

# Anand Swaroop

## List of Publications by Year in descending order

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

Version: 2024-02-01

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	Next-generation genotype imputation service and methods. <i>Nature Genetics</i> , 2016, 48, 1284-1287.	21.4	2,828
2	A reference panel of 64,976 haplotypes for genotype imputation. <i>Nature Genetics</i> , 2016, 48, 1279-1283.	21.4	2,421
3	A large genome-wide association study of age-related macular degeneration highlights contributions of rare and common variants. <i>Nature Genetics</i> , 2016, 48, 134-143.	21.4	1,167
4	Retinal repair by transplantation of photoreceptor precursors. <i>Nature</i> , 2006, 444, 203-207.	27.8	999
5	Positional cloning and characterization of a paired box- and homeobox-containing gene from the aniridia region. <i>Cell</i> , 1991, 67, 1059-1074.	28.9	810
6	Nrl is required for rod photoreceptor development. <i>Nature Genetics</i> , 2001, 29, 447-452.	21.4	795
7	Seven new loci associated with age-related macular degeneration. <i>Nature Genetics</i> , 2013, 45, 433-439.	21.4	687
8	The centrosomal protein nephrocystin-6 is mutated in Joubert syndrome and activates transcription factor ATF4. <i>Nature Genetics</i> , 2006, 38, 674-681.	21.4	535
9	Genetic variants near <i>TIMP3</i> and high-density lipoprotein-associated loci influence susceptibility to age-related macular degeneration. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2010, 107, 7401-7406.	7.1	475
10	Transcriptional regulation of photoreceptor development and homeostasis in the mammalian retina. <i>Nature Reviews Neuroscience</i> , 2010, 11, 563-576.	10.2	465
11	A variant of mitochondrial protein LOC387715/ARMS2, not HTRA1, is strongly associated with age-related macular degeneration. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2007, 104, 16227-16232.	7.1	398
12	Age-Related Macular Degeneration: Genetics and Biology Coming Together. <i>Annual Review of Genomics and Human Genetics</i> , 2014, 15, 151-171.	6.2	394
13	Nephrocystin-5, a ciliary IQ domain protein, is mutated in Senior-Loken syndrome and interacts with RPGR and calmodulin. <i>Nature Genetics</i> , 2005, 37, 282-288.	21.4	367
14	Mutation of a Gene Encoding a Protein with Extracellular Matrix Motifs in Usher Syndrome Type IIa. <i>Science</i> , 1998, 280, 1753-1757.	12.6	366
15	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. <i>Human Molecular Genetics</i> , 2006, 15, 1847-1857.	2.9	353
16	Strong Association of the Y402H Variant in Complement Factor H at 1q32 with Susceptibility to Age-Related Macular Degeneration. <i>American Journal of Human Genetics</i> , 2005, 77, 149-153.	6.2	327
17	CFH haplotypes without the Y402H coding variant show strong association with susceptibility to age-related macular degeneration. <i>Nature Genetics</i> , 2006, 38, 1049-1054.	21.4	318
18	Targeting of GFP to newborn rods by Nrl promoter and temporal expression profiling of flow-sorted photoreceptors. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2006, 103, 3890-3895.	7.1	310

#	ARTICLE	IF	CITATIONS
19	A common allele in RRGRIPL1 is a modifier of retinal degeneration in ciliopathies. <i>Nature Genetics</i> , 2009, 41, 739-745.	21.4	255
20	Unraveling a Multifactorial Late-Onset Disease: From Genetic Susceptibility to Disease Mechanisms for Age-Related Macular Degeneration. <i>Annual Review of Genomics and Human Genetics</i> , 2009, 10, 19-43.	6.2	254
21	Mutations in the Cone-Rod Homeobox Gene Are Associated with the Cone-Rod Dystrophy Photoreceptor Degeneration. <i>Neuron</i> , 1997, 19, 1329-1336.	8.1	250
22	Transcriptome analysis and molecular signature of human retinal pigment epithelium. <i>Human Molecular Genetics</i> , 2010, 19, 2468-2486.	2.9	249
23	E2-2 Protein and Fuchs's Corneal Dystrophy. <i>New England Journal of Medicine</i> , 2010, 363, 1016-1024.	27.0	247
24	High-Resolution Imaging with Adaptive Optics in Patients with Inherited Retinal Degeneration. , 2007, 48, 3283.		241
25	Gene therapy rescues photoreceptor blindness in dogs and paves the way for treating human X-linked retinitis pigmentosa. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2012, 109, 2132-2137.	7.1	237
26	Nrl knockdown by AAV-delivered CRISPR/Cas9 prevents retinal degeneration in mice. <i>Nature Communications</i> , 2017, 8, 14716.	12.8	231
27	CP110 Suppresses Primary Cilia Formation through Its Interaction with CEP290, a Protein Deficient in Human Ciliary Disease. <i>Developmental Cell</i> , 2008, 15, 187-197.	7.0	228
28	Photoreceptor-specific nuclear receptor NR2E3 functions as a transcriptional activator in rod photoreceptors. <i>Human Molecular Genetics</i> , 2004, 13, 1563-1575.	2.9	226
29	A Comprehensive Mutation Analysis of RP2 and RPGR in a North American Cohort of Families with X-Linked Retinitis Pigmentosa. <i>American Journal of Human Genetics</i> , 2002, 70, 1545-1554.	6.2	224
30	Meta-analysis of genome scans of age-related macular degeneration. <i>Human Molecular Genetics</i> , 2005, 14, 2257-2264.	2.9	224
31	MicroRNA-204/211 alters epithelial physiology. <i>FASEB Journal</i> , 2010, 24, 1552-1571.	0.5	218
32	Biology and therapy of inherited retinal degenerative disease: insights from mouse models. <i>DMM Disease Models and Mechanisms</i> , 2015, 8, 109-129.	2.4	207
33	A mutation in NRL is associated with autosomal dominant retinitis pigmentosa. <i>Nature Genetics</i> , 1999, 21, 355-356.	21.4	205
34	Molecular Anatomy of the Developing Human Retina. <i>Developmental Cell</i> , 2017, 43, 763-779.e4.	7.0	205
35	Retinal transcriptome and eQTL analyses identify genes associated with age-related macular degeneration. <i>Nature Genetics</i> , 2019, 51, 606-610.	21.4	201
36	Cone-like Morphological, Molecular, and Electrophysiological Features of the Photoreceptors of the Nrl Knockout Mouse. , 2005, 46, 2156.		190

#	ARTICLE	IF	CITATIONS
37	The Leucine Zipper of NRL Interacts with the CRX Homeodomain. <i>Journal of Biological Chemistry</i> , 2000, 275, 29794-29799.	3.4	188
38	The basic motif-leucine zipper transcription factor Nrl can positively regulate rhodopsin gene expression.. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1996, 93, 191-195.	7.1	187
39	Toll-like receptor 4 variant D299G is associated with susceptibility to age-related macular degeneration. <i>Human Molecular Genetics</i> , 2005, 14, 1449-1455.	2.9	177
40	Gene expression signatures and biomarkers of noninvasive and invasive breast cancer cells: comprehensive profiles by representational difference analysis, microarrays and proteomics. <i>Oncogene</i> , 2006, 25, 2328-2338.	5.9	175
41	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. <i>Human Molecular Genetics</i> , 2003, 12, 2657-2667.	2.9	172
42	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. <i>Human Molecular Genetics</i> , 1999, 8, 299-305.	2.9	169
43	Genetic susceptibility to age-related macular degeneration: a paradigm for dissecting complex disease traits. <i>Human Molecular Genetics</i> , 2007, 16, R174-R182.	2.9	168
44	Accelerated and Improved Differentiation of Retinal Organoids from Pluripotent Stem Cells in Rotating-Wall Vessel Bioreactors. <i>Stem Cell Reports</i> , 2018, 10, 300-313.	4.8	168
45	Identification of a rare coding variant in complement 3 associated with age-related macular degeneration. <i>Nature Genetics</i> , 2013, 45, 1375-1379.	21.4	158
46	Expression profiling of the developing and mature Nrl <sup>+/+</sup> / <sub>+</sub> mouse retina: identification of retinal disease candidates and transcriptional regulatory targets of Nrl. <i>Human Molecular Genetics</i> , 2004, 13, 1487-1503.	2.9	157
47	Age-Related Macular Degeneration: A High-Resolution Genome Scan for Susceptibility Loci in a Population Enriched for Late-Stage Disease. <i>American Journal of Human Genetics</i> , 2004, 74, 482-494.	6.2	157
48	Molecular analysis of the cDNA for human SPARC/osteonectin/BM-40: Sequence, expression, and localization of the gene to chromosome 5q31-q33. <i>Genomics</i> , 1988, 2, 37-47.	2.9	154
49	RPGR-ORF15, Which Is Mutated in Retinitis Pigmentosa, Associates with SMC1, SMC3, and Microtubule Transport Proteins. <i>Journal of Biological Chemistry</i> , 2005, 280, 33580-33587.	3.4	154
50	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. <i>Stem Cells</i> , 2015, 33, 3504-3518.	3.2	153
51	Centrosomal-ciliary gene CEP290/NPHP6 mutations result in blindness with unexpected sparing of photoreceptors and visual brain: implications for therapy of Leber congenital amaurosis. <i>Human Mutation</i> , 2007, 28, 1074-1083.	2.5	148
52	Free radical scavenging, antioxidant and cancer chemoprevention by grape seed proanthocyanidin: An overview. <i>Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis</i> , 2014, 768, 69-73.	1.0	148
53	Hypomorphic CEP290/NPHP6 mutations result in anosmia caused by the selective loss of G proteins in cilia of olfactory sensory neurons. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2007, 104, 15917-15922.	7.1	144
54	Biomarkers of Cardiovascular Disease as Risk Factors for Age-Related Macular Degeneration. <i>Ophthalmology</i> , 2005, 112, 2076-2080.	5.2	143

#	ARTICLE	IF	CITATIONS
55	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.	7.1	136
56	Ancestry estimation and control of population stratification for sequence-based association studies. Nature Genetics, 2014, 46, 409-415.	21.4	136
57	Photoreceptor sensory cilia and ciliopathies: focus on CEP290, RPGR and their interacting proteins. Cilia, 2012, 1, 22.	1.8	131
58	Evidence of association of <i>APOE</i> with age-related macular degeneration - a pooled analysis of 15 studies. Human Mutation, 2011, 32, 1407-1416.	2.5	130
59	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
60	The bZIP Transcription Factor Nrl Stimulates Rhodopsin Promoter Activity in Primary Retinal Cell Cultures. Journal of Biological Chemistry, 1996, 271, 29612-29618.	3.4	128
61	Progression of Geographic Atrophy in Age-related Macular Degeneration. Ophthalmology, 2018, 125, 1913-1928.	5.2	127
62	Photoreceptors of <i>Nrl</i> <sup>-/-</sup> Mice Coexpress Functional S- and M-cone Opsins Having Distinct Inactivation Mechanisms. Journal of General Physiology, 2005, 125, 287-304.	1.9	125
63	In Vitro Modeling Using Ciliopathy-Patient-Derived Cells Reveals Distinct Cilia Dysfunctions Caused by CEP290 Mutations. Cell Reports, 2017, 20, 384-396.	6.4	120
64	Cone photoreceptors are the main targets for gene therapy of NPHP5 (IQCB1) or NPHP6 (CEP290) blindness: generation of an all-cone <i>Nphp6</i> hypomorph mouse that mimics the human retinal ciliopathy. Human Molecular Genetics, 2011, 20, 1411-1423.	2.9	115
65	Retinoid-related orphan nuclear receptor ROR $\gamma$ 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.	7.1	114
66	In vivo function of the orphan nuclear receptor NR2E3 in establishing photoreceptor identity during mammalian retinal development. Human Molecular Genetics, 2006, 15, 2588-2602.	2.9	113
67	Barrier to Autointegration Factor Interacts with the Cone-Rod Homeobox and Represses Its Transactivation Function. Journal of Biological Chemistry, 2002, 277, 43288-43300.	3.4	112
68	Premature Truncation of a Novel Protein, RD3, Exhibiting Subnuclear Localization Is Associated with Retinal Degeneration. American Journal of Human Genetics, 2006, 79, 1059-1070.	6.2	112
69	Mutations in <i>RPGR</i> and <i>RP2</i> Account for 15% of Males with Simplex Retinal Degenerative Disease. , 2012, 53, 8232.		108
70	Multiple Phosphorylated Isoforms of NRL Are Expressed in Rod Photoreceptors. Journal of Biological Chemistry, 2001, 276, 36824-36830.	3.4	107
71	Inner Retinal Abnormalities in X-linked Retinitis Pigmentosa with <i>RPGR</i> Mutations. , 2007, 48, 4759.		107
72	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.	3.6	105

#	ARTICLE	IF	CITATIONS
73	Rod differentiation factor NRL activates the expression of nuclear receptor NR2E3 to suppress the development of cone photoreceptors. <i>Brain Research</i> , 2008, 1236, 16-29.	2.2	105
74	CEP290 alleles in mice disrupt tissue-specific cilia biogenesis and recapitulate features of syndromic ciliopathies. <i>Human Molecular Genetics</i> , 2015, 24, 3775-3791.	2.9	105
75	Epigenetic control of gene regulation during development and disease: A view from the retina. <i>Progress in Retinal and Eye Research</i> , 2018, 65, 1-27.	15.5	105
76	Quantification of Oxygen Consumption in Retina Ex Vivo Demonstrates Limited Reserve Capacity of Photoreceptor Mitochondria. , 2015, 56, 8428.		104
77	NRL-Regulated Transcriptome Dynamics of Developing Rod Photoreceptors. <i>Cell Reports</i> , 2016, 17, 2460-2473.	6.4	104
78	Long-term survival and differentiation of retinal neurons derived from human embryonic stem cell lines in un-immunosuppressed mouse retina. <i>Molecular Vision</i> , 2012, 18, 920-36.	1.1	104
79	RPGR ORF15 isoform co-localizes with RCGRIP1 at centrioles and basal bodies and interacts with nucleophosmin. <i>Human Molecular Genetics</i> , 2005, 14, 1183-1197.	2.9	103
80	Recruitment of Rod Photoreceptors from Short-Wavelength-Sensitive Cones during the Evolution of Nocturnal Vision in Mammals. <i>Developmental Cell</i> , 2016, 37, 520-532.	7.0	103
81	Gene expression changes in aging retinal microglia: relationship to microglial support functions and regulation of activation. <i>Neurobiology of Aging</i> , 2013, 34, 2310-2321.	3.1	100
82	Transcriptional Regulation of Rod Photoreceptor Homeostasis Revealed by In Vivo NRL Targetome Analysis. <i>PLoS Genetics</i> , 2012, 8, e1002649.	3.5	99
83	MicroRNA Profile of the Developing Mouse Retina. , 2010, 51, 1823.		98
84	Developing Rods Transplanted into the Degenerating Retina of Crx-Knockout Mice Exhibit Neural Activity Similar to Native Photoreceptors. <i>Stem Cells</i> , 2013, 31, 1149-1159.	3.2	98
85	Defects in neural stem cell proliferation and olfaction in Chd7 deficient mice indicate a mechanism for hyposmia in human CHARGE syndrome. <i>Human Molecular Genetics</i> , 2009, 18, 1909-1923.	2.9	97
86	A comprehensive analysis of the expression of crystallins in mouse retina. <i>Molecular Vision</i> , 2003, 9, 410-9.	1.1	95
87	Nuclear receptor NR2E3 gene mutations distort human retinal laminar architecture and cause an unusual degeneration. <i>Human Molecular Genetics</i> , 2004, 13, 1893-1902.	2.9	94
88	Spectrum of Mutations in the RPGR Gene That Are Identified in 20% of Families with X-Linked Retinitis Pigmentosa. <i>American Journal of Human Genetics</i> , 1997, 61, 1287-1292.	6.2	93
89	Genome-wide association study and meta-analysis of intraocular pressure. <i>Human Genetics</i> , 2014, 133, 41-57.	3.8	93
90	Mutation analysis of NR2E3 and NRL genes in Enhanced S Cone Syndrome. <i>Human Mutation</i> , 2004, 24, 439-439.	2.5	92

#	ARTICLE	IF	CITATIONS
91	Complement Factor D in Age-Related Macular Degeneration. , 2011, 52, 8828.		92
92	Interaction of retinitis pigmentosa GTPase regulator (RPGR) with RAB8A GTPase: implications for cilia dysfunction and photoreceptor degeneration. Human Molecular Genetics, 2010, 19, 3591-3598.	2.9	91
93	Mutations in a BTB-Kelch Protein, KLHL7, Cause Autosomal-Dominant Retinitis Pigmentosa. American Journal of Human Genetics, 2009, 84, 792-800.	6.2	89
94	Next-generation sequencing facilitates quantitative analysis of wild-type and Nrl(-/-) retinal transcriptomes. Molecular Vision, 2011, 17, 3034-54.	1.1	89
95	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.	7.1	88
96	No Clinically Significant Association between CFH and ARMS2 Genotypes and Response to Nutritional Supplements. Ophthalmology, 2014, 121, 2173-2180.	5.2	86
97	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.	3.4	85
98	Chondroitinase ABC Treatment Enhances Synaptogenesis between Transplant and Host Neurons in Model of Retinal Degeneration. Cell Transplantation, 2007, 16, 493-503.	2.5	83
99	Pluripotent stem cell-derived retinal organoids for disease modeling and development of therapies. Stem Cells, 2020, 38, 1206-1215.	3.2	83
100	Expression of Photoreceptor-Specific Nuclear Receptor NR2E3 in Rod Photoreceptors of Fetal Human Retina. , 2004, 45, 2807.		82
101	Candidate Gene Association Study for Diabetic Retinopathy in Persons with Type 2 Diabetes: The Candidate Gene Association Resource (CARE). , 2011, 52, 7593.		82
102	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.	14.5	81
103	Differential expression of novel GsI± signal transduction protein cDNA species. Nucleic Acids Research, 1991, 19, 4725-4729.	14.5	80
104	Toll-like Receptor Polymorphisms and Age-Related Macular Degeneration. , 2008, 49, 1652.		79
105	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
106	Loss of RPGR glutamylation underlies the pathogenic mechanism of retinal dystrophy caused by <i>TTL5</i> mutations. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E2925-34.	7.1	79
107	Deep-learning-based prediction of late age-related macular degeneration progression. Nature Machine Intelligence, 2020, 2, 141-150.	16.0	79
108	Microarray analysis of gene expression in the aging human retina. Investigative Ophthalmology and Visual Science, 2002, 43, 2554-60.	3.3	79



#	ARTICLE	IF	CITATIONS
109	Protein-Truncation Mutations in the RP2 Gene in a North American Cohort of Families with X-Linked Retinitis Pigmentosa. <i>American Journal of Human Genetics</i> , 1999, 64, 897-900.	6.2	78
110	Centrosomal protein CP110 controls maturation of mother centriole during cilia biogenesis. <i>Development (Cambridge)</i> , 2016, 143, 1491-501.	2.5	78
111	Recruitment of the Rod Pathway by Cones in the Absence of Rods. <i>Journal of Neuroscience</i> , 2004, 24, 7576-7582.	3.6	77
112	Mutations in TOPORS Cause Autosomal Dominant Retinitis Pigmentosa with Perivascular Retinal Pigment Epithelium Atrophy. <i>American Journal of Human Genetics</i> , 2007, 81, 1098-1103.	6.2	77
113	Conditional knockdown of DNA methyltransferase 1 reveals a key role of retinal pigment epithelium integrity in photoreceptor outer segment morphogenesis. <i>Development (Cambridge)</i> , 2013, 140, 1330-1341.	2.5	77
114	Improved Retinal Organoid Differentiation by Modulating Signaling Pathways Revealed by Comparative Transcriptome Analyses with Development In Vivo. <i>Stem Cell Reports</i> , 2019, 13, 891-905.	4.8	77
115	Human retinopathy-associated ciliary protein retinitis pigmentosa GTPase regulator mediates cilia-dependent vertebrate development. <i>Human Molecular Genetics</i> , 2010, 19, 90-98.	2.9	76
116	Combining Cep290 and Mkks ciliopathy alleles in mice rescues sensory defects and restores ciliogenesis. <i>Journal of Clinical Investigation</i> , 2012, 122, 1233-1245.	8.2	75
117	Two Transcription Factors Can Direct Three Photoreceptor Outcomes from Rod Precursor Cells in Mouse Retinal Development. <i>Journal of Neuroscience</i> , 2011, 31, 11118-11125.	3.6	74
118	Rapid, Dynamic Activation of Müller Glial Stem Cell Responses in Zebrafish. , 2016, 57, 5148.		74
119	Biochemical Characterization and Subcellular Localization of the Mouse Retinitis Pigmentosa GTPase Regulator (mRpr). <i>Journal of Biological Chemistry</i> , 1998, 273, 19656-19663.	3.4	73
120	QRX, a novel homeobox gene, modulates photoreceptor gene expression. <i>Human Molecular Genetics</i> , 2004, 13, 1025-1040.	2.9	73
121	Genetic Studies of Age-related Macular Degeneration. <i>Ophthalmology</i> , 2012, 119, 2526-2536.	5.2	73
122	Localization of the Gene for Pigment Epithelium-Derived Factor (PEDF) to Chromosome 17p13.1 and Expression in Cultured Human Retinoblastoma Cells. <i>Genomics</i> , 1994, 19, 266-272.	2.9	70
123	Rd9 Is a Naturally Occurring Mouse Model of a Common Form of Retinitis Pigmentosa Caused by Mutations in RPGR-ORF15. <i>PLoS ONE</i> , 2012, 7, e35865.	2.5	69
124	Activation of Signaling Pathways and Stress-Response Genes in an Experimental Model of Retinal Detachment. , 2006, 47, 1691.		68
125	Deletion of Aryl Hydrocarbon Receptor AHR in Mice Leads to Subretinal Accumulation of Microglia and RPE Atrophy. , 2014, 55, 6031.		67
126	Genome-wide analysis of disease progression in age-related macular degeneration. <i>Human Molecular Genetics</i> , 2018, 27, 929-940.	2.9	67



#	ARTICLE	IF	CITATIONS
127	Expressed sequence tags and chromosomal localization of cDNA clones from a subtracted retinal pigment epithelium library. <i>Genomics</i> , 1992, 13, 873-876.	2.9	66
128	Subunit Dissociation and Diffusion Determine the Subcellular Localization of Rod and Cone Transducins. <i>Journal of Neuroscience</i> , 2007, 27, 5484-5494.	3.6	66
129	Ciliopathy-associated gene <i>Cc2d2a</i> promotes assembly of subdistal appendages on the mother centriole during cilia biogenesis. <i>Nature Communications</i> , 2014, 5, 4207.	12.8	66
130	Whole Exome Sequencing Reveals Mutations in Known Retinal Disease Genes in 33 out of 68 Israeli Families with Inherited Retinopathies. <i>Scientific Reports</i> , 2015, 5, 13187.	3.3	66
131	Pharmacologic fibroblast reprogramming into photoreceptors restores vision. <i>Nature</i> , 2020, 581, 83-88.	27.8	66
132	Dietary Nutrient Intake and Progression to Late Age-Related Macular Degeneration in the Age-Related Eye Disease Studies 1 and 2. <i>Ophthalmology</i> , 2021, 128, 425-442.	5.2	66
133	A Novel Locus (RP24) for X-linked Retinitis Pigmentosa Maps to Xq26-27. <i>American Journal of Human Genetics</i> , 1998, 63, 1439-1447.	6.2	65
134	Increased retinal mtDNA damage in the CFH variant associated with age-related macular degeneration. <i>Experimental Eye Research</i> , 2016, 145, 269-277.	2.6	64
135	The retinitis pigmentosa protein RP2 interacts with polycystin 2 and regulates cilia-mediated vertebrate development. <i>Human Molecular Genetics</i> , 2010, 19, 4330-4344.	2.9	63
136	A long-term efficacy study of gene replacement therapy for RPGR-associated retinal degeneration. <i>Human Molecular Genetics</i> , 2015, 24, 3956-3970.	2.9	63
137	A simple and efficient cDNA library subtraction procedure: isolation of human retina-specific cDNA clones. <i>Nucleic Acids Research</i> , 1991, 19, 1954-1954.	14.5	62
138	RP2 Phenotype and Pathogenetic Correlations in X-Linked Retinitis Pigmentosa. <i>JAMA Ophthalmology</i> , 2010, 128, 915.	2.4	62
139	Differential DNA methylation identified in the blood and retina of AMD patients. <i>Epigenetics</i> , 2015, 10, 698-707.	2.7	62
140	Analysis of the RPGR Gene in 11 Pedigrees with the Retinitis Pigmentosa Type 3 Genotype: Paucity of Mutations in the Coding Region but Splice Defects in Two Families. <i>American Journal of Human Genetics</i> , 1997, 61, 571-580.	6.2	61
141	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. <i>American Journal of Human Genetics</i> , 2000, 67, 1000-1003.	6.2	61
142	Clinical and Genetic Factors Associated with Progression of Geographic Atrophy Lesions in Age-Related Macular Degeneration. <i>PLoS ONE</i> , 2015, 10, e0126636.	2.5	61
143	Retinoic Acid Regulates the Expression of Photoreceptor Transcription Factor NRL. <i>Journal of Biological Chemistry</i> , 2006, 281, 27327-27334.	3.4	60
144	Primary cilia biogenesis and associated retinal ciliopathies. <i>Seminars in Cell and Developmental Biology</i> , 2021, 110, 70-88.	5.0	60

#	ARTICLE	IF	CITATIONS
145	Three-dimensional retinal organoids from mouse pluripotent stem cells mimic development with enhanced stratification and rod photoreceptor differentiation. <i>Molecular Vision</i> , 2016, 22, 1077-1094.	1.1	60
146	Autosomal Recessive Retinitis Pigmentosa with Early Macular Affection Caused by Premature Truncation in <i>PROM1</i> . , 2010, 51, 2656.		59
147	OTX2 loss causes rod differentiation defect in CRX-associated congenital blindness. <i>Journal of Clinical Investigation</i> , 2014, 124, 631-643.	8.2	59
148	Next generation sequencing technology and genomewide data analysis: Perspectives for retinal research. <i>Progress in Retinal and Eye Research</i> , 2016, 55, 1-31.	15.5	58
149	Retinitis Pigmentosa GTPase Regulator (RPGR) protein isoforms in mammalian retina: Insights into X-linked Retinitis Pigmentosa and associated ciliopathies. <i>Vision Research</i> , 2008, 48, 366-376.	1.4	57
150	<i>RPGR</i> -Associated Retinal Degeneration in Human X-Linked RP and a Murine Model. , 2012, 53, 5594.		56
151	Mechanisms of aging in senescence-accelerated mice. <i>Genome Biology</i> , 2005, 6, R48.	9.6	55
152	Associations of <i>CFHR1</i> and <i>CFHR3</i> deletion and a <i>CFH</i> SNP to age-related macular degeneration are not independent. <i>Nature Genetics</i> , 2010, 42, 553-555.	21.4	55
153	Loss of lysophosphatidylcholine acyltransferase 1 leads to photoreceptor degeneration in <i>rd11</i> mice. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2010, 107, 15523-15528.	7.1	55
154	Functional Analysis of the Rod Photoreceptor cGMP Phosphodiesterase $\beta$ -Subunit Gene Promoter. <i>Journal of Biological Chemistry</i> , 2004, 279, 19800-19807.	3.4	54
155	Ablation of the X-Linked Retinitis Pigmentosa 2 ( <i>Rp2</i> ) Gene in Mice Results in Opsin Mislocalization and Photoreceptor Degeneration. , 2013, 54, 4503.		54
156	Evaluation of the <i>ELOVL4</i> gene in patients with age-related macular degeneration. <i>Ophthalmic Genetics</i> , 2001, 22, 233-239.	1.2	53
157	Network constrained clustering for gene microarray data. <i>Bioinformatics</i> , 2005, 21, 4014-4020.	4.1	53
158	Canine RD3 mutation establishes rod-cone dysplasia type 2 ( <i>rcd2</i> ) as ortholog of human and murine <i>rd3</i> . <i>Mammalian Genome</i> , 2009, 20, 109-123.	2.2	53
159	Minireview: The Role of Nuclear Receptors in Photoreceptor Differentiation and Disease. <i>Molecular Endocrinology</i> , 2012, 26, 905-915.	3.7	53
160	Gene Therapy of Dominant CRX-Leber Congenital Amaurosis using Patient Stem Cell-Derived Retinal Organoids. <i>Stem Cell Reports</i> , 2021, 16, 252-263.	4.8	53
161	Altered Expression of Genes of the Bmp/Smad and Wnt/Calcium Signaling Pathways in the Cone-only <i>Nrl</i> <sup>-/-</sup> Mouse Retina, Revealed by Gene Profiling Using Custom cDNA Microarrays. <i>Journal of Biological Chemistry</i> , 2004, 279, 42211-42220.	3.4	52
162	An isoform of retinoid-related orphan receptor $\beta 2$ directs differentiation of retinal amacrine and horizontal interneurons. <i>Nature Communications</i> , 2013, 4, 1813.	12.8	52

#	ARTICLE	IF	CITATIONS
163	Rare and common variants in extracellular matrix gene Fibrillin 2 (FBN2) are associated with macular degeneration. <i>Human Molecular Genetics</i> , 2014, 23, 5827-5837.	2.9	52
164	Preservation of Cone Photoreceptors after a Rapid yet Transient Degeneration and Remodeling in Cone-Only <i>Nrl</i> Mouse Retina. <i>Journal of Neuroscience</i> , 2012, 32, 528-541.	3.6	51
165	The cellular and compartmental profile of mouse retinal glycolysis, tricarboxylic acid cycle, oxidative phosphorylation, and -P transferring kinases. <i>Molecular Vision</i> , 2016, 22, 847-85.	1.1	51
166	Inflammation in the pathogenesis of age-related macular degeneration. <i>British Journal of Ophthalmology</i> , 2008, 92, 448-450.	3.9	50
167	Two novel CRX mutant proteins causing autosomal dominant Leber congenital amaurosis interact differently with NRL. <i>Human Mutation</i> , 2010, 31, E1472-83.	2.5	50
168	Vision from next generation sequencing: Multi-dimensional genome-wide analysis for producing gene regulatory networks underlying retinal development, aging and disease. <i>Progress in Retinal and Eye Research</i> , 2015, 46, 1-30.	15.5	50
169	Maturation arrest in early postnatal sensory receptors by deletion of the miR-183/96/182 cluster in mouse. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017, 114, E4271-E4280.	7.1	50
170	TOPORS, implicated in retinal degeneration, is a cilia-centrosomal protein. <i>Human Molecular Genetics</i> , 2011, 20, 975-987.	2.9	49
171	Knockdown of Bardet-Biedl Syndrome Gene BBS9/PTHB1 Leads to Cilia Defects. <i>PLoS ONE</i> , 2012, 7, e34389.	2.5	49
172	Discordant Phenotypes in Fraternal Twins Having an Identical Mutation in Exon ORF15 of the RPGR Gene. <i>JAMA Ophthalmology</i> , 2008, 126, 379.	2.4	48
173	Ciliary Neurotrophic Factor Induces Genes Associated with Inflammation and Gliosis in the Retina: A Gene Profiling Study of Flow-Sorted, Müller Cells. <i>PLoS ONE</i> , 2011, 6, e20326.	2.5	48
174	High Throughput Screening of Co-Expressed Gene Pairs with Controlled False Discovery Rate (FDR) and Minimum Acceptable Strength (MAS). <i>Journal of Computational Biology</i> , 2005, 12, 1029-1045.	1.6	47
175	XIAP Therapy Increases Survival of Transplanted Rod Precursors in a Degenerating Host Retina. , 2011, 52, 1567.		47
176	Distinct nuclear localization patterns of DNA methyltransferases in developing and mature mammalian retina. <i>Journal of Comparative Neurology</i> , 2011, 519, 1914-1930.	1.6	47
177	Null and hypomorph <i>Prickle1</i> alleles in mice phenocopy human Robinow syndrome and disrupt signaling downstream of Wnt5a. <i>Biology Open</i> , 2014, 3, 861-870.	1.2	47
178	Lens-Specific Gene Recruitment of $\beta$ -Crystallin through Pax6, Nrl-Maf, and Brain Suppressor Sites. <i>Molecular and Cellular Biology</i> , 1998, 18, 2067-2076.	2.3	46
179	Rdh12 Activity and Effects on Retinoid Processing in the Murine Retina. <i>Journal of Biological Chemistry</i> , 2009, 284, 21468-21477.	3.4	46
180	A 32 kb Critical Region Excluding Y402H in CFH Mediates Risk for Age-Related Macular Degeneration. <i>PLoS ONE</i> , 2011, 6, e25598.	2.5	46

#	ARTICLE	IF	CITATIONS
181	Bivariate Analysis of Age-Related Macular Degeneration Progression Using Genetic Risk Scores. <i>Genetics</i> , 2017, 206, 119-133.	2.9	46
182	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. <i>Human Molecular Genetics</i> , 2011, 20, 4102-4115.	2.9	45
183	Prickle1 is expressed in distinct cell populations of the central nervous system and contributes to neuronal morphogenesis. <i>Human Molecular Genetics</i> , 2013, 22, 2234-2246.	2.9	45
184	The transcription-splicing protein NonO/p54nrb and three NonO-interacting proteins bind to distal enhancer region and augment rhodopsin expression. <i>Human Molecular Genetics</i> , 2014, 23, 2132-2144.	2.9	45
185	Evaluation and optimization of procedures for target labeling and hybridization of cDNA microarrays. <i>Molecular Vision</i> , 2002, 8, 130-7.	1.1	44
186	Cloning and functional expression of human retinal Kir2.4, a pH-sensitive inwardly rectifying K <sup>+</sup> channel. <i>American Journal of Physiology - Cell Physiology</i> , 2000, 279, C771-C784.	4.6	42
187	Hypomethylation of the IL17RC Promoter in Peripheral Blood Leukocytes Is Not A Hallmark of Age-Related Macular Degeneration. <i>Cell Reports</i> , 2013, 5, 1527-1535.	6.4	42
188	Retinopathy mutations in the bZIP protein NRL alter phosphorylation and transcriptional activity. <i>Human Mutation</i> , 2007, 28, 589-598.	2.5	41
189	Development and Plasticity of Outer Retinal Circuitry Following Genetic Removal of Horizontal Cells. <i>Journal of Neuroscience</i> , 2013, 33, 17847-17862.	3.6	41
190	Regulation of retinal progenitor expansion by Frizzled receptors: implications for microphthalmia and retinal coloboma. <i>Human Molecular Genetics</i> , 2012, 21, 1848-1860.	2.9	40
191	A secreted WNT-ligand-binding domain of FZD5 generated by a frameshift mutation causes autosomal dominant coloboma. <i>Human Molecular Genetics</i> , 2016, 25, 1382-1391.	2.9	40
192	Age-related macular degeneration-associated variants at chromosome 10q26 do not significantly alter ARMS2 and HTRA1 transcript levels in the human retina. <i>Molecular Vision</i> , 2010, 16, 1317-23.	1.1	40
193	Mouse eye gene microarrays for investigating ocular development and disease. <i>Vision Research</i> , 2002, 42, 463-470.	1.4	39
194	<i>Nrl</i> -Knockout Mice Deficient in Rpe65 Fail to Synthesize 11- <i>cis</i> Retinal and Cone Outer Segments. , 2008, 49, 1126.		39
195	Sumoylation of bZIP Transcription Factor NRL Modulates Target Gene Expression during Photoreceptor Differentiation. <i>Journal of Biological Chemistry</i> , 2010, 285, 25637-25644.	3.4	39
196	Retinal Organoids derived from hiPSCs of an AIPL1-LCA Patient Maintain Cytoarchitecture despite Reduced levels of Mutant AIPL1. <i>Scientific Reports</i> , 2020, 10, 5426.	3.3	39
197	Afferent Control of Horizontal Cell Morphology Revealed by Genetic Respecification of Rods and Cones. <i>Journal of Neuroscience</i> , 2007, 27, 3540-3547.	3.6	38
198	Long-term rescue of cone photoreceptor degeneration in retinitis pigmentosa 2 ( <i>RP2</i> )-knockout mice by gene replacement therapy. <i>Human Molecular Genetics</i> , 2015, 24, 6446-6458.	2.9	38

#	ARTICLE	IF	CITATIONS
199	Transcriptome analysis using next generation sequencing reveals molecular signatures of diabetic retinopathy and efficacy of candidate drugs. <i>Molecular Vision</i> , 2012, 18, 1123-46.	1.1	38
200	Retinal Histopathology of an XLRP Carrier with a Mutation in the RPGR Exon ORF15. <i>Experimental Eye Research</i> , 2002, 75, 431-443.	2.6	37
201	Interaction of retinal bZIP transcription factor NRL with Flt3-interacting zinc-finger protein Fiz1: possible role of Fiz1 as a transcriptional repressor. <i>Human Molecular Genetics</i> , 2003, 12, 365-373.	2.9	37
202	The Transcription Factor Neural Retina Leucine Zipper (NRL) Controls Photoreceptor-specific Expression of Myocyte Enhancer Factor Mef2c from an Alternative Promoter. <i>Journal of Biological Chemistry</i> , 2011, 286, 34893-34902.	3.4	35
203	Treatment Paradigms for Retinal and Macular Diseases Using 3-D Retina Cultures Derived From Human Reporter Pluripotent Stem Cell Lines. , 2016, 57, ORSF1.		35
204	Variegated yet non-random rod and cone photoreceptor disease patterns in RPGR-ORF15-associated retinal degeneration. <i>Human Molecular Genetics</i> , 2016, 25, 5444-5459.	2.9	35
205	EYS Mutations Causing Autosomal Recessive Retinitis Pigmentosa: Changes of Retinal Structure and Function with Disease Progression. <i>Genes</i> , 2017, 8, 178.	2.4	35
206	Distinct Signature of Altered Homeostasis in Aging Rod Photoreceptors: Implications for Retinal Diseases. <i>PLoS ONE</i> , 2010, 5, e13885.	2.5	35
207	Charon BS (+) and (â”), versatile ð» phage vectors for constructing directional cDNA libraries and their efficient transfer to plasmids. <i>Nucleic Acids Research</i> , 1988, 16, 8739-8739.	14.5	34
208	Annotation and analysis of 10,000 expressed sequence tags from developing mouse eye and adult retina. <i>Genome Biology</i> , 2003, 4, R65.	9.6	34
209	The Minimal Transactivation Domain of the Basic Motif-Leucine Zipper Transcription Factor NRL Interacts with TATA-binding Protein. <i>Journal of Biological Chemistry</i> , 2004, 279, 47233-47241.	3.4	34
210	Bi-allelic Truncating Mutations in CEP78 , Encoding Centrosomal Protein 78, Cause Cone-Rod Degeneration with Sensorineural Hearing Loss. <i>American Journal of Human Genetics</i> , 2016, 99, 777-784.	6.2	34
211	Adherence to the Mediterranean Diet and Progression to Late Age-Related Macular Degeneration in the Age-Related Eye Disease Studies 1 and 2. <i>Ophthalmology</i> , 2020, 127, 1515-1528.	5.2	34
212	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. <i>Human Molecular Genetics</i> , 1994, 3, 1281-1286.	2.9	33
213	Genetic association study of age-related macular degeneration in the Spanish population. <i>Acta Ophthalmologica</i> , 2011, 89, e12-e22.	1.1	33
214	Combinatorial Regulation of Photoreceptor Differentiation Factor, Neural Retina Leucine Zipper Gene Nrl, Revealed by in Vivo Promoter Analysis. <i>Journal of Biological Chemistry</i> , 2011, 286, 28247-28255.	3.4	33
215	Genetic architecture of retinal and macular degenerative diseases: the promise and challenges of next-generation sequencing. <i>Genome Medicine</i> , 2013, 5, 84.	8.2	33
216	Synergistically acting agonists and antagonists of G proteinâ€‘coupled receptors prevent photoreceptor cell degeneration. <i>Science Signaling</i> , 2016, 9, ra74.	3.6	33

#	ARTICLE	IF	CITATIONS
217	Transcriptome-based molecular staging of human stem cell-derived retinal organoids uncovers accelerated photoreceptor differentiation by 9-cis retinal. <i>Molecular Vision</i> , 2019, 25, 663-678.	1.1	33
218	De Novo Mutation in the RP1 Gene (Arg677Ter) Associated with Retinitis Pigmentosa. , 2003, 44, 3593.		32
219	Whole Exome Sequencing Reveals GUCY2D as a Major Gene Associated With Cone and Cone-Rod Dystrophy in Israel. <i>Investigative Ophthalmology and Visual Science</i> , 2015, 56, 420-430.	3.3	32
220	Mini and customized low-cost bioreactors for optimized high-throughput generation of tissue organoids. <i>Stem Cell Investigation</i> , 2018, 5, 33-33.	3.0	32
221	A CEP290 C-Terminal Domain Complements the Mutant CEP290 of Rd16 Mice In Trans and Rescues Retinal Degeneration. <i>Cell Reports</i> , 2018, 25, 611-623.e6.	6.4	32
222	Generation of hydrogen peroxide on oxidation of NADH by hepatic plasma membranes. <i>Journal of Bioenergetics and Biomembranes</i> , 1981, 13, 241-253.	2.3	31
223	Transcriptome analysis of the retina. <i>Genome Biology</i> , 2002, 3, reviews1022.1.	9.6	31
224	Phenotypic Conservation in Patients With X-Linked Retinitis Pigmentosa Caused by <i>RPGR</i> Mutations. <i>JAMA Ophthalmology</i> , 2013, 131, 1016.	2.5	31
225	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). <i>Experimental Eye Research</i> , 1991, 52, 549-561.	2.6	30
226	A Deep Phenotype Association Study Reveals Specific Phenotype Associations with Genetic Variants in Age-related Macular Degeneration. <i>Ophthalmology</i> , 2018, 125, 559-568.	5.2	30
227	Transgenic Mice Expressing Cre-Recombinase Specifically in M- or S-Cone Photoreceptors. , 2004, 45, 42.		29
228	Madeline 2.0 PDE: a new program for local and web-based pedigree drawing. <i>Bioinformatics</i> , 2007, 23, 1854-1856.	4.1	29
229	RNA Biology in Retinal Development and Disease. <i>Trends in Genetics</i> , 2018, 34, 341-351.	6.7	29
230	Association of Smoking, Alcohol Consumption, Blood Pressure, Body Mass Index, and Glycemic Risk Factors With Age-Related Macular Degeneration. <i>JAMA Ophthalmology</i> , 2021, 139, 1299.	2.5	29
231	Human bZIP Transcription Factor Gene <i>NRL</i> : Structure, Genomic Sequence, and Fine Linkage Mapping at 14q11.2 and Negative Mutation Analysis in Patients with Retinal Degeneration. <i>Genomics</i> , 1997, 45, 395-401.	2.9	28
232	RPGR-containing protein complexes in syndromic and non-syndromic retinal degeneration due to ciliary dysfunction. <i>Journal of Genetics</i> , 2009, 88, 399-407.	0.7	28
233	Assessment of Novel Genome-Wide Significant Gene Loci and Lesion Growth in Geographic Atrophy Secondary to Age-Related Macular Degeneration. <i>JAMA Ophthalmology</i> , 2019, 137, 867.	2.5	28
234	Adherence to a Mediterranean diet and cognitive function in the Age-Related Eye Disease Studies 1 & 2. <i>Alzheimer's and Dementia</i> , 2020, 16, 831-842.	0.8	28



#	ARTICLE	IF	CITATIONS
235	Aging of the Retina: Molecular and Metabolic Turbulences and Potential Interventions. Annual Review of Vision Science, 2021, 7, 633-664.	4.4	28
236	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
237	Molecular Characterization of the Murine Neural Retina Leucine Zipper Gene, Nrl. Genomics, 1993, 18, 216-222.	2.9	27
238	Long-term follow-up of a family with dominant X-linked retinitis pigmentosa. Eye, 2010, 24, 764-774.	2.1	27
239	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
240	Family-based exome sequencing identifies rare coding variants in age-related macular degeneration. Human Molecular Genetics, 2020, 29, 2022-2034.	2.9	26
241	Proneural genes define ground-state rules to regulate neurogenic patterning and cortical folding. Neuron, 2021, 109, 2847-2863.e11.	8.1	26
242	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.	1.6	26
243	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.	1.2	25
244	Multimomics 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.	2.9	25
245	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
246	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.	5.2	23
247	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
248	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
249	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
250	FGF19 Exhibits Neuroprotective Effects on Adult Mammalian Photoreceptors In Vitro. , 2008, 49, 1696.		22
251	Feedback Induction of a Photoreceptor-specific Isoform of Retinoid-related Orphan Nuclear Receptor $\hat{1}^2$ by the Rod Transcription Factor NRL. Journal of Biological Chemistry, 2014, 289, 32469-32480.	3.4	22
252	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



#	ARTICLE	IF	CITATIONS
253	Aberrant RNA splicing is the major pathogenic effect in a knock-in mouse model of the dominantly inherited c.1430A>G human <i>RPE65</i> mutation. <i>Human Mutation</i> , 2019, 40, 426-443.	2.5	22
254	A mega-analysis of expression quantitative trait loci in retinal tissue. <i>PLoS Genetics</i> , 2020, 16, e1008934.	3.5	22
255	A simple and efficient method for generating human retinal organoids. <i>Molecular Vision</i> , 2020, 26, 97-105.	1.1	21
256	Multicriteria Gene Screening for Analysis of Differential Expression with DNA Microarrays. <i>Eurasip Journal on Advances in Signal Processing</i> , 2004, 2004, 1.	1.7	20
257	<i>Tbx2a</i> Modulates Switching of <i>RH2</i> and <i>LWS</i> Opsin Gene Expression. <i>Molecular Biology and Evolution</i> , 2020, 37, 2002-2014.	8.9	20
258	Genome-wide Profiling Identifies DNA Methylation Signatures of Aging in Rod Photoreceptors Associated with Alterations in Energy Metabolism. <i>Cell Reports</i> , 2020, 31, 107525.	6.4	20
259	Phenotype Associated With Mutation in the Recently Identified Autosomal Dominant Retinitis Pigmentosa <i>KLHL7</i> Gene. <i>JAMA Ophthalmology</i> , 2010, 128, 772.	2.4	19
260	A role for prenylated rab acceptor 1 in vertebrate photoreceptor development. <i>BMC Neuroscience</i> , 2012, 13, 152.	1.9	19
261	Mouse model of human <i>RPE65</i> P25L hypomorph resembles wild type under normal light rearing but is fully resistant to acute light damage. <i>Human Molecular Genetics</i> , 2015, 24, 4417-4428.	2.9	19
262	Regulation of Noncoding Transcriptome in Developing Photoreceptors by Rod Differentiation Factor <i>NRL</i> . , 2017, 58, 4422.		19
263	Increasing evidence for syndromic phenotypes associated with <i>RPGR</i> mutations. <i>American Journal of Ophthalmology</i> , 2004, 137, 785-786.	3.3	19
264	Genetic components in diabetic retinopathy. <i>Indian Journal of Ophthalmology</i> , 2016, 64, 55.	1.1	19
265	Gene expression profile of native human retinal pigment epithelium. <i>Investigative Ophthalmology and Visual Science</i> , 2002, 43, 603-7.	3.3	19
266	Retinal histopathology of an <i>XLRP</i> carrier with a mutation in the <i>RPGR</i> exon ORF15. <i>Experimental Eye Research</i> , 2002, 75, 431-43.	2.6	19
267	<i>NRL</i> S50T mutation and the importance of "founder effects"™ in inherited retinal dystrophies. <i>European Journal of Human Genetics</i> , 2000, 8, 783-787.	2.8	18
268	Restoration of <i>RPGR</i> expression in vivo using CRISPR/Cas9 gene editing. <i>Gene Therapy</i> , 2022, 29, 81-93.	4.5	18
269	Integration of genomics and transcriptomics predicts diabetic retinopathy susceptibility genes. <i>ELife</i> , 2020, 9, .	6.0	18
270	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> . <i>Human Mutation</i> , 2015, 36, 836-841.	2.5	17

#	ARTICLE	IF	CITATIONS
271	Molecular Diagnosis of Inherited Retinal Diseases in Indigenous African Populations by Whole-Exome Sequencing. , 2016, 57, 6374.		17
272	Cone-rod homeobox CRX controls presynaptic active zone formation in photoreceptors of mammalian retina. Human Molecular Genetics, 2018, 27, 3555-3567.	2.9	17
273	Characterization and Sequence Analysis of the Human Homeobox-Containing Gene GBX2. Genomics, 1996, 31, 335-342.	2.9	16
274	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
275	Preventing polyglutamine-induced activation of c-Jun delays neuronal dysfunction in a mouse model of SCA7 retinopathy. Neurobiology of Disease, 2007, 25, 571-581.	4.4	15
276	Global expression profiling of peripheral Qa-1 <sup>hi</sup> restricted CD8 <sup>hi</sup> +TCR <sup>hi</sup> + regulatory T cells reveals innate-like features: Implications for immune-regulatory repertoire. Human Immunology, 2012, 73, 214-222.	2.4	15
277	No CFH or ARMS2 Interaction with Omega-3 Fatty Acids, Low versus High Zinc, or $\beta$ -Carotene versus Lutein and Zeaxanthin on Progression of Age-Related Macular Degeneration in the Age-Related Eye Disease Study 2. Ophthalmology, 2019, 126, 1541-1548.	5.2	15
278	The combination of whole-exome sequencing and clinical analysis allows better diagnosis of rare syndromic retinal dystrophies. Acta Ophthalmologica, 2019, 97, e877-e886.	1.1	15
279	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
280	Protective Gene Expression Changes Elicited by an Inherited Defect in Photoreceptor Structure. PLoS ONE, 2012, 7, e31371.	2.5	14
281	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
282	Geographic atrophy in age-related macular degeneration and TLR3. New England Journal of Medicine, 2009, 360, 2254-5; author reply 2255-6.	27.0	14
283	The golden era of ocular disease gene discovery: Race to the finish. Clinical Genetics, 2013, 84, 99-101.	2.0	13
284	A Novel ARL3 Gene Mutation Associated With Autosomal Dominant Retinal Degeneration. Frontiers in Cell and Developmental Biology, 2021, 9, 720782.	3.7	13
285	An optimized protocol for retina single-cell RNA sequencing. Molecular Vision, 2020, 26, 705-717.	1.1	13
286	Gene-Based Association Analysis for Censored Traits Via Fixed Effect Functional Regressions. Genetic Epidemiology, 2016, 40, 133-143.	1.3	12
287	Analysis of RP2 and RPCR Mutations in Five X-Linked Chinese Families with Retinitis Pigmentosa. Scientific Reports, 2017, 7, 44465.	3.3	12
288	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

#	ARTICLE	IF	CITATIONS
289	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
290	Neural retina-specific leucine zipper gene NRL (D14S46E) maps to human chromosome 14q11.1-q11.2. Genomics, 1992, 14, 491-492.	2.9	11
291	Five novelRPGR mutations in families with X-linked retinitis pigmentosa. Human Mutation, 2001, 17, 151-151.	2.5	11
292	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
293	pSH4: a mammalian cDNA expression vector. Nucleic Acids Research, 1990, 18, 3668-3668.	14.5	10
294	A New 2â€œBase Pair Deletion in the RPGR Gene in a Black Family With X-Linked Retinitis Pigmentosa. JAMA Ophthalmology, 1998, 116, 213-8.	2.4	10
295	Seeing the Unseen: Microarray-Based Gene Expression Profiling in Vision. , 2004, 45, 2457.		10
296	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
297	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.	1.1	9
298	Characterization of New Transcripts Enriched in the Mouse Retina and Identification of Candidate Retinal Disease Genes. , 2004, 45, 3313.		9
299	Gene discovery using Pareto depth sampling distributions. Journal of the Franklin Institute, 2004, 341, 55-75.	3.4	9
300	Genetic Analysis of the Rhodopsin Gene Identifies a Mosaic Dominant Retinitis Pigmentosa Mutation in a Healthy Individual. , 2016, 57, 940.		9
301	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
302	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
303	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
304	Chromosomal localization and cDNA sequence of humanralB, a GTP binding protein. Somatic Cell and Molecular Genetics, 1990, 16, 407-410.	0.7	7
305	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
306	Genetics and therapy for pediatric eye diseases. EBioMedicine, 2021, 67, 103360.	6.1	7

#	ARTICLE	IF	CITATIONS
307	Exome Sequencing: Capture and Sequencing of All Human Coding Regions for Disease Gene Discovery. <i>Methods in Molecular Biology</i> , 2012, 884, 335-351.	0.9	7
308	Senile panretinal cone dysfunction in age-related macular degeneration (AMD): a report of 52 amd patients compared to age-matched controls. <i>Transactions of the American Ophthalmological Society</i> , 2006, 104, 232-40.	1.4	7
309	Divergent Effects of HSP70 Overexpression in Photoreceptors During Inherited Retinal Degeneration. , 2020, 61, 25.		6
310	GATD3A, a mitochondrial deglycase with evolutionary origins from gammaproteobacteria, restricts the formation of advanced glycation end products. <i>BMC Biology</i> , 2022, 20, 68.	3.8	6
311	Nicotinamide Promotes Formation of Retinal Organoids From Human Pluripotent Stem Cells via Enhanced Neural Cell Fate Commitment. <i>Frontiers in Cellular Neuroscience</i> , 0, 16, .	3.7	6
312	Molecular dissection of cone photoreceptor-enriched genes encoding transmembrane and secretory proteins. <i>Journal of Neuroscience Research</i> , 2019, 97, 16-28.	2.9	5
313	Glycogen Synthase Kinase 3 Regulates the Genesis of Displaced Retinal Ganglion Cells3. <i>ENeuro</i> , 2021, 8, ENEURO.0171-21.2021.	1.9	5
314	Expression of deubiquitinating enzyme genes in the developing mammal retina. <i>Molecular Vision</i> , 2019, 25, 800-813.	1.1	5
315	A unique -associated variant in a Georgian Jewish family with probable North Carolina macular dystrophy and the possible contribution of a unique variant. <i>Molecular Vision</i> , 2020, 26, 299-310.	1.1	5
316	Clinical expression of X-linked retinitis pigmentosa in a Swedish family with the RP2 genotype. <i>Ophthalmic Genetics</i> , 1998, 19, 187-196.	1.2	4
317	Pias3 is necessary for dorso-ventral patterning and visual response of retinal cones but is not required for rod photoreceptor differentiation. <i>Biology Open</i> , 2017, 6, 881-890.	1.2	4
318	HIPRO: A High-Efficiency, Hypoxia-Induced Protocol for Generation of Photoreceptors in Retinal Organoids from Mouse Pluripotent Stem Cells. <i>STAR Protocols</i> , 2020, 1, 100018.	1.2	4
319	Retinal pigment epithelium transcriptome analysis in chronic smoking reveals a suppressed innate immune response and activation of differentiation pathways. <i>Free Radical Biology and Medicine</i> , 2020, 156, 176-189.	2.9	4
320	Retinal Phenotype of an X-Linked Pseudo-usher Syndrome in Association with the G173R Mutation in the RPGR Gene. <i>Advances in Experimental Medicine and Biology</i> , 2008, 613, 221-227.	1.6	4
321	X-Linked Retinitis Pigmentosa: Current Status. , 2001, , 11-22.		4
322	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. <i>Human Molecular Genetics</i> , 2022, 31, 3914-3933.	2.9	4
323	What's in a Name?RPGRMutations Redefine the Genetic and Phenotypic Landscape in Retinal Degenerative Diseases. , 2013, 54, 1417.		3
324	Determination of Mitochondrial Oxygen Consumption in the Retina Ex Vivo: Applications for Retinal Disease. <i>Methods in Molecular Biology</i> , 2018, 1753, 167-177.	0.9	3

#	ARTICLE	IF	CITATIONS
325	SSBP1 faux pas in mitonuclear tango causes optic neuropathy. <i>Journal of Clinical Investigation</i> , 2019, 130, 62-64.	8.2	3
326	A novel exome probe set captures phototransduction genes across birds (Aves) enabling efficient analysis of vision evolution. <i>Molecular Ecology Resources</i> , 2022, 22, 587-601.	4.8	3
327	Mutations associated with retinopathies alter mitogen-activated protein kinase-induced phosphorylation of neural retina leucine zipper. <i>Molecular Vision</i> , 2007, 13, 1114-20.	1.1	3
328	Mapping of the neural retina leucine zipper gene, Nrl, to mouse Chromosome 14. <i>Mammalian Genome</i> , 1993, 4, 618-620.	2.2	2
329	λ-SHK and λ-AASV: phage vectors for efficient cDNA cloning and expression in mammalian cells. <i>Gene</i> , 1993, 123, 287-288.	2.2	2
330	Dinucleotide repeat polymorphism at the DXS977 locus. <i>Human Molecular Genetics</i> , 1994, 3, 1030-1030.	2.9	2
331	Determination of Posttranslational Modifications of Photoreceptor Differentiation Factor NRL: Focus on SUMOylation. <i>Methods in Molecular Biology</i> , 2012, 884, 353-361.	0.9	2
332	Making Biological Sense of Genetic Studies of Age-Related Macular Degeneration. <i>Advances in Experimental Medicine and Biology</i> , 2021, 1256, 201-219.	1.6	2
333	Construction of Directional cDNA Libraries from Human Retinal Tissue/Cells and Their Enrichment for Specific Genes Using an Efficient Subtraction Procedure. <i>Methods in Neurosciences</i> , 1993, , 285-300.	0.5	2
334	Dinucleotide polymorphism at the DXS1178 locus is tightly linked to PGK1 at Xq13. <i>Human Genetics</i> , 1995, 95, 467-8.	3.8	1
335	[11]Isolation of candidate genes for inherited diseases: Application to X-linked retinal degenerations. <i>Methods in Molecular Genetics</i> , 1996, 8, 207-228.	0.6	0
336	Reply. <i>Ophthalmology</i> , 2020, 127, e19-e20.	5.2	0
337	Combining Cep290 and Mlks ciliopathy alleles in mice rescues sensory defects and restores ciliogenesis. <i>Journal of Clinical Investigation</i> , 2012, 122, 3025-3025.	8.2	0
338	Photoreceptor Degeneration: Molecular Mechanisms of Photoreceptor Degeneration. , 2014, , 275-308.		0
339	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
340	Photoreceptor Development: Early Steps/Fate . , 2017, , .		0
341	Title is missing!. , 2020, 16, e1009259.		0
342	Title is missing!. , 2020, 16, e1009259.		0

#	ARTICLE	IF	CITATIONS
343	Title is missing!. , 2020, 16, e1009259.		0
344	Title is missing!. , 2020, 16, e1009259.		0
345	Title is missing!.. , 2020, 16, e1009259.		0
346	Title is missing!.. , 2020, 16, e1009259.		0