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	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
2	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
3	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
4	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	27.8	338
5	Biallelic Mutation of BEST1 Causes a Distinct Retinopathy in Humans. American Journal of Human Genetics, 2008, 82, 19-31.	6.2	257
6	Mutations in the unfolded protein response regulator ATF6 cause the cone dysfunction disorder achromatopsia. Nature Genetics, 2015, 47, 757-765.	21.4	183
7	Germline selection shapes human mitochondrial DNA diversity. Science, 2019, 364, .	12.6	178
8	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
9	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
10	Retinal Structure and Function in Achromatopsia. Ophthalmology, 2014, 121, 234-245.	5.2	145
11	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
12	Mutations in REEP6 Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2016, 99, 1305-1315.	6.2	121
13	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
14	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
15	Phenotypic Variation in Enhanced S-cone Syndrome. , 2008, 49, 2082.		107
16	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
17	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
18	SERIAL IMAGING AND STRUCTURE-FUNCTION CORRELATES OF HIGH-DENSITY RINGS OF FUNDUS AUTOFLUORESCENCE IN RETINITIS PIGMENTOSA. Retina, 2011, 31, 1670-1679.	1.7	79

#	Article	IF	Citations
19	Detailed Phenotypic and Genotypic Characterization of Bietti Crystalline Dystrophy. Ophthalmology, 2014, 121, 1174-1184.	5.2	79
20	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
21	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
22	A Prospective Longitudinal Study of Retinal Structure and Function in Achromatopsia. , 2014, 55, 5733.		68
23	Genotype-Dependent Variability in Residual Cone Structure in Achromatopsia: Toward Developing Metrics for Assessing Cone Health. , 2014, 55, 7303.		67
24	Biallelic Variants in TTLL5, Encoding a Tubulin Glutamylase, Cause Retinal Dystrophy. American Journal of Human Genetics, 2014, 94, 760-769.	6.2	67
25	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
26	Childhood-onset Leber hereditary optic neuropathy. British Journal of Ophthalmology, 2017, 101, 1505-1509.	3.9	62
27	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
28	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
29	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
30	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
31	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
32	Prevalence of cystoid macular oedema, epiretinal membrane and cataract in retinitis pigmentosa. British Journal of Ophthalmology, 2019, 103, 1163-1166.	3.9	55
33	Detailed Phenotyping and Therapeutic Strategies for Intronic ABCA4 Variants in Stargardt Disease. Molecular Therapy - Nucleic Acids, 2020, 21, 412-427.	5.1	55
34	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
35	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
36	A clinical and molecular characterisation of CRB1-associated maculopathy. European Journal of Human Genetics, 2018, 26, 687-694.	2.8	51

#	Article	IF	Citations
37	RDH12 retinopathy: novel mutations and phenotypic description. Molecular Vision, 2011, 17, 2706-16.	1.1	47
38	Single-base substitutions in theCHMpromoter as a cause of choroideremia. Human Mutation, 2017, 38, 704-715.	2.5	45
39	Deep Phenotyping ofPDE6C-Associated Achromatopsia. , 2019, 60, 5112.		44
40	Association of Steroid 5Î±-Reductase Type 3 Congenital Disorder of Glycosylation With Early-Onset Retinal Dystrophy. JAMA Ophthalmology, 2017, 135, 339.	2.5	43
41	Lack of Interphotoreceptor Retinoid Binding Protein Caused by Homozygous Mutation ofRBP3Is Associated With High Myopia and Retinal Dystrophy. , 2015, 56, 2358.		42
42	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
43	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
44	The Effect on Retinal Structure and Function of 15 Specific ABCA4 Mutations: A Detailed Examination of 82 Hemizygous Patients. , 2016, 57, 5963.		41
45	SSBP1 mutations in dominant optic atrophy with variable retinal degeneration. Annals of Neurology, 2019, 86, 368-383.	5.3	41
46	Phenotype and Progression of Retinal Degeneration Associated With Nullizigosity of ABCA4. , 2016, 57, 4668.		40
47	Mechanism and evidence of nonsense suppression therapy for genetic eye disorders. Experimental Eye Research, 2017, 155, 24-37.	2.6	40
48	Phenopolis: an open platform for harmonization and analysis of genetic and phenotypic data. Bioinformatics, 2017, 33, 2421-2423.	4.1	40
49	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
50	Clinical utility gene card for: Choroideremia. European Journal of Human Genetics, 2014, 22, 572-572.	2.8	37
51	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
52	Phenotypic findings in C1QTNF5 retinopathy (lateâ€onset retinal degeneration). Acta Ophthalmologica, 2013, 91, e191-5.	1.1	35
53	Specific Alleles of CLN7/MFSD8, a Protein That Localizes to Photoreceptor Synaptic Terminals, Cause a Spectrum of Nonsyndromic Retinal Dystrophy. , 2017, 58, 2906.		35
54	QUANTITATIVE ANALYSIS OF HYPERAUTOFLUORESCENT RINGS TO CHARACTERIZE THE NATURAL HISTORY AND PROGRESSION IN RPGR-ASSOCIATED RETINOPATHY. Retina, 2018, 38, 2401-2414.	1.7	33

#	Article	IF	Citations
55	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
56	Differential Light-induced Responses in Sectorial Inherited Retinal Degeneration. Journal of Biological Chemistry, 2014, 289, 35918-35928.	3.4	32
57	The clinical features of retinal disease due to a dominant mutation in RPE65. Molecular Vision, 2016, 22, 626-35.	1.1	32
58	Autosomal Recessive Bestrophinopathy. Ophthalmology, 2021, 128, 706-718.	5.2	31
59	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
60	Mutations in CACNA2D4 Cause Distinctive Retinal Dysfunction in Humans. Ophthalmology, 2016, 123, 668-671.e2.	5.2	29
61	Expanding the Phenotype ofTRNT1-Related Immunodeficiency to Include Childhood Cataract and Inner Retinal Dysfunction. JAMA Ophthalmology, 2016, 134, 1049.	2.5	29
62	Phagosomal and mitochondrial alterations in RPE may contribute to KCNJ13 retinopathy. Scientific Reports, 2019, 9, 3793.	3.3	29
63	Natural History and Retinal Structure in Patients with Usher Syndrome Type 1 Owing to MYO7A Mutation. Ophthalmology, 2014, 121, 580-587.	5.2	27
64	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
65	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
66	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
67	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
68	Preserved visual function in retinal dystrophy due to hypomorphicRPE65mutations. British Journal of Ophthalmology, 2016, 100, 1499-1505.	3.9	25
69	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
70	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
71	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
72	CELLULAR IMAGING OF THE TAPETAL-LIKE REFLEX IN CARRIERS OF RPGR-ASSOCIATED RETINOPATHY. Retina, 2019, 39, 570-580.	1.7	25

#	Article	IF	Citations
73	Clinical and preclinical therapeutic outcome metrics for USH2A-related disease. Human Molecular Genetics, 2020, 29, 1882-1899.	2.9	24
74	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
75	The effect of the common c.2299delG mutation in USH2A on RNA splicing. Experimental Eye Research, 2014, 122, 9-12.	2.6	23
76	Clinical Characterization of CNGB1-Related Autosomal Recessive Retinitis Pigmentosa. JAMA Ophthalmology, 2017, 135, 137.	2.5	23
77	Duplication events downstream of IRX1 cause North Carolina macular dystrophy at the MCDR3 locus. Scientific Reports, 2017, 7, 7512.	3.3	23
78	Missense variants in the X-linked genePRPS1cause retinal degeneration in females. Human Mutation, 2018, 39, 80-91.	2.5	23
79	Benefit of an electronic headâ€mounted low vision aid. Ophthalmic and Physiological Optics, 2019, 39, 422-431.	2.0	23
80	PIGMENTED PARAVENOUS CHORIORETINAL ATROPHY. Retina, 2019, 39, 514-529.	1.7	23
81	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
82	Mutations in AGBL5, Encoding Î±-Tubulin Deglutamylase, Are Associated With Autosomal Recessive Retinitis Pigmentosa. , 2016, 57, 6180.		21
83	Clinical and Molecular Characterization of Familial Exudative Vitreoretinopathy Associated With Microcephaly. American Journal of Ophthalmology, 2019, 207, 87-98.	3.3	20
84	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
85	Sector Retinitis Pigmentosa: Extending the Molecular Genetics Basis and Elucidating the Natural History. American Journal of Ophthalmology, 2021, 221, 299-310.	3.3	20
86	Lamination of the Outer Plexiform Layer in Optic Atrophy Caused by Dominant WFS1 Mutations. Ophthalmology, 2016, 123, 1624-1626.	5.2	19
87	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
88	X-Linked Retinoschisis. Ophthalmology, 2022, 129, 542-551.	5.2	19
89	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
90	Unilateral pigmentary retinopathy: a retrospective case series. Acta Ophthalmologica, 2019, 97, e601-e617.	1.1	17

#	Article	IF	Citations
91	KCNV2-Associated Retinopathy: Genetics, Electrophysiology, and Clinical Courseâ€"KCNV2 Study Group Report 1. American Journal of Ophthalmology, 2021, 225, 95-107.	3.3	17
92	NORMAL ELECTROOCULOGRAPHY IN BEST DISEASE AND AUTOSOMAL RECESSIVE BESTROPHINOPATHY. Retina, 2018, 38, 379-386.	1.7	16
93	CNGB1 â€related rodâ€cone dystrophy: A mutation review and update. Human Mutation, 2021, 42, 641-666.	2.5	16
94	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
95	Expansion of Ocular Phenotypic Features Associated With Mutations inADAMTS18. JAMA Ophthalmology, 2014, 132, 996.	2.5	15
96	Retinal Architecture in â€‹RGS9- and â€‹R9AP-Associated Retinal Dysfunction (Bradyopsia). American Journal of Ophthalmology, 2015, 160, 1269-1275.e1.	3.3	15
97	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
98	Dark-Adaptation Functions in Molecularly Confirmed Achromatopsia and the Implications for Assessment in Retinal Therapy Trials. , 2014, 55, 6340.		14
99	Clinical Heterogeneity in a Family With Mutations inUSH2A. JAMA Ophthalmology, 2015, 133, 352.	2.5	14
100	Expanding the clinical phenotype in patients with disease causing variants associated with atypical Usher syndrome. Ophthalmic Genetics, 2021, 42, 664-673.	1.2	14
101	Disease Expression in Autosomal Recessive Retinal Dystrophy Associated With Mutations in theDRAM2Gene. , 2015, 56, 8083.		13
102	Association of C-Reactive Protein Genetic Polymorphisms With Late Age-Related Macular Degeneration. JAMA Ophthalmology, 2017, 135, 909.	2.5	13
103	DETAILED RETINAL IMAGING IN CARRIERS OF OCULAR ALBINISM. Retina, 2018, 38, 620-628.	1.7	13
104	Whole genome sequencing reveals novel mutations causing autosomal dominant inherited macular degeneration. Ophthalmic Genetics, 2018, 39, 763-770.	1.2	13
105	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
106	Benign Yellow Dot Maculopathy. Ophthalmology, 2017, 124, 1004-1013.	5.2	12
107	Retinopathy Associated with Biallelic Mutations in PYGM (McArdle Disease). Ophthalmology, 2019, 126, 320-322.	5.2	12
108	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

#	Article	IF	Citations
109	Macula-predominant retinopathy associated with biallelic variants in RDH12. Ophthalmic Genetics, 2020, 41, 612-615.	1.2	12
110	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
111	A new paradigm for delivering personalised care: integrating genetics with surgical interventions in BEST1 mutations. Eye, 2020, 34, 577-583.	2.1	11
112	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
113	Delineating the expanding phenotype associated with SCAPER gene mutation. American Journal of Medical Genetics, Part A, 2019, 179, 1665-1671.	1.2	10
114	Clinical Features of a Retinopathy Associated With a Dominant Allele of the RGR Gene. , 2018, 59, 4812.		9
115	Isolated rod dysfunction associated with a novel genotype of CNGB1. American Journal of Ophthalmology Case Reports, 2019, 14, 83-86.	0.7	9
116	Factors associated with visual acuity in patients with cystoid macular oedema and Retinitis Pigmentosa. Ophthalmic Epidemiology, 2018, 25, 183-186.	1.7	8
117	Clinical and Genetic Findings in CTNNA1-Associated Macular Pattern Dystrophy. Ophthalmology, 2021, 128, 952-955.	5.2	8
118	Broadening INPP5E phenotypic spectrum: detection of rare variants in syndromic and non-syndromic IRD. Npj Genomic Medicine, 2021, 6, 53.	3.8	8
119	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
120	WFS1-Associated Optic Neuropathy: Genotype-Phenotype Correlations and Disease Progression. American Journal of Ophthalmology, 2022, 241, 9-27.	3.3	8
121	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
122	Ceramide synthase TLCD3B is a novel gene associated with human recessive retinal dystrophy. Genetics in Medicine, 2021, 23, 488-497.	2.4	7
123	New variants and in silico analyses in GRK1 associated Oguchi disease. Human Mutation, 2021, 42, 164-176.	2.5	7
124	The treatment of refractory angle-closure glaucoma in a patient with X-linked juvenile retinoschisis. Ophthalmic Genetics, 2018, 39, 625-627.	1.2	6
125	Rod-cone dystrophy associated with the Gly167Asp variant in PRPH2. Ophthalmic Genetics, 2019, 40, 188-189.	1.2	6
126	Novel homozygous splicing mutations in cause autosomal recessive retinitis pigmentosa. Molecular Vision, 2018, 24, 603-612.	1.1	6

#	Article	IF	Citations
127	Axial Length Distributions in Patients With Genetically Confirmed Inherited Retinal Diseases. , 2022, 63, 15.		6
128	Retinal findings in a patient with mutations in ABCC6 and ABCA4. Eye, 2018, 32, 1542-1543.	2.1	5
129	Reanalysis of Association of Pro50Leu Substitution in Guanylate Cyclase Activating Protein-1 With Dominant Retinal Dystrophy. JAMA Ophthalmology, 2020, 138, 200.	2.5	5
130	A clinical study of patients with novel CDHR1 genotypes associated with late-onset macular dystrophy. Eye, 2021, 35, 1482-1489.	2.1	5
131	Monozygotic twins discordant for asymmetric pigmented paravenous chorioretinal atrophy. Retinal Cases and Brief Reports, 2020, Publish Ahead of Print, .	0.6	4
132	Exome sequencing revealsADAM9mutations in a child with cone-rod dystrophy. Acta Ophthalmologica, 2015, 93, e392-e393.	1.1	3
133	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
134	Macular maldevelopment inATF6-mediated retinal dysfunction. Ophthalmic Genetics, 2019, 40, 564-569.	1.2	3
135	Prevalence of electronegative electroretinograms in a healthy adult cohort. BMJ Open Ophthalmology, 2021, 6, e000751.	1.6	3
136	Infrared Fundus Photography in a Case of Acute Macular Neuroretinopathy. Neuro-Ophthalmology, 2008, 32, 200-202.	1.0	2
137	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
138	Awareness of olfactory impairment in a cohort of patients with CNGB1-associated retinitis pigmentosa. Eye, 2020, 34, 783-784.	2.1	2
139	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
140	A rare canonical splice-site variant in VPS13B causes attenuated Cohen syndrome. Ophthalmic Genetics, 2022, 43, 110-115.	1.2	2
141	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
142	Investigation of SLA4A3 as a candidate gene for human retinal disease. Journal of Negative Results in BioMedicine, 2016, 15, 11.	1.4	1
143	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
144	Variability of retinopathy consequent upon novel mutations in LAMA1. Ophthalmic Genetics, 2022, 43, 671-678.	1.2	1

#	Article	IF	Citations
145	Author reply. Ophthalmology, 2015, 122, e22.	5.2	0