Edwin M Stone

List of Publications by Year
in descending order

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376 papers	30,776 citations	2802 94 h-index	7348 152 g-index
379 all docs	379 docs citations	379 times ranked	18068 citing authors

#	Article	IF	Citations
1	Predominance of hyperopia in autosomal dominant Best vitelliform macular dystrophy. British Journal of Ophthalmology, 2022, 106, 522-527.	3.9	6
2	Long-Term Outcomes and Risk Factors for Severe Vision Loss in Autosomal Dominant Neovascular Inflammatory Vitreoretinopathy (ADNIV). American Journal of Ophthalmology, 2022, 233, 144-152.	3.3	4
3	Local factor H production by human choroidal endothelial cells mitigates complement deposition: implications for macular degeneration. Journal of Pathology, 2022, 257, 29-38.	4.5	12
4	Familial Glaucomaâ€"A Pedigree Revisited With Genetic Testing After 70 Years. JAMA Ophthalmology, 2022, 140, 543.	2.5	2
5	Biocompatibility of Human Induced Pluripotent Stem Cellâ€"Derived Retinal Progenitor Cell Grafts in Immunocompromised Rats. Cell Transplantation, 2022, 31, 096368972211044.	2.5	9
6	Patient derived stem cells for discovery and validation of novel pathogenic variants in inherited retinal disease. Progress in Retinal and Eye Research, 2021, 83, 100918.	15.5	16
7	Exome-based investigation of the genetic basis of human pigmentary glaucoma. BMC Genomics, 2021, 22, 477.	2.8	9
8	An Unusual Presentation of CLN3-Associated Batten Disease With Classic Histopathologic and Ultrastructural Findings. Journal of Neuropathology and Experimental Neurology, 2021, 80, 1081-1084.	1.7	2
9	Single-cell RNA sequencing in vision research: Insights into human retinal health and disease. Progress in Retinal and Eye Research, 2021, 83, 100934.	15.5	24
10	Intrafamilial Variability of Ocular Manifestations of von Hippel-Lindau Disease. Ophthalmology Retina, 2021, 6, 89-89.	2.4	1
11	Development and biological characterization of a clinical gene transfer vector for the treatment of MAK-associated retinitis pigmentosa. Gene Therapy, 2021, , .	4.5	5
12	Correlation of features on OCT with visual acuity and Gass lesion type in Best vitelliform macular dystrophy. BMJ Open Ophthalmology, 2021, 6, e000860.	1.6	5
13	Scleral pits represent degeneration around the posterior ciliary arteries and are signs of disease severity in choroideremia. Eye, 2020, 34, 746-754.	2.1	4
14	Subliminal Message: Outer Retinal Tubulations Resembling Mitochondria in Maternally Inherited Diabetes and Deafness. Ophthalmology Retina, 2020, 4, 1102.	2.4	4
15	Spectacle: An interactive resource for ocular single-cell RNA sequencing data analysis. Experimental Eye Research, 2020, 200, 108204.	2.6	47
16	Human iPSC Modeling Reveals Mutation-Specific Responses to Gene Therapy in a Genotypically Diverse Dominant Maculopathy. American Journal of Human Genetics, 2020, 107, 278-292.	6.2	35
17	Analysis of retinal sublayer thicknesses and rates of change in ABCA4-associated Stargardt disease. Scientific Reports, 2020, 10, 16576.	3.3	12
18	Stepwise differentiation and functional characterization of human induced pluripotent stem cell-derived choroidal endothelial cells. Stem Cell Research and Therapy, 2020, 11, 409.	5.5	19

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19	Retinal Tropism and Transduction of Adeno-Associated Virus Varies by Serotype and Route of Delivery (Intravitreal, Subretinal, or Suprachoroidal) in Rats. Human Gene Therapy, 2020, 31, 1288-1299.	2.7	28
20	Bulk and single-cell gene expression analyses reveal aging human choriocapillaris has pro-inflammatory phenotype. Microvascular Research, 2020, 131, 104031.	2.5	34
21	Autologous cell replacement: a noninvasive AI approach to clinical release testing. Journal of Clinical Investigation, 2020, 130, 608-611.	8.2	5
22	Single-cell transcriptomics of the human retinal pigment epithelium and choroid in health and macular degeneration. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 24100-24107.	7.1	234
23	Helper-Dependent Adenovirus Transduces the Human and Rat Retina but Elicits an Inflammatory Reaction When Delivered Subretinally in Rats. Human Gene Therapy, 2019, 30, 1371-1384.	2.7	19
24	Wide-Field Swept-Source OCT and Angiography in X-Linked Retinoschisis. Ophthalmology Retina, 2019, 3, 178-185.	2.4	30
25	Development of a Molecularly Stable Gene Therapy Vector for the Treatment of RPGR-Associated X-Linked Retinitis Pigmentosa. Human Gene Therapy, 2019, 30, 967-974.	2.7	16
26	Two-photon polymerized poly(caprolactone) retinal cell delivery scaffolds and their systemic and retinal biocompatibility. Acta Biomaterialia, 2019, 94, 204-218.	8.3	51
27	Optimizing Donor Cellular Dissociation and Subretinal Injection Parameters for Stem Cell-Based Treatments. Stem Cells Translational Medicine, 2019, 8, 797-809.	3.3	21
28	Two-color pupillometry in KCNV2 retinopathy. Documenta Ophthalmologica, 2019, 139, 11-20.	2.2	7
29	Choriocapillaris Degeneration in Geographic Atrophy. American Journal of Pathology, 2019, 189, 1473-1480.	3.8	48
30	Correction of NR2E3 Associated Enhanced S-cone Syndrome Patient-specific iPSCs using CRISPR-Cas9. Genes, 2019, 10, 278.	2.4	27
31	Myocilin Mutations in Patients With Normal-Tension Glaucoma. JAMA Ophthalmology, 2019, 137, 559.	2.5	17
32	A family with spinocerebellar ataxia and retinitis pigmentosa attributed to an ELOVL4 mutation. Neurology: Genetics, 2019, 5, e357.	1.9	25
33	Generation of an immortalized human choroid endothelial cell line (iChEC-1) using an endothelial cell specific promoter. Microvascular Research, 2019, 123, 50-57.	2.5	18
34	The ARMS2 A69S Polymorphism Is Associated with Delayed Rod-Mediated DarkÂAdaptation in Eyes at Risk for Incident Age-Related Macular Degeneration. Ophthalmology, 2019, 126, 591-600.	5.2	26
35	Disruption of RPGR protein interaction network is the common feature of RPGR missense variations that cause XLRP. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 1353-1360.	7.1	34
36	CRISPRâ€Cas9â€Based Genome Editing of Human Induced Pluripotent Stem Cells. Current Protocols in Stem Cell Biology, 2018, 44, 5B.7.1-5B.7.22.	3.0	25

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37	Expanded Retinal Disease Spectrum Associated With Autosomal Recessive Mutations in GUCY2D. American Journal of Ophthalmology, 2018, 190, 58-68.	3.3	20
38	CRISPR-Cas9 genome engineering: Treating inherited retinal degeneration. Progress in Retinal and Eye Research, 2018, 65, 28-49.	15.5	64
39	Evaluation of sFLT1 protein levels in human eyes with the FLT1 rs9943922 polymorphism. Ophthalmic Genetics, 2018, 39, 68-72.	1.2	2
40	Assessment of Adeno-Associated Virus Serotype Tropism in Human Retinal Explants. Human Gene Therapy, 2018, 29, 424-436.	2.7	53
41	Correlation of Optical Coherence Tomography and Retinal Histology in Normal and Pro23His Retinal Degeneration Pig. Translational Vision Science and Technology, 2018, 7, 18.	2.2	13
42	CHOROIDAL NEOVASCULARIZATION IN NORTH CAROLINA MACULAR DYSTROPHY RESPONSIVE TO ANTIâ€“VASCULAR ENDOTHELIAL GROWTH FACTOR THERAPY. Retinal Cases and Brief Reports, 2018, Publish Ahead of Print, 509-513.	0.6	12
43	Swept-Source OCT of a Macular Coloboma in NMNAT1-Leber Congenital Amaurosis. Ophthalmology Retina, 2018, 2, 1040.	2.4	9
44	Imidazole Compounds for Protecting Choroidal Endothelial Cells from Complement Injury. Scientific Reports, 2018, 8, 13387.	3.3	7
45	Effect of Molecular Weight and Functionality on Acrylated Poly(caprolactone) for Stereolithography and Biomedical Applications. Biomacromolecules, 2018, 19, 3682-3692.	5.4	51
46	Evaluation of serum and ocular levels of membrane attack complex and C-reactive protein in CFH-genotyped human donors. Eye, 2018, 32, 1740-1742.	2.1	14
47	Human Retinal Engineering using 3D PCL Scaffolds. FASEB Journal, 2018, 32, 816.12.	0.5	0
48	Reduced penetrance in a large Caucasian pedigree with Stickler syndrome. Ophthalmic Genetics, 2017, 38, 43-50.	1.2	7
49	Bestrophinopathy: An RPE-photoreceptor interface disease. Progress in Retinal and Eye Research, 2017, 58, 70-88.	15.5	85
50	Connective Tissue Growth Factor Promotes Efficient Generation of Human Induced Pluripotent Stem Cell-Derived Choroidal Endothelium. Stem Cells Translational Medicine, 2017, 6, 1533-1546.	3.3	30
51	Preparation and evaluation of human choroid extracellular matrix scaffolds for the study of cell replacement strategies. Acta Biomaterialia, 2017, 57, 293-303.	8.3	19
52	Clinically Focused Molecular Investigation of 1000 Consecutive Families with Inherited Retinal Disease. Ophthalmology, 2017, 124, 1314-1331.	5.2	312
53	Using CRISPR-Cas9 to Generate Gene-Corrected Autologous iPSCs for the Treatment of Inherited Retinal Degeneration. Molecular Therapy, 2017, 25, 1999-2013.	8.2	121
54	Patient-specific induced pluripotent stem cells to evaluate the pathophysiology of TRNT1 -associated Retinitis pigmentosa. Stem Cell Research, 2017, 21, 58-70.	0.7	45

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55	Two-photon polymerization for production of human iPSC-derived retinal cell grafts. Acta Biomaterialia, 2017, 55, 385-395.	8.3	76
56	Genomic Organization of TBK1 Copy Number Variations in Glaucoma Patients. Journal of Glaucoma, 2017, 26, 1063-1067.	1.6	6
57	Generation of Xenoâ€Free, cGMPâ€Compliant Patientâ€Specific iPSCs from Skin Biopsy. Current Protocols in Stem Cell Biology, 2017, 42, 4A.12.1-4A.12.14.	3.0	15
58	EYS Mutations Causing Autosomal Recessive Retinitis Pigmentosa: Changes of Retinal Structure and Function with Disease Progression. Genes, 2017, 8, 178.	2.4	35
59	Outcome Measures for Clinical Trials of Leber Congenital Amaurosis Caused by the Intronic Mutation in the CEP290 Gene. , 2017, 58, 2609.		46
60	Primary congenital and developmental glaucomas. Human Molecular Genetics, 2017, 26, R28-R36.	2.9	85
61	Confirmation of the OVOL2 Promoter Mutation c.-307T>C in Posterior Polymorphous Corneal Dystrophy 1. PLoS ONE, 2017, 12, e0169215.	2.5	20
62	Keeping an Eye on Bardet-Biedl Syndrome: A Comprehensive Review of the Role of Bardet-Biedl Syndrome Genes in the Eye. Medical Research Archives, 2017, 5, .	0.2	45
63	Autosomal Dominant Retinal Dystrophies Caused by a Founder Splice Site Mutation, c.828+3A>T, inPRPH2and Protein Haplotypes intransas Modifiers. , 2016, 57, 349.		17
64	SQSTM1 Mutations and Glaucoma. PLoS ONE, 2016, 11, e0156001.	2.5	9
65	A Method for Sectioning and Immunohistochemical Analysis of Stem Cellâ€"Derived 3â€D Organoids. Current Protocols in Stem Cell Biology, 2016, 37, 1C.19.1-1C.19.11.	3.0	11
66	Phenotypic Variation in a Family With Pseudodominant Stargardt Disease. JAMA Ophthalmology, 2016, 134, 580.	2.5	15
67	Two-color pupillometry in enhanced S-cone syndrome caused by NR2E3 mutations. Documenta Ophthalmologica, 2016, 132, 157-166.	2.2	16
68	Monomeric C-reactive protein and inflammation in age-related macular degeneration. Journal of Pathology, 2016, 240, 173-183.	4.5	43
69	Using Patient-Specific Induced Pluripotent Stem Cells and Wild-Type Mice to Develop a Gene Augmentation-Based Strategy to Treat CLN3-Associated Retinal Degeneration. Human Gene Therapy, 2016, 27, 835-846.	2.7	29
70	Abnormal 8-Hz flicker electroretinograms in carriers of X-linked retinoschisis. Documenta Ophthalmologica, 2016, 133, 61-70.	2.2	4
71	cGMP production of patient-specific iPSCs and photoreceptor precursor cells to treat retinal degenerative blindness. Scientific Reports, 2016, 6, 30742.	3.3	108
72	Distinguishing optic pathway glioma and retinitis pigmentosa with visual field testing. Canadian Journal of Ophthalmology, 2016, 51, e94-e96.	0.7	1

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73	Selective accumulation of the complement membrane attack complex in aging choriocapillaris. Experimental Eye Research, 2016, 146, 393-397.	2.6	51
74	North Carolina Macular Dystrophy Is Caused by Dysregulation of the Retinal Transcription Factor PRDM13. Ophthalmology, 2016, 123, 9-18.	5.2	105
75	Molecular response of chorioretinal endothelial cells to complement injury: implications for macular degeneration. Journal of Pathology, 2016, 238, 446-456.	4.5	47
76	Peroxisome biogenesis disorders in the Zellweger spectrum: An overview of current diagnosis, clinical manifestations, and treatment guidelines. Molecular Genetics and Metabolism, 2016, 117, 313-321.	1.1	314
77	Concise Review: Patient-Specific Stem Cells to Interrogate Inherited Eye Disease. Stem Cells Translational Medicine, 2016, 5, 132-140.	3.3	19
78	Clinically detectable drusen domains in fibulin-5-associated age-related macular degeneration (AMD). International Ophthalmology, 2016, 36, 569-575.	1.4	5
79	Hypomorphic mutations inTRNT1cause retinitis pigmentosa with erythrocytic microcytosis. Human Molecular Genetics, 2016, 25, 44-56.	2.9	64
80	Using patient-specific induced pluripotent stem cells to interrogate the pathogenicity of a novel retinal pigment epithelium-specific 65ÂkDa cryptic splice site mutation and confirm eligibility for enrollment into a clinical gene augmentation trial. Translational Research, 2015, 166, 740-749.e1.	5.0	30
81	Allogenic iPSC-derived RPE cell transplants induce immune response in pigs: a pilot study. Scientific Reports, 2015, 5, 11791.	3.3	48
82	Heterozygous Triplication of Upstream Regulatory Sequences Leads to Dysregulation of Matrix Metalloproteinase 19 in Patients with Cavitary Optic Disc Anomaly. Human Mutation, 2015, 36, 369-378.	2.5	10
83	Generating iPSC-Derived Choroidal Endothelial Cells to Study Age-Related Macular Degeneration. , 2015, 56, 8258.		36
84	Full-Field Pupillary Light Responses, Luminance Thresholds, and Light Discomfort Thresholds inCEP290Leber Congenital Amaurosis Patients. , 2015, 56, 7130.		26
85	Predicting Progression ofABCA4-Associated Retinal Degenerations Based on Longitudinal Measurements of the Leading Disease Front. , 2015, 56, 5946.		36
86	Basal exon skipping and genetic pleiotropy: A predictive model of disease pathogenesis. Science Translational Medicine, 2015, 7, 291ra97.	12.4	73
87	Genetic Testing for Age-Related Macular Degeneration. JAMA Ophthalmology, 2015, 133, 598.	2.5	32
88	Comparison of Retinal and Choriocapillaris Thicknesses Following Sitting to Supine Transition in Healthy Individuals and Patients With Age-Related Macular Degeneration. JAMA Ophthalmology, 2015, 133, 297.	2.5	33
89	Autosomal Dominant Microcephaly Associated With Congenital Lymphedema and Chorioretinopathy Due to a Novel Mutation inKIF11. JAMA Ophthalmology, 2015, 133, 720.	2.5	9
90	Complement activation and choriocapillaris loss in early AMD: Implications for pathophysiology and therapy. Progress in Retinal and Eye Research, 2015, 45, 1-29.	15.5	189

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91	Stem Cells as Tools for Studying the Genetics of Inherited Retinal Degenerations. Cold Spring Harbor Perspectives in Medicine, 2015, 5, a017160-a017160.	6.2	11
92	Protein misfolding and the pathogenesis of ABCA4-associated retinal degenerations. Human Molecular Genetics, 2015, 24, 3220-3237.	2.9	69
93	Apparent Usher Syndrome Caused by the Combination ofBBS1-Associated Retinitis Pigmentosa andSLC26A4-Associated Deafness. JAMA Ophthalmology, 2015, 133, 967.	2.5	6
94	Founder Effect of a c.828+3A>T Splice Site Mutation in Peripherin 2 (PRPH2) Causing Autosomal Dominant Retinal Dystrophies. JAMA Ophthalmology, 2015, 133, 511.	2.5	14
95	COMPARISON OF DRUSEN AND MODIFYING GENES IN AUTOSOMAL DOMINANT RADIAL DRUSEN AND AGE-RELATED MACULAR DEGENERATION. Retina, 2015, 35, 48-57.	1.7	34
96	Clinical and Electrophysiologic Characteristics of a Large Kindred with X-Linked Retinitis Pigmentosa Associated with the RPGR Locus. Ophthalmic Genetics, 2015, 36, 321-326.	1.2	7
97	Vitritis in Pediatric Genetic Retinal Disorders. Ophthalmology, 2015, 122, 192-199.	5.2	8
98	Heterozygous Deep-Intronic Variants and Deletions inABCA4in Persons with Retinal Dystrophies and One ExonicABCA4Variant. Human Mutation, 2015, 36, 43-47.	2.5	68
99	Patient-specific induced pluripotent stem cells (iPSCs) for the study and treatment of retinal degenerative diseases. Progress in Retinal and Eye Research, 2015, 44, 15-35.	15.5	108
100	Gene Therapy Using Stem Cells. Cold Spring Harbor Perspectives in Medicine, 2015, 5, a017434-a017434.	6.2	16
101	Novel TMEM98 mutations in pedigrees with autosomal dominant nanophthalmos. Molecular Vision, 2015, 21, 1017-23.	1.1	24
102	Structural and Biochemical Analyses of Choroidal Thickness in Human Donor Eyes. , 2014, 55, 1352.		77
103	Natural History of Cone Disease in the Murine Model of Leber Congenital Amaurosis Due to CEP290 Mutation: Determining the Timing and Expectation of Therapy. PLoS ONE, 2014, 9, e92928.	2.5	23
104	Posterior Amorphous Corneal Dystrophy Is Associated with a Deletion of Small Leucine-rich Proteoglycans on Chromosome 12. PLoS ONE, 2014, 9, e95037.	2.5	28
105	BBS mutations modify phenotypic expression of CEP290-related ciliopathies. Human Molecular Genetics, 2014, 23, 40-51.	2.9	164
106	Transcriptomic analysis across nasal, temporal, and macular regions of human neural retina and RPE/choroid by RNA-Seq. Experimental Eye Research, 2014, 129, 93-106.	2.6	122
107	TULP1Mutations Causing Early-Onset Retinal Degeneration: Preserved but Insensitive Macular Cones. , 2014, 55, 5354.		47
108	Resolution of Mid-Peripheral Schisis in X-Linked Retinoschisis with the Use of Dorzolamide. Ophthalmic Genetics, 2014, 35, 125-127.	1.2	11

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109	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. Genome Biology, 2014, 15, R53.	9.6	101
110	Photoreceptor Cells With Profound Structural Deficits Can Support Useful Vision in Mice. , 2014, 55, 1859.		15
111	THE VALUE OF RETINAL IMAGING WITH INFRARED SCANNING LASER OPHTHALMOSCOPY IN PATIENTS WITH STARGARDT DISEASE. Retina, 2014, 34, 1391-1399.	1.7	14
112	Is Age-Related Macular Degeneration a Microvascular Disease?. Advances in Experimental Medicine and Biology, 2014, 801, 283-289.	1.6	25
113	Outer Segment Length in Different Best Disease Genotypes. JAMA Ophthalmology, 2014, 132, 1152.	2.5	3
114	The Membrane Attack Complex in Aging Human Choriocapillaris. American Journal of Pathology, 2014, 184, 3142-3153.	3.8	174
115	Inner and Outer Retinal Changes in Retinal Degenerations Associated WithABCA4Mutations. , 2014, 55, 1810.		48
116	Stem cells for investigation and treatment of inherited retinal disease. Human Molecular Genetics, 2014, 23, R9-R16.	2.9	59
117	Mechanical properties of murine and porcine ocular tissues in compression. Experimental Eye Research, 2014, 121, 194-199.	2.6	51
118	Prioritization of Retinal Disease Genes: An Integrative Approach. Human Mutation, 2013, 34, 853-859.	2.5	7
119	Confirmation of the association between theTCF4risk allele and Fuchs endothelial corneal dystrophy in patients from the Midwestern United States. Ophthalmic Genetics, 2013, 34, 32-34.	1.2	22
120	Macular Dystrophies. , 2013, , 852-890.		7
121	A Longitudinal Study of Stargardt Disease: Clinical and Electrophysiologic Assessment, Progression, and Genotype Correlations. American Journal of Ophthalmology, 2013, 155, 1075-1088.e13.	3.3	121
122	Identification of Proteins that Interact with TANK Binding Kinase 1 and Testing for Mutations Associated with Glaucoma. Current Eye Research, 2013, 38, 310-315.	1.5	7
123	Aflibercept Therapy for Exudative Age-related Macular Degeneration Resistant to Bevacizumab and Ranibizumab. American Journal of Ophthalmology, 2013, 156, 15-22.e1.	3.3	217
124	Wide-Field Spectral-Domain Optical Coherence Tomography in Patients and Carriers of X-linked Retinoschisis. Ophthalmology, 2013, 120, 169-174.	5.2	56
125	Non-exomic and synonymous variants in ABCA4 are an important cause of Stargardt disease. Human Molecular Genetics, 2013, 22, 5136-5145.	2.9	159
126	BBS7 is required for BBSome formation and its absence in mice results in Bardet-Biedl syndrome phenotypes and selective abnormalities in membrane protein trafficking. Journal of Cell Science, 2013, 126, 2372-80.	2.0	115

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127	Determining consequences of retinal membrane guanylyl cyclase (RetGC1) deficiency in human Leber congenital amaurosis en route to therapy: residual cone-photoreceptor vision correlates with biochemical properties of the mutants. Human Molecular Genetics, 2013, 22, 168-183.	2.9	89
128	Use of a Synthetic Xeno-Free Culture Substrate for Induced Pluripotent Stem Cell Induction and Retinal Differentiation. Stem Cells Translational Medicine, 2013, 2, 16-24.	3.3	89
129	RETINAL DETACHMENT IN A PATIENT WITH LEBER CONGENITAL AMAUROSIS. Retinal Cases and Brief Reports, 2013, 7, 102-104.	0.6	3
130	Visual Acuity Changes in Patients With Leber Congenital Amaurosis and Mutations in CEP290. JAMA Ophthalmology, 2013, 131, 178.	2.5	44
131	Patient-specific iPSC-derived photoreceptor precursor cells as a means to investigate retinitis pigmentosa. ELife, 2013, 2, e00824.	6.0	168
132	Human Photoreceptor Outer Segments Shorten During Light Adaptation. , 2013, 54, 3721.		63
133	Subretinal Gene Therapy of Mice With Bardet-Biedl Syndrome Type 1. , 2013, 54, 6118.		79
134	A Longitudinal Study of Stargardt Disease: Quantitative Assessment of Fundus Autofluorescence, Progression, and Genotype Correlations. , 2013, 54, 8181.		119
135	A Genome-Wide Association Study for Primary Open Angle Glaucoma and Macular Degeneration Reveals Novel Loci. PLoS ONE, 2013, 8, e58657.	2.5	35
136	Calpain-5 Mutations Cause Autoimmune Uveitis, Retinal Neovascularization, and Photoreceptor Degeneration. PLoS Genetics, 2012, 8, e1003001.	3.5	76
137	BBS proteins interact genetically with the IFT pathway to influence SHH-related phenotypes. Human Molecular Genetics, 2012, 21, 1945-1953.	2.9	123
138	Three-dimensional Distribution of the Vitelliform Lesion, Photoreceptors, and Retinal Pigment Epithelium in the Macula of Patients With Best Vitelliform Macular Dystrophy. JAMA Ophthalmology, 2012, 130, 357.	2.4	54
139	Intravitreal Bevacizumab for Peripapillary Choroidal Neovascular Membranes. JAMA Ophthalmology, 2012, 130, 1073.	2.4	8
140	Analysis of ASB10 variants in open angle glaucoma. Human Molecular Genetics, 2012, 21, 4543-4548.	2.9	20
141	Topical Ocular Sodium 4-Phenylbutyrate Rescues Glaucoma in a Myocilin Mouse Model of Primary Open-Angle Glaucoma. , 2012, 53, 1557.		100
142	Intrinsic Protein-Protein Interaction-mediated and Chaperonin-assisted Sequential Assembly of Stable Bardet-Biedl Syndrome Protein Complex, the BBSome. Journal of Biological Chemistry, 2012, 287, 20625-20635.	3.4	142
143	Gene Therapy for Leber Congenital Amaurosis Caused by RPE65 Mutations. JAMA Ophthalmology, 2012, 130, 9.	2.4	580
144	PROTEOMIC ANALYSIS OF VITREOUS BIOPSY TECHNIQUES. Retina, 2012, 32, 2141-2149.	1.7	23

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145	Phenotypic expression of Bardetâ€“Biedl syndrome in patients homozygous for the common M390R mutation in the BBS1 gene. Vision Research, 2012, 75, 77-87.	1.4	34
146	Assessing Retinal Structure in Complete Congenital Stationary Night Blindness and Oguchi Disease. American Journal of Ophthalmology, 2012, 154, 987-1001.e1.	3.3	55
147	Recommendations for Genetic Testing of Inherited Eye Diseases. Ophthalmology, 2012, 119, 2408-2410.	5.2	157
148	TUDCA Slows Retinal Degeneration in Two Different Mouse Models of Retinitis Pigmentosa and Prevents Obesity in Bardet-Biedl Syndrome Type 1 Mice. , 2012, 53, 100.		84
149	Macular Function in Macular Degenerations: Repeatability of Microperimetry as a Potential Outcome Measure forABCA4-Associated Retinopathy Trials. , 2012, 53, 841.		105
150	Inhibition of Neovascularization but Not Fibrosis With the Fluocinolone Acetonide Implant in Autosomal Dominant Neovascular Inflammatory Vitreoretinopathy. JAMA Ophthalmology, 2012, 130, 1395.	2.4	15
151	Sequencing and disease variation detection tools and techniques. , 2011, , .		1
152	Photoreceptor Structure and Function in Patients with Congenital Achromatopsia. , 2011, 52, 7298.		142
153	Genomics and the Eye. New England Journal of Medicine, 2011, 364, 1932-1942.	27.0	81
154	Elevated membrane attack complex in human choroid with high risk complement factor H genotypes. Experimental Eye Research, 2011, 93, 565-567.	2.6	112
155	Autosomal Recessive Retinitis Pigmentosa Caused by Mutations in theMAKGene. , 2011, 52, 9665.		39
156	Computational Quantification of Complex Fundus Phenotypes in Age-Related Macular Degeneration and Stargardt Disease. , 2011, 52, 2976.		14
157	The Phenotype of Severe Early Childhood Onset Retinal Dystrophy (SECORD) from Mutation ofRPE65and Differentiation from Leber Congenital Amaurosis. , 2011, 52, 292.		65
158	Automated Discovery and Quantification of Image-Based Complex Phenotypes: A Twin Study of Drusen Phenotypes in Age-Related Macular Degeneration. , 2011, 52, 9195.		7
159	AUTOSOMAL RECESSIVE VITELLIFORM MACULAR DYSTROPHY IN A LARGE COHORT OF VITELLIFORM MACULAR DYSTROPHY PATIENTS. Retina, 2011, 31, 581-595.	1.7	75
160	BILATERAL INTRAVITREAL INJECTION OF ANTIVASCULAR ENDOTHELIAL GROWTH FACTOR THERAPY. Retina, 2011, 31, 31-35.	1.7	30
161	Variations in NPHP5 in Patients With Nonsyndromic Leber Congenital Amaurosis and Senior-Loken Syndrome. JAMA Ophthalmology, 2011, 129, 81.	2.4	62
162	Seroreactivity Against Aqueous-Soluble and Detergent-Soluble Retinal Proteins in Posterior Uveitis. JAMA Ophthalmology, 2011, 129, 415.	2.4	18

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163	Genome-wide analysis of copy number variants in age-related macular degeneration. Human Genetics, 2011, 129, 91-100.	3.8	36
164	Evaluation of variants in the selectin genes in age-related macular degeneration. BMC Medical Genetics, 2011, 12, 58.	2.1	15
165	Human Retinal Disease fromAIPL1Gene Mutations: Foveal Cone Loss with Minimal Macular Photoreceptors and Rod Function Remaining. , 2011, 52, 70.		59
166	HumanCRB1-Associated Retinal Degeneration: Comparison with therd8 Crb1-Mutant Mouse Model. , 2011, 52, 6898.		98
167	Residual Electroretinograms in Young Leber Congenital Amaurosis Patients with Mutations ofAIPL1. , 2011, 52, 8166.		32
168	Different Inner Retinal Pathways Mediate Rod-Cone Input in Irradiance Detection for the Pupillary Light Reflex and Regulation of Behavioral State in Mice. , 2011, 52, 618.		17
169	The N-terminal region of centrosomal protein 290 (CEP290) restores vision in a zebrafish model of human blindness. Human Molecular Genetics, 2011, 20, 1467-1477.	2.9	56
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