Andrew R Webster

List of Publications by Year
in descending order

145 papers	6,817 citations	87888 38 h-index	82547 72 g-index
149 all docs	149 docs citations	149 times ranked	7515 citing authors

#	Article	IF	Citations
1	Characterization of Retinal Function Using Microperimetry-Derived Metrics in Both Adults and Children With RPGR-Associated Retinopathy. American Journal of Ophthalmology, 2022, 234, 81-90.	3.3	8
2	A rare canonical splice-site variant in VPS13B causes attenuated Cohen syndrome. Ophthalmic Genetics, 2022, 43, 110-115.	1.2	2
3	Collaborative Research and Development of a Novel, Patient-Centered Digital Platform (MyEyeSite) for Rare Inherited Retinal Disease Data: Acceptability and Feasibility Study. JMIR Formative Research, 2022, 6, e21341.	1.4	2
4	X-Linked Retinoschisis. Ophthalmology, 2022, 129, 542-551.	5.2	19
5	WFS1-Associated Optic Neuropathy: Genotype-Phenotype Correlations and Disease Progression. American Journal of Ophthalmology, 2022, 241, 9-27.	3.3	8
6	Electrical responses from human retinal cone pathways associate with a common genetic polymorphism implicated in myopia. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	7.1	8
7	Variability of retinopathy consequent upon novel mutations in LAMA1. Ophthalmic Genetics, 2022, 43, 671-678.	1.2	1
8	Axial Length Distributions in Patients With Genetically Confirmed Inherited Retinal Diseases. , 2022, 63, 15.		6
9	A clinical study of patients with novel CDHR1 genotypes associated with late-onset macular dystrophy. Eye, 2021, 35, 1482-1489.	2.1	5
10	Clinical and Genetic Findings in CTNNA1-Associated Macular Pattern Dystrophy. Ophthalmology, 2021, 128, 952-955.	5.2	8
11	Sector Retinitis Pigmentosa: Extending the Molecular Genetics Basis and Elucidating the Natural History. American Journal of Ophthalmology, 2021, 221, 299-310.	3.3	20
12	The X-linked retinopathies: Physiological insights, pathogenic mechanisms, phenotypic features and novel therapies. Progress in Retinal and Eye Research, 2021, 82, 100898.	15.5	65
13	Ceramide synthase TLCD3B is a novel gene associated with human recessive retinal dystrophy. Genetics in Medicine, 2021, 23, 488-497.	2.4	7
14	Autosomal Recessive Bestrophinopathy. Ophthalmology, 2021, 128, 706-718.	5.2	31
15	New variants and in silico analyses in GRK1 associated Oguchi disease. Human Mutation, 2021, 42, 164-176.	2.5	7
16	KCNV2-Associated Retinopathy: Detailed Retinal Phenotype and Structural Endpointsâ€"KCNV2 Study Group Report 2. American Journal of Ophthalmology, 2021, 230, 1-11.	3.3	11
17	KCNV2-Associated Retinopathy: Genetics, Electrophysiology, and Clinical Courseâ€"KCNV2 Study Group Report 1. American Journal of Ophthalmology, 2021, 225, 95-107.	3.3	17
18	CNGB1 â€related rodâ€cone dystrophy: A mutation review and update. Human Mutation, 2021, 42, 641-666.	2.5	16

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19	Broadening INPP5E phenotypic spectrum: detection of rare variants in syndromic and non-syndromic IRD. Npj Genomic Medicine, 2021, 6, 53.	3.8	8
20	Expanding the clinical phenotype in patients with disease causing variants associated with atypical Usher syndrome. Ophthalmic Genetics, 2021, 42, 664-673.	1.2	14
21	Prevalence of electronegative electroretinograms in a healthy adult cohort. BMJ Open Ophthalmology, 2021, 6, e000751.	1.6	3
22	Comparison of in silico strategies to prioritize rare genomic variants impacting RNA splicing for the diagnosis of genomic disorders. Scientific Reports, 2021, 11, 20607.	3.3	37
23	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880.	27.0	352
24	Awareness of olfactory impairment in a cohort of patients with CNGB1-associated retinitis pigmentosa. Eye, 2020, 34, 783-784.	2.1	2
25	A new paradigm for delivering personalised care: integrating genetics with surgical interventions in BEST1 mutations. Eye, 2020, 34, 577-583.	2.1	11
26	GUCY2D-Associated Leber Congenital Amaurosis: A Retrospective Natural History Study in Preparation for Trials of Novel Therapies. American Journal of Ophthalmology, 2020, 210, 59-70.	3.3	39
27	Transcorneal Electrical Stimulation for the Treatment of Retinitis Pigmentosa: A Multicenter Safety Study of the OkuStimÂ® System (TESOLA-Study). Ophthalmic Research, 2020, 63, 234-243.	1.9	18
28	Reanalysis of Association of Pro50Leu Substitution in Guanylate Cyclase Activating Protein-1 With Dominant Retinal Dystrophy. JAMA Ophthalmology, 2020, 138, 200.	2.5	5
29	Detailed Phenotyping and Therapeutic Strategies for Intronic ABCA4 Variants in Stargardt Disease. Molecular Therapy - Nucleic Acids, 2020, 21, 412-427.	5.1	55
30	Structural Variants Create New Topological-Associated Domains and Ectopic Retinal Enhancer-Gene Contact in Dominant Retinitis Pigmentosa. American Journal of Human Genetics, 2020, 107, 802-814.	6.2	75
31	A genetic and clinical study of individuals with nonsyndromic retinopathy consequent upon sequence variants in <scp>HGSNAT</scp>, the gene associated with Sanfilippo C mucopolysaccharidosis. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 631-643.	1.6	12
32	Long-term follow-up of a case of posterior microphthalmos (PRSS56) with hyperautofluorescent retinal pigment epithelial deposits. European Journal of Ophthalmology, 2020, , 112067212094975.	1.3	2
33	Macula-predominant retinopathy associated with biallelic variants in RDH12. Ophthalmic Genetics, 2020, 41, 612-615.	1.2	12
34	Genetic Basis of Inherited Retinal Disease in a Molecularly Characterized Cohort of More Than 3000 Families from the United Kingdom. Ophthalmology, 2020, 127, 1384-1394.	5.2	131
35	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	27.8	338
36	Quantifying the Separation Between the Retinal Pigment Epithelium and Bruch's Membrane using Optical Coherence Tomography in Patients with Inherited Macular Degeneration. Translational Vision Science and Technology, 2020, 9, 26.	2.2	15

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37	Clinical and preclinical therapeutic outcome metrics for USH2A-related disease. Human Molecular Genetics, 2020, 29, 1882-1899.	2.9	24
38	The majority of autosomal recessive nanophthalmos and posterior microphthalmia can be attributed to biallelic sequence and structural variants in MFRP and PRSS56. Scientific Reports, 2020, 10, 1289.	3.3	24
39	Characterisation of microvascular abnormalities using OCT angiography in patients with biallelic variants in USH2A and MYO7A. British Journal of Ophthalmology, 2020, 104, 480-486.	3.9	12
40	An Improved Phenotype-Driven Tool for Rare Mendelian Variant Prioritization: Benchmarking Exomiser on Real Patient Whole-Exome Data. Genes, 2020, 11, 460.	2.4	42
41	Monozygotic twins discordant for asymmetric pigmented paravenous chorioretinal atrophy. Retinal Cases and Brief Reports, 2020, Publish Ahead of Print, .	0.6	4
42	SSBP1 mutations in dominant optic atrophy with variable retinal degeneration. Annals of Neurology, 2019, 86, 368-383.	5.3	41
43	Missense variants in the conserved transmembrane M2 protein domain of KCNJ13 associated with retinovascular changes in humans and zebrafish. Experimental Eye Research, 2019, 189, 107852.	2.6	13
44	Benefit of an electronic headâ€mounted low vision aid. Ophthalmic and Physiological Optics, 2019, 39, 422-431.	2.0	23
45	ABCA4-associated disease as a model for missing heritability in autosomal recessive disorders: novel noncoding splice, cis-regulatory, structural, and recurrent hypomorphic variants. Genetics in Medicine, 2019, 21, 1761-1771.	2.4	111
46	Nonsense-mediated mRNA decay efficiency varies in choroideremia providing a target to boost small molecule therapeutics. Human Molecular Genetics, 2019, 28, 1865-1871.	2.9	25
47	Unique noncoding variants upstream of PRDM13 are associated with a spectrum of developmental retinal dystrophies including progressive bifocal chorioretinal atrophy. Human Mutation, 2019, 40, 578-587.	2.5	19
48	Delineating the expanding phenotype associated with SCAPER gene mutation. American Journal of Medical Genetics, Part A, 2019, 179, 1665-1671.	1.2	10
49	Germline selection shapes human mitochondrial DNA diversity. Science, 2019, 364, .	12.6	178
50	Isolated rod dysfunction associated with a novel genotype of CNGB1. American Journal of Ophthalmology Case Reports, 2019, 14, 83-86.	0.7	9
51	Loss-of-Function Mutations in the CFH Gene Affecting Alternatively Encoded Factor H-like 1 Protein Cause Dominant Early-Onset Macular Drusen. Ophthalmology, 2019, 126, 1410-1421.	5.2	25
52	Clinical and Molecular Characterization of Familial Exudative Vitreoretinopathy Associated With Microcephaly. American Journal of Ophthalmology, 2019, 207, 87-98.	3.3	20
53	Rod-cone dystrophy associated with the Gly167Asp variant in PRPH2. Ophthalmic Genetics, 2019, 40, 188-189.	1.2	6
54	Phagosomal and mitochondrial alterations in RPE may contribute to KCNJ13 retinopathy. Scientific Reports, 2019, 9, 3793.	3.3	29

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55	Detailed clinical characterisation, unique features and natural history of autosomal recessive RDH12-associated retinal degeneration. British Journal of Ophthalmology, 2019, 103, bjophthalmol-2018-313580.	3.9	20
56	Macular maldevelopment inATF6-mediated retinal dysfunction. Ophthalmic Genetics, 2019, 40, 564-569.	1.2	3
57	Deep Phenotyping ofPDE6C-Associated Achromatopsia. , 2019, 60, 5112.		44
58	Unilateral pigmentary retinopathy: a retrospective case series. Acta Ophthalmologica, 2019, 97, e601-e617.	1.1	17
59	Deep-intronic ABCA4 variants explain missing heritability in Stargardt disease and allow correction of splice defects by antisense oligonucleotides. Genetics in Medicine, 2019, 21, 1751-1760.	2.4	147
60	Retinopathy Associated with Biallelic Mutations in PYGM (McArdle Disease). Ophthalmology, 2019, 126, 320-322.	5.2	12
61	Prevalence of cystoid macular oedema, epiretinal membrane and cataract in retinitis pigmentosa. British Journal of Ophthalmology, 2019, 103, 1163-1166.	3.9	55
62	CELLULAR IMAGING OF THE TAPETAL-LIKE REFLEX IN CARRIERS OF RPGR-ASSOCIATED RETINOPATHY. Retina, 2019, 39, 570-580.	1.7	25
63	PIGMENTED PARAVENOUS CHORIORETINAL ATROPHY. Retina, 2019, 39, 514-529.	1.7	23
64	A clinical and molecular characterisation of CRB1-associated maculopathy. European Journal of Human Genetics, 2018, 26, 687-694.	2.8	51
65	Leber Congenital Amaurosis Associated with Mutations in CEP290, Clinical Phenotype, and Natural History in Preparation for Trials of Novel Therapies. Ophthalmology, 2018, 125, 894-903.	5.2	58
66	DETAILED RETINAL IMAGING IN CARRIERS OF OCULAR ALBINISM. Retina, 2018, 38, 620-628.	1.7	13
67	NORMAL ELECTROOCULOGRAPHY IN BEST DISEASE AND AUTOSOMAL RECESSIVE BESTROPHINOPATHY. Retina, 2018, 38, 379-386.	1.7	16
68	Assessment of the incorporation of CNV surveillance into gene panel next-generation sequencing testing for inherited retinal diseases. Journal of Medical Genetics, 2018, 55, 114-121.	3.2	57
69	Missense variants in the X-linked genePRPS1cause retinal degeneration in females. Human Mutation, 2018, 39, 80-91.	2.5	23
70	QUANTITATIVE ANALYSIS OF HYPERAUTOFLUORESCENT RINGS TO CHARACTERIZE THE NATURAL HISTORY AND PROGRESSION IN RPGR-ASSOCIATED RETINOPATHY. Retina, 2018, 38, 2401-2414.	1.7	33
71	Factors associated with visual acuity in patients with cystoid macular oedema and Retinitis Pigmentosa. Ophthalmic Epidemiology, 2018, 25, 183-186.	1.7	8
72	Whole genome sequencing reveals novel mutations causing autosomal dominant inherited macular degeneration. Ophthalmic Genetics, 2018, 39, 763-770.	1.2	13

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73	Complex structural variants in Mendelian disorders: identification and breakpoint resolution using short- and long-read genome sequencing. Genome Medicine, 2018, 10, 95.	8.2	111
74	Clinical Features of a Retinopathy Associated With a Dominant Allele of the RGR Gene. , 2018, 59, 4812.		9
75	Retinal findings in a patient with mutations in ABCC6 and ABCA4. Eye, 2018, 32, 1542-1543.	2.1	5
76	Identification and characterization of the VAX2 p.Leu139Arg variant: possible involvement of VAX2 in cone dystrophy. Ophthalmic Genetics, 2018, 39, 539-543.	1.2	1
77	The treatment of refractory angle-closure glaucoma in a patient with X-linked juvenile retinoschisis. Ophthalmic Genetics, 2018, 39, 625-627.	1.2	6
78	Novel homozygous splicing mutations in cause autosomal recessive retinitis pigmentosa. Molecular Vision, 2018, 24, 603-612.	1.1	6
79	Clinical Characterization of CNGB1-Related Autosomal Recessive Retinitis Pigmentosa. JAMA Ophthalmology, 2017, 135, 137.	2.5	23
80	Mechanism and evidence of nonsense suppression therapy for genetic eye disorders. Experimental Eye Research, 2017, 155, 24-37.	2.6	40
81	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. American Journal of Human Genetics, 2017, 100, 334-342.	6.2	26
82	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. American Journal of Human Genetics, 2017, 100, 592-604.	6.2	61
83	Measurement and Reproducibility of Preserved Ellipsoid Zone Area and Preserved Retinal Pigment Epithelium Area in Eyes With Choroideremia. American Journal of Ophthalmology, 2017, 179, 110-117.	3.3	51
84	Genome-wide linkage and haplotype sharing analysis implicates the MCDR3 locus as a candidate region for a developmental macular disorder in association with digit abnormalities. Ophthalmic Genetics, 2017, 38, 511-519.	1.2	2
85	Phenopolis: an open platform for harmonization and analysis of genetic and phenotypic data. Bioinformatics, 2017, 33, 2421-2423.	4.1	40
86	Single-base substitutions in theCHMpromoter as a cause of choroideremia. Human Mutation, 2017, 38, 704-715.	2.5	45
87	Association of Steroid 5Î±-Reductase Type 3 Congenital Disorder of Glycosylation With Early-Onset Retinal Dystrophy. JAMA Ophthalmology, 2017, 135, 339.	2.5	43
88	Quantitative Analysis of Retinal Structure Using Spectral-Domain Optical Coherence Tomography in RPGR -Associated Retinopathy. American Journal of Ophthalmology, 2017, 178, 18-26.	3.3	30
89	Rescue of the MERTK phagocytic defect in a human iPSC disease model using translational read-through inducing drugs. Scientific Reports, 2017, 7, 51.	3.3	55
90	Childhood-onset Leber hereditary optic neuropathy. British Journal of Ophthalmology, 2017, 101, 1505-1509.	3.9	62

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91	Benign Yellow Dot Maculopathy. Ophthalmology, 2017, 124, 1004-1013.	5.2	12
92	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. American Journal of Human Genetics, 2017, 100, 75-90.	6.2	343
93	Duplication events downstream of IRX1 cause North Carolina macular dystrophy at the MCDR3 locus. Scientific Reports, 2017, 7, 7512.	3.3	23
94	Association of C-Reactive Protein Genetic Polymorphisms With Late Age-Related Macular Degeneration. JAMA Ophthalmology, 2017, 135, 909.	2.5	13
95	Specific Alleles of CLN7/MFSD8, a Protein That Localizes to Photoreceptor Synaptic Terminals, Cause a Spectrum of Nonsyndromic Retinal Dystrophy. , 2017, 58, 2906.		35
96	Psychophysical measures of visual function and everyday perceptual experience in a case of congenital stationary night blindness. Clinical Ophthalmology, 2016, Volume 10, 1593-1606.	1.8	3
97	Reevaluation of the Retinal Dystrophy Due to Recessive Alleles of RGR With the Discovery of a Cis-Acting Mutation in CDHR1. , 2016, 57, 4806.		25
98	Phenotype and Progression of Retinal Degeneration Associated With Nullizigosity of ABCA4. , 2016, 57, 4668.		40
99	Mutations in AGBL5, Encoding Î±-Tubulin Deglutamylase, Are Associated With Autosomal Recessive Retinitis Pigmentosa. , 2016, 57, 6180.		21
100	The Effect on Retinal Structure and Function of 15 Specific ABCA4 Mutations: A Detailed Examination of 82 Hemizygous Patients. , 2016, 57, 5963.		41
101	Preserved visual function in retinal dystrophy due to hypomorphicRPE65mutations. British Journal of Ophthalmology, 2016, 100, 1499-1505.	3.9	25
102	Mutations in CACNA2D4 Cause Distinctive Retinal Dysfunction in Humans. Ophthalmology, 2016, 123, 668-671.e2.	5.2	29
103	Lamination of the Outer Plexiform Layer in Optic Atrophy Caused by Dominant WFS1 Mutations. Ophthalmology, 2016, 123, 1624-1626.	5.2	19
104	Mutations in REEP6 Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2016, 99, 1305-1315.	6.2	121
105	Expanding the Phenotype ofTRNT1-Related Immunodeficiency to Include Childhood Cataract and Inner Retinal Dysfunction. JAMA Ophthalmology, 2016, 134, 1049.	2.5	29
106	Mislocalisation of BEST1 in iPSC-derived retinal pigment epithelial cells from a family with autosomal dominant vitreoretinochoroidopathy (ADVIRC). Scientific Reports, 2016, 6, 33792.	3.3	25
107	Functional rescue of REP1 following treatment with PTC124 and novel derivative PTC-414 in human choroideremia fibroblasts and the nonsense-mediated zebrafish model. Human Molecular Genetics, 2016, 25, 3416-3431.	2.9	69
108	Investigation of SLA4A3 as a candidate gene for human retinal disease. Journal of Negative Results in BioMedicine, 2016, 15, 11.	1.4	1

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109	The clinical features of retinal disease due to a dominant mutation in RPE65. Molecular Vision, 2016, 22, 626-35.	1.1	32
110	Retinal Architecture in â€‹RGS9- and â€‹R9AP-Associated Retinal Dysfunction (Bradyopsia). American Journal of Ophthalmology, 2015, 160, 1269-1275.e1.	3.3	15
111	Exome sequencing revealsADAM9mutations in a child with cone-rod dystrophy. Acta Ophthalmologica, 2015, 93, e392-e393.	1.1	3
112	Somatic mosaicism of a novel IKBKG mutation in a male patient with incontinentia pigmenti. American Journal of Medical Genetics, Part A, 2015, 167, 1601-1604.	1.2	27
113	Disease Expression in Autosomal Recessive Retinal Dystrophy Associated With Mutations in theDRAM2Gene. , 2015, 56, 8083.		13
114	Lack of Interphotoreceptor Retinoid Binding Protein Caused by Homozygous Mutation ofRBP3Is Associated With High Myopia and Retinal Dystrophy. , 2015, 56, 2358.		42
115	Efficacy and Prognostic Factors of Response to Carbonic Anhydrase Inhibitors in Management of Cystoid Macular Edema in Retinitis Pigmentosa. Investigative Ophthalmology and Visual Science, 2015, 56, 1531-1536.	3.3	54
116	Mutations in the unfolded protein response regulator ATF6 cause the cone dysfunction disorder achromatopsia. Nature Genetics, 2015, 47, 757-765.	21.4	183
117	Clinical Heterogeneity in a Family With Mutations inUSH2A. JAMA Ophthalmology, 2015, 133, 352.	2.5	14
118	Predominantly Cone-System Dysfunction as Rare Form of Retinal Degeneration in Patients With Molecularly Confirmed Bardet-Biedl Syndrome. American Journal of Ophthalmology, 2015, 160, 364-372.e1.	3.3	26
119	A detailed clinical and molecular survey of subjects with nonsyndromic USH2A retinopathy reveals an allelic hierarchy of disease-causing variants. European Journal of Human Genetics, 2015, 23, 1318-1327.	2.8	89
120	Author reply. Ophthalmology, 2015, 122, e22.	5.2	0
121	Biallelic Mutations in the Autophagy Regulator DRAM2 Cause Retinal Dystrophy with Early Macular Involvement. American Journal of Human Genetics, 2015, 96, 948-954.	6.2	42
122	Genotype-Dependent Variability in Residual Cone Structure in Achromatopsia: Toward Developing Metrics for Assessing Cone Health. , 2014, 55, 7303.		67
123	Dark-Adaptation Functions in Molecularly Confirmed Achromatopsia and the Implications for Assessment in Retinal Therapy Trials. , 2014, 55, 6340.		14
124	Clinical utility gene card for: Choroideremia. European Journal of Human Genetics, 2014, 22, 572-572.	2.8	37
125	Differential Light-induced Responses in Sectorial Inherited Retinal Degeneration. Journal of Biological Chemistry, 2014, 289, 35918-35928.	3.4	32
126	A Prospective Longitudinal Study of Retinal Structure and Function in Achromatopsia. , 2014, 55, 5733.		68

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127	Cone Dystrophy With â€œSupernormalâ€•Rod ERG: Psychophysical Testing Shows Comparable Rod and Cone Temporal Sensitivity Losses With No Gain in Rod Function. , 2014, 55, 832.		21
128	Expansion of Ocular Phenotypic Features Associated With Mutations inADAMTS18. JAMA Ophthalmology, 2014, 132, 996.	2.5	15
129	Detailed Phenotypic and Genotypic Characterization of Bietti Crystalline Dystrophy. Ophthalmology, 2014, 121, 1174-1184.	5.2	79
130	Retinal Structure and Function in Achromatopsia. Ophthalmology, 2014, 121, 234-245.	5.2	145
131	Retinal gene therapy in patients with choroideremia: initial findings from a phase 1/2 clinical trial. Lancet, The, 2014, 383, 1129-1137.	13.7	689
132	The Phenotypic Variability of Retinal Dystrophies Associated With Mutations in CRX, With Report of a Novel Macular Dystrophy Phenotype. Investigative Ophthalmology and Visual Science, 2014, 55, 6934-6944.	3.3	59
133	The effect of the common c.2299delG mutation in USH2A on RNA splicing. Experimental Eye Research, 2014, 122, 9-12.	2.6	23
134	Biallelic Variants in TTLL5, Encoding a Tubulin Glutamylase, Cause Retinal Dystrophy. American Journal of Human Genetics, 2014, 94, 760-769.	6.2	67
135	Natural History and Retinal Structure in Patients with Usher Syndrome Type 1 Owing to MYO7A Mutation. Ophthalmology, 2014, 121, 580-587.	5.2	27
136	Clinical and Molecular Analysis of Stargardt Disease With Preserved Foveal Structure and Function. American Journal of Ophthalmology, 2013, 156, 487-501.e1.	3.3	100
137	Phenotypic findings in C1QTNF5 retinopathy (lateâ€onset retinal degeneration). Acta Ophthalmologica, 2013, 91, e191-5.	1.1	35
138	Screening of a Large Cohort of Leber Congenital Amaurosis and Retinitis Pigmentosa Patients Identifies Novel LCA5 Mutations and New Genotype-Phenotype Correlations. Human Mutation, 2013, 34, 1537-1546.	2.5	32
139	Comprehensive sequence analysis of nine Usher syndrome genes in the UK National Collaborative Usher Study. Journal of Medical Genetics, 2012, 49, 27-36.	3.2	152
140	SERIAL IMAGING AND STRUCTURE-FUNCTION CORRELATES OF HIGH-DENSITY RINGS OF FUNDUS AUTOFLUORESCENCE IN RETINITIS PIGMENTOSA. Retina, 2011, 31, 1670-1679.	1.7	79
141	Autosomal dominant Best disease with an unusual electrooculographic light rise and risk of angle-closure glaucoma: a clinical and molecular genetic study. Molecular Vision, 2011, 17, 2272-82.	1.1	16
142	RDH12 retinopathy: novel mutations and phenotypic description. Molecular Vision, 2011, 17, 2706-16.	1.1	47
143	Biallelic Mutation of BEST1 Causes a Distinct Retinopathy in Humans. American Journal of Human Genetics, 2008, 82, 19-31.	6.2	257
144	Infrared Fundus Photography in a Case of Acute Macular Neuroretinopathy. Neuro-Ophthalmology, 2008, 32, 200-202.	1.0	2

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145	Phenotypic Variation in Enhanced S-cone Syndrome. , 2008, 49, 2082.		107