Anand Swaroop

List of Publications by Citations

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	A reference panel of 64,976 haplotypes for genotype imputation. *Nature Genetics*, **2016**, 48, 1279-83	36.3	1447
346	Next-generation genotype imputation service and methods. *Nature Genetics*, **2016**, 48, 1284-1287	36.3	1369
345	Retinal repair by transplantation of photoreceptor precursors. *Nature*, **2006**, 444, 203-7	50.4	847
344	A large genome-wide association study of age-related macular degeneration highlights contributions of rare and common variants. *Nature Genetics*, **2016**, 48, 134-43	36.3	769
343	Positional cloning and characterization of a paired box- and homeobox-containing gene from the aniridia region. *Cell*, **1991**, 67, 1059-74	56.2	737
342	Nrl is required for rod photoreceptor development. *Nature Genetics*, **2001**, 29, 447-52	36.3	706
341	Seven new loci associated with age-related macular degeneration. *Nature Genetics*, **2013**, 45, 433-9, 439e1-2	36.3	577
340	The centrosomal protein nephrocystin-6 is mutated in Joubert syndrome and activates transcription factor ATF4. *Nature Genetics*, **2006**, 38, 674-81	36.3	464
339	Genetic variants near TIMP3 and high-density lipoprotein-associated loci influence susceptibility to age-related macular degeneration. *Proceedings of the National Academy of Sciences of the United States of America*, **2010**, 107, 7401-6	11.5	417
338	Transcriptional regulation of photoreceptor development and homeostasis in the mammalian retina. *Nature Reviews Neuroscience*, **2010**, 11, 563-76	13.5	370
337	A variant of mitochondrial protein LOC387715/ARMS2, not HTRA1, is strongly associated with age-related macular degeneration. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 16227-32	11.5	356
336	Nephrocystin-5, a ciliary IQ domain protein, is mutated in Senior-Loken syndrome and interacts with RPGR and calmodulin. *Nature Genetics*, **2005**, 37, 282-8	36.3	331
335	Mutation of a gene encoding a protein with extracellular matrix motifs in Usher syndrome type IIa. *Science*, **1998**, 280, 1753-7	33.3	321
334	In-frame deletion in a novel centrosomal/ciliary protein CEP290/NPHP6 perturbs its interaction with RPGR and results in early-onset retinal degeneration in the rd16 mouse. *Human Molecular Genetics*, **2006**, 15, 1847-57	5.6	315
333	Strong association of the Y402H variant in complement factor H at 1q32 with susceptibility to age-related macular degeneration. *American Journal of Human Genetics*, **2005**, 77, 149-53	11	298
332	Age-related macular degeneration: genetics and biology coming together. *Annual Review of Genomics and Human Genetics*, **2014**, 15, 151-71	9.7	293
331	CFH haplotypes without the Y402H coding variant show strong association with susceptibility to age-related macular degeneration. *Nature Genetics*, **2006**, 38, 1049-54	36.3	291

330	Targeting of GFP to newborn rods by Nrl promoter and temporal expression profiling of flow-sorted photoreceptors. *Proceedings of the National Academy of Sciences of the United States of America*, **2006**, 103, 3890-5	11.5	251
329	A common allele in RPGRIP1L is a modifier of retinal degeneration in ciliopathies. *Nature Genetics*, **2009**, 41, 739-45	36.3	236
328	Mutations in the cone-rod homeobox gene are associated with the cone-rod dystrophy photoreceptor degeneration. *Neuron*, **1997**, 19, 1329-36	13.9	228
327	Unraveling a multifactorial late-onset disease: from genetic susceptibility to disease mechanisms for age-related macular degeneration. *Annual Review of Genomics and Human Genetics*, **2009**, 10, 19-43	9.7	217
326	High-resolution imaging with adaptive optics in patients with inherited retinal degeneration. *Investigative Ophthalmology and Visual Science*, **2007**, 48, 3283-91		213
325	Gene therapy rescues photoreceptor blindness in dogs and paves the way for treating human X-linked retinitis pigmentosa. *Proceedings of the National Academy of Sciences of the United States of America*, **2012**, 109, 2132-7	11.5	203
324	MicroRNA-204/211 alters epithelial physiology. *FASEB Journal*, **2010**, 24, 1552-71	0.9	198
323	E2-2 protein and Fuchs corneal dystrophy. *New England Journal of Medicine*, **2010**, 363, 1016-24	59.2	197
322	Meta-analysis of genome scans of age-related macular degeneration. *Human Molecular Genetics*, **2005**, 14, 2257-64	5.6	197
321	A comprehensive mutation analysis of RP2 and RPGR in a North American cohort of families with X-linked retinitis pigmentosa. *American Journal of Human Genetics*, **2002**, 70, 1545-54	11	194
320	Transcriptome analysis and molecular signature of human retinal pigment epithelium. *Human Molecular Genetics*, **2010**, 19, 2468-86	5.6	193
319	CP110 suppresses primary cilia formation through its interaction with CEP290, a protein deficient in human ciliary disease. *Developmental Cell*, **2008**, 15, 187-97	10.2	190
318	A mutation in NRL is associated with autosomal dominant retinitis pigmentosa. *Nature Genetics*, **1999**, 21, 355-6	36.3	180
317	Nrl knockdown by AAV-delivered CRISPR/Cas9 prevents retinal degeneration in mice. *Nature Communications*, **2017**, 8, 14716	17.4	176
316	Photoreceptor-specific nuclear receptor NR2E3 functions as a transcriptional activator in rod photoreceptors. *Human Molecular Genetics*, **2004**, 13, 1563-75	5.6	174
315	The basic motif-leucine zipper transcription factor Nrl can positively regulate rhodopsin gene expression. *Proceedings of the National Academy of Sciences of the United States of America*, **1996**, 93, 191-5	11.5	172
314	Cone-like morphological, molecular, and electrophysiological features of the photoreceptors of the Nrl knockout mouse. *Investigative Ophthalmology and Visual Science*, **2005**, 46, 2156-67		168
313	Biology and therapy of inherited retinal degenerative disease: insights from mouse models. *DMM Disease Models and Mechanisms*, **2015**, 8, 109-29	4.1	160

312	Toll-like receptor 4 variant D299G is associated with susceptibility to age-related macular degeneration. *Human Molecular Genetics*, **2005**, 14, 1449-55	5.6	158
311	Gene expression signatures and biomarkers of noninvasive and invasive breast cancer cells: comprehensive profiles by representational difference analysis, microarrays and proteomics. *Oncogene*, **2006**, 25, 2328-38	9.2	156
310	Mutation in a short-chain collagen gene, CTRP5, results in extracellular deposit formation in late-onset retinal degeneration: a genetic model for age-related macular degeneration. *Human Molecular Genetics*, **2003**, 12, 2657-67	5.6	150
309	The leucine zipper of NRL interacts with the CRX homeodomain. A possible mechanism of transcriptional synergy in rhodopsin regulation. *Journal of Biological Chemistry*, **2000**, 275, 29794-9	5.4	149
308	Genetic susceptibility to age-related macular degeneration: a paradigm for dissecting complex disease traits. *Human Molecular Genetics*, **2007**, 16 Spec No. 2, R174-82	5.6	146
307	Age-related macular degeneration: a high-resolution genome scan for susceptibility loci in a population enriched for late-stage disease. *American Journal of Human Genetics*, **2004**, 74, 482-94	11	141
306	Expression profiling of the developing and mature Nrl-/- mouse retina: identification of retinal disease candidates and transcriptional regulatory targets of Nrl. *Human Molecular Genetics*, **2004**, 13, 1487-503	5.6	138
305	Molecular analysis of the cDNA for human SPARC/osteonectin/BM-40: sequence, expression, and localization of the gene to chromosome 5q31-q33. *Genomics*, **1988**, 2, 37-47	4.3	138
304	Biomarkers of cardiovascular disease as risk factors for age-related macular degeneration. *Ophthalmology*, **2005**, 112, 2076-80	7.3	132
303	Identification of a rare coding variant in complement 3 associated with age-related macular degeneration. *Nature Genetics*, **2013**, 45, 1375-9	36.3	130
302	Hypomorphic CEP290/NPHP6 mutations result in anosmia caused by the selective loss of G proteins in cilia of olfactory sensory neurons. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 15917-22	11.5	128
301	Leber congenital amaurosis caused by a homozygous mutation (R90W) in the homeodomain of the retinal transcription factor CRX: direct evidence for the involvement of CRX in the development of photoreceptor function. *Human Molecular Genetics*, **1999**, 8, 299-305	5.6	128
300	Molecular Anatomy of the Developing Human Retina. *Developmental Cell*, **2017**, 43, 763-779.e4	10.2	125
299	RPGR-ORF15, which is mutated in retinitis pigmentosa, associates with SMC1, SMC3, and microtubule transport proteins. *Journal of Biological Chemistry*, **2005**, 280, 33580-7	5.4	125
298	Free radical scavenging, antioxidant and cancer chemoprevention by grape seed proanthocyanidin: an overview. *Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis*, **2014**, 768, 69-73	3.3	122
297	Centrosomal-ciliary gene CEP290/NPHP6 mutations result in blindness with unexpected sparing of photoreceptors and visual brain: implications for therapy of Leber congenital amaurosis. *Human Mutation*, **2007**, 28, 1074-83	4.7	121
296	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, 1679-84	11.5	121
295	Accelerated and Improved Differentiation of Retinal Organoids from Pluripotent Stem Cells in Rotating-Wall Vessel Bioreactors. *Stem Cell Reports*, **2018**, 10, 300-313	8	114

294	Transcriptome Dynamics of Developing Photoreceptors in Three-Dimensional Retina Cultures Recapitulates Temporal Sequence of Human Cone and Rod Differentiation Revealing Cell Surface Markers and Gene Networks. *Stem Cells*, **2015**, 33, 3504-18	5.8	112
293	Photoreceptors of Nrl -/- mice coexpress functional S- and M-cone opsins having distinct inactivation mechanisms. *Journal of General Physiology*, **2005**, 125, 287-304	3.4	111
292	The bZIP transcription factor Nrl stimulates rhodopsin promoter activity in primary retinal cell cultures. *Journal of Biological Chemistry*, **1996**, 271, 29612-8	5.4	106
291	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, 1306-10		101
290	Retinoid-related orphan nuclear receptor RORbeta is an early-acting factor in rod photoreceptor development. *Proceedings of the National Academy of Sciences of the United States of America*, **2009**, 106, 17534-9	11.5	100
289	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, 6152-60	6.6	100
288	Evidence of association of APOE with age-related macular degeneration: a pooled analysis of 15 studies. *Human Mutation*, **2011**, 32, 1407-16	4.7	99
287	Premature truncation of a novel protein, RD3, exhibiting subnuclear localization is associated with retinal degeneration. *American Journal of Human Genetics*, **2006**, 79, 1059-70	11	98
286	Photoreceptor sensory cilia and ciliopathies: focus on CEP290, RPGR and their interacting proteins. *Cilia*, **2012**, 1, 22	5.5	97
285	Cone photoreceptors are the main targets for gene therapy of NPHP5 (IQCB1) or NPHP6 (CEP290) blindness: generation of an all-cone Nphp6 hypomorph mouse that mimics the human retinal ciliopathy. *Human Molecular Genetics*, **2011**, 20, 1411-23	5.6	97
284	Barrier to autointegration factor interacts with the cone-rod homeobox and represses its transactivation function. *Journal of Biological Chemistry*, **2002**, 277, 43288-300	5.4	96
283	In vivo function of the orphan nuclear receptor NR2E3 in establishing photoreceptor identity during mammalian retinal development. *Human Molecular Genetics*, **2006**, 15, 2588-602	5.6	95
282	RPGR ORF15 isoform co-localizes with RPGRIP1 at centrioles and basal bodies and interacts with nucleophosmin. *Human Molecular Genetics*, **2005**, 14, 1183-97	5.6	95
281	Long-term survival and differentiation of retinal neurons derived from human embryonic stem cell lines in un-immunosuppressed mouse retina. *Molecular Vision*, **2012**, 18, 920-36	2.3	94
280	Retinal transcriptome and eQTL analyses identify genes associated with age-related macular degeneration. *Nature Genetics*, **2019**, 51, 606-610	36.3	93
279	Mutations in RPGR and RP2 account for 15% of males with simplex retinal degenerative disease **2012**, 53, 8232-7		91
278	Rod differentiation factor NRL activates the expression of nuclear receptor NR2E3 to suppress the development of cone photoreceptors. *Brain Research*, **2008**, 1236, 16-29	3.7	90
277	Inner retinal abnormalities in X-linked retinitis pigmentosa with RPGR mutations. *Investigative Ophthalmology and Visual Science*, **2007**, 48, 4759-65		89

276	Multiple phosphorylated isoforms of NRL are expressed in rod photoreceptors. *Journal of Biological Chemistry*, **2001**, 276, 36824-30	5.4	88
275	A comprehensive analysis of the expression of crystallins in mouse retina. *Molecular Vision*, **2003**, 9, 410-9	2.3	88
274	Spectrum of mutations in the RPGR gene that are identified in 20% of families with X-linked retinitis pigmentosa. *American Journal of Human Genetics*, **1997**, 61, 1287-92	11	86
273	Defects in neural stem cell proliferation and olfaction in Chd7 deficient mice indicate a mechanism for hyposmia in human CHARGE syndrome. *Human Molecular Genetics*, **2009**, 18, 1909-23	5.6	84
272	Mutations in a BTB-Kelch protein, KLHL7, cause autosomal-dominant retinitis pigmentosa. *American Journal of Human Genetics*, **2009**, 84, 792-800	11	84
271	Next-generation sequencing facilitates quantitative analysis of wild-type and Nrl(-/-) retinal transcriptomes. *Molecular Vision*, **2011**, 17, 3034-54	2.3	83
270	Ancestry estimation and control of population stratification for sequence-based association studies. *Nature Genetics*, **2014**, 46, 409-15	36.3	82
269	Developing rods transplanted into the degenerating retina of Crx-knockout mice exhibit neural activity similar to native photoreceptors. *Stem Cells*, **2013**, 31, 1149-59	5.8	81
268	MicroRNA profile of the developing mouse retina **2010**, 51, 1823-31		81
267	In Vitro Modeling Using Ciliopathy-Patient-Derived Cells Reveals Distinct Cilia Dysfunctions Caused by CEP290 Mutations. *Cell Reports*, **2017**, 20, 384-396	10.6	80
266	Transcriptional regulation of rod photoreceptor homeostasis revealed by in vivo NRL targetome analysis. *PLoS Genetics*, **2012**, 8, e1002649	6	80
265	CEP290 alleles in mice disrupt tissue-specific cilia biogenesis and recapitulate features of syndromic ciliopathies. *Human Molecular Genetics*, **2015**, 24, 3775-91	5.6	79
264	Genome-wide association study and meta-analysis of intraocular pressure. *Human Genetics*, **2014**, 133, 41-57	6.3	79
263	Interaction of retinitis pigmentosa GTPase regulator (RPGR) with RAB8A GTPase: implications for cilia dysfunction and photoreceptor degeneration. *Human Molecular Genetics*, **2010**, 19, 3591-8	5.6	79
262	Quantification of Oxygen Consumption in Retina Ex Vivo Demonstrates Limited Reserve Capacity of Photoreceptor Mitochondria **2015**, 56, 8428-36		78
261	Combining Cep290 and Mkks ciliopathy alleles in mice rescues sensory defects and restores ciliogenesis. *Journal of Clinical Investigation*, **2012**, 122, 3025-3025	15.9	78
260	No clinically significant association between CFH and ARMS2 genotypes and response to nutritional supplements: AREDS report number 38. *Ophthalmology*, **2014**, 121, 2173-80	7.3	76
259	Nuclear receptor NR2E3 gene mutations distort human retinal laminar architecture and cause an unusual degeneration. *Human Molecular Genetics*, **2004**, 13, 1893-902	5.6	76

258	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, 17819-24	11.5	75
257	Recruitment of Rod Photoreceptors from Short-Wavelength-Sensitive Cones during the Evolution of Nocturnal Vision in Mammals. *Developmental Cell*, **2016**, 37, 520-32	10.2	74
256	Chondroitinase ABC treatment enhances synaptogenesis between transplant and host neurons in model of retinal degeneration. *Cell Transplantation*, **2007**, 16, 493-503	4	74
255	Microarray analysis of gene expression in the aging human retina. *Investigative Ophthalmology and Visual Science*, **2002**, 43, 2554-60		74
254	Candidate gene association study for diabetic retinopathy in persons with type 2 diabetes: the Candidate gene Association Resource (CARe) **2011**, 52, 7593-602		73
253	Mutation analysis of NR2E3 and NRL genes in Enhanced S Cone Syndrome. *Human Mutation*, **2004**, 24, 439	4.7	73
252	Protein-truncation mutations in the RP2 gene in a North American cohort of families with X-linked retinitis pigmentosa. *American Journal of Human Genetics*, **1999**, 64, 897-900	11	73
251	Complement factor D in age-related macular degeneration **2011**, 52, 8828-34		72
250	Progression of Geographic Atrophy in Age-related Macular Degeneration: AREDS2 Report Number 16. *Ophthalmology*, **2018**, 125, 1913-1928	7.3	71
249	Toll-like receptor polymorphisms and age-related macular degeneration. *Investigative Ophthalmology and Visual Science*, **2008**, 49, 1652-9		71
248	NRL-Regulated Transcriptome Dynamics of Developing Rod Photoreceptors. *Cell Reports*, **2016**, 17, 2460-2473	8.47	70
247	Complement factor H genetic variant and age-related macular degeneration: effect size, modifiers and relationship to disease subtype. *International Journal of Epidemiology*, **2012**, 41, 250-62	7.8	70
246	Differential expression of novel Gs alpha signal transduction protein cDNA species. *Nucleic Acids Research*, **1991**, 19, 4725-9	20.1	70
245	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	20.1	70
244	Gene expression changes in aging retinal microglia: relationship to microglial support functions and regulation of activation. *Neurobiology of Aging*, **2013**, 34, 2310-21	5.6	69
243	Recruitment of the rod pathway by cones in the absence of rods. *Journal of Neuroscience*, **2004**, 24, 7576-82	6.82	69
242	Human retinopathy-associated ciliary protein retinitis pigmentosa GTPase regulator mediates cilia-dependent vertebrate development. *Human Molecular Genetics*, **2010**, 19, 90-8	5.6	68
241	Mutations in TOPORS cause autosomal dominant retinitis pigmentosa with perivascular retinal pigment epithelium atrophy. *American Journal of Human Genetics*, **2007**, 81, 1098-103	11	68

240	Variations in apolipoprotein E frequency with age in a pooled analysis of a large group of older people. *American Journal of Epidemiology*, **2011**, 173, 1357-64	3.8	67
239	Genetic studies of age-related macular degeneration: lessons, challenges, and opportunities for disease management. *Ophthalmology*, **2012**, 119, 2526-36	7.3	66
238	Expression of photoreceptor-specific nuclear receptor NR2E3 in rod photoreceptors of fetal human retina. *Investigative Ophthalmology and Visual Science*, **2004**, 45, 2807-12		66
237	Conditional knockdown of DNA methyltransferase 1 reveals a key role of retinal pigment epithelium integrity in photoreceptor outer segment morphogenesis. *Development (Cambridge)*, **2013**, 140, 1330-41	6.6	64
236	Combining Cep290 and Mkks ciliopathy alleles in mice rescues sensory defects and restores ciliogenesis. *Journal of Clinical Investigation*, **2012**, 122, 1233-45	15.9	64
235	A novel locus (RP24) for X-linked retinitis pigmentosa maps to Xq26-27. *American Journal of Human Genetics*, **1998**, 63, 1439-47	11	63
234	QRX, a novel homeobox gene, modulates photoreceptor gene expression. *Human Molecular Genetics*, **2004**, 13, 1025-40	5.6	62
233	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. *American Journal of Human Genetics*, **1997**, 61, 571-80	11	61
232	Expressed sequence tags and chromosomal localization of cDNA clones from a subtracted retinal pigment epithelium library. *Genomics*, **1992**, 13, 873-6	4.3	61
231	Epigenetic control of gene regulation during development and disease: A view from the retina. *Progress in Retinal and Eye Research*, **2018**, 65, 1-27	20.5	60
230	Two transcription factors can direct three photoreceptor outcomes from rod precursor cells in mouse retinal development. *Journal of Neuroscience*, **2011**, 31, 11118-25	6.6	60
229	Activation of signaling pathways and stress-response genes in an experimental model of retinal detachment. *Investigative Ophthalmology and Visual Science*, **2006**, 47, 1691-5		60
228	Rd9 is a naturally occurring mouse model of a common form of retinitis pigmentosa caused by mutations in RPGR-ORF15. *PLoS ONE*, **2012**, 7, e35865	3.7	59
227	The retinitis pigmentosa protein RP2 interacts with polycystin 2 and regulates cilia-mediated vertebrate development. *Human Molecular Genetics*, **2010**, 19, 4330-44	5.6	59
226	Biochemical characterization and subcellular localization of the mouse retinitis pigmentosa GTPase regulator (mRpgr). *Journal of Biological Chemistry*, **1998**, 273, 19656-63	5.4	59
225	Subunit dissociation and diffusion determine the subcellular localization of rod and cone transducins. *Journal of Neuroscience*, **2007**, 27, 5484-94	6.6	58
224	Localization of the gene for pigment epithelium-derived factor (PEDF) to chromosome 17p13.1 and expression in cultured human retinoblastoma cells. *Genomics*, **1994**, 19, 266-72	4.3	58
223	Remapping of the RP15 locus for X-linked cone-rod degeneration to Xp11.4-p21.1, and identification of a de novo insertion in the RPGR exon ORF15. *American Journal of Human Genetics*, **2000**, 67, 1000-3	11	57

222	Loss of RPGR glutamylation underlies the pathogenic mechanism of retinal dystrophy caused by TTLL5 mutations. *Proceedings of the National Academy of Sciences of the United States of America*, **2016**, 113, E2925-34	11.5	56
221	A simple and efficient cDNA library subtraction procedure: isolation of human retina-specific cDNA clones. *Nucleic Acids Research*, **1991**, 19, 1954	20.1	54
220	Deletion of aryl hydrocarbon receptor AHR in mice leads to subretinal accumulation of microglia and RPE atrophy **2014**, 55, 6031-40		53
219	A long-term efficacy study of gene replacement therapy for RPGR-associated retinal degeneration. *Human Molecular Genetics*, **2015**, 24, 3956-70	5.6	52
218	Retinoic acid regulates the expression of photoreceptor transcription factor NRL. *Journal of Biological Chemistry*, **2006**, 281, 27327-34	5.4	51
217	Ciliopathy-associated gene Cc2d2a promotes assembly of subdistal appendages on the mother centriole during cilia biogenesis. *Nature Communications*, **2014**, 5, 4207	17.4	50
216	Three-dimensional retinal organoids from mouse pluripotent stem cells mimic development with enhanced stratification and rod photoreceptor differentiation. *Molecular Vision*, **2016**, 22, 1077-1094	2.3	50
215	Improved Retinal Organoid Differentiation by Modulating Signaling Pathways Revealed by Comparative Transcriptome Analyses with Development In Vivo. *Stem Cell Reports*, **2019**, 13, 891-905	8	49
214	Whole Exome Sequencing Reveals Mutations in Known Retinal Disease Genes in 33 out of 68 Israeli Families with Inherited Retinopathies. *Scientific Reports*, **2015**, 5, 13187	4.9	49
213	RPGR-associated retinal degeneration in human X-linked RP and a murine model **2012**, 53, 5594-608		49
212	Loss of lysophosphatidylcholine acyltransferase 1 leads to photoreceptor degeneration in rd11 mice. *Proceedings of the National Academy of Sciences of the United States of America*, **2010**, 107, 15523-8	11.5	49
211	Canine RD3 mutation establishes rod-cone dysplasia type 2 (rcd2) as ortholog of human and murine rd3. *Mammalian Genome*, **2009**, 20, 109-23	3.2	49
210	Autosomal recessive retinitis pigmentosa with early macular affectation caused by premature truncation in PROM1 **2010**, 51, 2656-63		48
209	Functional analysis of the rod photoreceptor cGMP phosphodiesterase alpha-subunit gene promoter: Nrl and Crx are required for full transcriptional activity. *Journal of Biological Chemistry*, **2004**, 279, 19800-7	5.4	48
208	Altered expression of genes of the Bmp/Smad and Wnt/calcium signaling pathways in the cone-only Nrl-/- mouse retina, revealed by gene profiling using custom cDNA microarrays. *Journal of Biological Chemistry*, **2004**, 279, 42211-20	5.4	48
207	Increased retinal mtDNA damage in the CFH variant associated with age-related macular degeneration. *Experimental Eye Research*, **2016**, 145, 269-277	3.7	47
206	Ablation of the X-linked retinitis pigmentosa 2 (Rp2) gene in mice results in opsin mislocalization and photoreceptor degeneration **2013**, 54, 4503-11		47
205	Minireview: the role of nuclear receptors in photoreceptor differentiation and disease. *Molecular Endocrinology*, **2012**, 26, 905-15		47

204	Retinitis Pigmentosa GTPase Regulator (RPGR) protein isoforms in mammalian retina: insights into X-linked Retinitis Pigmentosa and associated ciliopathies. *Vision Research*, **2008**, 48, 366-76	2.1	47
203	Evaluation of the ELOVL4 gene in patients with age-related macular degeneration. *Ophthalmic Genetics*, **2001**, 22, 233-9	1.2	47
202	Differential DNA methylation identified in the blood and retina of AMD patients. *Epigenetics*, **2015**, 10, 698-707	5.7	46
201	Associations of CFHR1-CFHR3 deletion and a CFH SNP to age-related macular degeneration are not independent. *Nature Genetics*, **2010**, 42, 553-5; author reply 555-6	36.3	46
200	RP2 phenotype and pathogenetic correlations in X-linked retinitis pigmentosa. *JAMA Ophthalmology*, **2010**, 128, 915-23		45
199	Discordant phenotypes in fraternal twins having an identical mutation in exon ORF15 of the RPGR gene. *JAMA Ophthalmology*, **2008**, 126, 379-84		44
198	Mechanisms of aging in senescence-accelerated mice. *Genome Biology*, **2005**, 6, R48	18.3	44
197	Centrosomal protein CP110 controls maturation of the mother centriole during cilia biogenesis. *Development (Cambridge)*, **2016**, 143, 1491-501	6.6	43
196	An isoform of retinoid-related orphan receptor directs differentiation of retinal amacrine and horizontal interneurons. *Nature Communications*, **2013**, 4, 1813	17.4	43
195	Vision from next generation sequencing: multi-dimensional genome-wide analysis for producing gene regulatory networks underlying retinal development, aging and disease. *Progress in Retinal and Eye Research*, **2015**, 46, 1-30	20.5	43
194	XIAP therapy increases survival of transplanted rod precursors in a degenerating host retina **2011**, 52, 1567-72		43
193	TOPORS, implicated in retinal degeneration, is a cilia-centrosomal protein. *Human Molecular Genetics*, **2011**, 20, 975-87	5.6	43
192	OTX2 loss causes rod differentiation defect in CRX-associated congenital blindness. *Journal of Clinical Investigation*, **2014**, 124, 631-43	15.9	43
191	Clinical and genetic factors associated with progression of geographic atrophy lesions in age-related macular degeneration. *PLoS ONE*, **2015**, 10, e0126636	3.7	43
190	The cellular and compartmental profile of mouse retinal glycolysis, tricarboxylic acid cycle, oxidative phosphorylation, and ~P transferring kinases. *Molecular Vision*, **2016**, 22, 847-85	2.3	43
189	Preservation of cone photoreceptors after a rapid yet transient degeneration and remodeling in cone-only Nrl-/- mouse retina. *Journal of Neuroscience*, **2012**, 32, 528-41	6.6	42
188	Lens-specific gene recruitment of zeta-crystallin through Pax6, Nrl-Maf, and brain suppressor sites. *Molecular and Cellular Biology*, **1998**, 18, 2067-76	4.8	42
187	Ciliary neurotrophic factor induces genes associated with inflammation and gliosis in the retina: a gene profiling study of flow-sorted, Mller cells. *PLoS ONE*, **2011**, 6, e20326	3.7	41

186	A 32 kb critical region excluding Y402H in CFH mediates risk for age-related macular degeneration. *PLoS ONE*, **2011**, 6, e25598	3.7	41
185	Network constrained clustering for gene microarray data. *Bioinformatics*, **2005**, 21, 4014-20	7.2	41
184	Evaluation and optimization of procedures for target labeling and hybridization of cDNA microarrays. *Molecular Vision*, **2002**, 8, 130-7	2.3	41
183	Distinct nuclear localization patterns of DNA methyltransferases in developing and mature mammalian retina. *Journal of Comparative Neurology*, **2011**, 519, 1914-30	3.4	40
182	Rdh12 activity and effects on retinoid processing in the murine retina. *Journal of Biological Chemistry*, **2009**, 284, 21468-77	5.4	40
181	Next generation sequencing technology and genomewide data analysis: Perspectives for retinal research. *Progress in Retinal and Eye Research*, **2016**, 55, 1-31	20.5	39
180	Rapid, Dynamic Activation of Müller Glial Stem Cell Responses in Zebrafish **2016**, 57, 5148-5160		39
179	Excess cones in the retinal degeneration rd7 mouse, caused by the loss of function of orphan nuclear receptor Nr2e3, originate from early-born photoreceptor precursors. *Human Molecular Genetics*, **2011**, 20, 4102-15	5.6	38
178	High throughput screening of co-expressed gene pairs with controlled false discovery rate (FDR) and minimum acceptable strength (MAS). *Journal of Computational Biology*, **2005**, 12, 1029-45	1.7	38
177	Pluripotent stem cell-derived retinal organoids for disease modeling and development of therapies. *Stem Cells*, **2020**, 38, 1206-1215	5.8	37
176	Genome-wide analysis of disease progression in age-related macular degeneration. *Human Molecular Genetics*, **2018**, 27, 929-940	5.6	37
175	Hypomethylation of the IL17RC promoter in peripheral blood leukocytes is not a hallmark of age-related macular degeneration. *Cell Reports*, **2013**, 5, 1527-35	10.6	37
174	Knockdown of Bardet-Biedl syndrome gene BBS9/PTHB1 leads to cilia defects. *PLoS ONE*, **2012**, 7, e34389	3.7	37
173	Nrl-knockout mice deficient in Rpe65 fail to synthesize 11-cis retinal and cone outer segments. *Investigative Ophthalmology and Visual Science*, **2008**, 49, 1126-35		37
172	Cloning and functional expression of human retinal kir2.4, a pH-sensitive inwardly rectifying K(+) channel. *American Journal of Physiology - Cell Physiology*, **2000**, 279, C771-84	5.4	37
171	Pharmacologic fibroblast reprogramming into photoreceptors restores vision. *Nature*, **2020**, 581, 83-88	50.4	37
170	Two novel CRX mutant proteins causing autosomal dominant Leber congenital amaurosis interact differently with NRL. *Human Mutation*, **2010**, 31, E1472-83	4.7	36
169	Inflammation in the pathogenesis of age-related macular degeneration. *British Journal of Ophthalmology*, **2008**, 92, 448-50	5.5	36

168	Maturation arrest in early postnatal sensory receptors by deletion of the miR-183/96/182 cluster in mouse. *Proceedings of the National Academy of Sciences of the United States of America*, **2017**, 114, E4271-E4280	11.5	35
167	Deep-learning-based Prediction of Late Age-Related Macular Degeneration Progression. *Nature Machine Intelligence*, **2020**, 2, 141-150	22.5	35
166	Afferent control of horizontal cell morphology revealed by genetic respecification of rods and cones. *Journal of Neuroscience*, **2007**, 27, 3540-7	6.6	35
165	Mouse eye gene microarrays for investigating ocular development and disease. *Vision Research*, **2002**, 42, 463-70	2.1	35
164	Transcriptome analysis using next generation sequencing reveals molecular signatures of diabetic retinopathy and efficacy of candidate drugs. *Molecular Vision*, **2012**, 18, 1123-46	2.3	35
163	Rare and common variants in extracellular matrix gene Fibrillin 2 (FBN2) are associated with macular degeneration. *Human Molecular Genetics*, **2014**, 23, 5827-37	5.6	34
162	Retinal Histopathology of an XLRP Carrier with a Mutation in the RPGR Exon ORF15. *Experimental Eye Research*, **2002**, 75, 431-443	3.7	34
161	Age-related macular degeneration-associated variants at chromosome 10q26 do not significantly alter ARMS2 and HTRA1 transcript levels in the human retina. *Molecular Vision*, **2010**, 16, 1317-23	2.3	34
160	Null and hypomorph Prickle1 alleles in mice phenocopy human Robinow syndrome and disrupt signaling downstream of Wnt5a. *Biology Open*, **2014**, 3, 861-70	2.2	33
159	Prickle1 is expressed in distinct cell populations of the central nervous system and contributes to neuronal morphogenesis. *Human Molecular Genetics*, **2013**, 22, 2234-46	5.6	33
158	Sumoylation of bZIP transcription factor NRL modulates target gene expression during photoreceptor differentiation. *Journal of Biological Chemistry*, **2010**, 285, 25637-44	5.4	33
157	Development and plasticity of outer retinal circuitry following genetic removal of horizontal cells. *Journal of Neuroscience*, **2013**, 33, 17847-62	6.6	32
156	Regulation of retinal progenitor expansion by Frizzled receptors: implications for microphthalmia and retinal coloboma. *Human Molecular Genetics*, **2012**, 21, 1848-60	5.6	32
155	Bivariate Analysis of Age-Related Macular Degeneration Progression Using Genetic Risk Scores. *Genetics*, **2017**, 206, 119-133	4	31
154	Long-term rescue of cone photoreceptor degeneration in retinitis pigmentosa 2 (RP2)-knockout mice by gene replacement therapy. *Human Molecular Genetics*, **2015**, 24, 6446-58	5.6	31
153	Genetic association study of age-related macular degeneration in the Spanish population. *Acta Ophthalmologica*, **2011**, 89, e12-22	3.7	31
152	The transcription factor neural retina leucine zipper (NRL) controls photoreceptor-specific expression of myocyte enhancer factor Mef2c from an alternative promoter. *Journal of Biological Chemistry*, **2011**, 286, 34893-902	5.4	31
151	The minimal transactivation domain of the basic motif-leucine zipper transcription factor NRL interacts with TATA-binding protein. *Journal of Biological Chemistry*, **2004**, 279, 47233-41	5.4	31

150	Charon BS(+) and (-), versatile lambda phage vectors for constructing directional cDNA libraries and their efficient transfer to plasmids. *Nucleic Acids Research*, **1988**, 16, 8739	20.1	31
149	A secreted WNT-ligand-binding domain of FZD5 generated by a frameshift mutation causes autosomal dominant coloboma. *Human Molecular Genetics*, **2016**, 25, 1382-91	5.6	30
148	Phenotypic conservation in patients with X-linked retinitis pigmentosa caused by RPGR mutations. *JAMA Ophthalmology*, **2013**, 131, 1016-25	3.9	30
147	Retinopathy mutations in the bZIP protein NRL alter phosphorylation and transcriptional activity. *Human Mutation*, **2007**, 28, 589-98	4.7	30
146	Annotation and analysis of 10,000 expressed sequence tags from developing mouse eye and adult retina. *Genome Biology*, **2003**, 4, R65	18.3	30
145	Generation of hydrogen peroxide on oxidation of NADH by hepatic plasma membranes. *Journal of Bioenergetics and Biomembranes*, **1981**, 13, 241-53	3.7	30
144	Treatment Paradigms for Retinal and Macular Diseases Using 3-D Retina Cultures Derived From Human Reporter Pluripotent Stem Cell Lines **2016**, 57, ORSFl1-ORSFl11		30
143	Bi-allelic Truncating Mutations in CEP78, Encoding Centrosomal Protein 78, Cause Cone-Rod Degeneration with Sensorineural Hearing Loss. *American Journal of Human Genetics*, **2016**, 99, 777-784	11	30
142	Genetic architecture of retinal and macular degenerative diseases: the promise and challenges of next-generation sequencing. *Genome Medicine*, **2013**, 5, 84	14.4	28
141	Interaction of retinal bZIP transcription factor NRL with Flt3-interacting zinc-finger protein Fiz1: possible role of Fiz1 as a transcriptional repressor. *Human Molecular Genetics*, **2003**, 12, 365-73	5.6	28
140	Molecular characterization of a novel human gene, SEC13R, related to the yeast secretory pathway gene SEC13, and mapping to a conserved linkage group on human chromosome 3p24-p25 and mouse chromosome 6. *Human Molecular Genetics*, **1994**, 3, 1281-6	5.6	28
139	Sequence analysis, expression and chromosomal localization of a gene, isolated from a subtracted human retina cDNA library, that encodes an insulin-like growth factor binding protein (IGFBP2). *Experimental Eye Research*, **1991**, 52, 549-61	3.7	28
138	Variegated yet non-random rod and cone photoreceptor disease patterns in RPGR-ORF15-associated retinal degeneration. *Human Molecular Genetics*, **2016**, 25, 5444-5459	5.6	27
137	Combinatorial regulation of photoreceptor differentiation factor, neural retina leucine zipper gene NRL, revealed by in vivo promoter analysis. *Journal of Biological Chemistry*, **2011**, 286, 28247-55	5.4	27
136	De novo mutation in the RP1 gene (Arg677ter) associated with retinitis pigmentosa. *Investigative Ophthalmology and Visual Science*, **2003**, 44, 3593-7		26
135	Transgenic mice expressing Cre-recombinase specifically in M- or S-cone photoreceptors. *Investigative Ophthalmology and Visual Science*, **2004**, 45, 42-7		26
134	Distinct signature of altered homeostasis in aging rod photoreceptors: implications for retinal diseases. *PLoS ONE*, **2010**, 5, e13885	3.7	26
133	Long-term follow-up of a family with dominant X-linked retinitis pigmentosa. *Eye*, **2010**, 24, 764-74	4.4	25

132	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. *Genomics*, **1997**, 45, 395-401	4.3	25
131	Madeline 2.0 PDE: a new program for local and web-based pedigree drawing. *Bioinformatics*, **2007**, 23, 1854-6	7.2	25
130	A comprehensive analysis of sequence variants and putative disease-causing mutations in photoreceptor-specific nuclear receptor NR2E3. *Molecular Vision*, **2009**, 15, 2174-84	2.3	25
129	EYS Mutations Causing Autosomal Recessive Retinitis Pigmentosa: Changes of Retinal Structure and Function with Disease Progression. *Genes*, **2017**, 8,	4.2	24
128	Whole exome sequencing reveals GUCY2D as a major gene associated with cone and cone-rod dystrophy in Israel. *Investigative Ophthalmology and Visual Science*, **2014**, 56, 420-30		24
127	The transcription-splicing protein NonO/p54nrb and three NonO-interacting proteins bind to distal enhancer region and augment rhodopsin expression. *Human Molecular Genetics*, **2014**, 23, 2132-44	5.6	24
126	RPGR-containing protein complexes in syndromic and non-syndromic retinal degeneration due to ciliary dysfunction. *Journal of Genetics*, **2009**, 88, 399-407	1.2	24
125	Transcriptome analysis of the retina. *Genome Biology*, **2002**, 3, REVIEWS1022	18.3	23
124	Molecular characterization of the murine neural retina leucine zipper gene, Nrl. *Genomics*, **1993**, 18, 216-22	4.3	23
123	Transcriptome-based molecular staging of human stem cell-derived retinal organoids uncovers accelerated photoreceptor differentiation by 9-cis retinal. *Molecular Vision*, **2019**, 25, 663-678	2.3	23
122	Synergistically acting agonists and antagonists of G protein-coupled receptors prevent photoreceptor cell degeneration. *Science Signaling*, **2016**, 9, ra74	8.8	22
121	Retinal transcriptome profiling by directional next-generation sequencing using 100 ng of total RNA. *Methods in Molecular Biology*, **2012**, 884, 319-34	1.4	22
120	FGF19 exhibits neuroprotective effects on adult mammalian photoreceptors in vitro. *Investigative Ophthalmology and Visual Science*, **2008**, 49, 1696-704		22
119	A CEP290 C-Terminal Domain Complements the Mutant CEP290 of Rd16 Mice In Trans and Rescues Retinal Degeneration. *Cell Reports*, **2018**, 25, 611-623.e6	10.6	22
118	RNA Biology in Retinal Development and Disease. *Trends in Genetics*, **2018**, 34, 341-351	8.5	21
117	Regulation of a novel isoform of Receptor Expression Enhancing Protein REEP6 in rod photoreceptors by bZIP transcription factor NRL. *Human Molecular Genetics*, **2014**, 23, 4260-71	5.6	21
116	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. *Ophthalmology*, **1998**, 105, 2286-96	7.3	21
115	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, 215-23	1.2	21

114	Dietary Nutrient Intake and Progression to Late Age-Related Macular Degeneration in the Age-Related Eye Disease Studies 1 and 2. *Ophthalmology*, **2021**, 128, 425-442	7.3	21
113	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, 105-14	3.6	20
112	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. *Ophthalmology*, **2018**, 125, 559-568	7.3	19
111	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	3.7	19
110	Mouse model of human RPE65 P25L hypomorph resembles wild type under normal light rearing but is fully resistant to acute light damage. *Human Molecular Genetics*, **2015**, 24, 4417-28	5.6	18
109	Gene expression profile of native human retinal pigment epithelium. *Investigative Ophthalmology and Visual Science*, **2002**, 43, 603-7		18
108	Retinal histopathology of an XLRP carrier with a mutation in the RPGR exon ORF15. *Experimental Eye Research*, **2002**, 75, 431-43	3.7	18
107	Retinal Organoids derived from hiPSCs of an AIPL1-LCA Patient Maintain Cytoarchitecture despite Reduced levels of Mutant AIPL1. *Scientific Reports*, **2020**, 10, 5426	4.9	17
106	Patient iPSC-derived neural stem cells exhibit phenotypes in concordance with the clinical severity of mucopolysaccharidosis I. *Human Molecular Genetics*, **2018**, 27, 3612-3626	5.6	17
105	Phenotype associated with mutation in the recently identified autosomal dominant retinitis pigmentosa KLHL7 gene. *JAMA Ophthalmology*, **2010**, 128, 772-8		17
104	NRL S50T mutation and the importance of founder effectsNn inherited retinal dystrophies. *European Journal of Human Genetics*, **2000**, 8, 783-7	5.3	17
103	Genetic components in diabetic retinopathy. *Indian Journal of Ophthalmology*, **2016**, 64, 55-61	1.6	17
102	Primary cilia biogenesis and associated retinal ciliopathies. *Seminars in Cell and Developmental Biology*, **2021**, 110, 70-88	7.5	17
101	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	5.6	16
100	Feedback induction of a photoreceptor-specific isoform of retinoid-related orphan nuclear receptor by the rod transcription factor NRL. *Journal of Biological Chemistry*, **2014**, 289, 32469-80	5.4	16
99	Increasing evidence for syndromic phenotypes associated with RPGR mutations. *American Journal of Ophthalmology*, **2004**, 137, 785-6; author reply 786	4.9	16
98	Assessment of Novel Genome-Wide Significant Gene Loci and Lesion Growth in Geographic Atrophy Secondary to Age-Related Macular Degeneration. *JAMA Ophthalmology*, **2019**, 137, 867-876	3.9	15
97	Regulation of Noncoding Transcriptome in Developing Photoreceptors by Rod Differentiation Factor NRL **2017**, 58, 4422-4435		15

96	A role for prenylated rab acceptor 1 in vertebrate photoreceptor development. *BMC Neuroscience*, **2012**, 13, 152	3.2	14
95	Preventing polyglutamine-induced activation of c-Jun delays neuronal dysfunction in a mouse model of SCA7 retinopathy. *Neurobiology of Disease*, **2007**, 25, 571-81	7.5	14
94	Characterization and sequence analysis of the human homeobox-containing gene GBX2. *Genomics*, **1996**, 31, 335-42	4.3	14
93	Protective gene expression changes elicited by an inherited defect in photoreceptor structure. *PLoS ONE*, **2012**, 7, e31371	3.7	14
92	A simple and efficient method for generating human retinal organoids. *Molecular Vision*, **2020**, 26, 97-105	2.3	14
91	Aberrant RNA splicing is the major pathogenic effect in a knock-in mouse model of the dominantly inherited c.1430A>G human RPE65 mutation. *Human Mutation*, **2019**, 40, 426-443	4.7	14
90	Adherence to a Mediterranean diet and cognitive function in the Age-Related Eye Disease Studies 1 & 2. *Alzheimer's and Dementia*, **2020**, 16, 831-842	1.2	14
89	Adherence to the Mediterranean Diet and Progression to Late Age-Related Macular Degeneration in the Age-Related Eye Disease Studies 1 and 2. *Ophthalmology*, **2020**, 127, 1515-1528	7.3	13
88	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. *Human Mutation*, **2015**, 36, 836-41	4.7	13
87	Gene Therapy of Dominant CRX-Leber Congenital Amaurosis using Patient Stem Cell-Derived Retinal Organoids. *Stem Cell Reports*, **2021**, 16, 252-263	8	13
86	Geographic atrophy in age-related macular degeneration and TLR3. *New England Journal of Medicine*, **2009**, 360, 2254-5; author reply 2255-6	59.2	13
85	No CFH or ARMS2 Interaction with Omega-3 Fatty Acids, Low versus High Zinc, or β-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. *Ophthalmology*, 2019, 126, 1541-1548	7.3	12
84	Global expression profiling of peripheral Qa-1-restricted CD8αβ TCRαβ regulatory T cells reveals innate-like features: implications for immune-regulatory repertoire. *Human Immunology*, **2012**, 73, 214-22	2.3	12
83	The combination of whole-exome sequencing and clinical analysis allows better diagnosis of rare syndromic retinal dystrophies. *Acta Ophthalmologica*, **2019**, 97, e877-e886	3.7	11
82	Cone-rod homeobox CRX controls presynaptic active zone formation in photoreceptors of mammalian retina. *Human Molecular Genetics*, **2018**, 27, 3555-3567	5.6	11
81	Retinal disease in ciliopathies: Recent advances with a focus on stem cell-based therapies. *Translational Science of Rare Diseases*, **2019**, 4, 97-115	3.3	11
80	Multicriteria Gene Screening for Analysis of Differential Expression with DNA Microarrays. *Eurasip Journal on Advances in Signal Processing*, **2004**, 2004, 1	1.9	11
79	Neural retina-specific leucine zipper gene NRL (D14S46E) maps to human chromosome 14q11.1-q11.2. *Genomics*, **1992**, 14, 491-2	4.3	11

78	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	2.3	11
77	Molecular Diagnosis of Inherited Retinal Diseases in Indigenous African Populations by Whole-Exome Sequencing **2016**, 57, 6374-6381		11
76	The golden era of ocular disease gene discovery: race to the finish. *Clinical Genetics*, **2013**, 84, 99-101	4	10
75	Five novel RPGR mutations in families with X-linked retinitis pigmentosa. *Human Mutation*, **2001**, 17, 151	4.7	10
74	Gene-Based Association Analysis for Censored Traits Via Fixed Effect Functional Regressions. *Genetic Epidemiology*, **2016**, 40, 133-43	2.6	10
73	Tbx2a Modulates Switching of RH2 and LWS Opsin Gene Expression. *Molecular Biology and Evolution*, **2020**, 37, 2002-2014	8.3	9
72	Family-based exome sequencing identifies rare coding variants in age-related macular degeneration. *Human Molecular Genetics*, **2020**, 29, 2022-2034	5.6	9
71	Characterization of new transcripts enriched in the mouse retina and identification of candidate retinal disease genes. *Investigative Ophthalmology and Visual Science*, **2004**, 45, 3313-9		9
70	pSH4: a mammalian cDNA expression vector. *Nucleic Acids Research*, **1990**, 18, 3668	20.1	9
69	Analysis of RP2 and RPGR Mutations in Five X-Linked Chinese Families with Retinitis Pigmentosa. *Scientific Reports*, **2017**, 7, 44465	4.9	8
68	A new 2-base pair deletion in the RPGR gene in a black family with X-linked retinitis pigmentosa. *JAMA Ophthalmology*, **1998**, 116, 213-8		8
67	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, 71-4	1.9	8
66	Genetic Analysis of the Rhodopsin Gene Identifies a Mosaic Dominant Retinitis Pigmentosa Mutation in a Healthy Individual **2016**, 57, 940-7		8
65	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	2.3	8
64	Gene discovery using Pareto depth sampling distributions. *Journal of the Franklin Institute*, **2004**, 341, 55-75	4	7
63	Senile panretinal cone dysfunction in age-related macular degeneration (AMD): a report of 52 amd patients compared to age-matched controls. *Transactions of the American Ophthalmological Society*, **2006**, 104, 232-40		7
62	Association of Rare Predicted Loss-of-Function Variants in Cellular Pathways with Sub-Phenotypes in Age-Related Macular Degeneration. *Ophthalmology*, **2018**, 125, 398-406	7.3	7
61	Genome-wide Profiling Identifies DNA Methylation Signatures of Aging in Rod Photoreceptors Associated with Alterations in Energy Metabolism. *Cell Reports*, **2020**, 31, 107525	10.6	6

60	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	10.2	6
59	Seeing the unseen: Microarray-based gene expression profiling in vision. *Investigative Ophthalmology and Visual Science*, **2004**, 45, 2457-62		6
58	Exome sequencing: capture and sequencing of all human coding regions for disease gene discovery. *Methods in Molecular Biology*, **2012**, 884, 335-51	1.4	6
57	The transcription factor GTF2IRD1 regulates the topology and function of photoreceptors by modulating photoreceptor gene expression across the retina. *Journal of Neuroscience*, **2014**, 34, 15356-68	6.6	5
56	Chromosomal localization and cDNA sequence of human ralB, a GTP binding protein. *Somatic Cell and Molecular Genetics*, **1990**, 16, 407-10		5
55	Integration of genomics and transcriptomics predicts diabetic retinopathy susceptibility genes. *ELife*, **2020**, 9,	8.9	5
54	An optimized protocol for retina single-cell RNA sequencing. *Molecular Vision*, **2020**, 26, 705-717	2.3	5
53	A mega-analysis of expression quantitative trait loci in retinal tissue. *PLoS Genetics*, **2020**, 16, e1008934	6	5
52	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	5.6	5
51	Proneural genes define ground-state rules to regulate neurogenic patterning and cortical folding. *Neuron*, **2021**, 109, 2847-2863.e11	13.9	5
50	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	2.9	4
49	Pias3 is necessary for dorso-ventral patterning and visual response of retinal cones but is not required for rod photoreceptor differentiation. *Biology Open*, **2017**, 6, 881-890	2.2	4
48	Clinical expression of X-linked retinitis pigmentosa in a Swedish family with the RP2 genotype. *Ophthalmic Genetics*, **1998**, 19, 187-96	1.2	4
47	Loss of endocytosis-associated RabGEF1 causes aberrant morphogenesis and altered autophagy in photoreceptors leading to retinal degeneration. *PLoS Genetics*, **2020**, 16, e1009259	6	4
46	Restoration of RPGR expression in vivo using CRISPR/Cas9 gene editing. *Gene Therapy*, **2021**,	4	4
45	Molecular dissection of cone photoreceptor-enriched genes encoding transmembrane and secretory proteins. *Journal of Neuroscience Research*, **2019**, 97, 16-28	4.4	4
44	Genome-Wide Association Studies-Based Machine Learning for Prediction of Age-Related Macular Degeneration Risk. *Translational Vision Science and Technology*, **2021**, 10, 29	3.3	4
43	Aging of the Retina: Molecular and Metabolic Turbulences and Potential Interventions. *Annual Review of Vision Science*, **2021**, 7, 633-664	8.2	4

42	Retinal phenotype of an X-linked pseudo-Usher syndrome in association with the G173R mutation in the RPGR gene. *Advances in Experimental Medicine and Biology*, **2008**, 613, 221-7	3.6	4
41	What's in a name? RPGR mutations redefine the genetic and phenotypic landscape in retinal degenerative diseases **2013**, 54, 1417		3
40	Mutations associated with retinopathies alter mitogen-activated protein kinase-induced phosphorylation of neural retina leucine-zipper. *Molecular Vision*, **2007**, 13, 1114-20	2.3	3
39	Expression of deubiquitinating enzyme genes in the developing mammal retina. *Molecular Vision*, **2019**, 25, 800-813	2.3	3
38	A unique -associated variant in a Georgian Jewish family with probable North Carolina macular dystrophy and the possible contribution of a unique variant. *Molecular Vision*, **2020**, 26, 299-310	2.3	3
37	Proneural genes define ground state rules to regulate neurogenic patterning and cortical folding		3
36	Accelerated Development of Rod Photoreceptors in Retinal Organoids Derived from Human Pluripotent Stem Cells by Supplementation with 9- Retinal. *STAR Protocols*, **2020**, 1, 100033-100033	1.4	3
35	Retinal pigment epithelium transcriptome analysis in chronic smoking reveals a suppressed innate immune response and activation of differentiation pathways. *Free Radical Biology and Medicine*, **2020**, 156, 176-189	7.8	2
34	Determination of Mitochondrial Oxygen Consumption in the Retina Ex Vivo: Applications for Retinal Disease. *Methods in Molecular Biology*, **2018**, 1753, 167-177	1.4	2
33	lambda SHK and lambda AASV: phage vectors for efficient cDNA cloning and expression in mammalian cells. *Gene*, **1993**, 123, 287-8	3.8	2
32	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.. *Human Molecular Genetics*, **2022**,	5.6	2
31	SSBP1 faux pas in mitonuclear tango causes optic neuropathy. *Journal of Clinical Investigation*, **2020** , 130, 62-64	15.9	2
30	Association of Smoking, Alcohol Consumption, Blood Pressure, Body Mass Index, and Glycemic Risk Factors With Age-Related Macular Degeneration: A Mendelian Randomization Study. *JAMA Ophthalmology*, **2021**,	3.9	2
29	Construction of Directional cDNA Libraries from Human Retinal Tissue/Cells and Their Enrichment for Specific Genes Using an Efficient Subtraction Procedure. *Methods in Neurosciences*, **1993**, 285-300		2
28	GWAS-based Machine Learning for Prediction of Age-Related Macular Degeneration Risk		2
27	Transcriptome-based molecular staging of human stem cell-derived retinal organoids uncovers accelerated photoreceptor differentiation by 9-cis retinal		2
26	HIPRO: A High-Efficiency, Hypoxia-Induced Protocol for Generation of Photoreceptors in Retinal Organoids from Mouse Pluripotent Stem Cells. *STAR Protocols*, **2020**, 1, 100018-100018	1.4	2
25	Divergent Effects of HSP70 Overexpression in Photoreceptors During Inherited Retinal Degeneration **2020**, 61, 25		2

24	Glycogen Synthase Kinase 3 regulates the genesis of the rare displaced ganglion cell retinal subtype		2
23	A Novel Gene Mutation Associated With Autosomal Dominant Retinal Degeneration. *Frontiers in Cell and Developmental Biology*, **2021**, 9, 720782	5.7	2
22	Dinucleotide polymorphism at the DXS1178 locus is tightly linked to PGK1 at Xq13. *Human Genetics* , **1995**, 95, 467-8	6.3	1
21	Dinucleotide repeat polymorphism at the DXS977 locus. *Human Molecular Genetics*, **1994**, 3, 1030	5.6	1
20	HiCRes: a computational method to estimate and predict the resolution of HiC libraries		1
19	Deep-learning-based Prediction of Late Age-Related Macular Degeneration Progression		1
18	Glycogen Synthase Kinase 3 Regulates the Genesis of Displaced Retinal Ganglion Cells3. *ENeuro*, **2021**, 8,	3.9	1
17	Making Biological Sense of Genetic Studies of Age-Related Macular Degeneration. *Advances in Experimental Medicine and Biology*, **2021**, 1256, 201-219	3.6	1
16	X-Linked Retinitis Pigmentosa: Current Status **2001**, 11-22		1
15	Determination of posttranslational modifications of photoreceptor differentiation factor NRL: focus on SUMOylation. *Methods in Molecular Biology*, **2012**, 884, 353-61	1.4	0
14	Mapping of the neural retina leucine zipper gene, Nrl, to mouse chromosome 14. *Mammalian Genome*, **1993**, 4, 618-20	3.2	0
13	Genetics and therapy for pediatric eye diseases. *EBioMedicine*, **2021**, 67, 103360	8.8	0
12	GATD3A, a mitochondrial deglycase with evolutionary origins from gammaproteobacteria, restricts the formation of advanced glycation end products.. *BMC Biology*, **2022**, 20, 68	7.3	0
11	Reply. *Ophthalmology*, **2020**, 127, e19-e20	7.3	
10	[11]Isolation of candidate genes for inherited diseases: Application to X-linked retinal degenerations. *Methods in Molecular Genetics*, **1996**, 8, 207-228		
9	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		
8	A Sandwich-Hybridization Method for Specific and Efficient Selection of cDNA Clones from Genomic Regions **1994**, 91-99		
7	Photoreceptor Degeneration: Molecular Mechanisms of Photoreceptor Degeneration **2014**, 275-308		

6 Loss of endocytosis-associated RabGEF1 causes aberrant morphogenesis and altered autophagy in photoreceptors leading to retinal degeneration **2020**, 16, e1009259

5 Loss of endocytosis-associated RabGEF1 causes aberrant morphogenesis and altered autophagy in photoreceptors leading to retinal degeneration **2020**, 16, e1009259

4 Loss of endocytosis-associated RabGEF1 causes aberrant morphogenesis and altered autophagy in photoreceptors leading to retinal degeneration **2020**, 16, e1009259

3 Loss of endocytosis-associated RabGEF1 causes aberrant morphogenesis and altered autophagy in photoreceptors leading to retinal degeneration **2020**, 16, e1009259

2 Loss of endocytosis-associated RabGEF1 causes aberrant morphogenesis and altered autophagy in photoreceptors leading to retinal degeneration **2020**, 16, e1009259

1 Loss of endocytosis-associated RabGEF1 causes aberrant morphogenesis and altered autophagy in photoreceptors leading to retinal degeneration **2020**, 16, e1009259