## **Anand Swaroop**

## List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/3341370/publications.pdf

Version: 2024-02-01

6512 5782 34,100 346 84 162 citations g-index h-index papers 363 363 363 36381 docs citations times ranked citing authors all docs

#	Article	IF	CITATIONS
1	Restoration of RPGR expression in vivo using CRISPR/Cas9 gene editing. Gene Therapy, 2022, 29, 81-93.	2.3	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.	2.2	3
3	Multiomics analyses reveal early metabolic imbalance and mitochondrial stress in neonatal photoreceptors leading to cell death in <i>Pde6brd1/rd1</i> mouse model of retinal degeneration. Human Molecular Genetics, 2022, 31, 2137-2154.	1.4	25
4	GATD3A, a mitochondrial deglycase with evolutionary origins from gammaproteobacteria, restricts the formation of advanced glycation end products. BMC Biology, 2022, 20, 68.	1.7	6
5	HiCRes: a computational method to estimate and predict the genomic resolution of Hi-C libraries. Nucleic Acids Research, 2022, 50, e35-e35.	6.5	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.	1.4	4
7	Primary cilia biogenesis and associated retinal ciliopathies. Seminars in Cell and Developmental Biology, 2021, 110, 70-88.	2.3	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.	2.5	66
9	Gene Therapy of Dominant CRX-Leber Congenital Amaurosis using Patient Stem Cell-Derived Retinal Organoids. Stem Cell Reports, 2021, 16, 252-263.	2.3	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.	1.1	14
11	Genetics and therapy for pediatric eye diseases. EBioMedicine, 2021, 67, 103360.	2.7	7
12	A Novel ARL3 Gene Mutation Associated With Autosomal Dominant Retinal Degeneration. Frontiers in Cell and Developmental Biology, 2021, 9, 720782.	1.8	13
13	Glycogen Synthase Kinase 3 Regulates the Genesis of Displaced Retinal Ganglion Cells3. ENeuro, 2021, 8, ENEURO.0171-21.2021.	0.9	5
14	Aging of the Retina: Molecular and Metabolic Turbulences and Potential Interventions. Annual Review of Vision Science, 2021, 7, 633-664.	2.3	28
15	Proneural genes define ground-state rules to regulate neurogenic patterning and cortical folding. Neuron, 2021, 109, 2847-2863.e11.	3.8	26
16	Making Biological Sense of Genetic Studies of Age-Related Macular Degeneration. Advances in Experimental Medicine and Biology, 2021, 1256, 201-219.	0.8	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.	1.4	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.	0.5	4

#	Article	IF	Citations
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.	0.5	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.	1.5	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.	2.5	34
23	Pluripotent stem cell-derived retinal organoids for disease modeling and development of therapies. Stem Cells, 2020, 38, 1206-1215.	1.4	83
24	<i>Tbx2a</i> Modulates Switching of <i>RH2</i> and <i>LWS</i> Opsin Gene Expression. Molecular Biology and Evolution, 2020, 37, 2002-2014.	3.5	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.	1.6	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.	1.3	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.	1.1	16
28	Reply. Ophthalmology, 2020, 127, e19-e20.	2.5	0
29	Deep-learning-based prediction of late age-related macular degeneration progression. Nature Machine Intelligence, 2020, 2, 141-150.	8.3	79
30	Family-based exome sequencing identifies rare coding variants in age-related macular degeneration. Human Molecular Genetics, 2020, 29, 2022-2034.	1.4	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.	2.9	20
32	Pharmacologic fibroblast reprogramming into photoreceptors restores vision. Nature, 2020, 581, 83-88.	13.7	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.	0.4	28
34	Loss of endocytosis-associated RabGEF1 causes aberrant morphogenesis and altered autophagy in photoreceptors leading to retinal degeneration. PLoS Genetics, 2020, 16, e1009259.	1.5	11
35	Integration of genomics and transcriptomics predicts diabetic retinopathy susceptibility genes. ELife, 2020, 9, .	2.8	18
36	A simple and efficient method for generating human retinal organoids. Molecular Vision, 2020, 26, 97-105.	1.1	21

#	Article	IF	Citations
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.		O
40	Title is missing!. , 2020, 16, e1009259.		0
41	Title is missing!. , 2020, 16, e1009259.		O
42	Title is missing!. , 2020, 16, e1009259.		0
43	Title is missing!. , 2020, 16, e1009259.		O
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.	1.6	22
46	No CFH or ARMS2 Interaction with Omega-3 Fatty Acids, Low versus High Zinc, or Î <sup>2</sup> -Carotene versus Lutein and Zeaxanthin onÂProgression of Age-Related Macular Degeneration in the Age-Related Eye DiseaseÂStudy 2. Ophthalmology, 2019, 126, 1541-1548.	2.5	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.	2.3	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.	1.4	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.	0.6	15
50	Retinal transcriptome and eQTL analyses identify genes associated with age-related macular degeneration. Nature Genetics, 2019, 51, 606-610.	9.4	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.	1.4	9
52	Aberrant RNA splicing is the major pathogenic effect in a knockâ€in mouse model of the dominantly inherited c.1430A>G human <i>RPE65</i> mutation. Human Mutation, 2019, 40, 426-443.	1.1	22
53	Molecular dissection of cone photoreceptorâ€enriched genes encoding transmembrane and secretory proteins. Journal of Neuroscience Research, 2019, 97, 16-28.	1.3	5
54	SSBP1 faux pas in mitonuclear tango causes optic neuropathy. Journal of Clinical Investigation, 2019, 130, 62-64.	3.9	3

#	Article	IF	CITATIONS
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.	2.9	29
59	Genome-wide analysis of disease progression in age-related macular degeneration. Human Molecular Genetics, 2018, 27, 929-940.	1.4	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.	2.3	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.	7.3	105
62	Determination of Mitochondrial Oxygen Consumption in the Retina Ex Vivo: Applications for Retinal Disease. Methods in Molecular Biology, 2018, 1753, 167-177.	0.4	3
63	A Deep Phenotype Association Study Reveals Specific Phenotype Associations with Genetic Variants in Age-related Macular Degeneration. Ophthalmology, 2018, 125, 559-568.	2.5	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.	2.5	12
65	Mini and customized low-cost bioreactors for optimized high-throughput generation of tissue organoids. Stem Cell Investigation, 2018, 5, 33-33.	1.3	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.	2.9	32
67	Progression of Geographic Atrophy in Age-related Macular Degeneration. Ophthalmology, 2018, 125, 1913-1928.	2.5	127
68	Cone-rod homeobox CRX controls presynaptic active zone formation in photoreceptors of mammalian retina. Human Molecular Genetics, 2018, 27, 3555-3567.	1.4	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.	1.4	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.	3.3	50
71	Bivariate Analysis of Age-Related Macular Degeneration Progression Using Genetic Risk Scores. Genetics, 2017, 206, 119-133.	1.2	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.	1.4	23

#	Article	IF	CITATIONS
73	Analysis of RP2 and RPGR Mutations in Five X-Linked Chinese Families with Retinitis Pigmentosa. Scientific Reports, 2017, 7, 44465.	1.6	12
74	Nrl knockdown by AAV-delivered CRISPR/Cas9 prevents retinal degeneration in mice. Nature Communications, 2017, 8, 14716.	5.8	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.	0.6	4
76	Molecular Anatomy of the Developing Human Retina. Developmental Cell, 2017, 43, 763-779.e4.	3.1	205
77	InÂVitro Modeling Using Ciliopathy-Patient-Derived Cells Reveals Distinct Cilia Dysfunctions Caused by CEP290 Mutations. Cell Reports, 2017, 20, 384-396.	2.9	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.	3.1	7
79	EYS Mutations Causing Autosomal Recessive Retinitis Pigmentosa: Changes of Retinal Structure and Function with Disease Progression. Genes, 2017, 8, 178.	1.0	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, ORSFI1.		35
83	Rapid, Dynamic Activation of Müller 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.	0.6	12
87	Loss of RPGR glutamylation underlies the pathogenic mechanism of retinal dystrophy caused by <i>TTLL5</i> mutations. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E2925-34.	3.3	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.	2.6	34
89	Next-generation genotype imputation service and methods. Nature Genetics, 2016, 48, 1284-1287.	9.4	2,828
90	A reference panel of 64,976 haplotypes for genotype imputation. Nature Genetics, 2016, 48, 1279-1283.	9.4	2,421

#	Article	IF	Citations
91	NRL-Regulated Transcriptome Dynamics of Developing Rod Photoreceptors. Cell Reports, 2016, 17, 2460-2473.	2.9	104
92	Synergistically acting agonists and antagonists of G protein–coupled receptors prevent photoreceptor cell degeneration. Science Signaling, 2016, 9, ra74.	1.6	33
93	Variegated yet non-random rod and cone photoreceptor disease patterns in <i>RPGR-ORF15</i> -associated retinal degeneration. Human Molecular Genetics, 2016, 25, 5444-5459.	1.4	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.	3.1	103
95	Next generation sequencing technology and genomewide data analysis: Perspectives for retinal research. Progress in Retinal and Eye Research, 2016, 55, 1-31.	7.3	58
96	Increased retinal mtDNA damage in the CFH variant associated with age-related macular degeneration. Experimental Eye Research, 2016, 145, 269-277.	1.2	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.	1.4	40
98	Centrosomal protein CP110 controls maturation of mother centriole during cilia biogenesis. Development (Cambridge), 2016, 143, 1491-501.	1.2	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.	9.4	1,167
100	Genetic components in diabetic retinopathy. Indian Journal of Ophthalmology, 2016, 64, 55.	0.5	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.	1.4	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.	1.6	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 in <i>ALMS1</i> and <i>DYSF</i> . Human Mutation, 2015, 36, 836-841.	1.1	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.	7.3	50

#	Article	IF	CITATIONS
109	Differential DNA methylation identified in the blood and retina of AMD patients. Epigenetics, 2015, 10, 698-707.	1.3	62
110	A long-term efficacy study of gene replacement therapy for RPGR-associated retinal degeneration. Human Molecular Genetics, 2015, 24, 3956-3970.	1.4	63
111	CEP290 alleles in mice disrupt tissue-specific cilia biogenesis and recapitulate features of syndromic ciliopathies. Human Molecular Genetics, 2015, 24, 3775-3791.	1.4	105
112	Long-term rescue of cone photoreceptor degeneration in retinitis pigmentosa 2 ( <i>RP2</i> )-knockout mice by gene replacement therapy. Human Molecular Genetics, 2015, 24, 6446-6458.	1.4	38
113	Mouse model of human <i>RPE65</i> P25L hypomorph resembles wild type under normal light rearing but is fully resistant to acute light damage. Human Molecular Genetics, 2015, 24, 4417-4428.	1.4	19
114	Biology and therapy of inherited retinal degenerative disease: insights from mouse models. DMM Disease Models and Mechanisms, 2015, 8, 109-129.	1.2	207
115	Clinical and Genetic Factors Associated with Progression of Geographic Atrophy Lesions in Age-Related Macular Degeneration. PLoS ONE, 2015, 10, e0126636.	1.1	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.	1.1	23
117	Feedback Induction of a Photoreceptor-specific Isoform of Retinoid-related Orphan Nuclear Receptor β by the Rod Transcription Factor NRL. Journal of Biological Chemistry, 2014, 289, 32469-32480.	1.6	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.	1.7	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.	1.4	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.	1.4	45
121	Genome-wide association study and meta-analysis of intraocular pressure. Human Genetics, 2014, 133, 41-57.	1.8	93
122	Ancestry estimation and control of population stratification for sequence-based association studies. Nature Genetics, 2014, 46, 409-415.	9.4	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.	0.4	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.	2.5	86
126	Null and hypomorph <i>Prickle1</i> alleles in mice phenocopy human Robinow syndrome and disrupt signaling downstream of Wnt5a. Biology Open, 2014, 3, 861-870.	0.6	47

#	Article	IF	Citations
127	Rare and common variants in extracellular matrix gene Fibrillin 2 (FBN2) are associated with macular degeneration. Human Molecular Genetics, 2014, 23, 5827-5837.	1.4	52
128	Ciliopathy-associated gene Cc2d2a promotes assembly of subdistal appendages on the mother centriole during cilia biogenesis. Nature Communications, 2014, 5, 4207.	5.8	66
129	Age-Related Macular Degeneration: Genetics and Biology Coming Together. Annual Review of Genomics and Human Genetics, 2014, 15, 151-171.	2.5	394
130	OTX2 loss causes rod differentiation defect in CRX-associated congenital blindness. Journal of Clinical Investigation, 2014, 124, 631-643.	3.9	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.	1.5	100
133	An isoform of retinoid-related orphan receptor $\hat{l}^2$ directs differentiation of retinal amacrine and horizontal interneurons. Nature Communications, 2013, 4, 1813.	5.8	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.	2.9	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.	1.4	45
136	Genetic architecture of retinal and macular degenerative diseases: the promise and challenges of next-generation sequencing. Genome Medicine, 2013, 5, 84.	3.6	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.	1.2	77
138	Seven new loci associated with age-related macular degeneration. Nature Genetics, 2013, 45, 433-439.	9.4	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.	1.4	98
140	Identification of a rare coding variant in complement 3 associated with age-related macular degeneration. Nature Genetics, 2013, 45, 1375-1379.	9.4	158
141	Ablation of the X-Linked Retinitis Pigmentosa 2 ( $\langle i\rangle$ Rp2 $\langle i\rangle$ ) 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.	1.7	41
143	The golden era of ocular disease gene discovery: Race to the finish. Clinical Genetics, 2013, 84, 99-101.	1.0	13
144	Phenotypic Conservation in Patients With X-Linked Retinitis Pigmentosa Caused by <i>RPGR </i> Mutations. JAMA Ophthalmology, 2013, 131, 1016.	1.4	31

#	Article	IF	CITATIONS
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.	1.5	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.	3.3	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.	0.9	79
150	Regulation of retinal progenitor expansion by Frizzled receptors: implications for microphthalmia and retinal coloboma. Human Molecular Genetics, 2012, 21, 1848-1860.	1.4	40
151	Retinal Transcriptome Profiling by Directional Next-Generation Sequencing Using 100 ng of Total RNA. Methods in Molecular Biology, 2012, 884, 319-334.	0.4	24
152	Global expression profiling of peripheral Qa-1–restricted CD8αα+TCRαβ+ regulatory T cells reveals innate-like features: Implications for immune-regulatory repertoire. Human Immunology, 2012, 73, 214-222.	1.2	15
153	Determination of Posttranslational Modifications of Photoreceptor Differentiation Factor NRL: Focus on SUMOylation. Methods in Molecular Biology, 2012, 884, 353-361.	0.4	2
154	Genetic Studies of Age-related Macular Degeneration. Ophthalmology, 2012, 119, 2526-2536.	2.5	73
155	Preservation of Cone Photoreceptors after a Rapid yet Transient Degeneration and Remodeling in Cone-Only <i>Nrl</i> <sup>â^'/â^'</sup> Mouse Retina. Journal of Neuroscience, 2012, 32, 528-541.	1.7	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.	0.8	19
158	Knockdown of Bardet-Biedl Syndrome Gene BBS9/PTHB1 Leads to Cilia Defects. PLoS ONE, 2012, 7, e34389.	1.1	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.	1.1	69
160	Mutations in <i>RPGR</i> and <i>RP2</i> Account for 15% of Males with Simplex Retinal Degenerative Disease., 2012, 53, 8232.		108
161	<i>RPGR-</i> 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.	0.4	7

#	Article	IF	CITATIONS
163	Combining Cep290 and Mkks ciliopathy alleles in mice rescues sensory defects and restores ciliogenesis. Journal of Clinical Investigation, 2012, 122, 1233-1245.	3.9	75
164	Protective Gene Expression Changes Elicited by an Inherited Defect in Photoreceptor Structure. PLoS ONE, 2012, 7, e31371.	1.1	14
165	Combining Cep290 and Mkks ciliopathy alleles in mice rescues sensory defects and restores ciliogenesis. Journal of Clinical Investigation, 2012, 122, 3025-3025.	3.9	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.	0.6	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, Mýller Cells. PLoS ONE, 2011, 6, e20326.	1.1	48
171	A 32 kb Critical Region Excluding Y402H in CFH Mediates Risk for Age-Related Macular Degeneration. PLoS ONE, 2011, 6, e25598.	1.1	46
172	Distinct nuclear localization patterns of DNA methyltransferases in developing and mature mammalian retina. Journal of Comparative Neurology, 2011, 519, 1914-1930.	0.9	47
173	Evidence of association of <i> APOE &lt; /i &gt; with age-related macular degeneration - a pooled analysis of 15 studies. Human Mutation, 2011, 32, 1407-1416.</i>	1.1	130
174	Combinatorial Regulation of Photoreceptor Differentiation Factor, Neural Retina Leucine Zipper Gene Nrl, Revealed by in Vivo Promoter Analysis. Journal of Biological Chemistry, 2011, 286, 28247-28255.	1.6	33
175	Variations in Apolipoprotein E Frequency With Age in a Pooled Analysis of a Large Group of Older People. American Journal of Epidemiology, 2011, 173, 1357-1364.	1.6	85
176	Excess cones in the retinal degeneration rd7 mouse, caused by the loss of function of orphan nuclear receptor Nr2e3, originate from early-born photoreceptor precursors. Human Molecular Genetics, 2011, 20, 4102-4115.	1.4	45
177	Candidate Gene Association Study for Diabetic Retinopathy in Persons with Type 2 Diabetes: The Candidate Gene Association Resource (CARe)., 2011, 52, 7593.		82
178	Complement Factor D in Age-Related Macular Degeneration. , 2011, 52, 8828.		92
179	Two Transcription Factors Can Direct Three Photoreceptor Outcomes from Rod Precursor Cells in Mouse Retinal Development. Journal of Neuroscience, 2011, 31, 11118-11125.	1.7	74
180	TOPORS, implicated in retinal degeneration, is a cilia-centrosomal protein. Human Molecular Genetics, 2011, 20, 975-987.	1.4	49

#	Article	IF	Citations
181	The Transcription Factor Neural Retina Leucine Zipper (NRL) Controls Photoreceptor-specific Expression of Myocyte Enhancer Factor Mef2c from an Alternative Promoter. Journal of Biological Chemistry, 2011, 286, 34893-34902.	1.6	35
182	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
183	Next-generation sequencing facilitates quantitative analysis of wild-type and NrI(-/-) retinal transcriptomes. Molecular Vision, 2011, 17, 3034-54.	1.1	89
184	Two novel CRX mutant proteins causing autosomal dominant Leber congenital amaurosis interact differently with NRL. Human Mutation, 2010, 31, E1472-83.	1.1	50
185	Transcriptional regulation of photoreceptor development and homeostasis in the mammalian retina. Nature Reviews Neuroscience, $2010, 11, 563-576$ .	4.9	465
186	Long-term follow-up of a family with dominant X-linked retinitis pigmentosa. Eye, 2010, 24, 764-774.	1.1	27
187	Associations of CFHR1–CFHR3 deletion and a CFH SNP to age-related macular degeneration are not independent. Nature Genetics, 2010, 42, 553-555.	9.4	55
188	MicroRNA Profile of the Developing Mouse Retina. , 2010, 51, 1823.		98
189	Autosomal Recessive Retinitis Pigmentosa with Early Macular Affectation Caused by Premature Truncation in <i> PROM1   2010, 51, 2656.</i>		59
190	Human retinopathy-associated ciliary protein retinitis pigmentosa GTPase regulator mediates cilia-dependent vertebrate development. Human Molecular Genetics, 2010, 19, 90-98.	1.4	76
191	Phenotype Associated With Mutation in the Recently Identified Autosomal Dominant Retinitis Pigmentosa KLHL7 Gene. JAMA Ophthalmology, 2010, 128, 772.	2.6	19
192	Interaction of retinitis pigmentosa GTPase regulator (RPGR) with RAB8A GTPase: implications for cilia dysfunction and photoreceptor degeneration. Human Molecular Genetics, 2010, 19, 3591-3598.	1.4	91
193	The retinitis pigmentosa protein RP2 interacts with polycystin 2 and regulates cilia-mediated vertebrate development. Human Molecular Genetics, 2010, 19, 4330-4344.	1.4	63
194	RP2 Phenotype and Pathogenetic Correlations in X-Linked Retinitis Pigmentosa. JAMA Ophthalmology, 2010, 128, 915.	2.6	62
195	Sumoylation of bZIP Transcription Factor NRL Modulates Target Gene Expression during Photoreceptor Differentiation. Journal of Biological Chemistry, 2010, 285, 25637-25644.	1.6	39
196	Loss of lysophosphatidylcholine acyltransferase 1 leads to photoreceptor degeneration in <i>rd11</i> mice. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 15523-15528.	3.3	55
197	E2-2 Protein and Fuchs's Corneal Dystrophy. New England Journal of Medicine, 2010, 363, 1016-1024.	13.9	247
198	Transcriptome analysis and molecular signature of human retinal pigment epithelium. Human Molecular Genetics, 2010, 19, 2468-2486.	1.4	249

#	Article	IF	CITATIONS
199	MicroRNAâ€⊋04/211 alters epithelial physiology. FASEB Journal, 2010, 24, 1552-1571.	0.2	218
200	Genetic variants near <i>TIMP3</i> and high-density lipoprotein–associated loci influence susceptibility to age-related macular degeneration. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 7401-7406.	3.3	475
201	Multiprotein Complexes of Retinitis Pigmentosa GTPase Regulator (RPGR), a Ciliary Protein Mutated in X-Linked Retinitis Pigmentosa (XLRP). Advances in Experimental Medicine and Biology, 2010, 664, 105-114.	0.8	26
202	Distinct Signature of Altered Homeostasis in Aging Rod Photoreceptors: Implications for Retinal Diseases. PLoS ONE, 2010, 5, e13885.	1.1	35
203	Age-related macular degeneration-associated variants at chromosome 10q26 do not significantly alter ARMS2 and HTRA1 transcript levels in the human retina. Molecular Vision, 2010, 16, 1317-23.	1.1	40
204	Retinoid-related orphan nuclear receptor $ROR\hat{I}^2$ is an early-acting factor in rod photoreceptor development. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 17534-17539.	3.3	114
205	Rdh12 Activity and Effects on Retinoid Processing in the Murine Retina. Journal of Biological Chemistry, 2009, 284, 21468-21477.	1.6	46
206	Defects in neural stem cell proliferation and olfaction in Chd7 deficient mice indicate a mechanism for hyposmia in human CHARGE syndrome. Human Molecular Genetics, 2009, 18, 1909-1923.	1.4	97
207	RPGR-containing protein complexes in syndromic and non-syndromic retinal degeneration due to ciliary dysfunction. Journal of Genetics, 2009, 88, 399-407.	0.4	28
208	Canine RD3 mutation establishes rod-cone dysplasia type 2 (rcd2) as ortholog of human and murine rd3. Mammalian Genome, 2009, 20, 109-123.	1.0	53
209	A common allele in RPGRIP1L is a modifier of retinal degeneration in ciliopathies. Nature Genetics, 2009, 41, 739-745.	9.4	255
210	Mutations in a BTB-Kelch Protein, KLHL7, Cause Autosomal-Dominant Retinitis Pigmentosa. American Journal of Human Genetics, 2009, 84, 792-800.	2.6	89
211	Unraveling a Multifactorial Late-Onset Disease: From Genetic Susceptibility to Disease Mechanisms for Age-Related Macular Degeneration. Annual Review of Genomics and Human Genetics, 2009, 10, 19-43.	2.5	254
212	A comprehensive analysis of sequence variants and putative disease-causing mutations in photoreceptor-specific nuclear receptor NR2E3. Molecular Vision, 2009, 15, 2174-84.	1.1	28
213	Geographic atrophy in age-related macular degeneration and TLR3. New England Journal of Medicine, 2009, 360, 2254-5; author reply 2255-6.	13.9	14
214	Retinitis Pigmentosa GTPase Regulator (RPGR) protein isoforms in mammalian retina: Insights into X-linked Retinitis Pigmentosa and associated ciliopathies. Vision Research, 2008, 48, 366-376.	0.7	57
215	Rod differentiation factor NRL activates the expression of nuclear receptor NR2E3 to suppress the development of cone photoreceptors. Brain Research, 2008, 1236, 16-29.	1.1	105
216	CP110 Suppresses Primary Cilia Formation through Its Interaction with CEP290, a Protein Deficient in Human Ciliary Disease. Developmental Cell, 2008, 15, 187-197.	3.1	228

#	Article	IF	CITATIONS
217	Discordant Phenotypes in Fraternal Twins Having an Identical Mutation in Exon ORF15 of the RPGR Gene. JAMA Ophthalmology, 2008, 126, 379.	2.6	48
218	Inflammation in the pathogenesis of age-related macular degeneration. British Journal of Ophthalmology, 2008, 92, 448-450.	2.1	50
219	<i>Nrl</i> -Knockout Mice Deficient in Rpe65 Fail to Synthesize 11- <i>cis</i> Retinal and Cone Outer Segments., 2008, 49, 1126.		39
220	Toll-like Receptor Polymorphisms and Age-Related Macular Degeneration., 2008, 49, 1652.		79
221	FGF19 Exhibits Neuroprotective Effects on Adult Mammalian Photoreceptors In Vitro., 2008, 49, 1696.		22
222	Retinal Phenotype of an X-Linked Pseudo-usher Syndrome in Association with the G173R Mutation in the RPGR Gene. Advances in Experimental Medicine and Biology, 2008, 613, 221-227.	0.8	4
223	Subunit Dissociation and Diffusion Determine the Subcellular Localization of Rod and Cone Transducins. Journal of Neuroscience, 2007, 27, 5484-5494.	1.7	66
224	Hypomorphic CEP290/NPHP6 mutations result in anosmia caused by the selective loss of G proteins in cilia of olfactory sensory neurons. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 15917-15922.	3.3	144
225	Madeline 2.0 PDE: a new program for local and web-based pedigree drawing. Bioinformatics, 2007, 23, 1854-1856.	1.8	29
226	A variant of mitochondrial protein LOC387715/ARMS2, not HTRA1, is strongly associated with age-related macular degeneration. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 16227-16232.	3.3	398
227	Genetic susceptibility to age-related macular degeneration: a paradigm for dissecting complex disease traits. Human Molecular Genetics, 2007, 16, R174-R182.	1.4	168
228	Transformation of cone precursors to functional rod photoreceptors by bZIP transcription factor NRL. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 1679-1684.	3.3	136
229	Afferent Control of Horizontal Cell Morphology Revealed by Genetic Respecification of Rods and Cones. Journal of Neuroscience, 2007, 27, 3540-3547.	1.7	38
230	Chondroitinase ABC Treatment Enhances Synaptogenesis between Transplant and Host Neurons in Model of Retinal Degeneration. Cell Transplantation, 2007, 16, 493-503.	1.2	83
231	Mutations in TOPORS Cause Autosomal Dominant Retinitis Pigmentosa with Perivascular Retinal Pigment Epithelium Atrophy. American Journal of Human Genetics, 2007, 81, 1098-1103.	2.6	77
232	High-Resolution Imaging with Adaptive Optics in Patients with Inherited Retinal Degeneration. , 2007, 48, 3283.		241
233	Inner Retinal Abnormalities in X-linked Retinitis Pigmentosa with <i>RPGR </i> Mutations., 2007, 48, 4759.		107
234	Retinopathy mutations in the bZIP protein NRL alter phosphorylation and transcriptional activity. Human Mutation, 2007, 28, 589-598.	1.1	41

#	Article	IF	Citations
235	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
236	Preventing polyglutamine-induced activation of c-Jun delays neuronal dysfunction in a mouse model of SCA7 retinopathy. Neurobiology of Disease, 2007, 25, 571-581.	2.1	15
237	Mutations associated with retinopathies alter mitogen-activated protein kinase-induced phosphorylation of neural retina leucine-zipper. Molecular Vision, 2007, 13, 1114-20.	1.1	3
238	Premature Truncation of a Novel Protein, RD3, Exhibiting Subnuclear Localization Is Associated with Retinal Degeneration. American Journal of Human Genetics, 2006, 79, 1059-1070.	2.6	112
239	The centrosomal protein nephrocystin-6 is mutated in Joubert syndrome and activates transcription factor ATF4. Nature Genetics, 2006, 38, 674-681.	9.4	535
240	CFH haplotypes without the Y402H coding variant show strong association with susceptibility to age-related macular degeneration. Nature Genetics, 2006, 38, 1049-1054.	9.4	318
241	Retinal repair by transplantation of photoreceptor precursors. Nature, 2006, 444, 203-207.	13.7	999
242	Gene expression signatures and biomarkers of noninvasive and invasive breast cancer cells: comprehensive profiles by representational difference analysis, microarrays and proteomics. Oncogene, 2006, 25, 2328-2338.	2.6	175
243	In vivo function of the orphan nuclear receptor NR2E3 in establishing photoreceptor identity during mammalian retinal development. Human Molecular Genetics, 2006, 15, 2588-2602.	1.4	113
244	Targeting of GFP to newborn rods by Nrl promoter and temporal expression profiling of flow-sorted photoreceptors. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 3890-3895.	3.3	310
245	Retinoic Acid Regulates the Expression of Photoreceptor Transcription Factor NRL. Journal of Biological Chemistry, 2006, 281, 27327-27334.	1.6	60
246	Activation of Signaling Pathways and Stress-Response Genes in an Experimental Model of Retinal Detachment., 2006, 47, 1691.		68
247	In-frame deletion in a novel centrosomal/ciliary protein CEP290/NPHP6 perturbs its interaction with RPGR and results in early-onset retinal degeneration in the rd16 mouse. Human Molecular Genetics, 2006, 15, 1847-1857.	1.4	353
248	Senile panretinal cone dysfunction in age-related macular degeneration (AMD): a report of 52 amd patients compared to age-matched controls. Transactions of the American Ophthalmological Society, 2006, 104, 232-40.	1.4	7
249	Nephrocystin-5, a ciliary IQ domain protein, is mutated in Senior-Loken syndrome and interacts with RPGR and calmodulin. Nature Genetics, 2005, 37, 282-288.	9.4	367
250	RPGR ORF15 isoform co-localizes with RPGRIP1 at centrioles and basal bodies and interacts with nucleophosmin. Human Molecular Genetics, 2005, 14, 1183-1197.	1.4	103
251	Cone-like Morphological, Molecular, and Electrophysiological Features of the Photoreceptors of the NrlKnockout Mouse., 2005, 46, 2156.		190
252	High Throughput Screening of Co-Expressed Gene Pairs with Controlled False Discovery Rate (FDR) and Minimum Acceptable Strength (MAS). Journal of Computational Biology, 2005, 12, 1029-1045.	0.8	47

#	Article	IF	Citations
253	RPGR-ORF15, Which Is Mutated in Retinitis Pigmentosa, Associates with SMC1, SMC3, and Microtubule Transport Proteins. Journal of Biological Chemistry, 2005, 280, 33580-33587.	1.6	154
254	Photoreceptors of Nrl $\hat{a}^2/\hat{a}^2$ Mice Coexpress Functional S- and M-cone Opsins Having Distinct Inactivation Mechanisms. Journal of General Physiology, 2005, 125, 287-304.	0.9	125
255	Network constrained clustering for gene microarray data. Bioinformatics, 2005, 21, 4014-4020.	1.8	53
256	Strong Association of the Y402H Variant in Complement Factor H at 1q32 with Susceptibility to Age-Related Macular Degeneration. American Journal of Human Genetics, 2005, 77, 149-153.	2.6	327
257	Meta-analysis of genome scans of age-related macular degeneration. Human Molecular Genetics, 2005, 14, 2257-2264.	1.4	224
258	Toll-like receptor 4 variant D299G is associated with susceptibility to age-related macular degeneration. Human Molecular Genetics, 2005, 14, 1449-1455.	1.4	177
259	Biomarkers of Cardiovascular Disease as Risk Factors for Age-Related Macular Degeneration. Ophthalmology, 2005, 112, 2076-2080.	2.5	143
260	Mechanisms of aging in senescence-accelerated mice. Genome Biology, 2005, 6, R48.	13.9	55
261	Expression of Photoreceptor-Specific Nuclear Receptor NR2E3 in Rod Photoreceptors of Fetal Human Retina., 2004, 45, 2807.		82
262	Characterization of New Transcripts Enriched in the Mouse Retina and Identification of Candidate Retinal Disease Genes., 2004, 45, 3313.		9
263	Seeing the Unseen: Microarray-Based Gene Expression Profiling in Vision. , 2004, 45, 2457.		10
264	Association of Apolipoprotein E Alleles with Susceptibility to Age-Related Macular Degeneration in a Large Cohort from a Single Center. Investigative Ophthalmology and Visual Science, 2004, 45, 1306-1310.	3.3	129
265	Expression profiling of the developing and mature Nrl â^'/â^' mouse retina: identification of retinal disease candidates and transcriptional regulatory targets of Nrl. Human Molecular Genetics, 2004, 13, 1487-1503.	1.4	157
266	Transgenic Mice Expressing Cre-Recombinase Specifically in M- or S-Cone Photoreceptors., 2004, 45, 42.		29
267	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
268	Functional Analysis of the Rod Photoreceptor cGMP Phosphodiesterase α-Subunit Gene Promoter. Journal of Biological Chemistry, 2004, 279, 19800-19807.	1.6	54
269	QRX, a novel homeobox gene, modulates photoreceptor gene expression. Human Molecular Genetics, 2004, 13, 1025-1040.	1.4	<b>7</b> 3
270	Photoreceptor-specific nuclear receptor NR2E3 functions as a transcriptional activator in rod photoreceptors. Human Molecular Genetics, 2004, 13, 1563-1575.	1.4	226

#	Article	IF	Citations
271	The Minimal Transactivation Domain of the Basic Motif-Leucine Zipper Transcription Factor NRL Interacts with TATA-binding Protein. Journal of Biological Chemistry, 2004, 279, 47233-47241.	1.6	34
272	Recruitment of the Rod Pathway by Cones in the Absence of Rods. Journal of Neuroscience, 2004, 24, 7576-7582.	1.7	77
273	Altered Expression of Genes of the Bmp/Smad and Wnt/Calcium Signaling Pathways in the Cone-only Nrl-/- Mouse Retina, Revealed by Gene Profiling Using Custom cDNA Microarrays. Journal of Biological Chemistry, 2004, 279, 42211-42220.	1.6	52
274	Recessive NRL mutations in patients with clumped pigmentary retinal degeneration and relative preservation of blue cone function. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 17819-17824.	3.3	88
275	Mutation analysis of NR2E3 and NRL genes in Enhanced S Cone Syndrome. Human Mutation, 2004, 24, 439-439.	1.1	92
276	Gene discovery using Pareto depth sampling distributions. Journal of the Franklin Institute, 2004, 341, 55-75.	1.9	9
277	Age-Related Macular Degeneration: A High-Resolution Genome Scan for Susceptibility Loci in a Population Enriched for Late-Stage Disease. American Journal of Human Genetics, 2004, 74, 482-494.	2.6	157
278	Multicriteria Gene Screening for Analysis of Differential Expression with DNA Microarrays. Eurasip Journal on Advances in Signal Processing, 2004, 2004, 1.	1.0	20
279	Increasing evidence for syndromic phenotypes associated with RPGR mutations. American Journal of Ophthalmology, 2004, 137, 785-786.	1.7	19
280	Annotation and analysis of 10,000 expressed sequence tags from developing mouse eye and adult retina. Genome Biology, 2003, 4, R65.	13.9	34
281	Interaction of retinal bZIP transcription factor NRL with Flt3-interacting zinc-finger protein Fiz1: possible role of Fiz1 as a transcriptional repressor. Human Molecular Genetics, 2003, 12, 365-373.	1.4	37
282	Mutation in a short-chain collagen gene, CTRP5, results in extracellular deposit formation in late-onset retinal degeneration: a genetic model for age-related macular degeneration. Human Molecular Genetics, 2003, 12, 2657-2667.	1.4	172
283	Clinical studies of X-linked retinitis pigmentosa in three Swedish families with newly identified mutations in the RP2 and RPGR-ORF15 genes. Ophthalmic Genetics, 2003, 24, 215-223.	0.5	25
284	GRK1-Dependent Phosphorylation of S and M Opsins and Their Binding to Cone Arrestin during Cone Phototransduction in the Mouse Retina. Journal of Neuroscience, 2003, 23, 6152-6160.	1.7	105
285	De Novo Mutation in theRP1Gene (Arg677ter) Associated with Retinitis Pigmentosa., 2003, 44, 3593.		32
286	Mutation screening of patients with Leber Congenital Amaurosis or the enhanced S-Cone Syndrome reveals a lack of sequence variations in the NRL gene. Molecular Vision, 2003, 9, 14-7.	1.1	8
287	A comprehensive analysis of the expression of crystallins in mouse retina. Molecular Vision, 2003, 9, 410-9.	1.1	95
288	Barrier to Autointegration Factor Interacts with the Cone-Rod Homeobox and Represses Its Transactivation Function. Journal of Biological Chemistry, 2002, 277, 43288-43300.	1.6	112

#	Article	IF	CITATIONS
289	Retinal Histopathology of an XLRP Carrier with a Mutation in the RPGR Exon ORF15. Experimental Eye Research, 2002, 75, 431-443.	1.2	37
290	Transcriptome analysis of the retina. Genome Biology, 2002, 3, reviews1022.1.	13.9	31
291	Mouse eye gene microarrays for investigating ocular development and disease. Vision Research, 2002, 42, 463-470.	0.7	39
292	A Comprehensive Mutation Analysis of RP2 and RPGR in a North American Cohort of Families with X-Linked Retinitis Pigmentosa. American Journal of Human Genetics, 2002, 70, 1545-1554.	2.6	224
293	Gene expression profile of native human retinal pigment epithelium. Investigative Ophthalmology and Visual Science, 2002, 43, 603-7.	3.3	19
294	Evaluation and optimization of procedures for target labeling and hybridization of cDNA microarrays. Molecular Vision, 2002, 8, 130-7.	1.1	44
295	Microarray analysis of gene expression in the aging human retina. Investigative Ophthalmology and Visual Science, 2002, 43, 2554-60.	3.3	79
296	Retinal histopathology of an XLRP carrier with a mutation in the RPGR exon ORF15. Experimental Eye Research, 2002, 75, 431-43.	1.2	19
297	Evaluation of the ELOVL4 gene in patients with age-related macular degeneration. Ophthalmic Genetics, 2001, 22, 233-239.	0.5	53
298	Five novelRPGR mutations in families with X-linked retinitis pigmentosa. Human Mutation, 2001, 17, 151-151.	1.1	11
299	Nrl is required for rod photoreceptor development. Nature Genetics, 2001, 29, 447-452.	9.4	795
300	Multiple Phosphorylated Isoforms of NRL Are Expressed in Rod Photoreceptors. Journal of Biological Chemistry, 2001, 276, 36824-36830.	1.6	107
301	X-Linked Retinitis Pigmentosa: Current Status. , 2001, , 11-22.		4
302	NRL S50T mutation and the importance of †founder effects' in inherited retinal dystrophies. European Journal of Human Genetics, 2000, 8, 783-787.	1.4	18
303	Cloning and functional expression of human retinal Kir2.4, a pH-sensitive inwardly rectifying K+ channel. American Journal of Physiology - Cell Physiology, 2000, 279, C771-C784.	2.1	42
304	The Leucine Zipper of NRL Interacts with the CRX Homeodomain. Journal of Biological Chemistry, 2000, 275, 29794-29799.	1.6	188
305	Remapping of the RP15 Locus for X-Linked Cone-Rod Degeneration to Xp11.4-p21.1, and Identification of a De Novo Insertion in the RPGR Exon ORF15. American Journal of Human Genetics, 2000, 67, 1000-1003.	2.6	61
306	Leber congenital amaurosis caused by a homozygous mutation (R90W) in the homeodomain of the retinal transcription factor CRX: direct evidence for the involvement of CRX in the development of photoreceptor function. Human Molecular Genetics, 1999, 8, 299-305.	1.4	169

#	Article	IF	CITATIONS
307	A mutation in NRL is associated with autosomal dominant retinitis pigmentosa. Nature Genetics, 1999, 21, 355-356.	9.4	205
308	Protein-Truncation Mutations in the RP2 Gene in a North American Cohort of Families with X-Linked Retinitis Pigmentosa. American Journal of Human Genetics, 1999, 64, 897-900.	2.6	78
309	X-linked retinitis pigmentosa in two families with a missense mutation in the RPGR gene and putative change of glycine to valine at codon 6011Dr. Buraczynska is currently on a sabbatical leave from Medical School, Lublin, Poland Ophthalmology, 1998, 105, 2286-2296.	2.5	23
310	A Novel Locus (RP24) for X-linked Retinitis Pigmentosa Maps to Xq26-27. American Journal of Human Genetics, 1998, 63, 1439-1447.	2.6	65
311	Clinical expression of X-linked retinitis pigmentosa in a Swedish family with the RP2 genotype. Ophthalmic Genetics, 1998, 19, 187-196.	0.5	4
312	Mutation of a Gene Encoding a Protein with Extracellular Matrix Motifs in Usher Syndrome Type IIa. Science, 1998, 280, 1753-1757.	6.0	366
313	Biochemical Characterization and Subcellular Localization of the Mouse Retinitis Pigmentosa GTPase Regulator (mRpgr). Journal of Biological Chemistry, 1998, 273, 19656-19663.	1.6	73
314	A New 2–Base Pair Deletion in the RPGR Gene in a Black Family With X-Linked Retinitis Pigmentosa. JAMA Ophthalmology, 1998, 116, 213-8.	2.6	10
315	Lens-Specific Gene Recruitment of ζ-Crystallin through Pax6, Nrl-Maf, and Brain Suppressor Sites. Molecular and Cellular Biology, 1998, 18, 2067-2076.	1.1	46
316	Human bZIP Transcription Factor GeneNRL:Structure, Genomic Sequence, and Fine Linkage Mapping at 14q11.2 and Negative Mutation Analysis in Patients with Retinal Degeneration. Genomics, 1997, 45, 395-401.	1.3	28
317	Mutations in the Cone-Rod Homeobox Gene Are Associated with the Cone-Rod Dystrophy Photoreceptor Degeneration. Neuron, 1997, 19, 1329-1336.	3.8	250
318	Spectrum of Mutations in the RPGR Gene That Are Identified in 20% of Families with X-Linked Retinitis Pigmentosa. American Journal of Human Genetics, 1997, 61, 1287-1292.	2.6	93
319	Analysis of the RPGRGene in 11 Pedigrees with the Retinitis Pigmentosa Type 3 Genotype: Paucity of Mutations in the Coding Region but Splice Defects in Two Families. American Journal of Human Genetics, 1997, 61, 571-580.	2.6	61
320	Characterization and Sequence Analysis of the Human Homeobox-Containing GeneGBX2. Genomics, 1996, 31, 335-342.	1.3	16
321	The basic motif-leucine zipper transcription factor Nrl can positively regulate rhodopsin gene expression Proceedings of the National Academy of Sciences of the United States of America, 1996, 93, 191-195.	3.3	187
322	[11]Isolation of candidate genes for inherited diseases: Application to X-linked retinal degenerations. Methods in Molecular Genetics, 1996, 8, 207-228.	0.6	0
323	The bZIP Transcription Factor Nrl Stimulates Rhodopsin Promoter Activity in Primary Retinal Cell Cultures. Journal of Biological Chemistry, 1996, 271, 29612-29618.	1.6	128
324	Dinucleotide polymorphism at the DXS1178 locus is tightly linked to PGK1 at Xq13. Human Genetics, 1995, 95, 467-8.	1.8	1

#	Article	IF	Citations
325	Expression and chromosomal localization of cDNA clones from an enriched human retinal pigment epithelial (RPE) cell line library: identification of two RPE-specific genes. Cytogenetic and Genome Research, 1995, 69, 71-74.	0.6	9
326	Dinucleotide repeat polymorphism at the DXS977 locus. Human Molecular Genetics, 1994, 3, 1030-1030.	1.4	2
327	Localization of the Gene for Pigment Epithelium-Derived Factor (PEDF) to Chromosome 17p13.1 and Expression in Cultured Human Retinoblastoma Cells. Genomics, 1994, 19, 266-272.	1.3	70
328	Molecular characterization of a novel human gene, SEC13R, related to the yeast secretory pathway gene SEC13, and mapping to a conserved linkage group on human chromosome 3p24-p25 and mouse chromosome 6. Human Molecular Genetics, 1994, 3, 1281-1286.	1.4	33
329	Mapping of the neural retina leucine zipper gene, Nrl, to mouse Chromosome 14. Mammalian Genome, 1993, 4, 618-620.	1.0	2
330	Molecular Characterization of the Murine Neural Retina Leucine Zipper Gene, Nrl. Genomics, 1993, 18, 216-222.	1.3	27
331	λSHK and λAASV: phage vectors for efficient cDNA cloning and expression in mammalian cells. Gene, 1993, 123, 287-288.	1.0	2
332	Construction of Directional cDNA Libraries from Human Retinal Tissue/Cells and Their Enrichment for Specific Genes Using an Efficient Subtraction Procedure. Methods in Neurosciences, 1993, , 285-300.	0.5	2
333	Identification of Candidate Genes for Eye Diseases: Studies on a Neural Retina-Specific Gene Encoding a Putative Dna Binding Protein of Leucine Zipper Family. , 1993, , 171-180.		0
334	Expressed sequence tags and chromosomal localization of cDNA clones from a subtracted retinal pigment epithelium library. Genomics, 1992, 13, 873-876.	1.3	66
335	Neural retina-specific leucine zipper gene NRL (D14S46E) maps to human chromosome 14q11.1-q11.2. Genomics, 1992, 14, 491-492.	1.3	11
336	Sequence analysis, expression and chromosomal localization of a gene, isolated from a subtracted human retina cDNA library, that encodes an insulin-like growth factor binding protein (IGFBP2). Experimental Eye Research, 1991, 52, 549-561.	1.2	30
337	Positional cloning and characterization of a paired box- and homeobox-containing gene from the aniridia region. Cell, 1991, 67, 1059-1074.	13.5	810
338	A simple and efficient cDNA library subtraction procedure: isolation of human retina-specific cDNA clones. Nucleic Acids Research, 1991, 19, 1954-1954.	6.5	62
339	Differential expression of novel $Gsl$ ± signal transduction protein cDNA species. Nucleic Acids Research, 1991, 19, 4725-4729.	6.5	80
340	Chromosomal localization and cDNA sequence of humanralB, a GTP binding protein. Somatic Cell and Molecular Genetics, 1990, 16, 407-410.	0.7	7
341	pSH4: a mammalian cDNA expression vector. Nucleic Acids Research, 1990, 18, 3668-3668.	6.5	10
342	The mRNA of a human class I gene HLA G/HLA 6.0 exhibits a restricted pattern of expression. Nucleic Acids Research, 1990, 18, 2189-2189.	6.5	81

#	Article	IF	CITATIONS
343	Molecular analysis of the cDNA for human SPARC/osteonectin/BM-40: Sequence, expression, and localization of the gene to chromosome 5q31-q33. Genomics, 1988, 2, 37-47.	1.3	154
344	Charon BS (+) and ( $\hat{a}^{-}$ ), versatile $\hat{l}$ » phage vectors for constructing directional cDNA libraries and their efficient transfer to plasmids. Nucleic Acids Research, 1988, 16, 8739-8739.	6.5	34
345	Generation of hydrogen peroxide on oxidation of NADH by hepatic plasma membranes. Journal of Bioenergetics and Biomembranes, 1981, 13, 241-253.	1.0	31
346	Nicotinamide Promotes Formation of Retinal Organoids From Human Pluripotent Stem Cells via Enhanced Neural Cell Fate Commitment. Frontiers in Cellular Neuroscience, 0, 16, .	1.8	6