Edwin M Stone

List of Publications by Year
in descending order

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376 papers	30,776 citations	2795 94 h-index	7333 152 g-index
379 all docs	379 docs citations	379 times ranked	18068 citing authors

#	Article	IF	Citations
1	Mutations in the SMAD4/DPC4 Gene in Juvenile Polyposis. Science, 1998, 280, 1086-1088.	6.0	866
2	The Sensitivity of Single-Strand Conformation Polymorphism Analysis for the Detection of Single Base Substitutions. Genomics, 1993, 16, 325-332.	1.3	653
3	Human gene therapy for RPE65 isomerase deficiency activates the retinoid cycle of vision but with slow rod kinetics. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 15112-15117.	3.3	639
4	Gene Therapy for Leber Congenital Amaurosis Caused by RPE65 Mutations. JAMA Ophthalmology, 2012, 130, 9.	2.6	580
5	Cloning and sequencing of a deoxyribonucleic acid copy of glyceraldehyde 3-phosphate dehydrogenase messenger ribonucleic acid isolated from chicken muscle. Biochemistry, 1983, 22, 1605-1613.	1.2	475
6	A single EFEMP1 mutation associated with both Malattia Leventinese and Doyne honeycomb retinal dystrophy. Nature Genetics, 1999, 22, 199-202.	9.4	453
7	Mutation of a nuclear receptor gene, NR2E3, causes enhanced S cone syndrome, a disorder of retinal cell fate. Nature Genetics, 2000, 24, 127-131.	9.4	439
8	Clinical Features Associated with Mutations in the Chromosome 1 Open-Angle Glaucoma Gene (GLC1A). New England Journal of Medicine, 1998, 338, 1022-1027.	13.9	423
9	The forkhead transcription factor gene FKHL7 is responsible for glaucoma phenotypes which map to 6p25. Nature Genetics, 1998, 19, 140-147.	9.4	416
10	Genetic linkage of familial open angle glaucoma to chromosome 1q21â€“q31. Nature Genetics, 1993, 4, 47-50.	9.4	410
11	Homozygosity mapping with SNP arrays identifies TRIM32, an E3 ubiquitin ligase, as a Bardet-Biedl syndrome gene (BBS11). Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 6287-6292.	3.3	378
12	Bbs2-null mice have neurosensory deficits, a defect in social dominance, and retinopathy associated with mislocalization of rhodopsin. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 16588-16593.	3.3	345
13	Identification of the gene (BBS1) most commonly involved in Bardet-Biedl syndrome, a complex human obesity syndrome. Nature Genetics, 2002, 31, 435-438.	9.4	327
14	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.	0.5	314
15	Clinically Focused Molecular Investigation of 1000 Consecutive Families with Inherited Retinal Disease. Ophthalmology, 2017, 124, 1314-1331.	2.5	312
16	Bardet-Biedl syndrome type 4 (BBS4)-null mice implicate Bbs4 in flagella formation but not global cilia assembly. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 8664-8669.	3.3	309
17	Missense Variations in the Fibulin 5 Gene and Age-Related Macular Degeneration. New England Journal of Medicine, 2004, 351, 346-353.	13.9	298
18	De novo mutations in the CRX homeobox gene associated with Leber congenital amaurosis. Nature Genetics, 1998, 18, 311-312.	9.4	276

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19	Leber Congenital Amaurosisâ€“A Model for Efficient Genetic Testing of Heterogeneous Disorders: LXIV Edward Jackson Memorial Lecture. American Journal of Ophthalmology, 2007, 144, 791-811.e6.	1.7	273
20	Butterflyâ€“shaped pigment dystrophy of the fovea caused by a point mutation in codon 167 of the RDS gene. Nature Genetics, 1993, 3, 202-207.	9.4	272
21	Mutations in MKKS cause Bardet-Biedl syndrome. Nature Genetics, 2000, 26, 15-16.	9.4	256
22	Identification of the gene that, when mutated, causes the human obesity syndrome BBS4. Nature Genetics, 2001, 28, 188-191.	9.4	254
23	Identifying photoreceptors in blind eyes caused by RPE65 mutations: Prerequisite for human gene therapy success. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 6177-6182.	3.3	249
24	Reduction of ER stress via a chemical chaperone prevents disease phenotypes in a mouse model of primary open angle glaucoma. Journal of Clinical Investigation, 2011, 121, 3542-3553.	3.9	249
25	A knockin mouse model of the Bardetâ€“Biedl syndrome 1 M390R mutation has cilia defects, ventriculomegaly, retinopathy, and obesity. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 19422-19427.	3.3	237
26	Genetic linkage of vitelliform macular degeneration (Best's disease) to chromosome 11q13. Nature Genetics, 1992, 1, 246-250.	9.4	234
27	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.	3.3	234
28	Mutations in ABCA4 result in accumulation of lipofuscin before slowing of the retinoid cycle: a reappraisal of the human disease sequence. Human Molecular Genetics, 2004, 13, 525-534.	1.4	231
29	Comparative Genomic Analysis Identifies an ADP-Ribosylation Factorâ€“like Gene as the Cause of Bardet-Biedl Syndrome (BBS3). American Journal of Human Genetics, 2004, 75, 475-484.	2.6	220
30	The nuclear receptor NR2E3 plays a role in human retinal photoreceptor differentiation and degeneration. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 473-478.	3.3	218
31	Aflibercept Therapy for Exudative Age-related Macular Degeneration Resistant to Bevacizumab and Ranibizumab. American Journal of Ophthalmology, 2013, 156, 15-22.e1.	1.7	217
32	Identification of a Bardet-Biedl syndrome locus on chromosome 3 and evaluation of an efficient approach to homozygosity mapping. Human Molecular Genetics, 1994, 3, 1331-1335.	1.4	216
33	Crumbs homolog 1 (CRB1) mutations result in a thick human retina with abnormal lamination. Human Molecular Genetics, 2003, 12, 1073-1078.	1.4	205
34	Myocilin Glaucoma. Survey of Ophthalmology, 2002, 47, 547-561.	1.7	201
35	Frequency of Usher syndrome in two pediatric populations: Implications for genetic screening of deaf and hard of hearing children. Genetics in Medicine, 2010, 12, 512-516.	1.1	198
36	Night blindness in Sorsby's fundus dystrophy reversed by vitamin A. Nature Genetics, 1995, 11, 27-32.	9.4	197

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37	Allelic variation in ABCR associated with Stargardt disease but not age-related macular degeneration. Nature Genetics, 1998, 20, 328-329.	9.4	194
38	Comparative Genomics and Gene Expression Analysis Identifies BBS9, a New Bardet-Biedl Syndrome Gene. American Journal of Human Genetics, 2005, 77, 1021-1033.	2.6	194
39	Regulation of gene expression in the mammalian eye and its relevance to eye disease. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 14429-14434.	3.3	190
40	Copy number variations on chromosome 12q14 in patients with normal tension glaucoma. Human Molecular Genetics, 2011, 20, 2482-2494.	1.4	189
41	Complement activation and choriocapillaris loss in early AMD: Implications for pathophysiology and therapy. Progress in Retinal and Eye Research, 2015, 45, 1-29.	7.3	189
42	Exome sequencing and analysis of induced pluripotent stem cells identify the cilia-related gene male germ cell-associated kinase (MAK) as a cause of retinitis pigmentosa. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, E569-76.	3.3	186
43	A Spectrum of FOXC1 Mutations Suggests Gene Dosage as a Mechanism for Developmental Defects of the Anterior Chamber of the Eye. American Journal of Human Genetics, 2001, 68, 364-372.	2.6	185
44	Mkks-null mice have a phenotype resembling Bardetâ€“Biedl syndrome. Human Molecular Genetics, 2005, 14, 1109-1118.	1.4	181
45	Linkage of Bardetâ€“Biedl syndrome to chromosome 16q and evidence for nonâ€“allelic genetic heterogeneity. Nature Genetics, 1993, 5, 392-396.	9.4	176
46	Bardetâ€“Biedl syndrome genes are important in retrograde intracellular trafficking and Kupffer's vesicle cilia function. Human Molecular Genetics, 2006, 15, 667-677.	1.4	176
47	Regional Distribution of Retinal Degeneration in Patients with the Proline to Histidine Mutation in Codon 23 of the Rhodopsion Gene. Ophthalmology, 1991, 98, 1806-1813.	2.5	175
48	The Membrane Attack Complex in Aging Human Choriocapillaris. American Journal of Pathology, 2014, 184, 3142-3153.	1.9	174
49	Patient-specific iPSC-derived photoreceptor precursor cells as a means to investigate retinitis pigmentosa. ELife, 2013, 2, e00824.	2.8	168
50	Evaluation of optineurin sequence variations in 1,048 patients with open-angle glaucoma. American Journal of Ophthalmology, 2003, 136, 904-910.	1.7	164
51	BBS mutations modify phenotypic expression of CEP290-related ciliopathies. Human Molecular Genetics, 2014, 23, 40-51.	1.4	164
52	ABCA4 disease progression and a proposed strategy for gene therapy. Human Molecular Genetics, 2009, 18, 931-941.	1.4	163
53	Identical mutation in a novel retinal gene causes progressive rodâ€“cone degeneration in dogs and retinitis pigmentosa in humans. Genomics, 2006, 88, 551-563.	1.3	161
54	Three autosomal dominant corneal dystrophies map to chromosome 5q. Nature Genetics, 1994, 6, 47-51.	9.4	159

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55	Pendred syndrome maps to chromosome 7q21-34 and is caused by an intrinsic defect in thyroid iodine organification. Nature Genetics, 1996, 12, 424-426.	9.4	159
56	Non-exomic and synonymous variants in ABCA4 are an important cause of Stargardt disease. Human Molecular Genetics, 2013, 22, 5136-5145.	1.4	159
57	Clinical Features of a Stargardt-Like Dominant Progressive Macular Dystrophy With Genetic Linkage to Chromosome 6q. JAMA Ophthalmology, 1994, 112, 765.	2.6	158
58	Recommendations for Genetic Testing of Inherited Eye Diseases. Ophthalmology, 2012, 119, 2408-2410.	2.5	157
59	Remodeling of the Human Retina in Choroideremia: Rab Escort Protein 1 (REP-1) Mutations. , 2006, 47, 4113.		156
60	Centrosomal-ciliary geneCEP290/NPHP6 mutations result in blindness with unexpected sparing of photoreceptors and visual brain: implications for therapy of Leber congenital amaurosis. Human Mutation, 2007, 28, 1074-1083.	1.1	148
61	Increased expression of the WNT antagonist sFRP-1 in glaucoma elevates intraocular pressure. Journal of Clinical Investigation, 2008, 118, 1056-64.	3.9	143
62	Photoreceptor Structure and Function in Patients with Congenital Achromatopsia. , 2011, 52, 7298.		142
63	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.	1.6	142
64	Retinal synthesis and deposition of complement components induced by ocular hypertension. Experimental Eye Research, 2006, 83, 620-628.	1.2	139
65	Avellino Corneal Dystrophy. Ophthalmology, 1992, 99, 1564-1568.	2.5	137
66	Linkage of posterior polymorphous corneal dystrophy to 20q11. Human Molecular Genetics, 1995, 4, 485-488.	1.4	135
67	Human cone photoreceptor dependence on RPE65 isomerase. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 15123-15128.	3.3	135
68	Bardet-Biedl syndrome 3 (Bbs3) knockout mouse model reveals common BBS-associated phenotypes and Bbs3 unique phenotypes. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 20678-20683.	3.3	135
69	Reduced-illuminance autofluorescence imaging in ABCA4-associated retinal degenerations. Journal of the Optical Society of America A: Optics and Image Science, and Vision, 2007, 24, 1457.	0.8	131
70	Autosomal dominant iris hypoplasia is caused by a mutation in the rieger syndrome (rieg/pitx2) gene. American Journal of Ophthalmology, 1998, 125, 98-100.	1.7	130
71	The R345W mutation in EFEMP1 is pathogenic and causes AMD-like deposits in mice. Human Molecular Genetics, 2007, 16, 2411-2422.	1.4	129
72	A family with Axenfeldâ€“Rieger syndrome and Peters Anomaly caused by a point mutation (Phe112Ser) in the FOXC1 gene. American Journal of Ophthalmology, 2003, 135, 368-375.	1.7	128

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73	A Gene for Familial Juvenile Polyposis Maps to Chromosome 18q21.1. American Journal of Human Genetics, 1998, 62, 1129-1136.	2.6	123
74	BBS proteins interact genetically with the IFT pathway to influence SHH-related phenotypes. Human Molecular Genetics, 2012, 21, 1945-1953.	1.4	123
75	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.	1.2	122
76	Use of isolated inbred human populations for identification of disease genes. Trends in Genetics, 1998, 14, 391-396.	2.9	121
77	A Longitudinal Study of Stargardt Disease: Clinical and Electrophysiologic Assessment, Progression, and Genotype Correlations. American Journal of Ophthalmology, 2013, 155, 1075-1088.e13.	1.7	121
78	Using CRISPR-Cas9 to Generate Gene-Corrected Autologous iPSCs for the Treatment of Inherited Retinal Degeneration. Molecular Therapy, 2017, 25, 1999-2013.	3.7	121
79	A Longitudinal Study of Stargardt Disease: Quantitative Assessment of Fundus Autofluorescence, Progression, and Genotype Correlations. , 2013, 54, 8181.		119
80	Retinal Laminar Architecture in Human Retinitis Pigmentosa Caused byRhodopsinGene Mutations. , 2008, 49, 1580.		118
81	Evaluation of Complex Inheritance Involving the Most Common Bardet-Biedl Syndrome Locus (BBS1). American Journal of Human Genetics, 2003, 72, 429-437.	2.6	117
82	ABCA4-Associated Retinal Degenerations Spare Structure and Function of the Human Parapapillary Retina. , 2005, 46, 4739.		117
83	Cone photoreceptors are the main targets for gene therapy of NPHP5 (IQCB1) or NPHP6 (CEP290) blindness: generation of an all-cone Nphp6 hypomorph mouse that mimics the human retinal ciliopathy. Human Molecular Genetics, 2011, 20, 1411-1423.	1.4	115
84	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.	1.2	115
85	Clinical Features and Linkage Analysis of a Family with Autosomal Dominant Juvenile Glaucoma. Ophthalmology, 1993, 100, 524-529.	2.5	114
86	Elevated membrane attack complex in human choroid with high risk complement factor H genotypes. Experimental Eye Research, 2011, 93, 565-567.	1.2	112
87	LOXL1 Mutations Are Associated with Exfoliation Syndrome in Patients from the Midwestern United States. American Journal of Ophthalmology, 2007, 144, 974-975.e1.	1.7	111
88	A case-control study of tobacco and alcohol consumption in leber hereditary optic neuropathy. American Journal of Ophthalmology, 2000, 130, 803-812.	1.7	109
89	Differential Macular and Peripheral Expression of Bestrophin in Human Eyes and Its Implication for Best Disease. , 2007, 48, 3372.		109
90	Defining the Residual Vision in Leber Congenital Amaurosis Caused byRPE65Mutations. , 2009, 50, 2368.		109

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91	Bestrophin Gene Mutations Cause Canine Multifocal Retinopathy: A Novel Animal Model for Best Disease. , 2007, 48, 1959.		108
92	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.	7.3	108
93	cGMP production of patient-specific iPSCs and photoreceptor precursor cells to treat retinal degenerative blindness. Scientific Reports, 2016, 6, 30742.	1.6	108
94	Evidence for genetic heterogeneity within eight glaucoma families, with the GLC1A Gln368STOP mutation being an important phenotypic modifier11None of the authors has a financial interest relating to this article.. Ophthalmology, 2001, 108, 1607-1620.	2.5	106
95	Evidence for a Novel X-Linked Modifier Locus for Leber Hereditary Optic Neuropathy. Ophthalmic Genetics, 2008, 29, 17-24.	0.5	105
96	Macular Function in Macular Degenerations: Repeatability of Microperimetry as a Potential Outcome Measure forABCA4-Associated Retinopathy Trials. , 2012, 53, 841.		105
97	North Carolina Macular Dystrophy Is Caused by Dysregulation of the Retinal Transcription Factor PRDM13. Ophthalmology, 2016, 123, 9-18.	2.5	105
98	Characterization and Comparison of the Human and MouseGLC1A Glaucomaâ€‰Genes. Genome Research, 1998, 8, 377-384.	2.4	103
99	Glaucoma-causing myocilin mutants require the Peroxisomal targeting signal-1 receptor (PTS1R) to elevate intraocular pressure. Human Molecular Genetics, 2007, 16, 609-617.	1.4	101
100	Complement Component C5a Activates ICAM-1 Expression on Human Choroidal Endothelial Cells. , 2010, 51, 5336.		101
101	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.	13.9	101
102	Topical Ocular Sodium 4-Phenylbutyrate Rescues Glaucoma in a Myocilin Mouse Model of Primary Open-Angle Glaucoma. , 2012, 53, 1557.		100
103	Connexin mutations and hearing loss. Nature, 1998, 391, 32-32.	13.7	98
104	HumanCRB1-Associated Retinal Degeneration: Comparison with therd8 Crb1-Mutant Mouse Model. , 2011, 52, 6898.		98
105	Phenotypic differences among patients with Bardet-Biedl syndrome linked to three different chromosome loci. American Journal of Medical Genetics Part A, 1995, 59, 199-203.	2.4	96
106	Macular Pigment and Lutein Supplementation in Choroideremia. Experimental Eye Research, 2002, 74, 371-381.	1.2	96
107	Adeno-Associated Virus Type 5: Transduction Efficiency and Cell-Type Specificity in the Primate Retina. Human Gene Therapy, 2003, 14, 1663-1671.	1.4	95
108	Late Development of Vitelliform Lesions and Flecks in a Patient With Best Disease. JAMA Ophthalmology, 2005, 123, 1588.	2.6	95

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109	Visual Acuity in Patients with Leber's Congenital Amaurosis and Early Childhood-Onset Retinitis Pigmentosa. Ophthalmology, 2010, 117, 1190-1198.	2.5	95
110	Nuclear receptor NR2E3 gene mutations distort human retinal laminar architecture and cause an unusual degeneration. Human Molecular Genetics, 2004, 13, 1893-1902.	1.4	94
111	ABCA4 Gene Sequence Variations in Patients With Autosomal Recessive Cone-Rod Dystrophy. JAMA Ophthalmology, 2003, 121, 851.	2.6	89
112	Differential Macular Morphology in Patients withRPE65-,CEP290-,GUCY2D-, andAIPL1-Related Leber Congenital Amaurosis. , 2010, 51, 2608.		89
113	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.	1.4	89
114	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.	1.6	89
115	Photoreceptor Layer Topography in Children with Leber Congenital Amaurosis Caused byRPE65Mutations. , 2008, 49, 4573.		86
116	Bestrophinopathy: An RPE-photoreceptor interface disease. Progress in Retinal and Eye Research, 2017, 58, 70-88.	7.3	85
117	Primary congenital and developmental glaucomas. Human Molecular Genetics, 2017, 26, R28-R36.	1.4	85
118	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
119	Intron-dependent evolution of chicken glyceraldehyde phosphate dehydrogenase gene. Nature, 1985, 313, 498-500.	13.7	83
120	Retinal Disease Expression in Bardet-Biedl Syndrome-1 (BBS1) Is a Spectrum from Maculopathy to Retina-Wide Degeneration. , 2006, 47, 5004.		83
121	CERKLMutations Cause an Autosomal Recessive Cone-Rod Dystrophy with Inner Retinopathy. , 2009, 50, 5944.		83
122	Genome-wide identification of pseudogenes capable of disease-causing gene conversion. Human Mutation, 2006, 27, 545-552.	1.1	82
123	Genomics and the Eye. New England Journal of Medicine, 2011, 364, 1932-1942.	13.9	81
124	Ranibizumab Therapy for Neovascular Age-Related Macular Degeneration. New England Journal of Medicine, 2010, 363, 1648-1655.	13.9	80
125	Subretinal Gene Therapy of Mice With Bardet-Biedl Syndrome Type 1. , 2013, 54, 6118.		79
126	Structural and Biochemical Analyses of Choroidal Thickness in Human Donor Eyes. , 2014, 55, 1352.		77

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127	Calpain-5 Mutations Cause Autoimmune Uveitis, Retinal Neovascularization, and Photoreceptor Degeneration. PLoS Genetics, 2012, 8, e1003001.	1.5	76
128	Two-photon polymerization for production of human iPSC-derived retinal cell grafts. Acta Biomaterialia, 2017, 55, 385-395.	4.1	76
129	EVALUATION OF GENOTYPEâ€“PHENOTYPE ASSOCIATIONS IN LEBER CONGENITAL AMAUROSIS. Retina, 2005, 25, 919-929.	1.0	75
130	Retinal Disease in Usher Syndrome III Caused by Mutations in the Clarin-1 Gene. , 2008, 49, 2651.		75
131	Identification and Functional Analysis of the Vision-Specific BBS3 (ARL6) Long Isoform. PLoS Genetics, 2010, 6, e1000884.	1.5	75
132	AUTOSOMAL RECESSIVE VITELLIFORM MACULAR DYSTROPHY IN A LARGE COHORT OF VITELLIFORM MACULAR DYSTROPHY PATIENTS. Retina, 2011, 31, 581-595.	1.0	75
133	QRX, a novel homeobox gene, modulates photoreceptor gene expression. Human Molecular Genetics, 2004, 13, 1025-1040.	1.4	73
134	Basal exon skipping and genetic pleiotropy: A predictive model of disease pathogenesis. Science Translational Medicine, 2015, 7, 291ra97.	5.8	73
135	Pitfalls in Homozygosity Mapping. American Journal of Human Genetics, 2000, 67, 1348-1351.	2.6	72
136	Linkage of autosomal dominant iris hypoplasia to the region of the Rieger syndrome locus (4q25). Human Molecular Genetics, 1995, 4, 1435-1439.	1.4	70
137	Discovery and Functional Analysis of a Retinitis Pigmentosa Gene, C2ORF71. American Journal of Human Genetics, 2010, 86, 686-695.	2.6	70
138	Protein misfolding and the pathogenesis of ABCA4-associated retinal degenerations. Human Molecular Genetics, 2015, 24, 3220-3237.	1.4	69
139	In vivo micropathology of Best macular dystrophy with optical coherence tomography. Experimental Eye Research, 2003, 76, 203-211.	1.2	68
140	Heterozygous Deep-Intronic Variants and Deletions inABCA4in Persons with Retinal Dystrophies and One ExonicABCA4Variant. Human Mutation, 2015, 36, 43-47.	1.1	68
141	Ocular phenotypes of three genetic variants of Bardet-Biedl syndrome. , 2005, 132A, 283-287.		66
142	Ethnic variation in AMD-associated complement factor H polymorphism p.Tyr402His. Human Mutation, 2006, 27, 921-925.	1.1	66
143	A Very Effective Treatment for Neovascular Macular Degeneration. New England Journal of Medicine, 2006, 355, 1493-1495.	13.9	66
144	RDH12andRPE65, Visual Cycle Genes Causing Leber Congenital Amaurosis, Differ in Disease Expression. , 2007, 48, 332.		66

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145	The Phenotype of Severe Early Childhood Onset Retinal Dystrophy (SECORD) from Mutation ofRPE65and Differentiation from Leber Congenital Amaurosis. , 2011, 52, 292.		65
146	Hypomorphic mutations inTRNT1cause retinitis pigmentosa with erythrocytic microcytosis. Human Molecular Genetics, 2016, 25, 44-56.	1.4	64
147	CRISPR-Cas9 genome engineering: Treating inherited retinal degeneration. Progress in Retinal and Eye Research, 2018, 65, 28-49.	7.3	64
148	Macular Pigment and Lutein Supplementation inABCA4-Associated Retinal Degenerations. , 2007, 48, 1319.		63
149	Human Photoreceptor Outer Segments Shorten During Light Adaptation. , 2013, 54, 3721.		63
150	Mutation analysis of the ROM1 gene in retinitis pigmentosa. Human Molecular Genetics, 1995, 4, 1895-1902.	1.4	62
151	Variations in NPHP5 in Patients With Nonsyndromic Leber Congenital Amaurosis and Senior-Loken Syndrome. JAMA Ophthalmology, 2011, 129, 81.	2.6	62
152	A Novel GCAP1 Missense Mutation (L151F) in a Large Family with Autosomal Dominant Cone-Rod Dystrophy (adCORD). , 2005, 46, 1124.		61
153	A Peripherin/Retinal Degeneration Slow Mutation (Pro-210-Arg) Associated with Macular and Peripheral Retinal Degeneration. Ophthalmology, 1995, 102, 246-255.	2.5	60
154	Disease Expression in Usher Syndrome Caused byVLGR1Gene Mutation (USH2C) and Comparison withUSH2APhenotype. , 2005, 46, 734.		60
155	CRB1 mutations may result in retinitis pigmentosa without para-arteriolar RPE preservation. Ophthalmic Genetics, 2001, 22, 163-169.	0.5	59
156	Human Retinal Disease fromAIPL1Gene Mutations: Foveal Cone Loss with Minimal Macular Photoreceptors and Rod Function Remaining. , 2011, 52, 70.		59
157	Autosomal Recessive Best Vitelliform Macular Dystrophy. JAMA Ophthalmology, 2011, 129, 211.	2.6	59
158	Stem cells for investigation and treatment of inherited retinal disease. Human Molecular Genetics, 2014, 23, R9-R16.	1.4	59
159	Gene Expression Analysis of Photoreceptor Cell Loss inBbs4-Knockout Mice Reveals an Early Stress Gene Response and Photoreceptor Cell Damage. , 2007, 48, 3329.		57
160	Novel approaches to linkage mapping. Current Opinion in Genetics and Development, 1995, 5, 335-341.	1.5	56
161	Visual Impairment in the Absence of Dystroglycan. Journal of Neuroscience, 2009, 29, 13136-13146.	1.7	56
162	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.	1.4	56

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163	Wide-Field Spectral-Domain Optical Coherence Tomography in Patients and Carriers of X-linked Retinoschisis. Ophthalmology, 2013, 120, 169-174.	2.5	56
164	Assessing Retinal Structure in Complete Congenital Stationary Night Blindness and Oguchi Disease. American Journal of Ophthalmology, 2012, 154, 987-1001.e1.	1.7	55
165	Expression of theMf1 gene in developing mouse hearts: Implication in the development of human congenital heart defects. , 1999, 216, 16-27.		54
166	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.	2.6	54
167	Relation of Response to Treatment with Dorzolamide in X-Linked Retinoschisis to the Mechanism of Functional Loss in Retinoschisin. American Journal of Ophthalmology, 2009, 147, 111-115.e1.	1.7	53
168	Deducing the pathogenic contribution of recessive ABCA4 alleles in an outbred population. Human Molecular Genetics, 2010, 19, 3693-3701.	1.4	53
169	Assessment of Adeno-Associated Virus Serotype Tropism in Human Retinal Explants. Human Gene Therapy, 2018, 29, 424-436.	1.4	53
170	Erosive Vitreoretinopathy. Ophthalmology, 1994, 101, 694-704.	2.5	52
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