Anand Swaroop

List of Publications by Year
in descending order

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346 papers	34,100 citations	4960 84 h-index	5679 162 g-index
363 all docs	363 docs citations	363 times ranked	33132 citing authors

#	Article	IF	Citations
1	Restoration of RPGR expression in vivo using CRISPR/Cas9 gene editing. Gene Therapy, 2022, 29, 81-93.	4.5	18
2	A novel exome probe set captures phototransduction genes across birds (Aves) enabling efficient analysis of vision evolution. Molecular Ecology Resources, 2022, 22, 587-601.	4.8	3
3	Multiomics analyses reveal early metabolic imbalance and mitochondrial stress in neonatal photoreceptors leading to cell death in Pde6brd1/rd1 mouse model of retinal degeneration. Human Molecular Genetics, 2022, 31, 2137-2154.	2.9	25
4	GATD3A, a mitochondrial deglycase with evolutionary origins from gammaproteobacteria, restricts the formation of advanced glycation end products. BMC Biology, 2022, 20, 68.	3.8	6
5	HiCRes: a computational method to estimate and predict the genomic resolution of Hi-C libraries. Nucleic Acids Research, 2022, 50, e35-e35.	14.5	8
6	Developmental genome-wide occupancy analysis of bZIP transcription factor NRL uncovers the role of c-Jun in early differentiation of rod photoreceptors in the mammalian retina. Human Molecular Genetics, 2022, 31, 3914-3933.	2.9	4
7	Primary cilia biogenesis and associated retinal ciliopathies. Seminars in Cell and Developmental Biology, 2021, 110, 70-88.	5.0	60
8	Dietary Nutrient Intake and Progression to Late Age-Related Macular Degeneration in the Age-Related Eye Disease Studies 1 and 2. Ophthalmology, 2021, 128, 425-442.	5.2	66
9	Gene Therapy of Dominant CRX-Leber Congenital Amaurosis using Patient Stem Cell-Derived Retinal Organoids. Stem Cell Reports, 2021, 16, 252-263.	4.8	53
10	Genome-Wide Association Studies-Based Machine Learning for Prediction of Age-Related Macular Degeneration Risk. Translational Vision Science and Technology, 2021, 10, 29.	2.2	14
11	Genetics and therapy for pediatric eye diseases. EBioMedicine, 2021, 67, 103360.	6.1	7
12	A Novel ARL3 Gene Mutation Associated With Autosomal Dominant Retinal Degeneration. Frontiers in Cell and Developmental Biology, 2021, 9, 720782.	3.7	13
13	Glycogen Synthase Kinase 3 Regulates the Genesis of Displaced Retinal Ganglion Cells3. ENeuro, 2021, 8, ENEURO.0171-21.2021.	1.9	5
14	Aging of the Retina: Molecular and Metabolic Turbulences and Potential Interventions. Annual Review of Vision Science, 2021, 7, 633-664.	4.4	28
15	Proneural genes define ground-state rules to regulate neurogenic patterning and cortical folding. Neuron, 2021, 109, 2847-2863.e11.	8.1	26
16	Making Biological Sense of Genetic Studies of Age-Related Macular Degeneration. Advances in Experimental Medicine and Biology, 2021, 1256, 201-219.	1.6	2
17	Association of Smoking, Alcohol Consumption, Blood Pressure, Body Mass Index, and Glycemic Risk Factors With Age-Related Macular Degeneration. JAMA Ophthalmology, 2021, 139, 1299.	2.5	29
18	HIPRO: A High-Efficiency, Hypoxia-Induced Protocol for Generation of Photoreceptors in Retinal Organoids from Mouse Pluripotent Stem Cells. STAR Protocols, 2020, 1, 100018.	1.2	4

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19	Accelerated Development of Rod Photoreceptors in Retinal Organoids Derived from Human Pluripotent Stem Cells by Supplementation with 9-cis Retinal. STAR Protocols, 2020, 1, 100033.	1.2	12
20	Divergent Effects of HSP70 Overexpression in Photoreceptors During Inherited Retinal Degeneration. , 2020, 61, 25.		6
21	A mega-analysis of expression quantitative trait loci in retinal tissue. PLoS Genetics, 2020, 16, e1008934.	3.5	22
22	Adherence to the Mediterranean Diet and Progression to Late Age-Related Macular Degeneration in the Age-Related Eye Disease Studies 1 and 2. Ophthalmology, 2020, 127, 1515-1528.	5.2	34
23	Pluripotent stem cell-derived retinal organoids for disease modeling and development of therapies. Stem Cells, 2020, 38, 1206-1215.	3.2	83
24	Tbx2aModulates Switching ofRH2andLWSOpsin Gene Expression. Molecular Biology and Evolution, 2020, 37, 2002-2014.	8.9	20
25	Retinal Organoids derived from hiPSCs of an AIPL1-LCA Patient Maintain Cytoarchitecture despite Reduced levels of Mutant AIPL1. Scientific Reports, 2020, 10, 5426.	3.3	39
26	Retinal pigment epithelium transcriptome analysis in chronic smoking reveals a suppressed innate immune response and activation of differentiation pathways. Free Radical Biology and Medicine, 2020, 156, 176-189.	2.9	4
27	Soy Protein Nanofiber Scaffolds for Uniform Maturation of Human Induced Pluripotent Stem Cell-Derived Retinal Pigment Epithelium. Tissue Engineering - Part C: Methods, 2020, 26, 433-446.	2.1	16
28	Reply. Ophthalmology, 2020, 127, e19-e20.	5.2	0
29	Deep-learning-based prediction of late age-related macular degeneration progression. Nature Machine Intelligence, 2020, 2, 141-150.	16.0	79
30	Family-based exome sequencing identifies rare coding variants in age-related macular degeneration. Human Molecular Genetics, 2020, 29, 2022-2034.	2.9	26
31	Genome-wide Profiling Identifies DNA Methylation Signatures of Aging in Rod Photoreceptors Associated with Alterations in Energy Metabolism. Cell Reports, 2020, 31, 107525.	6.4	20
32	Pharmacologic fibroblast reprogramming into photoreceptors restores vision. Nature, 2020, 581, 83-88.	27.8	66
33	Adherence to a Mediterranean diet and cognitive function in the Ageâ€Related Eye Disease Studies 1 & 2. Alzheimer's and Dementia, 2020, 16, 831-842.	0.8	28
34	Loss of endocytosis-associated RabGEF1 causes aberrant morphogenesis and altered autophagy in photoreceptors leading to retinal degeneration. PLoS Genetics, 2020, 16, e1009259.	3.5	11
35	Integration of genomics and transcriptomics predicts diabetic retinopathy susceptibility genes. ELife, 2020, 9, .	6.0	18
36	A simple and efficient method for generating human retinal organoids. Molecular Vision, 2020, 26, 97-105.	1.1	21

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37	A unique -associated variant in a Georgian Jewish family with probable North Carolina macular dystrophy and the possible contribution of a unique variant. Molecular Vision, 2020, 26, 299-310.	1.1	5
38	An optimized protocol for retina single-cell RNA sequencing. Molecular Vision, 2020, 26, 705-717.	1.1	13
39	Title is missing!. , 2020, 16, e1009259.		0
40	Title is missing!. , 2020, 16, e1009259.		0
41	Title is missing!. , 2020, 16, e1009259.		0
42	Title is missing!. , 2020, 16, e1009259.		0
43	Title is missing!. , 2020, 16, e1009259.		0
44	Title is missing!. , 2020, 16, e1009259.		0
45	Retinal disease in ciliopathies: Recent advances with a focus on stem cell-based therapies. Translational Science of Rare Diseases, 2019, 4, 97-115.	1.5	22
46	No CFH or ARMS2 Interaction with Omega-3 Fatty Acids, Low versus High Zinc, or Î²-Carotene versus Lutein and Zeaxanthin onÂProgression of Age-Related Macular Degeneration in the Age-Related Eye DiseaseÂStudy 2. Ophthalmology, 2019, 126, 1541-1548.	5.2	15
47	Improved Retinal Organoid Differentiation by Modulating Signaling Pathways Revealed by Comparative Transcriptome Analyses with Development InÂVivo. Stem Cell Reports, 2019, 13, 891-905.	4.8	77
48	Assessment of Novel Genome-Wide Significant Gene Loci and Lesion Growth in Geographic Atrophy Secondary to Age-Related Macular Degeneration. JAMA Ophthalmology, 2019, 137, 867.	2.5	28
49	The combination of wholeâ€exome sequencing and clinical analysis allows better diagnosis of rare syndromic retinal dystrophies. Acta Ophthalmologica, 2019, 97, e877-e886.	1.1	15
50	Retinal transcriptome and eQTL analyses identify genes associated with age-related macular degeneration. Nature Genetics, 2019, 51, 606-610.	21.4	201
51	Targeted deletion of an NRL- and CRX-regulated alternative promoter specifically silences FERM and PDZ domain containing 1 (Frmpd1) in rod photoreceptors. Human Molecular Genetics, 2019, 28, 804-817.	2.9	9
52	Aberrant RNA splicing is the major pathogenic effect in a knockâ€in mouse model of the dominantly inherited c.1430A>G humanRPE65mutation. Human Mutation, 2019, 40, 426-443.	2.5	22
53	Molecular dissection of cone photoreceptorâ€enriched genes encoding transmembrane and secretory proteins. Journal of Neuroscience Research, 2019, 97, 16-28.	2.9	5
54	SSBP1 faux pas in mitonuclear tango causes optic neuropathy. Journal of Clinical Investigation, 2019, 130, 62-64.	8.2	3

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55	Association of age-related macular degeneration with complement activation products, smoking, and single nucleotide polymorphisms in South Carolinians of European and African descent. Molecular Vision, 2019, 25, 79-92.	1.1	14
56	Transcriptome-based molecular staging of human stem cell-derived retinal organoids uncovers accelerated photoreceptor differentiation by 9-cis retinal. Molecular Vision, 2019, 25, 663-678.	1.1	33
57	Expression of deubiquitinating enzyme genes in the developing mammal retina. Molecular Vision, 2019, 25, 800-813.	1.1	5
58	RNA Biology in Retinal Development and Disease. Trends in Genetics, 2018, 34, 341-351.	6.7	29
59	Genome-wide analysis of disease progression in age-related macular degeneration. Human Molecular Genetics, 2018, 27, 929-940.	2.9	67
60	Accelerated and Improved Differentiation of Retinal Organoids from Pluripotent Stem Cells in Rotating-Wall Vessel Bioreactors. Stem Cell Reports, 2018, 10, 300-313.	4.8	168
61	Epigenetic control of gene regulation during development and disease: A view from the retina. Progress in Retinal and Eye Research, 2018, 65, 1-27.	15.5	105
62	Determination of Mitochondrial Oxygen Consumption in the Retina Ex Vivo: Applications for Retinal Disease. Methods in Molecular Biology, 2018, 1753, 167-177.	0.9	3
63	A Deep Phenotype Association Study Reveals Specific Phenotype Associations with Genetic Variants in Age-related Macular Degeneration. Ophthalmology, 2018, 125, 559-568.	5.2	30
64	Association of Rare Predicted Loss-of-Function Variants in Cellular Pathways with Sub-Phenotypes in Age-Related Macular Degeneration. Ophthalmology, 2018, 125, 398-406.	5.2	12
65	Mini and customized low-cost bioreactors for optimized high-throughput generation of tissue organoids. Stem Cell Investigation, 2018, 5, 33-33.	3.0	32
66	A CEP290 C-Terminal Domain Complements the Mutant CEP290 of Rd16 Mice In Trans and Rescues Retinal Degeneration. Cell Reports, 2018, 25, 611-623.e6.	6.4	32
67	Progression of Geographic Atrophy in Age-related Macular Degeneration. Ophthalmology, 2018, 125, 1913-1928.	5.2	127
68	Cone-rod homeobox CRX controls presynaptic active zone formation in photoreceptors of mammalian retina. Human Molecular Genetics, 2018, 27, 3555-3567.	2.9	17
69	Patient iPSC-derived neural stem cells exhibit phenotypes in concordance with the clinical severity of mucopolysaccharidosis I. Human Molecular Genetics, 2018, 27, 3612-3626.	2.9	23
70	Maturation arrest in early postnatal sensory receptors by deletion of the miR-183/96/182 cluster in mouse. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E4271-E4280.	7.1	50
71	Bivariate Analysis of Age-Related Macular Degeneration Progression Using Genetic Risk Scores. Genetics, 2017, 206, 119-133.	2.9	46
72	REEP6 mediates trafficking of a subset of Clathrin-coated vesicles and is critical for rod photoreceptor function and survival. Human Molecular Genetics, 2017, 26, 2218-2230.	2.9	23

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73	Analysis of RP2 and RPGR Mutations in Five X-Linked Chinese Families with Retinitis Pigmentosa. Scientific Reports, 2017, 7, 44465.	3.3	12
74	Nrl knockdown by AAV-delivered CRISPR/Cas9 prevents retinal degeneration in mice. Nature Communications, 2017, 8, 14716.	12.8	231
75	Pias3 is necessary for dorso-ventral patterning and visual response of retinal cones but is not required for rod photoreceptor differentiation. Biology Open, 2017, 6, 881-890.	1.2	4
76	Molecular Anatomy of the Developing Human Retina. Developmental Cell, 2017, 43, 763-779.e4.	7.0	205
77	InÂVitro Modeling Using Ciliopathy-Patient-Derived Cells Reveals Distinct Cilia Dysfunctions Caused by CEP290 Mutations. Cell Reports, 2017, 20, 384-396.	6.4	120
78	Transcriptome profiling of NIH3T3 cell lines expressing opsin and the P23H opsin mutant identifies candidate drugs for the treatment of retinitis pigmentosa. Pharmacological Research, 2017, 115, 1-13.	7.1	7
79	EYS Mutations Causing Autosomal Recessive Retinitis Pigmentosa: Changes of Retinal Structure and Function with Disease Progression. Genes, 2017, 8, 178.	2.4	35
80	Regulation of Noncoding Transcriptome in Developing Photoreceptors by Rod Differentiation Factor NRL. , 2017, 58, 4422.		19
81	Photoreceptor Development: Early Steps/Fate â˜†. , 2017, , .		0
82	Treatment Paradigms for Retinal and Macular Diseases Using 3-D Retina Cultures Derived From Human Reporter Pluripotent Stem Cell Lines. , 2016, 57, ORSFl1.		35
83	Rapid, Dynamic Activation of MÃ¼ller Glial Stem Cell Responses in Zebrafish. , 2016, 57, 5148.		74
84	Genetic Analysis of the Rhodopsin Gene Identifies a Mosaic Dominant Retinitis Pigmentosa Mutation in a Healthy Individual. , 2016, 57, 940.		9
85	Molecular Diagnosis of Inherited Retinal Diseases in Indigenous African Populations by Whole-Exome Sequencing. , 2016, 57, 6374.		17
86	Gene-Based Association Analysis for Censored Traits Via Fixed Effect Functional Regressions. Genetic Epidemiology, 2016, 40, 133-143.	1.3	12
87	Loss of RPGR glutamylation underlies the pathogenic mechanism of retinal dystrophy caused by TTLL5 mutations. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E2925-34.	7.1	79
88	Bi-allelic Truncating Mutations in CEP78 , Encoding Centrosomal Protein 78, Cause Cone-Rod Degeneration with Sensorineural Hearing Loss. American Journal of Human Genetics, 2016, 99, 777-784.	6.2	34
89	Next-generation genotype imputation service and methods. Nature Genetics, 2016, 48, 1284-1287.	21.4	2,828
90	A reference panel of 64,976 haplotypes for genotype imputation. Nature Genetics, 2016, 48, 1279-1283.	21.4	2,421

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91	NRL-Regulated Transcriptome Dynamics of Developing Rod Photoreceptors. Cell Reports, 2016, 17, 2460-2473.	6.4	104
92	Synergistically acting agonists and antagonists of G proteinâ€“coupled receptors prevent photoreceptor cell degeneration. Science Signaling, 2016, 9, ra74.	3.6	33
93	Variegated yet non-random rod and cone photoreceptor disease patterns inRPGR-ORF15-associated retinal degeneration. Human Molecular Genetics, 2016, 25, 5444-5459.	2.9	35
94	Recruitment of Rod Photoreceptors from Short-Wavelength-Sensitive Cones during the Evolution of Nocturnal Vision in Mammals. Developmental Cell, 2016, 37, 520-532.	7.0	103
95	Next generation sequencing technology and genomewide data analysis: Perspectives for retinal research. Progress in Retinal and Eye Research, 2016, 55, 1-31.	15.5	58
96	Increased retinal mtDNA damage in the CFH variant associated with age-related macular degeneration. Experimental Eye Research, 2016, 145, 269-277.	2.6	64
97	A secreted WNT-ligand-binding domain of FZD5 generated by a frameshift mutation causes autosomal dominant coloboma. Human Molecular Genetics, 2016, 25, 1382-1391.	2.9	40
98	Centrosomal protein CP110 controls maturation of mother centriole during cilia biogenesis. Development (Cambridge), 2016, 143, 1491-501.	2.5	78
99	A large genome-wide association study of age-related macular degeneration highlights contributions of rare and common variants. Nature Genetics, 2016, 48, 134-143.	21.4	1,167
100	Genetic components in diabetic retinopathy. Indian Journal of Ophthalmology, 2016, 64, 55.	1.1	19
101	The cellular and compartmental profile of mouse retinal glycolysis, tricarboxylic acid cycle, oxidative phosphorylation, and ~P transferring kinases. Molecular Vision, 2016, 22, 847-85.	1.1	51
102	Three-dimensional retinal organoids from mouse pluripotent stem cells mimic development with enhanced stratification and rod photoreceptor differentiation. Molecular Vision, 2016, 22, 1077-1094.	1.1	60
103	Transcriptome Dynamics of Developing Photoreceptors in Three-Dimensional Retina Cultures Recapitulates Temporal Sequence of Human Cone and Rod Differentiation Revealing Cell Surface Markers and Gene Networks. Stem Cells, 2015, 33, 3504-3518.	3.2	153
104	Whole Exome Sequencing Reveals Mutations in Known Retinal Disease Genes in 33 out of 68 Israeli Families with Inherited Retinopathies. Scientific Reports, 2015, 5, 13187.	3.3	66
105	Nonsyndromic Early-Onset Cone-Rod Dystrophy and Limb-Girdle Muscular Dystrophy in a Consanguineous Israeli Family are Caused by Two Independent yet Linked Mutations inALMS1andDYSF. Human Mutation, 2015, 36, 836-841.	2.5	17
106	Whole Exome Sequencing Reveals GUCY2D as a Major Gene Associated With Cone and Cone-Rod Dystrophy in Israel. Investigative Ophthalmology and Visual Science, 2015, 56, 420-430.	3.3	32
107	Quantification of Oxygen Consumption in Retina Ex Vivo Demonstrates Limited Reserve Capacity of Photoreceptor Mitochondria. , 2015, 56, 8428.		104
108	Vision from next generation sequencing: Multi-dimensional genome-wide analysis for producing gene regulatory networks underlying retinal development, aging and disease. Progress in Retinal and Eye Research, 2015, 46, 1-30.	15.5	50

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109	Differential DNA methylation identified in the blood and retina of AMD patients. Epigenetics, 2015, 10, 698-707.	2.7	62
110	A long-term efficacy study of gene replacement therapy for RPGR-associated retinal degeneration. Human Molecular Genetics, 2015, 24, 3956-3970.	2.9	63
111	CEP290 alleles in mice disrupt tissue-specific cilia biogenesis and recapitulate features of syndromic ciliopathies. Human Molecular Genetics, 2015, 24, 3775-3791.	2.9	105
112	Long-term rescue of cone photoreceptor degeneration in retinitis pigmentosa 2 (RP2)-knockout mice by gene replacement therapy. Human Molecular Genetics, 2015, 24, 6446-6458.	2.9	38
113	Mouse model of humanRPE65P25L hypomorph resembles wild type under normal light rearing but is fully resistant to acute light damage. Human Molecular Genetics, 2015, 24, 4417-4428.	2.9	19
114	Biology and therapy of inherited retinal degenerative disease: insights from mouse models. DMM Disease Models and Mechanisms, 2015, 8, 109-129.	2.4	207
115	Clinical and Genetic Factors Associated with Progression of Geographic Atrophy Lesions in Age-Related Macular Degeneration. PLoS ONE, 2015, 10, e0126636.	2.5	61
116	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
117	Feedback Induction of a Photoreceptor-specific Isoform of Retinoid-related Orphan Nuclear Receptor Î² by the Rod Transcription Factor NRL. Journal of Biological Chemistry, 2014, 289, 32469-32480.	3.4	22
118	The Transcription Factor GTF2IRD1 Regulates the Topology and Function of Photoreceptors by Modulating Photoreceptor Gene Expression across the Retina. Journal of Neuroscience, 2014, 34, 15356-15368.	3.6	10
119	Regulation of a novel isoform of Receptor Expression Enhancing Protein REEP6 in rod photoreceptors by bZIP transcription factor NRL. Human Molecular Genetics, 2014, 23, 4260-4271.	2.9	27
120	The transcription-splicing protein NonO/p54nrb and three NonO-interacting proteins bind to distal enhancer region and augment rhodopsin expression. Human Molecular Genetics, 2014, 23, 2132-2144.	2.9	45
121	Genome-wide association study and meta-analysis of intraocular pressure. Human Genetics, 2014, 133, 41-57.	3.8	93
122	Ancestry estimation and control of population stratification for sequence-based association studies. Nature Genetics, 2014, 46, 409-415.	21.4	136
123	Free radical scavenging, antioxidant and cancer chemoprevention by grape seed proanthocyanidin: An overview. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2014, 768, 69-73.	1.0	148
124	Deletion of Aryl Hydrocarbon Receptor AHR in Mice Leads to Subretinal Accumulation of Microglia and RPE Atrophy. , 2014, 55, 6031.		67
125	No Clinically Significant Association between CFH and ARMS2 Genotypes and Response to Nutritional Supplements. Ophthalmology, 2014, 121, 2173-2180.	5.2	86
126	Null and hypomorphPrickle1alleles in mice phenocopy human Robinow syndrome and disrupt signaling downstream of Wnt5a. Biology Open, 2014, 3, 861-870.	1.2	47

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127	Rare and common variants in extracellular matrix gene Fibrillin 2 (FBN2) are associated with macular degeneration. Human Molecular Genetics, 2014, 23, 5827-5837.	2.9	52
128	Ciliopathy-associated gene Cc2d2a promotes assembly of subdistal appendages on the mother centriole during cilia biogenesis. Nature Communications, 2014, 5, 4207.	12.8	66
129	Age-Related Macular Degeneration: Genetics and Biology Coming Together. Annual Review of Genomics and Human Genetics, 2014, 15, 151-171.	6.2	394
130	OTX2 loss causes rod differentiation defect in CRX-associated congenital blindness. Journal of Clinical Investigation, 2014, 124, 631-643.	8.2	59
131	Photoreceptor Degeneration: Molecular Mechanisms of Photoreceptor Degeneration. , 2014, , 275-308.		0
132	Gene expression changes in aging retinal microglia: relationship to microglial support functions and regulation of activation. Neurobiology of Aging, 2013, 34, 2310-2321.	3.1	100
133	An isoform of retinoid-related orphan receptor Î² directs differentiation of retinal amacrine and horizontal interneurons. Nature Communications, 2013, 4, 1813.	12.8	52
134	Hypomethylation of the IL17RC Promoter in Peripheral Blood Leukocytes Is Not A Hallmark of Age-Related Macular Degeneration. Cell Reports, 2013, 5, 1527-1535.	6.4	42
135	Prickle1 is expressed in distinct cell populations of the central nervous system and contributes to neuronal morphogenesis. Human Molecular Genetics, 2013, 22, 2234-2246.	2.9	45
136	Genetic architecture of retinal and macular degenerative diseases: the promise and challenges of next-generation sequencing. Genome Medicine, 2013, 5, 84.	8.2	33
137	Conditional knockdown of DNA methyltransferase 1 reveals a key role of retinal pigment epithelium integrity in photoreceptor outer segment morphogenesis. Development (Cambridge), 2013, 140, 1330-1341.	2.5	77
138	Seven new loci associated with age-related macular degeneration. Nature Genetics, 2013, 45, 433-439.	21.4	687
139	Developing Rods Transplanted into the Degenerating Retina of Crx-Knockout Mice Exhibit Neural Activity Similar to Native Photoreceptors. Stem Cells, 2013, 31, 1149-1159.	3.2	98
140	Identification of a rare coding variant in complement 3 associated with age-related macular degeneration. Nature Genetics, 2013, 45, 1375-1379.	21.4	158
141	Ablation of the X-Linked Retinitis Pigmentosa 2 (Rp2) Gene in Mice Results in Opsin Mislocalization and Photoreceptor Degeneration. , 2013, 54, 4503.		54
142	Development and Plasticity of Outer Retinal Circuitry Following Genetic Removal of Horizontal Cells. Journal of Neuroscience, 2013, 33, 17847-17862.	3.6	41
143	The golden era of ocular disease gene discovery: Race to the finish. Clinical Genetics, 2013, 84, 99-101.	2.0	13
144	Phenotypic Conservation in Patients With X-Linked Retinitis Pigmentosa Caused byRPGRMutations. JAMA Ophthalmology, 2013, 131, 1016.	2.5	31

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145	What's in a Name?RPGRMutations Redefine the Genetic and Phenotypic Landscape in Retinal Degenerative Diseases. , 2013, 54, 1417.		3
146	Transcriptional Regulation of Rod Photoreceptor Homeostasis Revealed by In Vivo NRL Targetome Analysis. PLoS Genetics, 2012, 8, e1002649.	3.5	99
147	Minireview: The Role of Nuclear Receptors in Photoreceptor Differentiation and Disease. Molecular Endocrinology, 2012, 26, 905-915.	3.7	53
148	Gene therapy rescues photoreceptor blindness in dogs and paves the way for treating human X-linked retinitis pigmentosa. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 2132-2137.	7.1	237
149	Complement factor H genetic variant and age-related macular degeneration: effect size, modifiers and relationship to disease subtype. International Journal of Epidemiology, 2012, 41, 250-262.	1.9	79
150	Regulation of retinal progenitor expansion by Frizzled receptors: implications for microphthalmia and retinal coloboma. Human Molecular Genetics, 2012, 21, 1848-1860.	2.9	40
151	Retinal Transcriptome Profiling by Directional Next-Generation Sequencing Using 100 ng of Total RNA. Methods in Molecular Biology, 2012, 884, 319-334.	0.9	24
152	Global expression profiling of peripheral Qa-1â€“restricted CD8Î±Î±+TCRÎ±Î²+ regulatory T cells reveals innate-like features: Implications for immune-regulatory repertoire. Human Immunology, 2012, 73, 214-222.	2.4	15
153	Determination of Posttranslational Modifications of Photoreceptor Differentiation Factor NRL: Focus on SUMOylation. Methods in Molecular Biology, 2012, 884, 353-361.	0.9	2
154	Genetic Studies of Age-related Macular Degeneration. Ophthalmology, 2012, 119, 2526-2536.	5.2	73
155	Preservation of Cone Photoreceptors after a Rapid yet Transient Degeneration and Remodeling in Cone-OnlyNrl^{â^'/â^'}Mouse Retina. Journal of Neuroscience, 2012, 32, 528-541.	3.6	51
156	Photoreceptor sensory cilia and ciliopathies: focus on CEP290, RPGR and their interacting proteins. Cilia, 2012, 1, 22.	1.8	131
157	A role for prenylated rab acceptor 1 in vertebrate photoreceptor development. BMC Neuroscience, 2012, 13, 152.	1.9	19
158	Knockdown of Bardet-Biedl Syndrome Gene BBS9/PTHB1 Leads to Cilia Defects. PLoS ONE, 2012, 7, e34389.	2.5	49
159	Rd9 Is a Naturally Occurring Mouse Model of a Common Form of Retinitis Pigmentosa Caused by Mutations in RPGR-ORF15. PLoS ONE, 2012, 7, e35865.	2.5	69
160	Mutations inRPGRandRP2Account for 15% of Males with Simplex Retinal Degenerative Disease. , 2012, 53, 8232.		108
161	RPGR-Associated Retinal Degeneration in Human X-Linked RP and a Murine Model. , 2012, 53, 5594.		56
162	Exome Sequencing: Capture and Sequencing of All Human Coding Regions for Disease Gene Discovery. Methods in Molecular Biology, 2012, 884, 335-351.	0.9	7

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163	Combining Cep290 and Mkks ciliopathy alleles in mice rescues sensory defects and restores ciliogenesis. Journal of Clinical Investigation, 2012, 122, 1233-1245.	8.2	75
164	Protective Gene Expression Changes Elicited by an Inherited Defect in Photoreceptor Structure. PLoS ONE, 2012, 7, e31371.	2.5	14
165	Combining Cep290 and Mkks ciliopathy alleles in mice rescues sensory defects and restores ciliogenesis. Journal of Clinical Investigation, 2012, 122, 3025-3025.	8.2	0
166	Long-term survival and differentiation of retinal neurons derived from human embryonic stem cell lines in un-immunosuppressed mouse retina. Molecular Vision, 2012, 18, 920-36.	1.1	104
167	Transcriptome analysis using next generation sequencing reveals molecular signatures of diabetic retinopathy and efficacy of candidate drugs. Molecular Vision, 2012, 18, 1123-46.	1.1	38
168	Genetic association study of age-related macular degeneration in the Spanish population. Acta Ophthalmologica, 2011, 89, e12-e22.	1.1	33
169	XIAP Therapy Increases Survival of Transplanted Rod Precursors in a Degenerating Host Retina. , 2011, 52, 1567.		47
170	Ciliary Neurotrophic Factor Induces Genes Associated with Inflammation and Gliosis in the Retina: A Gene Profiling Study of Flow-Sorted, MÃ¼ller Cells. PLoS ONE, 2011, 6, e20326.	2.5	48
171	A 32 kb Critical Region Excluding Y402H in CFH Mediates Risk for Age-Related Macular Degeneration. PLoS ONE, 2011, 6, e25598.	2.5	46
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