ€“environment-wide association scans accounting for education level identifies additional loci for refractive error. Nature Communications, 2016, 7, 11008.	12.8	104
59	Retinopathy of prematurity: recent advances in our understanding. British Journal of Ophthalmology, 2002, 86, 696-700.	3.9	99
60	Identity-by-descent approach to gene localisation in eight individuals affected by keratoconus from north-west Tasmania, Australia. Human Genetics, 2002, 110, 462-470.	3.8	97
61	A common variant mapping to CACNA1A is associated with susceptibility to exfoliation syndrome. Nature Genetics, 2015, 47, 387-392.	21.4	97
62	Mutations of theEPHA2receptor tyrosine kinase gene cause autosomal dominant congenital cataract. Human Mutation, 2009, 30, E603-E611.	2.5	96
63	The Path to Open-Angle Glaucoma Gene Discovery: Endophenotypic Status of Intraocular Pressure, Cup-to-Disc Ratio, and Central Corneal Thickness. , 2010, 51, 3509.		94
64	Meta-analysis of human methylation data for evidence of sex-specific autosomal patterns. BMC Genomics, 2014, 15, 981.	2.8	94
65	How does spending time outdoors protect against myopia? A review. British Journal of Ophthalmology, 2020, 104, 593-599.	3.9	94
66	Angiopoietin-1 is required for Schlemmâ€™s canal development in mice and humans. Journal of Clinical Investigation, 2017, 127, 4421-4436.	8.2	94
67	Spectrum and Frequency ofFZD4Mutations in Familial Exudative Vitreoretinopathy. , 2004, 45, 2083.		89
68	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
69	Drusen in patient-derived hiPSC-RPE models of macular dystrophies. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E8214-E8223.	7.1	88
70	Mutations in a protein target of the Pim-1 kinase associated with the RP9 form of autosomal dominant retinitis pigmentosa. European Journal of Human Genetics, 2002, 10, 245-249.	2.8	87
71	Novel pleiotropic risk loci for melanoma and nevus density implicate multiple biological pathways. Nature Communications, 2018, 9, 4774.	12.8	87
72	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.	21.4	86

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73	Evidence for Paleolithic and Neolithic Gene Flow in Europe. American Journal of Human Genetics, 1998, 62, 488-491.	6.2	85
74	Myopia Is Associated With Lower Vitamin D Status in Young Adults. , 2014, 55, 4552.		84
75	Identification ofKIF21AMutations as a Rare Cause of Congenital Fibrosis of the Extraocular Muscles Type 3 (CFEOM3). , 2004, 45, 2218.		83
76	Leber hereditary optic neuropathy in Australia. Australian and New Zealand Journal of Ophthalmology, 1992, 20, 177-184.	0.4	80
77	Myopia in Young Adults Is Inversely Related to an Objective Marker of Ocular Sun Exposure: The Western Australian Raine Cohort Study. American Journal of Ophthalmology, 2014, 158, 1079-1085.e2.	3.3	80
78	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.	3.3	80
79	The Association between Time Spent Outdoors and Myopia Using a Novel Biomarker of Outdoor Light Exposure. , 2012, 53, 4363.		79
80	ARHGEF12 influences the risk of glaucoma by increasing intraocular pressure. Human Molecular Genetics, 2015, 24, 2689-2699.	2.9	79
81	Leber's hereditary optic neuropathy triggered by antiretroviral therapy for human immunodeficiency virus. Eye, 2003, 17, 312-317.	2.1	77
82	Mutations in the NDP gene: contribution to Norrie disease, familial exudative vitreoretinopathy and retinopathy of prematurity. Clinical and Experimental Ophthalmology, 2006, 34, 682-688.	2.6	76
83	Investigation of crystallin genes in familial cataract, and report of two disease associated mutations. British Journal of Ophthalmology, 2004, 88, 79-83.	3.9	75
84	AUTOSOMAL RECESSIVE VITELLIFORM MACULAR DYSTROPHY IN A LARGE COHORT OF VITELLIFORM MACULAR DYSTROPHY PATIENTS. Retina, 2011, 31, 581-595.	1.7	75
85	Inherited retinal diseases are the most common cause of blindness in the working-age population in Australia. Ophthalmic Genetics, 2021, 42, 431-439.	1.2	75
86	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
87	Prevalence of Keratoconus Based on Scheimpflug Imaging. Ophthalmology, 2021, 128, 515-521.	5.2	73
88	Metaâ€analysis of Genomeâ€Wide Association Studies Identifies Novel Loci Associated With Optic Disc Morphology. Genetic Epidemiology, 2015, 39, 207-216.	1.3	72
89	Complex genetics of complex traits: the case of primary open-angle glaucoma. Clinical and Experimental Ophthalmology, 2006, 34, 472-484.	2.6	71
90	Recessive Mutations in SLC38A8 Cause Foveal Hypoplasia and Optic Nerve Misrouting without Albinism. American Journal of Human Genetics, 2013, 93, 1143-1150.	6.2	71

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91	How significant is a family history of glaucoma? Experience from the Glaucoma Inheritance Study in Tasmania. Clinical and Experimental Ophthalmology, 2007, 35, 793-799.	2.6	70
92	Identification of myopia-associated WNT7B polymorphisms provides insights into the mechanism underlying the development of myopia. Nature Communications, 2015, 6, 6689.	12.8	70
93	Copy Number Variations of TBK1 in Australian Patients With Primary Open-Angle Glaucoma. American Journal of Ophthalmology, 2015, 159, 124-130.e1.	3.3	68
94	Large scale international replication and meta-analysis study confirms association of the 15q14 locus with myopia. The CREAM consortium. Human Genetics, 2012, 131, 1467-1480.	3.8	67
95	Measurement of Systemic Mitochondrial Function in Advanced Primary Open-Angle Glaucoma and Leber Hereditary Optic Neuropathy. PLoS ONE, 2015, 10, e0140919.	2.5	66
96	mtDNA Mutations That Cause Optic Neuropathy: How Do We Know?. American Journal of Human Genetics, 1998, 62, 196-202.	6.2	65
97	Deletion of the OPA1 gene in a dominant optic atrophy family: evidence that haploinsufficiency is the cause of disease. Journal of Medical Genetics, 2002, 39, 47e-47.	3.2	65
98	An International Collaborative Family-Based Whole-Genome Linkage Scan for High-Grade Myopia. , 2009, 50, 3116.		65
99	Australian and New Zealand Registry of Advanced Glaucoma: methodology and recruitment. Clinical and Experimental Ophthalmology, 2012, 40, 569-575.	2.6	64
100	Evidence for a novel glaucoma locus at chromosome 3p21-22. Human Genetics, 2005, 117, 249-257.	3.8	63
101	Birth of a cohort â€" the first 20 years of the Raine study. Medical Journal of Australia, 2012, 197, 608-610.	1.7	63
102	Cross-ancestry genome-wide association analysis of corneal thickness strengthens link between complex and Mendelian eye diseases. Nature Communications, 2018, 9, 1864.	12.8	63
103	Emerging Mitochondrial Therapeutic Targets in Optic Neuropathies. , 2016, 165, 132-152.		62
104	Description of X-Linked Megalocornea With Identification of the Gene Locus. JAMA Ophthalmology, 1991, 109, 829.	2.4	61
105	The natural history of OPA1-related autosomal dominant optic atrophy. British Journal of Ophthalmology, 2008, 92, 1333-1336.	3.9	61
106	What is the appropriate age cutâ€off for cycloplegia in refraction?. Acta Ophthalmologica, 2014, 92, e458-62.	1.1	61
107	The apolipoprotein epsilon4 gene is associated with elevated risk of normal tension glaucoma. Molecular Vision, 2002, 8, 389-93.	1.1	61
108	Primary infantile glaucoma in an Australian population. Clinical and Experimental Ophthalmology, 2004, 32, 14-18.	2.6	60

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109	Nail-patella syndrome and its association with glaucoma: a review of eight families. British Journal of Ophthalmology, 2006, 90, 1505-1509.	3.9	60
110	The Heritability of Corneal Hysteresis and Ocular Pulse Amplitude. Ophthalmology, 2008, 115, 1545-1549.	5.2	60
111	LTBP2 and CYP1B1 mutations and associated ocular phenotypes in the Roma/Gypsy founder population. European Journal of Human Genetics, 2011, 19, 326-333.	2.8	60
112	A Glaucoma Case-control Study of the WDR36 Gene D658G Sequence Variant. American Journal of Ophthalmology, 2006, 142, 324-325.	3.3	59
113	COL1A1andCOL2A1Genes and Myopia Susceptibility: Evidence of Association and Suggestive Linkage to theCOL2A1Locus. , 2009, 50, 4080.		59
114	GWAS Findings for Human Iris Patterns: Associations with Variants in Genes that Influence Normal Neuronal Pattern Development. American Journal of Human Genetics, 2011, 89, 334-343.	6.2	59
115	X-Linked Megalocornea Caused by Mutations in CHRDL1 Identifies an Essential Role for Ventroptin in Anterior Segment Development. American Journal of Human Genetics, 2012, 90, 247-259.	6.2	59
116	No Association Between Variations in the WDR36 Gene and Primary Open-Angle Glaucoma. JAMA Ophthalmology, 2007, 125, 434.	2.4	58
117	WNT10A exonic variant increases the risk of keratoconus by decreasing corneal thickness. Human Molecular Genetics, 2015, 24, 5060-5068.	2.9	58
118	Hereditary Hyperferritinemia-Cataract Syndrome. JAMA Ophthalmology, 2003, 121, 1753.	2.4	57
119	Genetic Association of Insulin-like Growth Factor-1 Polymorphisms with High-Grade Myopia in an International Family Cohort. , 2010, 51, 4476.		57
120	The association between pterygium and conjunctival ultraviolet autofluorescence: The Norfolk Island Eye Study. Acta Ophthalmologica, 2013, 91, 363-370.	1.1	57
121	Quantitative Analysis of Retinal Vessel Attenuation in Eyes with Retinitis Pigmentosa. , 2012, 53, 4306.		56
122	Assessing the Genetic Predisposition of Education on Myopia: A Mendelian Randomization Study. Genetic Epidemiology, 2016, 40, 66-72.	1.3	56
123	Low-Penetrance Branches in Matrilineal Pedigrees with Leber Hereditary Optic Neuropathy. American Journal of Human Genetics, 1998, 63, 1220-1224.	6.2	55
124	Low penetrance of the 14484 LHON mutation when it arises in a non-haplogroup J mtDNA background. American Journal of Medical Genetics Part A, 2003, 119A, 147-151.	2.4	55
125	Giant cell arteritis: ophthalmic manifestations of a systemic disease. Graefe's Archive for Clinical and Experimental Ophthalmology, 2016, 254, 2291-2306.	1.9	54
126	Mitochondrial replacement in an iPSC model of Leberâ€™s hereditary optic neuropathy. Aging, 2017, 9, 1341-1350.	3.1	54

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127	A novel mutation in the Connexin 46 gene causes autosomal dominant congenital cataract with incomplete penetrance. Journal of Medical Genetics, 2004, 41, e106-e106.	3.2	53
128	PAX6Mutations May Be Associated with High Myopia. Ophthalmic Genetics, 2007, 28, 179-182.	1.2	53
129	Incidence and Progression of Myopia in Early Adulthood. JAMA Ophthalmology, 2022, 140, 162.	2.5	53
130	Raine Eye Health Study: Design, Methodology and Baseline Prevalence of Ophthalmic Disease in a Birth-cohort Study of Young Adults. Ophthalmic Genetics, 2013, 34, 199-208.	1.2	51
131	Genetic variation affects morphological retinal phenotypes extracted from UK Biobank optical coherence tomography images. PLoS Genetics, 2021, 17, e1009497.	3.5	50
132	Heritability of Central Corneal Thickness in Nuclear Families. , 2009, 50, 4087.		49
133	Optic disc morphology - Rethinking shape. Progress in Retinal and Eye Research, 2009, 28, 227-248.	15.5	49
134	Mitochondrial DNA haplogroup distribution within Leber hereditary optic neuropathy pedigrees. Journal of Medical Genetics, 2004, 41, e41-e41.	3.2	48
135	Prevalence of CYP1B1 mutations in Australian patients with primary congenital glaucoma. Clinical Genetics, 2007, 72, 255-260.	2.0	48
136	Genetic Dissection of Myopia. Ophthalmology, 2008, 115, 1053-1057.e2.	5.2	48
137	Higher Prevalence of Myocilin Mutations in Advanced Glaucoma in Comparison with Less Advanced Disease in an Australasian Disease Registry. Ophthalmology, 2013, 120, 1135-1143.	5.2	48
138	Genome-Wide Association Shows thatÂPigmentation Genes Play a Role in SkinÂAging. Journal of Investigative Dermatology, 2017, 137, 1887-1894.	0.7	48
139	Glaucoma Phenotype in Pedigrees With the Myocilin Thr377Met Mutation. JAMA Ophthalmology, 2003, 121, 1172.	2.4	47
140	Genetically low vitamin D concentrations and myopic refractive error: a Mendelian randomization study. International Journal of Epidemiology, 2017, 46, 1882-1890.	1.9	47
141	The genetic profile of Leber congenital amaurosis in an Australian cohort. Molecular Genetics & Genomic Medicine, 2017, 5, 652-667.	1.2	47
142	Mobile touch screen device use and associations with musculoskeletal symptoms and visual health in a nationally representative sample of Singaporean adolescents. Ergonomics, 2019, 62, 778-793.	2.1	47
143	Mitochondrial Oxidative Phosphorylation Compensation May Preserve Vision in Patients with OPA1-Linked Autosomal Dominant Optic Atrophy. PLoS ONE, 2011, 6, e21347.	2.5	47
144	Congenital fibrosis of the vertically acting extraocular muscles maps to the FEOM3 locus. Human Genetics, 2002, 110, 510-512.	3.8	46

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145	Heritability of Anterior Chamber Depth as an Intermediate Phenotype of Angle-Closure in Chinese: The Guangzhou Twin Eye Study. , 2008, 49, 81.		46
146	Three subgroups of patients from the United Kingdom with Leber hereditary optic neuropathy. Eye, 1994, 8, 431-436.	2.1	45
147	Rationale, design and methods for a community-based study of clustering and cumulative effects of chronic disease processes and their effects on ageing: the Busselton healthy ageing study. BMC Public Health, 2013, 13, 936.	2.9	45
148	Distribution of astigmatism as a function of age in an Australian population. Acta Ophthalmologica, 2015, 93, e377-85.	1.1	44
149	Classification of iris colour: review and refinement of a classification schema. Clinical and Experimental Ophthalmology, 2011, 39, 462-471.	2.6	43
150	Reliability and validity of conjunctival ultraviolet autofluorescence measurement. British Journal of Ophthalmology, 2012, 96, 801-805.	3.9	43
151	Haplotype reference consortium panel: Practical implications of imputations with large reference panels. Human Mutation, 2017, 38, 1025-1032.	2.5	43
152	Glaucoma spectrum and age-related prevalence of individuals with FOXC1 and PITX2 variants. European Journal of Human Genetics, 2017, 25, 839-847.	2.8	43
153	Linkage to 10q22 for Maximum Intraocular Pressure and 1p32 for Maximum Cup-to-Disc Ratio in an Extended Primary Open-Angle Glaucoma Pedigree. , 2005, 46, 3723.		42
154	Hypomethylation of the IL17RC Promoter in Peripheral Blood Leukocytes Is Not A Hallmark of Age-Related Macular Degeneration. Cell Reports, 2013, 5, 1527-1535.	6.4	42
155	Study of mitochondrial respiratory defects on reprogramming to human induced pluripotent stem cells. Aging, 2016, 8, 945-957.	3.1	42
156	Disease Severity of Familial Glaucoma Compared With Sporadic Glaucoma. JAMA Ophthalmology, 2006, 124, 950.	2.4	41
157	Retinal microvessels reflect familial vulnerability to psychotic symptoms: A comparison of twins discordant for psychotic symptoms and controls. Schizophrenia Research, 2015, 164, 47-52.	2.0	41
158	Association of Birth Weight With Type 2 Diabetes and Glycemic Traits. JAMA Network Open, 2019, 2, e1910915.	5.9	41
159	Nanceâ€“Horan syndrome protein, NHS, associates with epithelial cell junctions. Human Molecular Genetics, 2006, 15, 1972-1983.	2.9	40
160	Analysis of 15 primary open-angle glaucoma families from Australia identifies a founder effect for the Q368STOP mutation of myocilin. Human Genetics, 2003, 112, 110-116.	3.8	39
161	Effect of Birth Parameters on Retinal Vascular Caliber. Hypertension, 2009, 53, 487-493.	2.7	39
162	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, 3343-3348.	2.9	39

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163	Low-dose (0.01%) atropine eye-drops to reduce progression of myopia in children: a multicentre placebo-controlled randomised trial in the UK (CHAMP-UK)â€"study protocol. British Journal of Ophthalmology, 2020, 104, 950-955.	3.9	39
164	X-linked megalocornea: close linkage to DXS87 and DXS94. Human Genetics, 1989, 83, 292-294.	3.8	38
165	Quantitative Genetic Analysis of the Retinal Vascular Caliber. Hypertension, 2009, 54, 788-795.	2.7	38
166	Comparison of three methods of intraocular pressure measurement and their relation to central corneal thickness. Eye, 2010, 24, 1165-1170.	2.1	38
167	Optic Disc Evaluation in Optic Neuropathies. Ophthalmology, 2011, 118, 964-970.	5.2	38
168	Association of Genetic Variants in theTMCO1Gene with Clinical Parameters Related to Glaucoma and Characterization of the Protein in the Eye. , 2012, 53, 4917.		38
169	Establishment and evolution of the <scp>A</scp>ustralian <scp>I</scp>nherited <scp>R</scp>etinal <scp>D</scp>isease <scp>R</scp>egister and <scp>DNA B</scp>ank. Clinical and Experimental Ophthalmology, 2013, 41, 476-483.	2.6	37
170	Pterygium and conjunctival ultraviolet autofluorescence in young <scp>A</scp>ustralian adults: the <scp>R</scp>aine study. Clinical and Experimental Ophthalmology, 2015, 43, 300-307.	2.6	37
171	A COL17A1 Splice-Altering Mutation Is Prevalent in Inherited Recurrent Corneal Erosions. Ophthalmology, 2016, 123, 709-722.	5.2	37
172	Twins Eye Study in Tasmania (TEST): Rationale and Methodology to Recruit and Examine Twins. Twin Research and Human Genetics, 2009, 12, 441-454.	0.6	36
173	Novel quantitative trait loci for central corneal thickness identified by candidate gene analysis of osteogenesis imperfecta genes. Human Genetics, 2010, 127, 33-44.	3.8	36
174	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.	2.9	36
175	Intrasession Repeatability and Interocular Symmetry of Foveal Avascular Zone and Retinal Vessel Density in OCT Angiography. Translational Vision Science and Technology, 2018, 7, 6.	2.2	36
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