

# Anand Swaroop

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

347 papers	26,240 citations	80 h-index	151 g-index
363 ext. papers	30,821 ext. citations	8.7 avg, IF	6.52 L-index

#	Paper	IF	Citations
347	Multi-omics analyses reveal early metabolic imbalance and mitochondrial stress in neonatal photoreceptors leading to cell death in Pde6brd1/rd1 mouse model of retinal degeneration.. <i>Human Molecular Genetics</i> , <b>2022</b> ,	5.6	2
346	GATD3A, a mitochondrial deglycase with evolutionary origins from gammaproteobacteria, restricts the formation of advanced glycation end products.. <i>BMC Biology</i> , <b>2022</b> , 20, 68	7.3	0
345	Association of Smoking, Alcohol Consumption, Blood Pressure, Body Mass Index, and Glycemic Risk Factors With Age-Related Macular Degeneration: A Mendelian Randomization Study. <i>JAMA Ophthalmology</i> , <b>2021</b> ,	3.9	2
344	Genetics and therapy for pediatric eye diseases. <i>EBioMedicine</i> , <b>2021</b> , 67, 103360	8.8	0
343	Restoration of RPGR expression in vivo using CRISPR/Cas9 gene editing. <i>Gene Therapy</i> , <b>2021</b> ,	4	4
342	Primary cilia biogenesis and associated retinal ciliopathies. <i>Seminars in Cell and Developmental Biology</i> , <b>2021</b> , 110, 70-88	7.5	17
341	Dietary Nutrient Intake and Progression to Late Age-Related Macular Degeneration in the Age-Related Eye Disease Studies 1 and 2. <i>Ophthalmology</i> , <b>2021</b> , 128, 425-442	7.3	21
340	Gene Therapy of Dominant CRX-Leber Congenital Amaurosis using Patient Stem Cell-Derived Retinal Organoids. <i>Stem Cell Reports</i> , <b>2021</b> , 16, 252-263	8	13
339	Genome-Wide Association Studies-Based Machine Learning for Prediction of Age-Related Macular Degeneration Risk. <i>Translational Vision Science and Technology</i> , <b>2021</b> , 10, 29	3.3	4
338	A Novel Gene Mutation Associated With Autosomal Dominant Retinal Degeneration. <i>Frontiers in Cell and Developmental Biology</i> , <b>2021</b> , 9, 720782	5.7	2
337	Glycogen Synthase Kinase 3 Regulates the Genesis of Displaced Retinal Ganglion Cells3. <i>ENeuro</i> , <b>2021</b> , 8,	3.9	1
336	Aging of the Retina: Molecular and Metabolic Turbulences and Potential Interventions. <i>Annual Review of Vision Science</i> , <b>2021</b> , 7, 633-664	8.2	4
335	Proneural genes define ground-state rules to regulate neurogenic patterning and cortical folding. <i>Neuron</i> , <b>2021</b> , 109, 2847-2863.e11	13.9	5
334	Making Biological Sense of Genetic Studies of Age-Related Macular Degeneration. <i>Advances in Experimental Medicine and Biology</i> , <b>2021</b> , 1256, 201-219	3.6	1
333	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> , <b>2020</b> , 127, 1515-1528	7.3	13
332	Pluripotent stem cell-derived retinal organoids for disease modeling and development of therapies. <i>Stem Cells</i> , <b>2020</b> , 38, 1206-1215	5.8	37
331	Tbx2a Modulates Switching of RH2 and LWS Opsin Gene Expression. <i>Molecular Biology and Evolution</i> , <b>2020</b> , 37, 2002-2014	8.3	9

330	Retinal Organoids derived from hiPSCs of an AIPL1-LCA Patient Maintain Cytoarchitecture despite Reduced levels of Mutant AIPL1. <i>Scientific Reports</i> , <b>2020</b> , 10, 5426	4.9	17
329	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> , <b>2020</b> , 156, 176-189	7.8	2
328	Soy Protein Nanofiber Scaffolds for Uniform Maturation of Human Induced Pluripotent Stem Cell-Derived Retinal Pigment Epithelium. <i>Tissue Engineering - Part C: Methods</i> , <b>2020</b> , 26, 433-446	2.9	4
327	Reply. <i>Ophthalmology</i> , <b>2020</b> , 127, e19-e20	7.3	
326	Deep-learning-based Prediction of Late Age-Related Macular Degeneration Progression. <i>Nature Machine Intelligence</i> , <b>2020</b> , 2, 141-150	22.5	35
325	Family-based exome sequencing identifies rare coding variants in age-related macular degeneration. <i>Human Molecular Genetics</i> , <b>2020</b> , 29, 2022-2034	5.6	9
324	Genome-wide Profiling Identifies DNA Methylation Signatures of Aging in Rod Photoreceptors Associated with Alterations in Energy Metabolism. <i>Cell Reports</i> , <b>2020</b> , 31, 107525	10.6	6
323	SSBP1 faux pas in mitonuclear tango causes optic neuropathy. <i>Journal of Clinical Investigation</i> , <b>2020</b> , 130, 62-64	15.9	2
322	Integration of genomics and transcriptomics predicts diabetic retinopathy susceptibility genes. <i>ELife</i> , <b>2020</b> , 9,	8.9	5
321	A simple and efficient method for generating human retinal organoids. <i>Molecular Vision</i> , <b>2020</b> , 26, 97-105	5.3	14
320	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> , <b>2020</b> , 26, 299-310	2.3	3
319	An optimized protocol for retina single-cell RNA sequencing. <i>Molecular Vision</i> , <b>2020</b> , 26, 705-717	2.3	5
318	Loss of endocytosis-associated RabGEF1 causes aberrant morphogenesis and altered autophagy in photoreceptors leading to retinal degeneration. <i>PLoS Genetics</i> , <b>2020</b> , 16, e1009259	6	4
317	HIPRO: A High-Efficiency, Hypoxia-Induced Protocol for Generation of Photoreceptors in Retinal Organoids from Mouse Pluripotent Stem Cells. <i>STAR Protocols</i> , <b>2020</b> , 1, 100018-100018	1.4	2
316	Accelerated Development of Rod Photoreceptors in Retinal Organoids Derived from Human Pluripotent Stem Cells by Supplementation with 9- Retinal. <i>STAR Protocols</i> , <b>2020</b> , 1, 100033-100033	1.4	3
315	Divergent Effects of HSP70 Overexpression in Photoreceptors During Inherited Retinal Degeneration <b>2020</b> , 61, 25		2
314	A mega-analysis of expression quantitative trait loci in retinal tissue. <i>PLoS Genetics</i> , <b>2020</b> , 16, e1008934	6	5
313	Pharmacologic fibroblast reprogramming into photoreceptors restores vision. <i>Nature</i> , <b>2020</b> , 581, 83-88	50.4	37

312	Adherence to a Mediterranean diet and cognitive function in the Age-Related Eye Disease Studies 1 & 2. <i>Alzheimer's and Dementia</i> , <b>2020</b> , 16, 831-842	1.2	14
311	Loss of endocytosis-associated RabGEF1 causes aberrant morphogenesis and altered autophagy in photoreceptors leading to retinal degeneration <b>2020</b> , 16, e1009259		
310	Loss of endocytosis-associated RabGEF1 causes aberrant morphogenesis and altered autophagy in photoreceptors leading to retinal degeneration <b>2020</b> , 16, e1009259		
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306	Loss of endocytosis-associated RabGEF1 causes aberrant morphogenesis and altered autophagy in photoreceptors leading to retinal degeneration <b>2020</b> , 16, e1009259		
305	Assessment of Novel Genome-Wide Significant Gene Loci and Lesion Growth in Geographic Atrophy Secondary to Age-Related Macular Degeneration. <i>JAMA Ophthalmology</i> , <b>2019</b> , 137, 867-876	3.9	15
304	The combination of whole-exome sequencing and clinical analysis allows better diagnosis of rare syndromic retinal dystrophies. <i>Acta Ophthalmologica</i> , <b>2019</b> , 97, e877-e886	3.7	11
303	Retinal transcriptome and eQTL analyses identify genes associated with age-related macular degeneration. <i>Nature Genetics</i> , <b>2019</b> , 51, 606-610	36.3	93
302	Retinal disease in ciliopathies: Recent advances with a focus on stem cell-based therapies. <i>Translational Science of Rare Diseases</i> , <b>2019</b> , 4, 97-115	3.3	11
301	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: Age-Related Eye Disease Study 2 Report No. 18. <i>Ophthalmology</i> , <b>2019</b> , 126, 1511-1518	7.3	12
300	Improved Retinal Organoid Differentiation by Modulating Signaling Pathways Revealed by Comparative Transcriptome Analyses with Development In Vivo. <i>Stem Cell Reports</i> , <b>2019</b> , 13, 891-905	8	49
299	Association of age-related macular degeneration with complement activation products, smoking, and single nucleotide polymorphisms in South Carolinians of European and African descent. <i>Molecular Vision</i> , <b>2019</b> , 25, 79-92	2.3	11
298	Transcriptome-based molecular staging of human stem cell-derived retinal organoids uncovers accelerated photoreceptor differentiation by 9-cis retinal. <i>Molecular Vision</i> , <b>2019</b> , 25, 663-678	2.3	23
297	Expression of deubiquitinating enzyme genes in the developing mammal retina. <i>Molecular Vision</i> , <b>2019</b> , 25, 800-813	2.3	3
296	Targeted deletion of an NRL- and CRX-regulated alternative promoter specifically silences FERM and PDZ domain containing 1 (Frmpd1) in rod photoreceptors. <i>Human Molecular Genetics</i> , <b>2019</b> , 28, 804-817	5.6	5
295	Aberrant RNA splicing is the major pathogenic effect in a knock-in mouse model of the dominantly inherited c.1430A>G human RPE65 mutation. <i>Human Mutation</i> , <b>2019</b> , 40, 426-443	4.7	14

294	Molecular dissection of cone photoreceptor-enriched genes encoding transmembrane and secretory proteins. <i>Journal of Neuroscience Research</i> , <b>2019</b> , 97, 16-28	4.4	4
293	RNA Biology in Retinal Development and Disease. <i>Trends in Genetics</i> , <b>2018</b> , 34, 341-351	8.5	21
292	Genome-wide analysis of disease progression in age-related macular degeneration. <i>Human Molecular Genetics</i> , <b>2018</b> , 27, 929-940	5.6	37
291	Accelerated and Improved Differentiation of Retinal Organoids from Pluripotent Stem Cells in Rotating-Wall Vessel Bioreactors. <i>Stem Cell Reports</i> , <b>2018</b> , 10, 300-313	8	114
290	Epigenetic control of gene regulation during development and disease: A view from the retina. <i>Progress in Retinal and Eye Research</i> , <b>2018</b> , 65, 1-27	20.5	60
289	Determination of Mitochondrial Oxygen Consumption in the Retina Ex Vivo: Applications for Retinal Disease. <i>Methods in Molecular Biology</i> , <b>2018</b> , 1753, 167-177	1.4	2
288	A Deep Phenotype Association Study Reveals Specific Phenotype Associations with Genetic Variants in Age-related Macular Degeneration: Age-Related Eye Disease Study 2 (AREDS2) Report No. 14. <i>Ophthalmology</i> , <b>2018</b> , 125, 559-568	7.3	19
287	Progression of Geographic Atrophy in Age-related Macular Degeneration: AREDS2 Report Number 16. <i>Ophthalmology</i> , <b>2018</b> , 125, 1913-1928	7.3	71
286	Cone-rod homeobox CRX controls presynaptic active zone formation in photoreceptors of mammalian retina. <i>Human Molecular Genetics</i> , <b>2018</b> , 27, 3555-3567	5.6	11
285	Patient iPSC-derived neural stem cells exhibit phenotypes in concordance with the clinical severity of mucopolysaccharidosis I. <i>Human Molecular Genetics</i> , <b>2018</b> , 27, 3612-3626	5.6	17
284	Association of Rare Predicted Loss-of-Function Variants in Cellular Pathways with Sub-Phenotypes in Age-Related Macular Degeneration. <i>Ophthalmology</i> , <b>2018</b> , 125, 398-406	7.3	7
283	A CEP290 C-Terminal Domain Complements the Mutant CEP290 of Rd16 Mice In Trans and Rescues Retinal Degeneration. <i>Cell Reports</i> , <b>2018</b> , 25, 611-623.e6	10.6	22
282	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> , <b>2017</b> , 114, E4271-E4280	11.5	35
281	Bivariate Analysis of Age-Related Macular Degeneration Progression Using Genetic Risk Scores. <i>Genetics</i> , <b>2017</b> , 206, 119-133	4	31
280	REEP6 mediates trafficking of a subset of Clathrin-coated vesicles and is critical for rod photoreceptor function and survival. <i>Human Molecular Genetics</i> , <b>2017</b> , 26, 2218-2230	5.6	16
279	Analysis of RP2 and RPGR Mutations in Five X-Linked Chinese Families with Retinitis Pigmentosa. <i>Scientific Reports</i> , <b>2017</b> , 7, 44465	4.9	8
278	Nrl knockdown by AAV-delivered CRISPR/Cas9 prevents retinal degeneration in mice. <i>Nature Communications</i> , <b>2017</b> , 8, 14716	17.4	176
277	Regulation of Noncoding Transcriptome in Developing Photoreceptors by Rod Differentiation Factor NRL <b>2017</b> , 58, 4422-4435		15

276	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> , <b>2017</b> , 6, 881-890	2.2	4
275	Molecular Anatomy of the Developing Human Retina. <i>Developmental Cell</i> , <b>2017</b> , 43, 763-779.e4	10.2	125
274	In Vitro Modeling Using Ciliopathy-Patient-Derived Cells Reveals Distinct Cilia Dysfunctions Caused by CEP290 Mutations. <i>Cell Reports</i> , <b>2017</b> , 20, 384-396	10.6	80
273	Transcriptome profiling of NIH3T3 cell lines expressing opsin and the P23H opsin mutant identifies candidate drugs for the treatment of retinitis pigmentosa. <i>Pharmacological Research</i> , <b>2017</b> , 115, 1-13	10.2	6
272	EYS Mutations Causing Autosomal Recessive Retinitis Pigmentosa: Changes of Retinal Structure and Function with Disease Progression. <i>Genes</i> , <b>2017</b> , 8,	4.2	24
271	Next-generation genotype imputation service and methods. <i>Nature Genetics</i> , <b>2016</b> , 48, 1284-1287	36.3	1369
270	A reference panel of 64,976 haplotypes for genotype imputation. <i>Nature Genetics</i> , <b>2016</b> , 48, 1279-83	36.3	1447
269	NRL-Regulated Transcriptome Dynamics of Developing Rod Photoreceptors. <i>Cell Reports</i> , <b>2016</b> , 17, 2460-2473	14.7	70
268	Synergistically acting agonists and antagonists of G protein-coupled receptors prevent photoreceptor cell degeneration. <i>Science Signaling</i> , <b>2016</b> , 9, ra74	8.8	22
267	Variegated yet non-random rod and cone photoreceptor disease patterns in RPGR-ORF15-associated retinal degeneration. <i>Human Molecular Genetics</i> , <b>2016</b> , 25, 5444-5459	5.6	27
266	Recruitment of Rod Photoreceptors from Short-Wavelength-Sensitive Cones during the Evolution of Nocturnal Vision in Mammals. <i>Developmental Cell</i> , <b>2016</b> , 37, 520-32	10.2	74
265	Next generation sequencing technology and genomewide data analysis: Perspectives for retinal research. <i>Progress in Retinal and Eye Research</i> , <b>2016</b> , 55, 1-31	20.5	39
264	Increased retinal mtDNA damage in the CFH variant associated with age-related macular degeneration. <i>Experimental Eye Research</i> , <b>2016</b> , 145, 269-277	3.7	47
263	A secreted WNT-ligand-binding domain of FZD5 generated by a frameshift mutation causes autosomal dominant coloboma. <i>Human Molecular Genetics</i> , <b>2016</b> , 25, 1382-91	5.6	30
262	Centrosomal protein CP110 controls maturation of the mother centriole during cilia biogenesis. <i>Development (Cambridge)</i> , <b>2016</b> , 143, 1491-501	6.6	43
261	A large genome-wide association study of age-related macular degeneration highlights contributions of rare and common variants. <i>Nature Genetics</i> , <b>2016</b> , 48, 134-43	36.3	769
260	The cellular and compartmental profile of mouse retinal glycolysis, tricarboxylic acid cycle, oxidative phosphorylation, and ~P transferring kinases. <i>Molecular Vision</i> , <b>2016</b> , 22, 847-85	2.3	43
259	Three-dimensional retinal organoids from mouse pluripotent stem cells mimic development with enhanced stratification and rod photoreceptor differentiation. <i>Molecular Vision</i> , <b>2016</b> , 22, 1077-1094	2.3	50

258	Genetic components in diabetic retinopathy. <i>Indian Journal of Ophthalmology</i> , <b>2016</b> , 64, 55-61	1.6	17
257	Treatment Paradigms for Retinal and Macular Diseases Using 3-D Retina Cultures Derived From Human Reporter Pluripotent Stem Cell Lines <b>2016</b> , 57, ORSFL1-ORSFL11		30
256	Rapid, Dynamic Activation of Müller Glial Stem Cell Responses in Zebrafish <b>2016</b> , 57, 5148-5160		39
255	Genetic Analysis of the Rhodopsin Gene Identifies a Mosaic Dominant Retinitis Pigmentosa Mutation in a Healthy Individual <b>2016</b> , 57, 940-7		8
254	Molecular Diagnosis of Inherited Retinal Diseases in Indigenous African Populations by Whole-Exome Sequencing <b>2016</b> , 57, 6374-6381		11
253	Gene-Based Association Analysis for Censored Traits Via Fixed Effect Functional Regressions. <i>Genetic Epidemiology</i> , <b>2016</b> , 40, 133-43	2.6	10
252	Loss of RPGR glutamylation underlies the pathogenic mechanism of retinal dystrophy caused by TTLL5 mutations. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2016</b> , 113, E2925-34	11.5	56
251	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> , <b>2016</b> , 99, 777-784	11	30
250	Differential DNA methylation identified in the blood and retina of AMD patients. <i>Epigenetics</i> , <b>2015</b> , 10, 698-707	5.7	46
249	A long-term efficacy study of gene replacement therapy for RPGR-associated retinal degeneration. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 3956-70	5.6	52
248	CEP290 alleles in mice disrupt tissue-specific cilia biogenesis and recapitulate features of syndromic ciliopathies. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 3775-91	5.6	79
247	Long-term rescue of cone photoreceptor degeneration in retinitis pigmentosa 2 (RP2)-knockout mice by gene replacement therapy. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 6446-58	5.6	31
246	Mouse model of human RPE65 P25L hypomorph resembles wild type under normal light rearing but is fully resistant to acute light damage. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 4417-28	5.6	18
245	Biology and therapy of inherited retinal degenerative disease: insights from mouse models. <i>DMM Disease Models and Mechanisms</i> , <b>2015</b> , 8, 109-29	4.1	160
244	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> , <b>2015</b> , 33, 3504-18	5.8	112
243	Whole Exome Sequencing Reveals Mutations in Known Retinal Disease Genes in 33 out of 68 Israeli Families with Inherited Retinopathies. <i>Scientific Reports</i> , <b>2015</b> , 5, 13187	4.9	49
242	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 ALMS1 and DYSF. <i>Human Mutation</i> , <b>2015</b> , 36, 836-41	4.7	13
241	Quantification of Oxygen Consumption in Retina Ex Vivo Demonstrates Limited Reserve Capacity of Photoreceptor Mitochondria <b>2015</b> , 56, 8428-36		78



240	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> , <b>2015</b> , 46, 1-30	20.5	43
239	Clinical and genetic factors associated with progression of geographic atrophy lesions in age-related macular degeneration. <i>PLoS ONE</i> , <b>2015</b> , 10, e0126636	3.7	43
238	Free radical scavenging, antioxidant and cancer chemoprevention by grape seed proanthocyanidin: an overview. <i>Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis</i> , <b>2014</b> , 768, 69-73	2.3	122
237	Deletion of aryl hydrocarbon receptor AHR in mice leads to subretinal accumulation of microglia and RPE atrophy <b>2014</b> , 55, 6031-40		53
236	No clinically significant association between CFH and ARMS2 genotypes and response to nutritional supplements: AREDS report number 38. <i>Ophthalmology</i> , <b>2014</b> , 121, 2173-80	7.3	76
235	Null and hypomorph Prickle1 alleles in mice phenocopy human Robinow syndrome and disrupt signaling downstream of Wnt5a. <i>Biology Open</i> , <b>2014</b> , 3, 861-70	2.2	33
234	Rare and common variants in extracellular matrix gene Fibrillin 2 (FBN2) are associated with macular degeneration. <i>Human Molecular Genetics</i> , <b>2014</b> , 23, 5827-37	5.6	34
233	Ciliopathy-associated gene Cc2d2a promotes assembly of subdistal appendages on the mother centriole during cilia biogenesis. <i>Nature Communications</i> , <b>2014</b> , 5, 4207	17.4	50
232	Age-related macular degeneration: genetics and biology coming together. <i>Annual Review of Genomics and Human Genetics</i> , <b>2014</b> , 15, 151-71	9.7	293
231	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> , <b>2014</b> , 56, 420-30		24
230	Natural history of cone disease in the murine model of Leber congenital amaurosis due to CEP290 mutation: determining the timing and expectation of therapy. <i>PLoS ONE</i> , <b>2014</b> , 9, e92928	3.7	19
229	Feedback induction of a photoreceptor-specific isoform of retinoid-related orphan nuclear receptor by the rod transcription factor NRL. <i>Journal of Biological Chemistry</i> , <b>2014</b> , 289, 32469-80	5.4	16
228	The transcription factor GTF2IRD1 regulates the topology and function of photoreceptors by modulating photoreceptor gene expression across the retina. <i>Journal of Neuroscience</i> , <b>2014</b> , 34, 15356-68	6.6	5
227	Regulation of a novel isoform of Receptor Expression Enhancing Protein REEP6 in rod photoreceptors by bZIP transcription factor NRL. <i>Human Molecular Genetics</i> , <b>2014</b> , 23, 4260-71	5.6	21
226	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> , <b>2014</b> , 23, 2132-44	5.6	24
225	Genome-wide association study and meta-analysis of intraocular pressure. <i>Human Genetics</i> , <b>2014</b> , 133, 41-57	6.3	79
224	Ancestry estimation and control of population stratification for sequence-based association studies. <i>Nature Genetics</i> , <b>2014</b> , 46, 409-15	36.3	82
223	OTX2 loss causes rod differentiation defect in CRX-associated congenital blindness. <i>Journal of Clinical Investigation</i> , <b>2014</b> , 124, 631-43	15.9	43



222	Photoreceptor Degeneration: Molecular Mechanisms of Photoreceptor Degeneration <b>2014</b> , 275-308		
221	Gene expression changes in aging retinal microglia: relationship to microglial support functions and regulation of activation. <i>Neurobiology of Aging</i> , <b>2013</b> , 34, 2310-21	5.6	69
220	An isoform of retinoid-related orphan receptor $\alpha$ directs differentiation of retinal amacrine and horizontal interneurons. <i>Nature Communications</i> , <b>2013</b> , 4, 1813	17.4	43
219	Hypomethylation of the IL17RC promoter in peripheral blood leukocytes is not a hallmark of age-related macular degeneration. <i>Cell Reports</i> , <b>2013</b> , 5, 1527-35	10.6	37
218	Prickle1 is expressed in distinct cell populations of the central nervous system and contributes to neuronal morphogenesis. <i>Human Molecular Genetics</i> , <b>2013</b> , 22, 2234-46	5.6	33
217	Genetic architecture of retinal and macular degenerative diseases: the promise and challenges of next-generation sequencing. <i>Genome Medicine</i> , <b>2013</b> , 5, 84	14.4	28
216	Conditional knockdown of DNA methyltransferase 1 reveals a key role of retinal pigment epithelium integrity in photoreceptor outer segment morphogenesis. <i>Development (Cambridge)</i> , <b>2013</b> , 140, 1330-41	6.6	64
215	Seven new loci associated with age-related macular degeneration. <i>Nature Genetics</i> , <b>2013</b> , 45, 433-9, 439e1-3	56.3	577
214	Developing rods transplanted into the degenerating retina of Crx-knockout mice exhibit neural activity similar to native photoreceptors. <i>Stem Cells</i> , <b>2013</b> , 31, 1149-59	5.8	81
213	Identification of a rare coding variant in complement 3 associated with age-related macular degeneration. <i>Nature Genetics</i> , <b>2013</b> , 45, 1375-9	36.3	130
212	Ablation of the X-linked retinitis pigmentosa 2 (Rp2) gene in mice results in opsin mislocalization and photoreceptor degeneration <b>2013</b> , 54, 4503-11		47
211	Development and plasticity of outer retinal circuitry following genetic removal of horizontal cells. <i>Journal of Neuroscience</i> , <b>2013</b> , 33, 17847-62	6.6	32
210	The golden era of ocular disease gene discovery: race to the finish. <i>Clinical Genetics</i> , <b>2013</b> , 84, 99-101	4	10
209	Phenotypic conservation in patients with X-linked retinitis pigmentosa caused by RPGR mutations. <i>JAMA Ophthalmology</i> , <b>2013</b> , 131, 1016-25	3.9	30
208	What's in a name? RPGR mutations redefine the genetic and phenotypic landscape in retinal degenerative diseases <b>2013</b> , 54, 1417		3
207	Retinal transcriptome profiling by directional next-generation sequencing using 100 ng of total RNA. <i>Methods in Molecular Biology</i> , <b>2012</b> , 884, 319-34	1.4	22
206	Global expression profiling of peripheral Qa-1-restricted CD8 $\alpha$ TCR $\beta$ regulatory T cells reveals innate-like features: implications for immune-regulatory repertoire. <i>Human Immunology</i> , <b>2012</b> , 73, 214-223	2.3	12
205	Determination of posttranslational modifications of photoreceptor differentiation factor NRL: focus on SUMOylation. <i>Methods in Molecular Biology</i> , <b>2012</b> , 884, 353-61	1.4	0

204	Genetic studies of age-related macular degeneration: lessons, challenges, and opportunities for disease management. <i>Ophthalmology</i> , <b>2012</b> , 119, 2526-36	7.3	66
203	Preservation of cone photoreceptors after a rapid yet transient degeneration and remodeling in cone-only <i>Nrl</i> <sup>-/-</sup> mouse retina. <i>Journal of Neuroscience</i> , <b>2012</b> , 32, 528-41	6.6	42
202	Photoreceptor sensory cilia and ciliopathies: focus on CEP290, RPGR and their interacting proteins. <i>Cilia</i> , <b>2012</b> , 1, 22	5.5	97
201	A role for prenylated rab acceptor 1 in vertebrate photoreceptor development. <i>BMC Neuroscience</i> , <b>2012</b> , 13, 152	3.2	14
200	Knockdown of Bardet-Biedl syndrome gene BBS9/PTHB1 leads to cilia defects. <i>PLoS ONE</i> , <b>2012</b> , 7, e34389	3.7	37
199	Rd9 is a naturally occurring mouse model of a common form of retinitis pigmentosa caused by mutations in RPGR-ORF15. <i>PLoS ONE</i> , <b>2012</b> , 7, e35865	3.7	59
198	Mutations in RPGR and RP2 account for 15% of males with simplex retinal degenerative disease <b>2012</b> , 53, 8232-7		91
197	RPGR-associated retinal degeneration in human X-linked RP and a murine model <b>2012</b> , 53, 5594-608		49
196	Transcriptional regulation of rod photoreceptor homeostasis revealed by in vivo NRL targetome analysis. <i>PLoS Genetics</i> , <b>2012</b> , 8, e1002649	6	80
195	Minireview: the role of nuclear receptors in photoreceptor differentiation and disease. <i>Molecular Endocrinology</i> , <b>2012</b> , 26, 905-15		47
194	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> , <b>2012</b> , 109, 2132-7	11.5	203
193	Complement factor H genetic variant and age-related macular degeneration: effect size, modifiers and relationship to disease subtype. <i>International Journal of Epidemiology</i> , <b>2012</b> , 41, 250-62	7.8	70
192	Regulation of retinal progenitor expansion by Frizzled receptors: implications for microphthalmia and retinal coloboma. <i>Human Molecular Genetics</i> , <b>2012</b> , 21, 1848-60	5.6	32
191	Combining Cep290 and Mkks ciliopathy alleles in mice rescues sensory defects and restores ciliogenesis. <i>Journal of Clinical Investigation</i> , <b>2012</b> , 122, 1233-45	15.9	64
190	Protective gene expression changes elicited by an inherited defect in photoreceptor structure. <i>PLoS ONE</i> , <b>2012</b> , 7, e31371	3.7	14
189	Long-term survival and differentiation of retinal neurons derived from human embryonic stem cell lines in un-immunosuppressed mouse retina. <i>Molecular Vision</i> , <b>2012</b> , 18, 920-36	2.3	94
188	Transcriptome analysis using next generation sequencing reveals molecular signatures of diabetic retinopathy and efficacy of candidate drugs. <i>Molecular Vision</i> , <b>2012</b> , 18, 1123-46	2.3	35
187	Exome sequencing: capture and sequencing of all human coding regions for disease gene discovery. <i>Methods in Molecular Biology</i> , <b>2012</b> , 884, 335-51	1.4	6

186	Combining Cep290 and Mkks ciliopathy alleles in mice rescues sensory defects and restores ciliogenesis. <i>Journal of Clinical Investigation</i> , <b>2012</b> , 122, 3025-3025	15.9	78
185	Genetic association study of age-related macular degeneration in the Spanish population. <i>Acta Ophthalmologica</i> , <b>2011</b> , 89, e12-22	3.7	31
184	XIAP therapy increases survival of transplanted rod precursors in a degenerating host retina <b>2011</b> , 52, 1567-72		43
183	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> , <b>2011</b> , 6, e20326	3.7	41
182	A 32 kb critical region excluding Y402H in CFH mediates risk for age-related macular degeneration. <i>PLoS ONE</i> , <b>2011</b> , 6, e25598	3.7	41
181	Distinct nuclear localization patterns of DNA methyltransferases in developing and mature mammalian retina. <i>Journal of Comparative Neurology</i> , <b>2011</b> , 519, 1914-30	3.4	40
180	Evidence of association of APOE with age-related macular degeneration: a pooled analysis of 15 studies. <i>Human Mutation</i> , <b>2011</b> , 32, 1407-16	4.7	99
179	Combinatorial regulation of photoreceptor differentiation factor, neural retina leucine zipper gene NRL, revealed by in vivo promoter analysis. <i>Journal of Biological Chemistry</i> , <b>2011</b> , 286, 28247-55	5.4	27
178	Variations in apolipoprotein E frequency with age in a pooled analysis of a large group of older people. <i>American Journal of Epidemiology</i> , <b>2011</b> , 173, 1357-64	3.8	67
177	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> , <b>2011</b> , 20, 4102-15	5.6	38
176	Candidate gene association study for diabetic retinopathy in persons with type 2 diabetes: the Candidate gene Association Resource (CARE) <b>2011</b> , 52, 7593-602		73
175	Complement factor D in age-related macular degeneration <b>2011</b> , 52, 8828-34		72
174	Two transcription factors can direct three photoreceptor outcomes from rod precursor cells in mouse retinal development. <i>Journal of Neuroscience</i> , <b>2011</b> , 31, 11118-25	6.6	60
173	TOPORS, implicated in retinal degeneration, is a cilia-centrosomal protein. <i>Human Molecular Genetics</i> , <b>2011</b> , 20, 975-87	5.6	43
172	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> , <b>2011</b> , 286, 34893-902	5.4	31
171	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. <i>Human Molecular Genetics</i> , <b>2011</b> , 20, 1411-23	5.6	97
170	Next-generation sequencing facilitates quantitative analysis of wild-type and Nrl(-/-) retinal transcriptomes. <i>Molecular Vision</i> , <b>2011</b> , 17, 3034-54	2.3	83
169	Transcriptional regulation of photoreceptor development and homeostasis in the mammalian retina. <i>Nature Reviews Neuroscience</i> , <b>2010</b> , 11, 563-76	13.5	370

168	Long-term follow-up of a family with dominant X-linked retinitis pigmentosa. <i>Eye</i> , <b>2010</b> , 24, 764-74	4.4	25
167	Associations of CFHR1-CFHR3 deletion and a CFH SNP to age-related macular degeneration are not independent. <i>Nature Genetics</i> , <b>2010</b> , 42, 553-5; author reply 555-6	36.3	46
166	MicroRNA profile of the developing mouse retina <b>2010</b> , 51, 1823-31		81
165	Autosomal recessive retinitis pigmentosa with early macular affection caused by premature truncation in PROM1 <b>2010</b> , 51, 2656-63		48
164	Human retinopathy-associated ciliary protein retinitis pigmentosa GTPase regulator mediates cilia-dependent vertebrate development. <i>Human Molecular Genetics</i> , <b>2010</b> , 19, 90-8	5.6	68
163	Phenotype associated with mutation in the recently identified autosomal dominant retinitis pigmentosa KLHL7 gene. <i>JAMA Ophthalmology</i> , <b>2010</b> , 128, 772-8		17
162	Interaction of retinitis pigmentosa GTPase regulator (RPGR) with RAB8A GTPase: implications for cilia dysfunction and photoreceptor degeneration. <i>Human Molecular Genetics</i> , <b>2010</b> , 19, 3591-8	5.6	79
161	The retinitis pigmentosa protein RP2 interacts with polycystin 2 and regulates cilia-mediated vertebrate development. <i>Human Molecular Genetics</i> , <b>2010</b> , 19, 4330-44	5.6	59
160	RP2 phenotype and pathogenetic correlations in X-linked retinitis pigmentosa. <i>JAMA Ophthalmology</i> , <b>2010</b> , 128, 915-23		45
159	Sumoylation of bZIP transcription factor NRL modulates target gene expression during photoreceptor differentiation. <i>Journal of Biological Chemistry</i> , <b>2010</b> , 285, 25637-44	5.4	33
158	Loss of lysophosphatidylcholine acyltransferase 1 leads to photoreceptor degeneration in rd11 mice. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2010</b> , 107, 15523-8	11.5	49
157	E2-2 protein and Fuchs' corneal dystrophy. <i>New England Journal of Medicine</i> , <b>2010</b> , 363, 1016-24	59.2	197
156	Transcriptome analysis and molecular signature of human retinal pigment epithelium. <i>Human Molecular Genetics</i> , <b>2010</b> , 19, 2468-86	5.6	193
155	MicroRNA-204/211 alters epithelial physiology. <i>FASEB Journal</i> , <b>2010</b> , 24, 1552-71	0.9	198
154	Genetic variants near TIMP3 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> , <b>2010</b> , 107, 7401-6	11.5	417
153	Two novel CRX mutant proteins causing autosomal dominant Leber congenital amaurosis interact differently with NRL. <i>Human Mutation</i> , <b>2010</b> , 31, E1472-83	4.7	36
152	Distinct signature of altered homeostasis in aging rod photoreceptors: implications for retinal diseases. <i>PLoS ONE</i> , <b>2010</b> , 5, e13885	3.7	26
151	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> , <b>2010</b> , 16, 1317-23	2.3	34

150	Multiprotein complexes of Retinitis Pigmentosa GTPase regulator (RPGR), a ciliary protein mutated in X-linked Retinitis Pigmentosa (XLRP). <i>Advances in Experimental Medicine and Biology</i> , <b>2010</b> , 664, 105-14	3.6	20
149	Retinoid-related orphan nuclear receptor RORbeta is an early-acting factor in rod photoreceptor development. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2009</b> , 106, 17534-9	11.5	100
148	Rdh12 activity and effects on retinoid processing in the murine retina. <i>Journal of Biological Chemistry</i> , <b>2009</b> , 284, 21468-77	5.4	40
147	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> , <b>2009</b> , 18, 1909-23	5.6	84
146	RPGR-containing protein complexes in syndromic and non-syndromic retinal degeneration due to ciliary dysfunction. <i>Journal of Genetics</i> , <b>2009</b> , 88, 399-407	1.2	24
145	Canine RD3 mutation establishes rod-cone dysplasia type 2 (rcd2) as ortholog of human and murine rd3. <i>Mammalian Genome</i> , <b>2009</b> , 20, 109-23	3.2	49
144	A common allele in RPGRIP1L is a modifier of retinal degeneration in ciliopathies. <i>Nature Genetics</i> , <b>2009</b> , 41, 739-45	36.3	236
143	Mutations in a BTB-Kelch protein, KLHL7, cause autosomal-dominant retinitis pigmentosa. <i>American Journal of Human Genetics</i> , <b>2009</b> , 84, 792-800	11	84
142	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> , <b>2009</b> , 10, 19-43	9.7	217
141	A comprehensive analysis of sequence variants and putative disease-causing mutations in photoreceptor-specific nuclear receptor NR2E3. <i>Molecular Vision</i> , <b>2009</b> , 15, 2174-84	2.3	25
140	Geographic atrophy in age-related macular degeneration and TLR3. <i>New England Journal of Medicine</i> , <b>2009</b> , 360, 2254-5; author reply 2255-6	59.2	13
139	Retinitis Pigmentosa GTPase Regulator (RPGR) protein isoforms in mammalian retina: insights into X-linked Retinitis Pigmentosa and associated ciliopathies. <i>Vision Research</i> , <b>2008</b> , 48, 366-76	2.1	47
138	Rod differentiation factor NRL activates the expression of nuclear receptor NR2E3 to suppress the development of cone photoreceptors. <i>Brain Research</i> , <b>2008</b> , 1236, 16-29	3.7	90
137	CP110 suppresses primary cilia formation through its interaction with CEP290, a protein deficient in human ciliary disease. <i>Developmental Cell</i> , <b>2008</b> , 15, 187-97	10.2	190
136	Discordant phenotypes in fraternal twins having an identical mutation in exon ORF15 of the RPGR gene. <i>JAMA Ophthalmology</i> , <b>2008</b> , 126, 379-84		44
135	Inflammation in the pathogenesis of age-related macular degeneration. <i>British Journal of Ophthalmology</i> , <b>2008</b> , 92, 448-50	5.5	36
134	Nrl-knockout mice deficient in Rpe65 fail to synthesize 11-cis retinal and cone outer segments. <i>Investigative Ophthalmology and Visual Science</i> , <b>2008</b> , 49, 1126-35		37
133	Toll-like receptor polymorphisms and age-related macular degeneration. <i>Investigative Ophthalmology and Visual Science</i> , <b>2008</b> , 49, 1652-9		71

132	FGF19 exhibits neuroprotective effects on adult mammalian photoreceptors in vitro. <i>Investigative Ophthalmology and Visual Science</i> , <b>2008</b> , 49, 1696-704		22
131	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> , <b>2008</b> , 613, 221-7	3.6	4
130	High-resolution imaging with adaptive optics in patients with inherited retinal degeneration. <i>Investigative Ophthalmology and Visual Science</i> , <b>2007</b> , 48, 3283-91		213
129	Inner retinal abnormalities in X-linked retinitis pigmentosa with RPGR mutations. <i>Investigative Ophthalmology and Visual Science</i> , <b>2007</b> , 48, 4759-65		89
128	Retinopathy mutations in the bZIP protein NRL alter phosphorylation and transcriptional activity. <i>Human Mutation</i> , <b>2007</b> , 28, 589-98	4.7	30
127	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> , <b>2007</b> , 28, 1074-83	4.7	121
126	Preventing polyglutamine-induced activation of c-Jun delays neuronal dysfunction in a mouse model of SCA7 retinopathy. <i>Neurobiology of Disease</i> , <b>2007</b> , 25, 571-81	7.5	14
125	Subunit dissociation and diffusion determine the subcellular localization of rod and cone transducins. <i>Journal of Neuroscience</i> , <b>2007</b> , 27, 5484-94	6.6	58
124	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> , <b>2007</b> , 104, 15917-22	11.5	128
123	Madeline 2.0 PDE: a new program for local and web-based pedigree drawing. <i>Bioinformatics</i> , <b>2007</b> , 23, 1854-6	7.2	25
122	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> , <b>2007</b> , 104, 16227-32	11.5	356
121	Genetic susceptibility to age-related macular degeneration: a paradigm for dissecting complex disease traits. <i>Human Molecular Genetics</i> , <b>2007</b> , 16 Spec No. 2, R174-82	5.6	146
120	Transformation of cone precursors to functional rod photoreceptors by bZIP transcription factor NRL. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2007</b> , 104, 1679-84	11.5	121
119	Afferent control of horizontal cell morphology revealed by genetic respecification of rods and cones. <i>Journal of Neuroscience</i> , <b>2007</b> , 27, 3540-7	6.6	35
118	Chondroitinase ABC treatment enhances synaptogenesis between transplant and host neurons in model of retinal degeneration. <i>Cell Transplantation</i> , <b>2007</b> , 16, 493-503	4	74
117	Mutations in TOPORS cause autosomal dominant retinitis pigmentosa with perivascular retinal pigment epithelium atrophy. <i>American Journal of Human Genetics</i> , <b>2007</b> , 81, 1098-103	11	68
116	Mutations associated with retinopathies alter mitogen-activated protein kinase-induced phosphorylation of neural retina leucine-zipper. <i>Molecular Vision</i> , <b>2007</b> , 13, 1114-20	2.3	3
115	In vivo function of the orphan nuclear receptor NR2E3 in establishing photoreceptor identity during mammalian retinal development. <i>Human Molecular Genetics</i> , <b>2006</b> , 15, 2588-602	5.6	95



114	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> , <b>2006</b> , 103, 3890-5	11.5	251
113	Retinoic acid regulates the expression of photoreceptor transcription factor NRL. <i>Journal of Biological Chemistry</i> , <b>2006</b> , 281, 27327-34	5.4	51
112	Activation of signaling pathways and stress-response genes in an experimental model of retinal detachment. <i>Investigative Ophthalmology and Visual Science</i> , <b>2006</b> , 47, 1691-5		60
111	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> , <b>2006</b> , 15, 1847-57	5.6	315
110	Premature truncation of a novel protein, RD3, exhibiting subnuclear localization is associated with retinal degeneration. <i>American Journal of Human Genetics</i> , <b>2006</b> , 79, 1059-70	11	98
109	The centrosomal protein nephrocystin-6 is mutated in Joubert syndrome and activates transcription factor ATF4. <i>Nature Genetics</i> , <b>2006</b> , 38, 674-81	36.3	464
108	CFH haplotypes without the Y402H coding variant show strong association with susceptibility to age-related macular degeneration. <i>Nature Genetics</i> , <b>2006</b> , 38, 1049-54	36.3	291
107	Retinal repair by transplantation of photoreceptor precursors. <i>Nature</i> , <b>2006</b> , 444, 203-7	50.4	847
106	Gene expression signatures and biomarkers of noninvasive and invasive breast cancer cells: comprehensive profiles by representational difference analysis, microarrays and proteomics. <i>Oncogene</i> , <b>2006</b> , 25, 2328-38	9.2	156
105	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> , <b>2006</b> , 104, 232-40		7
104	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> , <b>2005</b> , 77, 149-53	11	298
103	Meta-analysis of genome scans of age-related macular degeneration. <i>Human Molecular Genetics</i> , <b>2005</b> , 14, 2257-64	5.6	197
102	Toll-like receptor 4 variant D299G is associated with susceptibility to age-related macular degeneration. <i>Human Molecular Genetics</i> , <b>2005</b> , 14, 1449-55	5.6	158
101	Biomarkers of cardiovascular disease as risk factors for age-related macular degeneration. <i>Ophthalmology</i> , <b>2005</b> , 112, 2076-80	7.3	132
100	Mechanisms of aging in senescence-accelerated mice. <i>Genome Biology</i> , <b>2005</b> , 6, R48	18.3	44
99	Nephrocystin-5, a ciliary IQ domain protein, is mutated in Senior-Loken syndrome and interacts with RPGR and calmodulin. <i>Nature Genetics</i> , <b>2005</b> , 37, 282-8	36.3	331
98	RPGR ORF15 isoform co-localizes with RPGRIP1 at centrioles and basal bodies and interacts with nucleophosmin. <i>Human Molecular Genetics</i> , <b>2005</b> , 14, 1183-97	5.6	95
97	Cone-like morphological, molecular, and electrophysiological features of the photoreceptors of the Nrl knockout mouse. <i>Investigative Ophthalmology and Visual Science</i> , <b>2005</b> , 46, 2156-67		168

96	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> , <b>2005</b> , 12, 1029-45	1.7	38
95	RPGR-ORF15, which is mutated in retinitis pigmentosa, associates with SMC1, SMC3, and microtubule transport proteins. <i>Journal of Biological Chemistry</i> , <b>2005</b> , 280, 33580-7	5.4	125
94	Photoreceptors of Nrl <sup>-/-</sup> mice coexpress functional S- and M-cone opsins having distinct inactivation mechanisms. <i>Journal of General Physiology</i> , <b>2005</b> , 125, 287-304	3.4	111
93	Network constrained clustering for gene microarray data. <i>Bioinformatics</i> , <b>2005</b> , 21, 4014-20	7.2	41
92	Expression of photoreceptor-specific nuclear receptor NR2E3 in rod photoreceptors of fetal human retina. <i>Investigative Ophthalmology and Visual Science</i> , <b>2004</b> , 45, 2807-12		66
91	Characterization of new transcripts enriched in the mouse retina and identification of candidate retinal disease genes. <i>Investigative Ophthalmology and Visual Science</i> , <b>2004</b> , 45, 3313-9		9
90	Seeing the unseen: Microarray-based gene expression profiling in vision. <i>Investigative Ophthalmology and Visual Science</i> , <b>2004</b> , 45, 2457-62		6
89	Association of apolipoprotein E alleles with susceptibility to age-related macular degeneration in a large cohort from a single center. <i>Investigative Ophthalmology and Visual Science</i> , <b>2004</b> , 45, 1306-10		101
88	Expression profiling of the developing and mature Nrl <sup>-/-</sup> mouse retina: identification of retinal disease candidates and transcriptional regulatory targets of Nrl. <i>Human Molecular Genetics</i> , <b>2004</b> , 13, 1487-503	5.6	138
87	Transgenic mice expressing Cre-recombinase specifically in M- or S-cone photoreceptors. <i>Investigative Ophthalmology and Visual Science</i> , <b>2004</b> , 45, 42-7		26
86	Nuclear receptor NR2E3 gene mutations distort human retinal laminar architecture and cause an unusual degeneration. <i>Human Molecular Genetics</i> , <b>2004</b> , 13, 1893-902	5.6	76
85	Functional analysis of the rod photoreceptor cGMP phosphodiesterase alpha-subunit gene promoter: Nrl and Crx are required for full transcriptional activity. <i>Journal of Biological Chemistry</i> , <b>2004</b> , 279, 19800-7	5.4	48
84	QRX, a novel homeobox gene, modulates photoreceptor gene expression. <i>Human Molecular Genetics</i> , <b>2004</b> , 13, 1025-40	5.6	62
83	Photoreceptor-specific nuclear receptor NR2E3 functions as a transcriptional activator in rod photoreceptors. <i>Human Molecular Genetics</i> , <b>2004</b> , 13, 1563-75	5.6	174
82	The minimal transactivation domain of the basic motif-leucine zipper transcription factor NRL interacts with TATA-binding protein. <i>Journal of Biological Chemistry</i> , <b>2004</b> , 279, 47233-41	5.4	31
81	Recruitment of the rod pathway by cones in the absence of rods. <i>Journal of Neuroscience</i> , <b>2004</b> , 24, 7576-82		69
80	Altered expression of genes of the Bmp/Smad and Wnt/calcium signaling pathways in the cone-only Nrl <sup>-/-</sup> mouse retina, revealed by gene profiling using custom cDNA microarrays. <i>Journal of Biological Chemistry</i> , <b>2004</b> , 279, 42211-20	5.4	48
79	Recessive NRL mutations in patients with clumped pigmentary retinal degeneration and relative preservation of blue cone function. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2004</b> , 101, 17819-24	11.5	75

78	Mutation analysis of NR2E3 and NRL genes in Enhanced S Cone Syndrome. <i>Human Mutation</i> , <b>2004</b> , 24, 439	4.7	73
77	Gene discovery using Pareto depth sampling distributions. <i>Journal of the Franklin Institute</i> , <b>2004</b> , 341, 55-75	4	7
76	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> , <b>2004</b> , 74, 482-94	11	141
75	Multicriteria Gene Screening for Analysis of Differential Expression with DNA Microarrays. <i>Eurasip Journal on Advances in Signal Processing</i> , <b>2004</b> , 2004, 1	1.9	11
74	Increasing evidence for syndromic phenotypes associated with RPGR mutations. <i>American Journal of Ophthalmology</i> , <b>2004</b> , 137, 785-6; author reply 786	4.9	16
73	GRK1-dependent phosphorylation of S and M opsins and their binding to cone arrestin during cone phototransduction in the mouse retina. <i>Journal of Neuroscience</i> , <b>2003</b> , 23, 6152-60	6.6	100
72	De novo mutation in the RP1 gene (Arg677ter) associated with retinitis pigmentosa. <i>Investigative Ophthalmology and Visual Science</i> , <b>2003</b> , 44, 3593-7		26
71	Annotation and analysis of 10,000 expressed sequence tags from developing mouse eye and adult retina. <i>Genome Biology</i> , <b>2003</b> , 4, R65	18.3	30
70	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> , <b>2003</b> , 12, 365-73	5.6	28
69	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> , <b>2003</b> , 12, 2657-67	5.6	150
68	Clinical studies of X-linked retinitis pigmentosa in three Swedish families with newly identified mutations in the RP2 and RPGR-ORF15 genes. <i>Ophthalmic Genetics</i> , <b>2003</b> , 24, 215-23	1.2	21
67	Mutation screening of patients with Leber Congenital Amaurosis or the enhanced S-Cone Syndrome reveals a lack of sequence variations in the NRL gene. <i>Molecular Vision</i> , <b>2003</b> , 9, 14-7	2.3	8
66	A comprehensive analysis of the expression of crystallins in mouse retina. <i>Molecular Vision</i> , <b>2003</b> , 9, 410-23	2.3	88
65	Barrier to autointegration factor interacts with the cone-rod homeobox and represses its transactivation function. <i>Journal of Biological Chemistry</i> , <b>2002</b> , 277, 43288-300	5.4	96
64	Retinal Histopathology of an XLRP Carrier with a Mutation in the RPGR Exon ORF15. <i>Experimental Eye Research</i> , <b>2002</b> , 75, 431-443	3.7	34
63	Transcriptome analysis of the retina. <i>Genome Biology</i> , <b>2002</b> , 3, REVIEWS1022	18.3	23
62	Mouse eye gene microarrays for investigating ocular development and disease. <i>Vision Research</i> , <b>2002</b> , 42, 463-70	2.1	35
61	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> , <b>2002</b> , 70, 1545-54	11	194

60	Gene expression profile of native human retinal pigment epithelium. <i>Investigative Ophthalmology and Visual Science</i> , <b>2002</b> , 43, 603-7		18
59	Evaluation and optimization of procedures for target labeling and hybridization of cDNA microarrays. <i>Molecular Vision</i> , <b>2002</b> , 8, 130-7	2.3	41
58	Microarray analysis of gene expression in the aging human retina. <i>Investigative Ophthalmology and Visual Science</i> , <b>2002</b> , 43, 2554-60		74
57	Retinal histopathology of an XLRP carrier with a mutation in the RPGR exon ORF15. <i>Experimental Eye Research</i> , <b>2002</b> , 75, 431-43	3.7	18
56	Five novel RPGR mutations in families with X-linked retinitis pigmentosa. <i>Human Mutation</i> , <b>2001</b> , 17, 151	4.7	10
55	Nrl is required for rod photoreceptor development. <i>Nature Genetics</i> , <b>2001</b> , 29, 447-52	36.3	706
54	Multiple phosphorylated isoforms of NRL are expressed in rod photoreceptors. <i>Journal of Biological Chemistry</i> , <b>2001</b> , 276, 36824-30	5.4	88
53	Evaluation of the ELOVL4 gene in patients with age-related macular degeneration. <i>Ophthalmic Genetics</i> , <b>2001</b> , 22, 233-9	1.2	47
52	X-Linked Retinitis Pigmentosa: Current Status <b>2001</b> , 11-22		1
51	NRL S50T mutation and the importance of founder effects in inherited retinal dystrophies. <i>European Journal of Human Genetics</i> , <b>2000</b> , 8, 783-7	5.3	17
50	Cloning and functional expression of human retinal kir2.4, a pH-sensitive inwardly rectifying K(+) channel. <i>American Journal of Physiology - Cell Physiology</i> , <b>2000</b> , 279, C771-84	5.4	37
49	The leucine zipper of NRL interacts with the CRX homeodomain. A possible mechanism of transcriptional synergy in rhodopsin regulation. <i>Journal of Biological Chemistry</i> , <b>2000</b> , 275, 29794-9	5.4	149
48	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> , <b>2000</b> , 67, 1000-3	11	57
47	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> , <b>1999</b> , 8, 299-305	5.6	128
46	A mutation in NRL is associated with autosomal dominant retinitis pigmentosa. <i>Nature Genetics</i> , <b>1999</b> , 21, 355-6	36.3	180
45	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> , <b>1999</b> , 64, 897-900	11	73
44	X-linked retinitis pigmentosa in two families with a missense mutation in the RPGR gene and putative change of glycine to valine at codon 60. <i>Ophthalmology</i> , <b>1998</b> , 105, 2286-96	7.3	21
43	A novel locus (RP24) for X-linked retinitis pigmentosa maps to Xq26-27. <i>American Journal of Human Genetics</i> , <b>1998</b> , 63, 1439-47	11	63

42	Clinical expression of X-linked retinitis pigmentosa in a Swedish family with the RP2 genotype. <i>Ophthalmic Genetics</i> , <b>1998</b> , 19, 187-96	1.2	4
41	Mutation of a gene encoding a protein with extracellular matrix motifs in Usher syndrome type IIa. <i>Science</i> , <b>1998</b> , 280, 1753-7	33.3	321
40	Biochemical characterization and subcellular localization of the mouse retinitis pigmentosa GTPase regulator (mRpg). <i>Journal of Biological Chemistry</i> , <b>1998</b> , 273, 19656-63	5.4	59
39	A new 2-base pair deletion in the RPGR gene in a black family with X-linked retinitis pigmentosa. <i>JAMA Ophthalmology</i> , <b>1998</b> , 116, 213-8		8
38	Lens-specific gene recruitment of zeta-crystallin through Pax6, Nrl-Maf, and brain suppressor sites. <i>Molecular and Cellular Biology</i> , <b>1998</b> , 18, 2067-76	4.8	42
37	Human bZIP transcription factor gene NRL: structure, genomic sequence, and fine linkage mapping at 14q11.2 and negative mutation analysis in patients with retinal degeneration. <i>Genomics</i> , <b>1997</b> , 45, 395-401	4.3	25
36	Mutations in the cone-rod homeobox gene are associated with the cone-rod dystrophy photoreceptor degeneration. <i>Neuron</i> , <b>1997</b> , 19, 1329-36	13.9	228
35	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> , <b>1997</b> , 61, 1287-92	11	86
34	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> , <b>1997</b> , 61, 571-80	11	61
33	Characterization and sequence analysis of the human homeobox-containing gene GBX2. <i>Genomics</i> , <b>1996</b> , 31, 335-42	4.3	14
32	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> , <b>1996</b> , 93, 191-5	11.5	172
31	[11]Isolation of candidate genes for inherited diseases: Application to X-linked retinal degenerations. <i>Methods in Molecular Genetics</i> , <b>1996</b> , 8, 207-228		
30	The bZIP transcription factor Nrl stimulates rhodopsin promoter activity in primary retinal cell cultures. <i>Journal of Biological Chemistry</i> , <b>1996</b> , 271, 29612-8	5.4	106
29	Dinucleotide polymorphism at the DXS1178 locus is tightly linked to PGK1 at Xq13. <i>Human Genetics</i> , <b>1995</b> , 95, 467-8	6.3	1
28	Expression and chromosomal localization of cDNA clones from an enriched human retinal pigment epithelial (RPE) cell line library: identification of two RPE-specific genes. <i>Cytogenetic and Genome Research</i> , <b>1995</b> , 69, 71-4	1.9	8
27	Dinucleotide repeat polymorphism at the DXS977 locus. <i>Human Molecular Genetics</i> , <b>1994</b> , 3, 1030	5.6	1
26	Localization of the gene for pigment epithelium-derived factor (PEDF) to chromosome 17p13.1 and expression in cultured human retinoblastoma cells. <i>Genomics</i> , <b>1994</b> , 19, 266-72	4.3	58
25	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> , <b>1994</b> , 3, 1281-6	5.6	28

24	A Sandwich-Hybridization Method for Specific and Efficient Selection of cDNA Clones from Genomic Regions <b>1994</b> , 91-99		
23	Molecular characterization of the murine neural retina leucine zipper gene, Nrl. <i>Genomics</i> , <b>1993</b> , 18, 216-223	4.3	23
22	lambda SHK and lambda AASV: phage vectors for efficient cDNA cloning and expression in mammalian cells. <i>Gene</i> , <b>1993</b> , 123, 287-8	3.8	2
21	Mapping of the neural retina leucine zipper gene, Nrl, to mouse chromosome 14. <i>Mammalian Genome</i> , <b>1993</b> , 4, 618-20	3.2	0
20	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> , <b>1993</b> , 285-300		2
19	Identification of Candidate Genes for Eye Diseases: Studies on a Neural Retina-Specific Gene Encoding a Putative Dna Binding Protein of Leucine Zipper Family <b>1993</b> , 171-180		
18	Expressed sequence tags and chromosomal localization of cDNA clones from a subtracted retinal pigment epithelium library. <i>Genomics</i> , <b>1992</b> , 13, 873-6	4.3	61
17	Neural retina-specific leucine zipper gene NRL (D14S46E) maps to human chromosome 14q11.1-q11.2. <i>Genomics</i> , <b>1992</b> , 14, 491-2	4.3	11
16	A simple and efficient cDNA library subtraction procedure: isolation of human retina-specific cDNA clones. <i>Nucleic Acids Research</i> , <b>1991</b> , 19, 1954	20.1	54
15	Differential expression of novel Gs alpha signal transduction protein cDNA species. <i>Nucleic Acids Research</i> , <b>1991</b> , 19, 4725-9	20.1	70
14	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> , <b>1991</b> , 52, 549-61	3.7	28
13	Positional cloning and characterization of a paired box- and homeobox-containing gene from the aniridia region. <i>Cell</i> , <b>1991</b> , 67, 1059-74	56.2	737
12	Chromosomal localization and cDNA sequence of human ralB, a GTP binding protein. <i>Somatic Cell and Molecular Genetics</i> , <b>1990</b> , 16, 407-10		5
11	pSH4: a mammalian cDNA expression vector. <i>Nucleic Acids Research</i> , <b>1990</b> , 18, 3668	20.1	9
10	The mRNA of a human class I gene HLA G/HLA 6.0 exhibits a restricted pattern of expression. <i>Nucleic Acids Research</i> , <b>1990</b> , 18, 2189	20.1	70
9	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> , <b>1988</b> , 2, 37-47	4.3	138
8	Charon BS(+) and (-), versatile lambda phage vectors for constructing directional cDNA libraries and their efficient transfer to plasmids. <i>Nucleic Acids Research</i> , <b>1988</b> , 16, 8739	20.1	31
7	Generation of hydrogen peroxide on oxidation of NADH by hepatic plasma membranes. <i>Journal of Bioenergetics and Biomembranes</i> , <b>1981</b> , 13, 241-53	3.7	30



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