David A. Mackey

List of Publications by Citations

472 papers	19,166 citations	67 h-index	122 g-index
507 ext. papers	22,494 ext. citations	6.6 avg, IF	6.12 L-index

#	Paper	IF	Citations
472	Identification of a gene that causes primary open angle glaucoma. *Science*, **1997**, 275, 668-70	33.3	1124
471	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
470	Analysis of myocilin mutations in 1703 glaucoma patients from five different populations. *Human Molecular Genetics*, **1999**, 8, 899-905	5.6	451
469	Human TUBB3 mutations perturb microtubule dynamics, kinesin interactions, and axon guidance. *Cell*, **2010**, 140, 74-87	56.2	418
468	Rare and low-frequency coding variants alter human adult height. *Nature*, **2017**, 542, 186-190	50.4	412
467	A single EFEMP1 mutation associated with both Malattia Leventinese and Doyne honeycomb retinal dystrophy. *Nature Genetics*, **1999**, 22, 199-202	36.3	384
466	Clinical features associated with mutations in the chromosome 1 open-angle glaucoma gene (GLC1A). *New England Journal of Medicine*, **1998**, 338, 1022-7	59.2	368
465	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
464	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
463	Common variants near CAV1 and CAV2 are associated with primary open-angle glaucoma. *Nature Genetics*, **2010**, 42, 906-9	36.3	303
462	Retinal vascular caliber: systemic, environmental, and genetic associations. *Survey of Ophthalmology*, **2009**, 54, 74-95	6.1	296
461	Mutations in LRP5 or FZD4 underlie the common familial exudative vitreoretinopathy locus on chromosome 11q. *American Journal of Human Genetics*, **2004**, 74, 721-30	11	287
460	The association between time spent outdoors and myopia in children and adolescents: a systematic review and meta-analysis. *Ophthalmology*, **2012**, 119, 2141-51	7.3	263
459	Genome-wide association analyses identify multiple loci associated with central corneal thickness and keratoconus. *Nature Genetics*, **2013**, 45, 155-63	36.3	222
458	Null mutations in LTBP2 cause primary congenital glaucoma. *American Journal of Human Genetics*, **2009**, 84, 664-71	11	208
457	Genetic influences on handedness: data from 25,732 Australian and Dutch twin families. *Neuropsychologia*, **2009**, 47, 330-7	3.2	205
456	Mutations in the pre-mRNA splicing factor gene PRPC8 in autosomal dominant retinitis pigmentosa (RP13). *Human Molecular Genetics*, **2001**, 10, 1555-62	5.6	198

455	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, 659-70	11	185
454	A genome-wide association study identifies a susceptibility locus for refractive errors and myopia at 15q14. *Nature Genetics*, **2010**, 42, 897-901	36.3	181
453	A genome-wide association study for myopia and refractive error identifies a susceptibility locus at 15q25. *Nature Genetics*, **2010**, 42, 902-5	36.3	179
452	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
451	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, 364-72	11	165
450	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
449	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	11	149
448	Evaluation of optineurin sequence variations in 1,048 patients with open-angle glaucoma. *American Journal of Ophthalmology*, **2003**, 136, 904-10	4.9	149
447	Axial Length Variation Impacts on Superficial Retinal Vessel Density and Foveal Avascular Zone Area Measurements Using Optical Coherence Tomography Angiography **2017**, 58, 3065-3072		148
446	Common variants near ABCA1, AFAP1 and GMDS confer risk of primary open-angle glaucoma. *Nature Genetics*, **2014**, 46, 1120-1125	36.3	141
445	Mutations in TSPAN12 cause autosomal-dominant familial exudative vitreoretinopathy. *American Journal of Human Genetics*, **2010**, 86, 248-53	11	137
444	Digital quantification of human eye color highlights genetic association of three new loci. *PLoS Genetics*, **2010**, 6, e1000934	6	135
443	Genome-wide association meta-analysis highlights light-induced signaling as a driver for refractive error. *Nature Genetics*, **2018**, 50, 834-848	36.3	135
442	Common genetic determinants of intraocular pressure and primary open-angle glaucoma. *PLoS Genetics*, **2012**, 8, e1002611	6	131
441	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
440	Directional dominance on stature and cognition in diverse human populations. *Nature*, **2015**, 523, 459-462	30.4	119
439	Genome-wide association identifies ATOH7 as a major gene determining human optic disc size. *Human Molecular Genetics*, **2010**, 19, 2716-24	5.6	118
438	Angiopoietin receptor TEK mutations underlie primary congenital glaucoma with variable expressivity. *Journal of Clinical Investigation*, **2016**, 126, 2575-87	15.9	117

437	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
436	Central corneal thickness is highly heritable: the twin eye studies. *Investigative Ophthalmology and Visual Science*, **2005**, 46, 3718-22		115
435	Four novel Loci (19q13, 6q24, 12q24, and 5q14) influence the microcirculation in vivo. *PLoS Genetics* , **2010**, 6, e1001184	6	111
434	The heritability of ocular traits. *Survey of Ophthalmology*, **2010**, 55, 561-83	6.1	111
433	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
432	Spectrum, frequency and penetrance of OPA1 mutations in dominant optic atrophy. *Human Molecular Genetics*, **2001**, 10, 1369-78	5.6	105
431	Aetiology of congenital and paediatric cataract in an Australian population. *British Journal of Ophthalmology*, **2002**, 86, 782-6	5.5	102
430	Autosomal dominant optic atrophy: penetrance and expressivity in patients with OPA1 mutations. *American Journal of Ophthalmology*, **2007**, 143, 656-62	4.9	101
429	Sequence analysis of the mitochondrial genomes from Dutch pedigrees with Leber hereditary optic neuropathy. *American Journal of Human Genetics*, **2003**, 72, 1460-9	11	98
428	Evidence for genetic heterogeneity within eight glaucoma families, with the GLC1A Gln368STOP mutation being an important phenotypic modifier. *Ophthalmology*, **2001**, 108, 1607-20	7.3	97
427	Inhaled corticosteroids, family history, and risk of glaucoma. *Ophthalmology*, **1999**, 106, 2301-6	7.3	96
426	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
425	Mutations of the EPHA2 receptor tyrosine kinase gene cause autosomal dominant congenital cataract. *Human Mutation*, **2009**, 30, E603-11	4.7	87
424	Genome-wide association study of intraocular pressure uncovers new pathways to glaucoma. *Nature Genetics*, **2018**, 50, 1067-1071	36.3	86
423	A common variant near TGFBR3 is associated with primary open angle glaucoma. *Human Molecular Genetics*, **2015**, 24, 3880-92	5.6	84
422	Myocilin allele-specific glaucoma phenotype database. *Human Mutation*, **2008**, 29, 207-11	4.7	84
421	Spectrum and frequency of FZD4 mutations in familial exudative vitreoretinopathy. *Investigative Ophthalmology and Visual Science*, **2004**, 45, 2083-90		81
420	Identity-by-descent approach to gene localisation in eight individuals affected by keratoconus from north-west Tasmania, Australia. *Human Genetics*, **2002**, 110, 462-70	6.3	81

419	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
418	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
417	The path to open-angle glaucoma gene discovery: endophenotypic status of intraocular pressure, cup-to-disc ratio, and central corneal thickness **2010**, 51, 3509-14		79
416	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
415	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, 245-9	5.3	76
414	Evidence for Paleolithic and Neolithic gene flow in Europe. *American Journal of Human Genetics*, **1998**, 62, 488-92	11	74
413	IMI - Myopia Genetics Report **2019**, 60, M89-M105		73
412	Genome-wide association study identifies seven novel susceptibility loci for primary open-angle glaucoma. *Human Molecular Genetics*, **2018**, 27, 1486-1496	5.6	72
411	Identification of KIF21A mutations as a rare cause of congenital fibrosis of the extraocular muscles type 3 (CFEOM3). *Investigative Ophthalmology and Visual Science*, **2004**, 45, 2218-23		72
410	Current state and future prospects of artificial intelligence in ophthalmology: a review. *Clinical and Experimental Ophthalmology*, **2019**, 47, 128-139	2.4	72
409	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
408	A common variant mapping to CACNA1A is associated with susceptibility to exfoliation syndrome. *Nature Genetics*, **2015**, 47, 387-92	36.3	70
407	Meta-analysis of 542,934 subjects of European ancestry identifies new genes and mechanisms predisposing to refractive error and myopia. *Nature Genetics*, **2020**, 52, 401-407	36.3	68
406	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
405	Leber's hereditary optic neuropathy triggered by antiretroviral therapy for human immunodeficiency virus. *Eye*, **2003**, 17, 312-7	4.4	67
404	The association between time spent outdoors and myopia using a novel biomarker of outdoor light exposure **2012**, 53, 4363-70		66
403	Myopia is associated with lower vitamin D status in young adults **2014**, 55, 4552-9		65
402	Meta-analysis of human methylation data for evidence of sex-specific autosomal patterns. *BMC Genomics*, **2014**, 15, 981	4.5	65

401	Retinopathy of prematurity: recent advances in our understanding. *British Journal of Ophthalmology*, **2002**, 86, 696-700	5.5	65
400	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
399	mtDNA mutations that cause optic neuropathy: how do we know?. *American Journal of Human Genetics*, **1998**, 62, 196-202	11	63
398	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, 1079-85	4.9	62
397	ARHGEF12 influences the risk of glaucoma by increasing intraocular pressure. *Human Molecular Genetics*, **2015**, 24, 2689-99	5.6	62
396	Leber hereditary optic neuropathy in Australia. *Australian and New Zealand Journal of Ophthalmology*, **1992**, 20, 177-84		62
395	Evidence for a novel glaucoma locus at chromosome 3p21-22. *Human Genetics*, **2005**, 117, 249-57	6.3	61
394	Autosomal recessive vitelliform macular dystrophy in a large cohort of vitelliform macular dystrophy patients. *Retina*, **2011**, 31, 581-95	3.6	60
393	An international collaborative family-based whole-genome linkage scan for high-grade myopia **2009**, 50, 3116-27		60
392	Complex genetics of complex traits: the case of primary open-angle glaucoma. *Clinical and Experimental Ophthalmology*, **2006**, 34, 472-84	2.4	60
391	Investigation of crystallin genes in familial cataract, and report of two disease associated mutations. *British Journal of Ophthalmology*, **2004**, 88, 79-83	5.5	60
390	Genome-wide association meta-analysis of individuals of European ancestry identifies new loci explaining a substantial fraction of hair color variation and heritability. *Nature Genetics*, **2018**, 50, 652-656	36.3	59
389	Meta-analysis of Genome-Wide Association Studies Identifies Novel Loci Associated With Optic Disc Morphology. *Genetic Epidemiology*, **2015**, 39, 207-16	2.6	58
388	A Glaucoma Case-control Study of the WDR36 Gene D658G sequence variant. *American Journal of Ophthalmology*, **2006**, 142, 324-5	4.9	58
387	Childhood gene-environment interactions and age-dependent effects of genetic variants associated with refractive error and myopia: The CREAM Consortium. *Scientific Reports*, **2016**, 6, 25853	4.9	57
386	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	11.5	57
385	Large scale international replication and meta-analysis study confirms association of the 15q14 locus with myopia. The CREAM consortium. *Human Genetics*, **2012**, 131, 1467-80	6.3	57
384	Angiopoietin-1 is required for Schlemm's canal development in mice and humans. *Journal of Clinical Investigation*, **2017**, 127, 4421-4436	15.9	57

383	Identification of myopia-associated WNT7B polymorphisms provides insights into the mechanism underlying the development of myopia. *Nature Communications*, **2015**, 6, 6689	17.4	56
382	Recessive mutations in SLC38A8 cause foveal hypoplasia and optic nerve misrouting without albinism. *American Journal of Human Genetics*, **2013**, 93, 1143-50	11	56
381	No association between variations in the WDR36 gene and primary open-angle glaucoma. *JAMA Ophthalmology*, **2007**, 125, 434-6		56
380	The natural history of OPA1-related autosomal dominant optic atrophy. *British Journal of Ophthalmology*, **2008**, 92, 1333-6	5.5	55
379	The apolipoprotein epsilon4 gene is associated with elevated risk of normal tension glaucoma. *Molecular Vision*, **2002**, 8, 389-93	2.3	55
378	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
377	The heritability of corneal hysteresis and ocular pulse amplitude: a twin study. *Ophthalmology*, **2008**, 115, 1545-9	7.3	54
376	How significant is a family history of glaucoma? Experience from the Glaucoma Inheritance Study in Tasmania. *Clinical and Experimental Ophthalmology*, **2007**, 35, 793-9	2.4	54
375	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, e47	5.8	54
374	Birth of a cohort--the first 20 years of the Raine study. *Medical Journal of Australia*, **2012**, 197, 608-10	4	53
373	Low-penetrance branches in matrilineal pedigrees with Leber hereditary optic neuropathy. *American Journal of Human Genetics*, **1998**, 63, 1220-4	11	52
372	COL1A1 and COL2A1 genes and myopia susceptibility: evidence of association and suggestive linkage to the COL2A1 locus **2009**, 50, 4080-6		51
371	WNT10A exonic variant increases the risk of keratoconus by decreasing corneal thickness. *Human Molecular Genetics*, **2015**, 24, 5060-8	5.6	50
370	LTBP2 and CYP1B1 mutations and associated ocular phenotypes in the Roma/Gypsy founder population. *European Journal of Human Genetics*, **2011**, 19, 326-33	5.3	50
369	Measurement of Systemic Mitochondrial Function in Advanced Primary Open-Angle Glaucoma and Leber Hereditary Optic Neuropathy. *PLoS ONE*, **2015**, 10, e0140919	3.7	47
368	GWAS findings for human iris patterns: associations with variants in genes that influence normal neuronal pattern development. *American Journal of Human Genetics*, **2011**, 89, 334-43	11	47
367	Primary infantile glaucoma in an Australian population. *Clinical and Experimental Ophthalmology*, **2004**, 32, 14-8	2.4	47
366	Description of X-linked megalocornea with identification of the gene locus. *JAMA Ophthalmology*, **1991**, 109, 829-33		47

365	Genome-wide meta-analysis identifies 127 open-angle glaucoma loci with consistent effect across ancestries. *Nature Communications*, **2021**, 12, 1258	17.4	47
364	Novel pleiotropic risk loci for melanoma and nevus density implicate multiple biological pathways. *Nature Communications*, **2018**, 9, 4774	17.4	47
363	Emerging Mitochondrial Therapeutic Targets in Optic Neuropathies. *Pharmacology & Therapeutics*, **2016**, 165, 132-52	13.9	46
362	The association between pterygium and conjunctival ultraviolet autofluorescence: the Norfolk Island Eye Study. *Acta Ophthalmologica*, **2013**, 91, 363-70	3.7	45
361	Australian and New Zealand Registry of Advanced Glaucoma: methodology and recruitment. *Clinical and Experimental Ophthalmology*, **2012**, 40, 569-75	2.4	45
360	PAX6 mutations may be associated with high myopia. *Ophthalmic Genetics*, **2007**, 28, 179-82	1.2	45
359	Nail-patella syndrome and its association with glaucoma: a review of eight families. *British Journal of Ophthalmology*, **2006**, 90, 1505-9	5.5	45
358	Hereditary hyperferritinemia-cataract syndrome: prevalence, lens morphology, spectrum of mutations, and clinical presentations. *JAMA Ophthalmology*, **2003**, 121, 1753-61		45
357	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
356	What is the appropriate age cut-off for cycloplegia in refraction?. *Acta Ophthalmologica*, **2014**, 92, e458-62	3.7	43
355	Genetic association of insulin-like growth factor-1 polymorphisms with high-grade myopia in an international family cohort **2010**, 51, 4476-9		43
354	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, 147-51		42
353	How does spending time outdoors protect against myopia? A review. *British Journal of Ophthalmology*, **2020**, 104, 593-599	5.5	41
352	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	41
351	Three subgroups of patients from the United Kingdom with Leber hereditary optic neuropathy. *Eye*, **1994**, 8 (Pt 4), 431-6	4.4	41
350	Quantitative analysis of retinal vessel attenuation in eyes with retinitis pigmentosa **2012**, 53, 4306-14		40
349	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, 247-59	11	40
348	Heritability of central corneal thickness in nuclear families **2009**, 50, 4087-90		40

347	Heritability of anterior chamber depth as an intermediate phenotype of angle-closure in Chinese: the Guangzhou Twin Eye Study. *Investigative Ophthalmology and Visual Science*, **2008**, 49, 81-6		40
346	Glaucoma phenotype in pedigrees with the myocilin Thr377Met mutation. *JAMA Ophthalmology*, **2003**, 121, 1172-80		40
345	Optic disc morphology--rethinking shape. *Progress in Retinal and Eye Research*, **2009**, 28, 227-48	20.5	39
344	Reliability and validity of conjunctival ultraviolet autofluorescence measurement. *British Journal of Ophthalmology*, **2012**, 96, 801-5	5.5	39
343	Genetic dissection of myopia: evidence for linkage of ocular axial length to chromosome 5q. *Ophthalmology*, **2008**, 115, 1053-1057.e2	7.3	39
342	Linkage to 10q22 for maximum intraocular pressure and 1p32 for maximum cup-to-disc ratio in an extended primary open-angle glaucoma pedigree. *Investigative Ophthalmology and Visual Science*, **2005**, 46, 3723-9		39
341	Assessing the Genetic Predisposition of Education on Myopia: A Mendelian Randomization Study. *Genetic Epidemiology*, **2016**, 40, 66-72	2.6	39
340	Prevalence of CYP1B1 mutations in Australian patients with primary congenital glaucoma. *Clinical Genetics*, **2007**, 72, 255-60	4	38
339	Hypomethylation of the IL17RC promoter in peripheral blood leukocytes is not a hallmark of age-related macular degeneration. *Cell Reports*, **2013**, 5, 1527-35	10.6	37
338	Congenital fibrosis of the vertically acting extraocular muscles maps to the FEOM3 locus. *Human Genetics*, **2002**, 110, 510-2	6.3	37
337	Mitochondrial oxidative phosphorylation compensation may preserve vision in patients with OPA1-linked autosomal dominant optic atrophy. *PLoS ONE*, **2011**, 6, e21347	3.7	37
336	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
335	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
334	Effect of birth parameters on retinal vascular caliber: the Twins Eye Study in Tasmania. *Hypertension*, **2009**, 53, 487-93	8.5	36
333	Mitochondrial DNA haplogroup distribution within Leber hereditary optic neuropathy pedigrees. *Journal of Medical Genetics*, **2004**, 41, e41	5.8	36
332	The genetic profile of Leber congenital amaurosis in an Australian cohort. *Molecular Genetics & Genomic Medicine*, **2017**, 5, 652-667	2.3	35
331	Mitochondrial replacement in an iPSC model of Leber's hereditary optic neuropathy. *Aging*, **2017**, 9, 1341-1350	5.6	35
330	Genetically low vitamin D concentrations and myopic refractive error: a Mendelian randomization study. *International Journal of Epidemiology*, **2017**, 46, 1882-1890	7.8	34

329	Quantitative genetic analysis of the retinal vascular caliber: the Australian Twins Eye Study. *Hypertension*, **2009**, 54, 788-95	8.5	34
328	Nance-Horan syndrome protein, NHS, associates with epithelial cell junctions. *Human Molecular Genetics*, **2006**, 15, 1972-83	5.6	34
327	Disease severity of familial glaucoma compared with sporadic glaucoma. *JAMA Ophthalmology*, **2006**, 124, 950-4		34
326	Analysis of 15 primary open-angle glaucoma families from Australia identifies a founder effect for the Q368STOP mutation of myocilin. *Human Genetics*, **2003**, 112, 110-6	6.3	34
325	Retinal microvessels reflect familial vulnerability to psychotic symptoms: A comparison of twins discordant for psychotic symptoms and controls. *Schizophrenia Research*, **2015**, 164, 47-52	3.6	33
324	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
323	Twins eye study in Tasmania (TEST): rationale and methodology to recruit and examine twins. *Twin Research and Human Genetics*, **2009**, 12, 441-54	2.2	33
322	Study of mitochondrial respiratory defects on reprogramming to human induced pluripotent stem cells. *Aging*, **2016**, 8, 945-57	5.6	33
321	Giant cell arteritis: ophthalmic manifestations of a systemic disease. *Graefe's Archive for Clinical and Experimental Ophthalmology*, **2016**, 254, 2291-2306	3.8	33
320	A genome-wide association study of intra-ocular pressure suggests a novel association in the gene FAM125B in the TwinsUK cohort. *Human Molecular Genetics*, **2014**, 23, 3343-8	5.6	32
319	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	4.1	31
318	Genetic variants near PDGFRA are associated with corneal curvature in Australians **2012**, 53, 7131-6		31
317	Mutations in the EPHA2 gene are a major contributor to inherited cataracts in South-Eastern Australia. *PLoS ONE*, **2013**, 8, e72518	3.7	31
316	Comparison of three methods of intraocular pressure measurement and their relation to central corneal thickness. *Eye*, **2010**, 24, 1165-70	4.4	31
315	Genome-Wide Association Shows that Pigmentation Genes Play a Role in Skin Aging. *Journal of Investigative Dermatology*, **2017**, 137, 1887-1894	4.3	30
314	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
313	Optic disc evaluation in optic neuropathies: the optic disc assessment project. *Ophthalmology*, **2011** , 118, 964-70	7.3	29
312	Classification of iris colour: review and refinement of a classification schema. *Clinical and Experimental Ophthalmology*, **2011**, 39, 462-71	2.4	29

311	Cardiac arrhythmia and Leber's hereditary optic neuropathy. *Lancet, The*, **1992**, 339, 1427-8	40	29
310	X-linked megalocornea: close linkage to DXS87 and DXS94. *Human Genetics*, **1989**, 83, 292-4	6.3	29
309	Intrasession Repeatability and Interocular Symmetry of Foveal Avascular Zone and Retinal Vessel Density in OCT Angiography. *Translational Vision Science and Technology*, **2018**, 7, 6	3.3	28
308	Genome-wide association study identifies nine novel loci for 2D:4D finger ratio, a putative retrospective biomarker of testosterone exposure in utero. *Human Molecular Genetics*, **2018**, 27, 2025-2038	5.6	27
307	Prognosis for splicing factor PRPF8 retinitis pigmentosa, novel mutations and correlation between human and yeast phenotypes. *Human Mutation*, **2010**, 31, E1361-76	4.7	27
306	The 'GIST' score: ranking glaucoma for genetic studies. Glaucoma Inheritance Study of Tasmania. *Ophthalmic Genetics*, **1996**, 17, 199-208	1.2	27
305	Pterygium and conjunctival ultraviolet autofluorescence in young Australian adults: the Raine study. *Clinical and Experimental Ophthalmology*, **2015**, 43, 300-7	2.4	26
304	Genotypic and phenotypic spectrum of X-linked retinoschisis in Australia. *Clinical and Experimental Ophthalmology*, **2005**, 33, 233-9	2.4	26
303	When do myopia genes have their effect? Comparison of genetic risks between children and adults. *Genetic Epidemiology*, **2016**, 40, 756-766	2.6	26
302	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
301	Analysis combining correlated glaucoma traits identifies five new risk loci for open-angle glaucoma. *Scientific Reports*, **2018**, 8, 3124	4.9	25
300	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
299	Rock, paper and scissors? Traumatic paediatric cataract in Victoria 1992-2006. *Clinical and Experimental Ophthalmology*, **2010**, 38, 237-41	2.4	25
298	Genetic analysis of the clusterin gene in pseudoexfoliation syndrome. *Molecular Vision*, **2008**, 14, 1727-36	2.3	25
297	Prevalence of Keratoconus Based on Scheimpflug Imaging: The Raine Study. *Ophthalmology*, **2021**, 128, 515-521	7.3	25
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295	Birth order and myopia. *Ophthalmic Epidemiology*, **2013**, 20, 375-84	1.9	24
294	Genome-wide association study of intraocular pressure identifies the GLCCI1/ICA1 region as a glaucoma susceptibility locus. *Human Molecular Genetics*, **2013**, 22, 4653-60	5.6	24

293	Establishment and evolution of the Australian Inherited Retinal Disease Register and DNA Bank. *Clinical and Experimental Ophthalmology*, **2013**, 41, 476-83	2.4	24
292	Broad phenotypic variability in a single pedigree with a novel 1410delC mutation in the PST domain of the PAX6 gene. *Human Mutation*, **2002**, 20, 322	4.7	24
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