Shomi S Bhattacharya

List of Publications by Year
in descending order

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173 papers	15,261 citations	22153 59 h-index	20358 116 g-index
174 all docs	174 docs citations	174 times ranked	12968 citing authors

#	Article	IF	Citations
1	Effect of Gene Therapy on Visual Function in Leber's Congenital Amaurosis. New England Journal of Medicine, 2008, 358, 2231-2239.	27.0	1,793
2	OPA1, encoding a dynamin-related GTPase, is mutated in autosomal dominant optic atrophy linked to chromosome 3q28. Nature Genetics, 2000, 26, 211-215.	21.4	1,169
3	Cone-Rod Dystrophy Due to Mutations in a Novel Photoreceptor-Specific Homeobox Gene (CRX) Essential for Maintenance of the Photoreceptor. Cell, 1997, 91, 543-553.	28.9	520
4	Photoreceptor degeneration: genetic and mechanistic dissection of a complex trait. Nature Reviews Genetics, 2010, 11, 273-284.	16.3	519
5	Mutations in a human homologue of Drosophila crumbs cause retinitis pigmentosa (RP12). Nature Genetics, 1999, 23, 217-221.	21.4	427
6	Fox's in development and disease. Trends in Genetics, 2003, 19, 339-344.	6.7	316
7	A Human Homolog of Yeast Pre-mRNA Splicing Gene, PRP31, Underlies Autosomal Dominant Retinitis Pigmentosa on Chromosome 19q13.4 (RP11). Molecular Cell, 2001, 8, 375-381.	9.7	305
8	Restoration of photoreceptor ultrastructure and function in retinal degeneration slow mice by gene therapy. Nature Genetics, 2000, 25, 306-310.	21.4	295
9	Mutations in a new photoreceptor-pineal gene on 17p cause Leber congenital amaurosis. Nature Genetics, 2000, 24, 79-83.	21.4	257
10	A common allele in RPGRIP1L is a modifier of retinal degeneration in ciliopathies. Nature Genetics, 2009, 41, 739-745.	21.4	255
11	Mutations in HPRP3, a third member ofpre-mRNA splicing factor genes, implicated in autosomal dominant retinitis pigmentosa. Human Molecular Genetics, 2002, 11, 87-92.	2.9	217
12	Gene transfer into the mouse retina mediated by an adeno-associated viral vector. Human Molecular Genetics, 1996, 5, 591-594.	2.9	209
13	Alpha-B Crystallin Gene (CRYAB) Mutation Causes Dominant Congenital Posterior Polar Cataract in Humans. American Journal of Human Genetics, 2001, 69, 1141-1145.	6.2	208
14	Molecular genetic basis of inherited cataract and associated phenotypes. Survey of Ophthalmology, 2004, 49, 300-315.	4.0	208
15	TRPM1 Is Mutated in Patients with Autosomal-Recessive Complete Congenital Stationary Night Blindness. American Journal of Human Genetics, 2009, 85, 720-729.	6.2	207
16	A mutation in NRL is associated with autosomal dominant retinitis pigmentosa. Nature Genetics, 1999, 21, 355-356.	21.4	205
17	Mutations ofVMD2Splicing Regulators Cause Nanophthalmos and Autosomal Dominant Vitreoretinochoroidopathy (ADVIRC). , 2004, 45, 3683.		205
18	Genome-wide association analyses identify three new susceptibility loci for primary angle closure glaucoma. Nature Genetics, 2012, 44, 1142-1146.	21.4	196

#	Article	IF	Citations
19	A new locus for autosomal dominant retinitis pigmentosa on chromosome 7p. Nature Genetics, 1993, 4, 51-53.	21.4	183
20	EYS, encoding an ortholog of Drosophila spacemaker, is mutated in autosomal recessive retinitis pigmentosa. Nature Genetics, 2008, 40, 1285-1287.	21.4	175
21	NMNAT1 mutations cause Leber congenital amaurosis. Nature Genetics, 2012, 44, 1040-1045.	21.4	171
22	CRB1 mutations in inherited retinal dystrophies. Human Mutation, 2012, 33, 306-315.	2.5	153
23	GCAP1(Y99C) Mutant Is Constitutively Active in Autosomal Dominant Cone Dystrophy. Molecular Cell, 1998, 2, 129-133.	9.7	150
24	Genome-wide association study identifies five new susceptibility loci for primary angle closure glaucoma. Nature Genetics, 2016, 48, 556-562.	21.4	147
25	Prevalence of AIPL1 Mutations in Inherited Retinal Degenerative Disease. Molecular Genetics and Metabolism, 2000, 70, 142-150.	1.1	144
26	Development and application of a next-generation-sequencing (NGS) approach to detect known and novel gene defects underlying retinal diseases. Orphanet Journal of Rare Diseases, 2012, 7, 8.	2.7	144
27	Connexin 50 mutation in a family with congenital "zonular nuclear" pulverulent cataract of Pakistani origin. Human Genetics, 1999, 105, 168-170.	3.8	139
28	Clinical Features and Course of Patients with Glaucoma with the E50K Mutation in the Optineurin Gene. , 2005, 46, 2816.		127
29	Chromosomal Duplication Involving the Forkhead Transcription Factor GeneFOXC1Causes Iris Hypoplasia and Glaucoma. American Journal of Human Genetics, 2000, 67, 1129-1135.	6.2	127
30	Expression ofPRPF31mRNA in Patients with Autosomal Dominant Retinitis Pigmentosa: A Molecular Clue for Incomplete Penetrance?. , 2003, 44, 4204.		125
31	A major marker for normal tension glaucoma: association with polymorphisms in the OPA1 gene. Human Genetics, 2002, 110, 52-56.	3.8	123
32	Whole-Exome Sequencing Identifies Mutations in GPR179 Leading to Autosomal-Recessive Complete Congenital Stationary Night Blindness. American Journal of Human Genetics, 2012, 90, 321-330.	6.2	121
33	Whole-Exome Sequencing Identifies LRIT3 Mutations as a Cause of Autosomal-Recessive Complete Congenital Stationary Night Blindness. American Journal of Human Genetics, 2013, 92, 67-75.	6.2	120
34	Adeno-Associated Virus Gene Transfer to Mouse Retina. Human Gene Therapy, 1998, 9, 81-86.	2.7	118
35	Identification of a sixth locus for autosomal dominant retinitis pigmentosa on chromosome 19. Human Molecular Genetics, 1994, 3, 351-354.	2.9	116
36	Identification and Functional Consequences of a New Mutation (E155G) in the Gene for GCAP1 That Causes Autosomal Dominant Cone Dystrophy. American Journal of Human Genetics, 2001, 69, 471-480.	6.2	115

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37	Premature Truncation of a Novel Protein, RD3, Exhibiting Subnuclear Localization Is Associated with Retinal Degeneration. American Journal of Human Genetics, 2006, 79, 1059-1070.	6.2	112
38	CNOT3 Is a Modifier of PRPF31 Mutations in Retinitis Pigmentosa with Incomplete Penetrance. PLoS Genetics, 2012, 8, e1003040.	3.5	109
39	Concise Review: Reactive Astrocytes and Stem Cells in Spinal Cord Injury: Good Guys or Bad Guys?. Stem Cells, 2015, 33, 1036-1041.	3.2	108
40	Chromosomal Duplication Involving the Forkhead Transcription Factor Gene FOXC1 Causes Iris Hypoplasia and Glaucoma. American Journal of Human Genetics, 2000, 67, 1129-1135.	6.2	105
41	The Phenotype of Leber Congenital Amaurosis in Patients With AIPL1 Mutations. JAMA Ophthalmology, 2004, 122, 1029.	2.4	105
42	An Integrated, Functionally Annotated Gene Map of the DXS8026â€“ELK1 Interval on Human Xp11.3â€“Xp11.23: Potential Hotspot for Neurogenetic Disorders. Genomics, 2002, 79, 560-572.	2.9	103
43	Mutations of theEPHA2receptor tyrosine kinase gene cause autosomal dominant congenital cataract. Human Mutation, 2009, 30, E603-E611.	2.5	96
44	Molecular genetics and prospects for therapy of the inherited retinal dystrophies. Current Opinion in Genetics and Development, 2001, 11, 307-316.	3.3	92
45	Mutations in a BTB-Kelch Protein, KLHL7, Cause Autosomal-Dominant Retinitis Pigmentosa. American Journal of Human Genetics, 2009, 84, 792-800.	6.2	89
46	Dominant cataract formation in association with a vimentin assembly disrupting mutation. Human Molecular Genetics, 2009, 18, 1052-1057.	2.9	88
47	Mutations in a protein target of the Pim-1 kinase associated with the RP9 form of autosomal dominant retinitis pigmentosa. European Journal of Human Genetics, 2002, 10, 245-249.	2.8	87
48	EYS is a major gene for rod-cone dystrophies in France. Human Mutation, 2010, 31, E1406-E1435.	2.5	86
49	Functional characterization of missense mutations at codon 838 in retinal guanylate cyclase correlates with disease severity in patients with autosomal dominant cone-rod dystrophy. Human Molecular Genetics, 2000, 9, 3065-3073.	2.9	83
50	Mutant carbonic anhydrase 4 impairs pH regulation and causes retinal photoreceptor degeneration. Human Molecular Genetics, 2005, 14, 255-265.	2.9	79
51	Complement factor H genetic variant and age-related macular degeneration: effect size, modifiers and relationship to disease subtype. International Journal of Epidemiology, 2012, 41, 250-262.	1.9	79
52	267 Spanish Exomes Reveal Population-Specific Differences in Disease-Related Genetic Variation. Molecular Biology and Evolution, 2016, 33, 1205-1218.	8.9	78
53	Mutations in TOPORS Cause Autosomal Dominant Retinitis Pigmentosa with Perivascular Retinal Pigment Epithelium Atrophy. American Journal of Human Genetics, 2007, 81, 1098-1103.	6.2	77
54	Retinitis pigmentosa associated with rhodopsin mutations: Correlation between phenotypic variability and molecular effects. Vision Research, 2006, 46, 4556-4567.	1.4	76

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55	Disease mechanism for retinitis pigmentosa (RP11) caused by mutations in the splicing factor gene PRPF31. Human Molecular Genetics, 2002, 11, 3209-3219.	2.9	75
56	Novel mutations in MERTK associated with childhood onset rod-cone dystrophy. Molecular Vision, 2010, 16, 369-77.	1.1	73
57	Three Gene-Targeted Mouse Models of RNA Splicing Factor RP Show Late-Onset RPE and Retinal Degeneration. , 2011, 52, 190.		70
58	Mutation spectrum of EYS in Spanish patients with autosomal recessive retinitis pigmentosa. Human Mutation, 2010, 31, E1772-E1800.	2.5	69
59	ABCC5, a Gene That Influences the Anterior Chamber Depth, Is Associated with Primary Angle Closure Glaucoma. PLoS Genetics, 2014, 10, e1004089.	3.5	68
60	High frequency of persistent hyperplastic primary vitreous and cataracts in p53-deficient mice. Cell Death and Differentiation, 1998, 5, 156-162.	11.2	67
61	Posterior Polymorphous Corneal Dystrophy in Czech Families Maps to Chromosome 20 and Excludes theVSX1Gene. , 2005, 46, 4480.		67
62	Novel mutations in theZEB1 gene identified in Czech and British patients with posterior polymorphous corneal dystrophy. Human Mutation, 2007, 28, 638-638.	2.5	67
63	Biallelic Variants in TTLL5, Encoding a Tubulin Glutamylase, Cause Retinal Dystrophy. American Journal of Human Genetics, 2014, 94, 760-769.	6.2	67
64	Ocular developmental abnormalities and glaucoma associated with interstitial 6p25 duplications and deletions. Investigative Ophthalmology and Visual Science, 2002, 43, 1843-9.	3.3	63
65	Mutations in Pre-mRNA Processing Factors 3, 8, and 31 Cause Dysfunction of the Retinal Pigment Epithelium. American Journal of Pathology, 2014, 184, 2641-2652.	3.8	62
66	Genetic blindness: current concepts in the pathogenesis of human outer retinal dystrophies. Trends in Genetics, 1998, 14, 103-108.	6.7	60
67	Mutations in the Gene Coding for the Pre-mRNA Splicing Factor,PRPF31, in Patients with Autosomal Dominant Retinitis Pigmentosa. , 2007, 48, 1330.		60
68	A frameshift mutation in exon 28 of the OPA1 gene explains the high prevalence of dominant optic atrophy in the Danish population: evidence for a founder effect. Human Genetics, 2001, 109, 498-502.	3.8	59
69	Identification of Novel Mutations in the Ortholog ofDrosophilaEyes Shut Gene (EYS) Causing Autosomal Recessive Retinitis Pigmentosa. , 2010, 51, 4266.		57
70	An Assessment of the Apex Microarray Technology in Genotyping Patients with Leber Congenital Amaurosis and Early-Onset Severe Retinal Dystrophy. , 2007, 48, 5684.		56
71	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, 15523-15528.	7.1	55
72	Confirmation of the rod cGMP phosphodiesterase Î² subunit (PDEÎ²) nonsense mutation in affected rcd-1 Irish setters in the UK and development of a diagnostic test. Current Eye Research, 1993, 12, 861-866.	1.5	54

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73	Study of Gene-Targeted Mouse Models of Splicing Factor GenePrpf31Implicated in Human Autosomal Dominant Retinitis Pigmentosa (RP). , 2009, 50, 5927.		52
74	Rare and common variants in extracellular matrix gene Fibrillin 2 (FBN2) are associated with macular degeneration. Human Molecular Genetics, 2014, 23, 5827-5837.	2.9	52
75	Hypoxia Enhances the Generation of Retinal Progenitor Cells from Human Induced Pluripotent and Embryonic Stem Cells. Stem Cells and Development, 2012, 21, 1344-1355.	2.1	51
76	Wolfram gene (WFS1) mutation causes autosomal dominant congenital nuclear cataract in humans. European Journal of Human Genetics, 2013, 21, 1356-1360.	2.8	50
77	Investigating the association between OPA1 polymorphisms and glaucoma: comparison between normal tension and high tension primary open angle glaucoma. Human Genetics, 2002, 110, 513-514.	3.8	49
78	Prevalence and novelty of PRPF31 mutations in French autosomal dominant rod-cone dystrophy patients and a review of published reports. BMC Medical Genetics, 2010, 11, 145.	2.1	49
79	TOPORS, implicated in retinal degeneration, is a cilia-centrosomal protein. Human Molecular Genetics, 2011, 20, 975-987.	2.9	49
80	Cleavage of Mer Tyrosine Kinase (MerTK) from the Cell Surface Contributes to the Regulation of Retinal Phagocytosis. Journal of Biological Chemistry, 2015, 290, 4941-4952.	3.4	49
81	A large deletion in the adRP gene PRPF31: evidence that haploinsufficiency is the cause of disease. Molecular Vision, 2006, 12, 384-8.	1.1	49
82	Maculopathy Due to the R345W Substitution in Fibulin-3: Distinct Clinical Features, Disease Variability, and Extent of Retinal Dysfunction. , 2006, 47, 3085.		48
83	Human iPSC derived disease model of MERTK-associated retinitis pigmentosa. Scientific Reports, 2015, 5, 12910.	3.3	47
84	RDH12 retinopathy: novel mutations and phenotypic description. Molecular Vision, 2011, 17, 2706-16.	1.1	47
85	Mutations in splicing factor PRPF3, causing retinal degeneration, form detrimental aggregates in photoreceptor cells. Human Molecular Genetics, 2007, 16, 1699-1707.	2.9	46
86	The ABCA4 2588G>C Stargardt mutation: single origin and increasing frequency from South-West to North-East Europe. European Journal of Human Genetics, 2002, 10, 197-203.	2.8	45
87	Spectrum of Rhodopsin Mutations in French Autosomal Dominant Rodâ€“Cone Dystrophy Patients. , 2010, 51, 3687.		45
88	Retinal pigment epithelium degeneration caused by aggregation of PRPF31 and the role of HSP70 family of proteins. Molecular Medicine, 2020, 26, 1.	4.4	45
89	An eighth locus for autosomal dominant retinitis pigmentosa is linked to chromosome 17q. Human Molecular Genetics, 1995, 4, 1459-1462.	2.9	44
90	Characterization of the Human TBX20 Gene, a New Member of the T-Box Gene Family Closely Related to the Drosophila H15 Gene. Genomics, 2000, 67, 317-332.	2.9	44

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91	Developmental Expression Profile of the Optic Atrophy Gene Product: OPA1 Is Not Localized Exclusively in the Mammalian Retinal Ganglion Cell Layer. , 2004, 45, 1667.		44
92	Founder Effect, Seen in the British Population, of the 172 Peripherin/RDS Mutationâ€"and Further Refinement of Genetic Positioning of the Peripherin/RDS Gene. American Journal of Human Genetics, 1998, 62, 192-195.	6.2	42
93	Transcriptional regulation of PRPF31 gene expression by MSR1 repeat elements causes incomplete penetrance in retinitis pigmentosa. Scientific Reports, 2016, 6, 19450.	3.3	42
94	A Locus for Autosomal Recessive Congenital Microphthalmia Maps to Chromosome 14q32. American Journal of Human Genetics, 1998, 62, 1113-1116.	6.2	41
95	Evidence for Keratoconus Susceptibility Locus on Chromosome 14. JAMA Ophthalmology, 2010, 128, 1191.	2.4	41
96	Cross species analysis of Prominin reveals a conserved cellular role in invertebrate and vertebrate photoreceptor cells. Developmental Biology, 2012, 371, 312-320.	2.0	41
97	Copy-Number Variations inEYS:A Significant Event in the Appearance of arRP. , 2011, 52, 5625.		40
98	Autosomal Dominant Retinitis Pigmentosa with Intrafamilial Variability and Incomplete Penetrance in Two Families Carrying Mutations inPRPF8. , 2011, 52, 9304.		38
99	The Roles ofPAX6andSOX2in Myopia: Lessons from the 1958 British Birth Cohort. , 2007, 48, 4421.		37
100	Hypoxia Increases the Yield of Photoreceptors Differentiating from Mouse Embryonic Stem Cells and Improves the Modeling of Retinogenesis In Vitro. Stem Cells, 2013, 31, 966-978.	3.2	36
101	Genotype and Phenotype Studies in Autosomal Dominant Retinitis Pigmentosa (adRP) of the French Canadian Founder Population. , 2015, 56, 8297.		36
102	EYS Is a Protein Associated with the Ciliary Axoneme in Rods and Cones. PLoS ONE, 2016, 11, e0166397.	2.5	36
103	Identification of novel mutations in the carbohydrate sulfotransferase gene (CHST6) causing macular corneal dystrophy. Investigative Ophthalmology and Visual Science, 2002, 43, 377-82.	3.3	36
104	Connexin 50 mutation in a family with congenital "zonular nuclear" pulverulent cataract of Pakistani origin. Human Genetics, 1999, 105, 168-170.	3.8	33
105	RP1 and autosomal dominant rod-cone dystrophy: Novel mutations, a review of published variants, and genotype-phenotype correlation. Human Mutation, 2012, 33, 73-80.	2.5	33
106	Refinement of the Locus for Autosomal Recessive Retinitis Pigmentosa (RP25) Linked to Chromosome 6q in a Family of Pakistani Origin. American Journal of Human Genetics, 1999, 65, 571-574.	6.2	32
107	NovelGUCA1AMutations Suggesting Possible Mechanisms of Pathogenesis in Cone, Cone-Rod, and Macular Dystrophy Patients. BioMed Research International, 2013, 2013, 1-15.	1.9	32
108	Demonstration of a founder effect and fine mapping of dominant optic atrophy locus on 3q28-qter by linkage disequilibrium method. Human Genetics, 1998, 102, 79-86.	3.8	31

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109	Prognosis for splicing factor PRPF8 retinitis pigmentosa, novel mutations and correlation between human and yeast phenotypes. Human Mutation, 2010, 31, E1361-E1376.	2.5	31
110	Novel C2orf71 mutations account for â^¼1% of cases in a large French arRP cohort. Human Mutation, 2011, 32, E2091-103.	2.5	29
111	The familial dementia gene revisited: a missense mutation revealed by whole-exome sequencing identifies ITM2B as a candidate gene underlying a novel autosomal dominant retinal dystrophy in a large family. Human Molecular Genetics, 2014, 23, 491-501.	2.9	29
112	Novel CHST6 nonsense and missense mutations responsible for macular corneal dystrophy. American Journal of Ophthalmology, 2005, 139, 192-193.	3.3	28
113	Derivation of Cerebellar Neurons from Human Pluripotent Stem Cells. Current Protocols in Stem Cell Biology, 2012, 20, Unit 1H.5.	3.0	28
114	A map of human microRNA variation uncovers unexpectedly high levels of variability. Genome Medicine, 2012, 4, 62.	8.2	28
115	DominantPRPF31Mutations Are Hypostatic to a RecessiveCNOT3Polymorphism in Retinitis Pigmentosa: A Novel Phenomenon of â€œLinkedTrans-Acting Epistasisâ€• Annals of Human Genetics, 2014, 78, 62-71.	0.8	28
116	ATR localizes to the photoreceptor connecting cilium and deficiency leads to severe photoreceptor degeneration in mice. Human Molecular Genetics, 2013, 22, 1507-1515.	2.9	27
117	Brief Report: Astrogliosis Promotes Functional Recovery of Completely Transected Spinal Cord Following Transplantation of hESC-Derived Oligodendrocyte and Motoneuron Progenitors. Stem Cells, 2014, 32, 594-599.	3.2	26
118	Disease mechanism for retinitis pigmentosa (RP11) caused by missense mutations in the splicing factor gene PRPF31. Molecular Vision, 2008, 14, 683-90.	1.1	26
119	A Clinical and Molecular Genetic Study of Egyptian and Saudi Arabian Patients With Primary Congenital Glaucoma (PCG). Journal of Glaucoma, 2007, 16, 104-111.	1.6	24
120	High Prevalence of Posterior Polymorphous Corneal Dystrophy in the Czech Republic; Linkage Disequilibrium Mapping and Dating an Ancestral Mutation. PLoS ONE, 2012, 7, e45495.	2.5	24
121	Absence of p53 delays apoptotic photoreceptor cell death in the rds mouse. Current Eye Research, 1998, 17, 917-923.	1.5	23
122	Novel frameshift mutations in theRP2 gene and polymorphic variants. Human Mutation, 2000, 15, 580-580.	2.5	22
123	A 112 kb Deletion in Chromosome 19q13.42 Leads to Retinitis Pigmentosa. , 2011, 52, 6597.		22
124	Non-coding RNAs in pluripotency and neural differentiation of human pluripotent stem cells. Frontiers in Genetics, 2014, 5, 132.	2.3	22
125	Segregation of a PRKCG Mutation in Two RP11 Families. American Journal of Human Genetics, 1998, 62, 1248-1252.	6.2	21
126	Phenotype of Retinitis Pigmentosa Associated With the Ser50Thr Mutation in the NRL Gene. JAMA Ophthalmology, 2003, 121, 793.	2.4	21

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127	Further Insights Into GPR179: Expression, Localization, and Associated Pathogenic Mechanisms Leading to Complete Congenital Stationary Night Blindness. , 2013, 54, 8041.		20
128	Molecular genetics of retinitis pigmentosa in two Romani (Gypsy) families. Molecular Vision, 2006, 12, 909-14.	1.1	20
129	NRL S50T mutation and the importance of â€˜founder effectsâ€™ in inherited retinal dystrophies. European Journal of Human Genetics, 2000, 8, 783-787.	2.8	18
130	A study of the nuclear trafficking of the splicing factor protein PRPF31 linked to autosomal dominant retinitis pigmentosa (ADRP). Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2006, 1762, 304-311.	3.8	18
131	Span poly-L-arginine nanoparticles are efficient non-viral vectors for PRPF31 gene delivery: An approach of gene therapy to treat retinitis pigmentosa. Nanomedicine: Nanotechnology, Biology, and Medicine, 2016, 12, 2251-2260.	3.3	18
132	A YAC Contig Spanning the Dominant Retinitis Pigmentosa Locus (RP9) on Chromosome 7p. Genomics, 1995, 28, 383-388.	2.9	17
133	Rasagiline delays retinal degeneration in a mouse model of retinitis pigmentosa via modulation of Bax/Bclâ€2 expression. CNS Neuroscience and Therapeutics, 2018, 24, 448-455.	3.9	17
134	A Clinical and Molecular Genetic Study of Autosomal-Dominant Stromal Corneal Dystrophy in British Population. Ophthalmic Research, 2005, 37, 310-317.	1.9	16
135	A missense mutation inASRGL1is involved in causing autosomal recessive retinal degeneration. Human Molecular Genetics, 2016, 25, ddw113.	2.9	16
136	A novel 1-bp deletion in PITX3 causing congenital posterior polar cataract. Molecular Vision, 2011, 17, 1249-53.	1.1	16
137	Autosomal dominant Best disease with an unusual electrooculographic light rise and risk of angle-closure glaucoma: a clinical and molecular genetic study. Molecular Vision, 2011, 17, 2272-82.	1.1	16
138	Expression of PRPF31 and TFPT: regulation in health and retinal disease. Human Molecular Genetics, 2012, 21, 4126-4137.	2.9	15
139	Genetic association and stress mediated down-regulation in trabecular meshwork implicates MPP7 as a novel candidate gene in primary open angle glaucoma. BMC Medical Genomics, 2016, 9, 15.	1.5	15
140	Subretinal Transplant of Induced Pluripotent Stem Cell-Derived Retinal Pigment Epithelium on Nanostructured Fibrin-Agarose. Tissue Engineering - Part A, 2019, 25, 799-808.	3.1	15
141	Disease-Causing Mutations in BEST1 Gene Are Associated with Altered Sorting of Bestrophin-1 Protein. International Journal of Molecular Sciences, 2013, 14, 15121-15140.	4.1	14
142	A new locus (RP31) for autosomal dominant retinitis pigmentosa maps to chromosome 9p. Human Genetics, 2005, 118, 501-503.	3.8	13
143	Novel mutations of theRPGR gene in RP3 families. Human Mutation, 2000, 15, 386-386.	2.5	12
144	Cloning and characterization of WDR17, a novel WD repeat-containing gene on chromosome 4q34. Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 2002, 1579, 18-25.	2.4	12

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145	Mutations in SPATA13/ASEF2 cause primary angle closure glaucoma. PLoS Genetics, 2020, 16, e1008721.	3.5	12
146	Exclusion of Four Candidate Genes, KHDRBS2, PTP4A1, KIAA1411 and OGFRL1, as Causative of Autosomal Recessive Retinitis Pigmentosa. Ophthalmic Research, 2006, 38, 19-23.	1.9	10
147	Phenotype Associated with the H626P Mutation and Other Changes in the <i>TGFBI</i> Gene in Czech Families. Ophthalmic Research, 2008, 40, 105-108.	1.9	10
148	Molecular genetic study of Egyptian patients with macular corneal dystrophy. British Journal of Ophthalmology, 2010, 94, 250-255.	3.9	10
149	TOPORS, a Dual E3 Ubiquitin and Sumo1 Ligase, Interacts with 26 S Protease Regulatory Subunit 4, Encoded by the PSMC1 Gene. PLoS ONE, 2016, 11, e0148678.	2.5	10
150	Effects of Ca2+ ions on bestrophin-1 surface films. Colloids and Surfaces B: Biointerfaces, 2017, 149, 226-232.	5.0	10
151	The Resveratrol Prodrug JC19 Delays Retinal Degeneration in rd10 Mice. Advances in Experimental Medicine and Biology, 2019, 1185, 457-462.	1.6	10
152	Sequence variation within theRPGR gene: Evidence for a founder complex allele. Human Mutation, 2000, 16, 273-274.	2.5	8
153	Purification, characterisation and intracellular localisation of aryl hydrocarbon interacting protein-like 1 (AIPL1) and effects of mutations associated with inherited retinal dystrophies. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2004, 1690, 141-149.	3.8	8
154	Molecular Genetic Analysis of Two Functional Candidate Genes in the Autosomal Recessive Retinitis Pigmentosa, RP25, Locus. Current Eye Research, 2005, 30, 1081-1087.	1.5	8
155	Mutations inTOPORS: A Rare Cause of Autosomal Dominant Retinitis Pigmentosa in Continental Europe?. Ophthalmic Genetics, 2009, 30, 96-98.	1.2	8
156	Characterization of the human diacylglycerol kinase Îµ gene and its assessment as a candidate for inherited retinitis pigmentosa. Gene, 1999, 239, 185-192.	2.2	7
157	Concise Review: Stem Cells for the Treatment of Cerebellar-Related Disorders. Stem Cells, 2011, 29, 564-569.	3.2	7
158	First insights into the expression of VAX2 in humans and its localization in the adult primate retina. Experimental Eye Research, 2016, 148, 24-29.	2.6	7
159	<scp>WDR34</scp>, a candidate gene for nonâ€syndromic rodâ€cone dystrophy. Clinical Genetics, 2021, 99, 298-302.	2.0	7
160	Localization of the aquaporin 1 (AQP1) gene within a YAC contig containing the polymorphic markers D7S632 and D7S526. Genomics, 1995, 25, 599-600.	2.9	6
161	Gene of the month:PRPF31. Journal of Clinical Pathology, 2017, 70, 729-732.	2.0	6
162	Dominant optic atrophy: exclusion and fine genetic mapping of the candidate gene, HRY. Mammalian Genome, 1998, 9, 784-787.	2.2	5

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163	Dominant Cone and Cone-Rod Dystrophies: Functional Analysis of Mutations in RetGC1 and GCAP1. Novartis Foundation Symposium, 2008, 255, 37-50.	1.1	5
164	Regional Assignment of 30 Expressed Sequence Tags on Human Chromosome 7 Using a Somatic Cell Hybrid Panel. Genomics, 1995, 30, 112-114.	2.9	4
165	A novel locus for autosomal dominant cone-rod dystrophy maps to chromosome 10q. European Journal of Human Genetics, 2013, 21, 338-342.	2.8	4
166	Common Polymorphisms in theSERPINI2Gene Are Associated with Refractive Error in the 1958 British Birth Cohort. , 2012, 53, 440.		3
167	Mutations in a protein target of the Pim-1 kinase associated with the RP9 form of autosomal dominant retinitis pigmentosa. , 0, .		2
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