Paul A Sieving

List of Publications by Year
in descending order

157 papers	9,626 citations	50276 46 h-index	48315 88 g-index
157 all docs	157 docs citations	157 times ranked	6827 citing authors

#	Article	IF	Citations
1	Nrl is required for rod photoreceptor development. Nature Genetics, 2001, 29, 447-452.	21.4	795
2	Retinopathy induced in mice by targeted disruption of the rhodopsin gene. Nature Genetics, 1997, 15, 216-219.	21.4	552
3	Ciliary neurotrophic factor (CNTF) for human retinal degeneration: Phase I trial of CNTF delivered by encapsulated cell intraocular implants. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 3896-3901.	7.1	548
4	Pushâ€“pull model of the primate photopic electroretinogram: A role for hyperpolarizing neurons in shaping the b-wave. Visual Neuroscience, 1994, 11, 519-532.	1.0	354
5	Treatment with isotretinoin inhibits lipofuscin accumulation in a mouse model of recessive Stargardt's macular degeneration. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 4742-4747.	7.1	271
6	PCDH15 is expressed in the neurosensory epithelium of the eye and ear and mutant alleles are responsible for both USH1F and DFNB23. Human Molecular Genetics, 2003, 12, 3215-3223.	2.9	255
7	Mutations in the Cone-Rod Homeobox Gene Are Associated with the Cone-Rod Dystrophy Photoreceptor Degeneration. Neuron, 1997, 19, 1329-1336.	8.1	250
8	A Comprehensive Mutation Analysis of RP2 and RPGR in a North American Cohort of Families with X-Linked Retinitis Pigmentosa. American Journal of Human Genetics, 2002, 70, 1545-1554.	6.2	224
9	CNGB3 mutations account for 50% of all cases with autosomal recessive achromatopsia. European Journal of Human Genetics, 2005, 13, 302-308.	2.8	216
10	Biology and therapy of inherited retinal degenerative disease: insights from mouse models. DMM Disease Models and Mechanisms, 2015, 8, 109-129.	2.4	207
11	RS-1Gene Delivery to an AdultRs1hKnockout Mouse Model Restores ERG b-Wave with Reversal of the Electronegative Waveform of X-Linked Retinoschisis. , 2004, 45, 3279.		181
12	Retinal AAV8-RS1 Gene Therapy for X-Linked Retinoschisis: Initial Findings from a Phase I/IIa Trial by Intravitreal Delivery. Molecular Therapy, 2018, 26, 2282-2294.	8.2	173
13	Mutations in the gene encoding lecithin retinol acyltransferase are associated with early-onset severe retinal dystrophy. Nature Genetics, 2001, 28, 123-124.	21.4	167
14	Age-Related Macular Degeneration: A High-Resolution Genome Scan for Susceptibility Loci in a Population Enriched for Late-Stage Disease. American Journal of Human Genetics, 2004, 74, 482-494.	6.2	157
15	X-Linked Recessive Atrophic Macular Degeneration from RPGR Mutation. Genomics, 2002, 80, 166-171.	2.9	124
16	Late-Onset Macular Degeneration and Long Anterior Lens Zonules Result from aCTRP5Gene Mutation. , 2005, 46, 3363.		119
17	Retinoschisin Gene Therapy and Natural History in theRs1h-KO Mouse: Long-term Rescue from Retinal Degeneration. , 2007, 48, 3837.		109
18	A Splice-Site Mutation in a Retina-Specific Exon of BBS8 Causes Nonsyndromic Retinitis Pigmentosa. American Journal of Human Genetics, 2010, 86, 805-812.	6.2	109

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19	Severe retinitis pigmentosa mapped to 4p15 and associated with a novel mutation in the PROM1 gene. Human Genetics, 2007, 122, 293-299.	3.8	97
20	Retinal degeneration associated with RDH12 mutations results from decreased 11- cis retinal synthesis due to disruption of the visual cycle. Human Molecular Genetics, 2005, 14, 3865-3875.	2.9	94
21	Spectrum of Mutations in the RPGR Gene That Are Identified in 20% of Families with X-Linked Retinitis Pigmentosa. American Journal of Human Genetics, 1997, 61, 1287-1292.	6.2	93
22	A Retinal Neuronal Developmental Wave of Retinoschisin Expression Begins in Ganglion Cells during Layer Formation. , 2004, 45, 3302.		93
23	CNGB3Achromatopsia with Progressive Loss of Residual Cone Function and Impaired Rod-Mediated Function. , 2007, 48, 3864.		90
24	Bestrophin Gene Mutations in Patients with Best Vitelliform Macular Dystrophy. Genomics, 1999, 58, 98-101.	2.9	89
25	The scotopic threshold response of the cat erg is suppressed selectively by GABA and glycine. Vision Research, 1991, 31, 1-15.	1.4	86
26	Characterization of mouse orthologue of ELOVL4: genomic organization and spatial and temporal expression. Genomics, 2004, 83, 626-635.	2.9	86
27	RanBP2 Modulates Cox11 and Hexokinase I Activities and Haploinsufficiency of RanBP2 Causes Deficits in Glucose Metabolism. PLoS Genetics, 2006, 2, e177.	3.5	86
28	CONGENITAL X-LINKED RETINOSCHISIS CLASSIFICATION SYSTEM. Retina, 2006, 26, S61-S64.	1.7	84
29	Synaptic pathology and therapeutic repair in adult retinoschisis mouse by AAV-RS1 transfer. Journal of Clinical Investigation, 2015, 125, 2891-2903.	8.2	84
30	The electroretinogram of the rhodopsin knockout mouse. Visual Neuroscience, 1999, 16, 391-398.	1.0	83
31	Regulation of Rod Phototransduction Machinery by Ciliary Neurotrophic Factor. Journal of Neuroscience, 2006, 26, 13523-13530.	3.6	83
32	Juvenile X-linked retinoschisis from XLRS1 Arg213Trp mutation with preservation of the electroretinogram scotopic b-wave. American Journal of Ophthalmology, 1999, 128, 179-184.	3.3	81
33	Synaptic Pathology in Retinoschisis Knockout (Rs1^{â^'/y}) Mouse Retina and Modification by rAAV-Rs1Gene Delivery. , 2008, 49, 3677.		81
34	Encapsulated Cell-Based Intraocular Delivery of Ciliary Neurotrophic Factor in Normal Rabbit: Dose-Dependent Effects on ERG and Retinal Histology. , 2004, 45, 2420.		80
35	MERTK Arginine-844-Cysteine in a Patient with Severe Rod-Cone Dystrophy: Loss of Mutant Protein Function in Transfected Cells. Investigative Ophthalmology and Visual Science, 2004, 45, 1456-1463.	3.3	79
36	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.	6.2	78

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37	Constitutive â€œLightâ€•Adaptation in Rods from G90D Rhodopsin: A Mechanism for Human Congenital Nightblindness without Rod Cell Loss. Journal of Neuroscience, 2001, 21, 5449-5460.	3.6	77
38	Analysis of photoreceptor function and inner retinal activity in juvenile X-linked retinoschisis. Vision Research, 2001, 41, 3931-3942.	1.4	71
39	A Mutation in SLC24A1 Implicated in Autosomal-Recessive Congenital Stationary Night Blindness. American Journal of Human Genetics, 2010, 87, 523-531.	6.2	67
40	A long-term efficacy study of gene replacement therapy for RPGR-associated retinal degeneration. Human Molecular Genetics, 2015, 24, 3956-3970.	2.9	63
41	Analysis of the RPGRGene in 11 Pedigrees with the Retinitis Pigmentosa Type 3 Genotype: Paucity of Mutations in the Coding Region but Splice Defects in Two Families. American Journal of Human Genetics, 1997, 61, 571-580.	6.2	61
42	Remapping of the RP15 Locus for X-Linked Cone-Rod Degeneration to Xp11.4-p21.1, and Identification of a De Novo Insertion in the RPGR Exon ORF15. American Journal of Human Genetics, 2000, 67, 1000-1003.	6.2	61
43	Primate Retinal Signaling Pathways: Suppressing on-Pathway Activity in Monkey With Glutamate Analogues Mimics Human CSNB1-NYX Genetic Night Blindness. Journal of Neurophysiology, 2005, 93, 481-492.	1.8	60
44	Preclinical Dose-Escalation Study of Intravitreal AAV-RS1 Gene Therapy in a Mouse Model of X-linked Retinoschisis: Dose-Dependent Expression and Improved Retinal Structure and Function. Human Gene Therapy, 2016, 27, 376-389.	2.7	60
45	Photopic ERG Negative Response from Amacrine Cell Signaling in RCS Rat Retinal Degeneration. , 2008, 49, 442.		59
46	Elovl45-bpâ€“Deletion Knock-in Mice Develop Progressive Photoreceptor Degeneration. , 2006, 47, 4558.		58
47	Constitutive Excitation by Gly90Asp Rhodopsin Rescues Rods from Degeneration Caused by Elevated Production of cGMP in the Dark. Journal of Neuroscience, 2007, 27, 8805-8815.	3.6	58
48	Assessment of foveal cone photoreceptors in Stargardt's macular dystrophy using a small dot detection task. Vision Research, 1993, 33, 1509-1524.	1.4	57
49	Loss of Circadian Photoentrainment and Abnormal Retinal Electrophysiology inMath5Mutant Mice. , 2005, 46, 2540.		56
50	Sequencing Arrays for Screening Multiple Genes Associated with Early-Onset Human Retinal Degenerations on a High-Throughput Platform. , 2005, 46, 3355.		54
51	Depleting Rac1 in mouse rod photoreceptors protects them from photo-oxidative stress without affecting their structure or function. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 9397-9402.	7.1	54
52	X-Linked Retinoschisis:RS1Mutation Severity and Age Affect the ERG Phenotype in a Cohort of 68 Affected Male Subjects. , 2011, 52, 9250.		54
53	Evaluation of the ELOVL4 gene in patients with age-related macular degeneration. Ophthalmic Genetics, 2001, 22, 233-239.	1.2	53
54	Atrophic macular degeneration mutations in ELOVL4 result in the intracellular misrouting of the protein. Genomics, 2004, 83, 615-625.	2.9	53

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55	Exome Analysis Identified a Novel Mutation in the RBP4 Gene in a Consanguineous Pedigree with Retinal Dystrophy and Developmental Abnormalities. PLoS ONE, 2012, 7, e50205.	2.5	53
56	Retinoschisin Is a Peripheral Membrane Protein with Affinity for Anionic Phospholipids and Affected by Divalent Cations. , 2007, 48, 991.		52
57	Electroretinographic Findings in Selected Pedigrees With Choroideremia. American Journal of Ophthalmology, 1986, 101, 361-367.	3.3	51
58	Long anterior zonules and pigment dispersion. American Journal of Ophthalmology, 2003, 136, 1176-1178.	3.3	50
59	Autosomal Recessive Retinitis Pigmentosa Is Associated with Mutations inRP1in Three Consanguineous Pakistani Families. , 2005, 46, 2264.		50
60	Paired octamer rings of retinoschisin suggest a junctional model for cellâ€“cell adhesion in the retina. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 5287-5292.	7.1	49
61	Neuroprotection for glaucoma: Requirements for clinical translation. Experimental Eye Research, 2017, 157, 34-37.	2.6	48
62	Quantitative Relationship of the Scotopic and Photopic ERG to Photoreceptor Cell Loss in Light Damaged Rats. Experimental Eye Research, 2000, 70, 693-705.	2.6	46
63	Haploinsufficiency Is Not the Key Mechanism of Pathogenesis in a HeterozygousElovl4Knockout Mouse Model of STGD3 Disease. , 2006, 47, 3603.		45
64	A Mutation in ZNF513, a Putative Regulator of Photoreceptor Development, Causes Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2010, 87, 400-409.	6.2	44
65	Molecular Mechanisms Leading to Null-Protein Product from Retinoschisin (RS1) Signal-Sequence Mutants in X-Linked Retinoschisis (XLRS) Disease. Human Mutation, 2010, 31, 1251-1260.	2.5	44
66	Post-photoreceptoral activity dominates primate photopic 32-Hz ERG for sine-, square-, and pulsed stimuli. Investigative Ophthalmology and Visual Science, 2002, 43, 2500-7.	3.3	44
67	Evidence for two sites of adaptation affecting the dark-adapted ERG of cats and primates. Vision Research, 1995, 35, 435-442.	1.4	43
68	Stargardt-like macular dystrophy protein ELOVL4 exerts a dominant negative effect by recruiting wild-type protein into aggresomes. Molecular Vision, 2005, 11, 665-76.	1.1	41
69	Novel mutations in XLRS1 causing retinoschisis, including first evidence of putative leader sequence change. Human Mutation, 1999, 14, 423-427.	2.5	40
70	Night Blindness and the Mechanism of Constitutive Signaling of Mutant G90D Rhodopsin. Journal of Neuroscience, 2008, 28, 11662-11672.	3.6	40
71	Ames Waltzer Deaf Mice Have Reduced Electroretinogram Amplitudes and Complex Alternative Splicing ofPcdh15Transcripts. , 2006, 47, 3074.		38
72	Contributions to the electroretinogram of currents originating in proximal retina. Visual Neuroscience, 1988, 1, 307-315.	1.0	37

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73	Long-term 12 year follow-up of X-linked congenital retinoschisis. Ophthalmic Genetics, 2010, 31, 114-125.	1.2	37
74	Retinoschisin expression and localization in rodent and human pineal and consequences of mouse RS1 gene knockout. Molecular Vision, 2006, 12, 1108-16.	1.1	37
75	Chapter 6 Negative components of the electroretinogram from proximal retina and photoreceptor. Progress in Retinal and Eye Research, 1991, 10, 121-160.	0.8	35
76	Mutations in RLBP1 associated with fundus albipunctatus in consanguineous Pakistani families. British Journal of Ophthalmology, 2011, 95, 1019-1024.	3.9	35
77	Limits to growth: why neuroscience needs large-scale science. Nature Neuroscience, 2004, 7, 426-427.	14.8	34
78	Homozygosity Mapping and Genetic Analysis of Autosomal Recessive Retinal Dystrophies in 144 Consanguineous Pakistani Families. , 2017, 58, 2218.		34
79	Spectrum of color gene deletions and phenotype in patients with blue cone monochromacy. Human Genetics, 2000, 107, 75-82.	3.8	33
80	Molecular modeling indicates distinct classes of missense variants with mild and severe XLRS phenotypes. Human Molecular Genetics, 2013, 22, 4756-4767.	2.9	32
81	Retinal Structure and Gene Therapy Outcome in Retinoschisin-Deficient Mice Assessed by Spectral-Domain Optical Coherence Tomography. , 2016, 57, OCT277.		32
82	Comparative structural and functional analysis of photoreceptor neurons of Rho-/- mice reveal increased survival on C57BL/6J in comparison to 129Sv genetic background. Visual Neuroscience, 2001, 18, 437-443.	1.0	31
83	Establishing the involvement of the novel geneAGBL5in retinitis pigmentosa by whole genome sequencing. Physiological Genomics, 2016, 48, 922-927.	2.3	29
84	Ocular and systemic safety of a recombinant AAV8 vector for X-linked retinoschisis gene therapy: GLP studies in rabbits and Rs1-KO mice. Molecular Therapy - Methods and Clinical Development, 2016, 3, 16011.	4.1	29
85	X-Linked Retinoschisis. Ophthalmology, 2022, 129, 191-202.	5.2	29
86	Human bZIP Transcription Factor GeneNRL:Structure, Genomic Sequence, and Fine Linkage Mapping at 14q11.2 and Negative Mutation Analysis in Patients with Retinal Degeneration. Genomics, 1997, 45, 395-401.	2.9	28
87	Optic nerve dysplasia and renal insufficiency in a family with a novel PAX2 mutation, Arg115X: further ophthalmologic delineation of the renal-coloboma syndrome. Ophthalmic Genetics, 2003, 24, 191-202.	1.2	28
88	Severe autosomal recessive retinitis pigmentosa maps to chromosome 1p13.3â€“p21.2 between D1S2896 and D1S457 but outside ABCA4. Human Genetics, 2005, 118, 356-365.	3.8	28
89	Probing potassium channel function in vivo by intracellular delivery of antibodies in a rat model of retinal neurodegeneration. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 12710-12715.	7.1	28
90	Nonsense mutation in MERTK causes autosomal recessive retinitis pigmentosa in a consanguineous Pakistani family. British Journal of Ophthalmology, 2010, 94, 1094-1099.	3.9	28

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91	Preclinical Safety Evaluation of a Recombinant AAV8 Vector for X-Linked Retinoschisis After Intravitreal Administration in Rabbits. Human Gene Therapy Clinical Development, 2014, 25, 202-211.	3.1	28
92	Host Immune Responses after Suprachoroidal Delivery of AAV8 in Nonhuman Primate Eyes. Human Gene Therapy, 2021, 32, 682-693.	2.7	27
93	Retinal cAMP levels during the progression of retinal degeneration in rhodopsin P23H and S334ter transgenic rats. Investigative Ophthalmology and Visual Science, 2002, 43, 1655-61.	3.3	26
94	Foveal Retinoschisis Associated With Senile Retinoschisis in a Woman. American Journal of Ophthalmology, 1988, 106, 107-109.	3.3	25
95	Mutation in the intracellular chloride channel CLCC1 associated with autosomal recessive retinitis pigmentosa. PLoS Genetics, 2018, 14, e1007504.	3.5	25
96	Accessory heterozygous mutations in cone photoreceptor CNGA3 exacerbate CNG channelâ€“associated retinopathy. Journal of Clinical Investigation, 2018, 128, 5663-5675.	8.2	25
97	Analysis of Anatomic and Functional Measures in X-Linked Retinoschisis. , 2018, 59, 2841.		24
98	Mutations in the gene encoding the alpha-subunit of rod phosphodiesterase in consanguineous Pakistani families. Molecular Vision, 2006, 12, 1283-91.	1.1	24
99	Clinical diagnoses that overlap with choroideremia. Canadian Journal of Ophthalmology, 2003, 38, 364-372.	0.7	23
100	A variant form of Oguchi disease mapped to 13q34 associated with partial deletion of GRK1 gene. Molecular Vision, 2005, 11, 977-85.	1.1	23
101	Identification and Characterization of the Human Homologue (RAI2) of a Mouse Retinoic Acid-Induced Gene in Xp22. Genomics, 1999, 55, 275-283.	2.9	21
102	Genetic Rescue of X-Linked Retinoschisis Mouse (Rs1^{â^'/y}) Retina Induces Quiescence of the Retinal Microglial Inflammatory State Following AAV8-RS1 Gene Transfer and Identifies Gene Networks Underlying Retinal Recovery. Human Gene Therapy, 2021, 32, 667-681.	2.7	21
103	Preventing blindness in retinal disease: ciliary neurotrophic factor intraocular implants. Canadian Journal of Ophthalmology, 2007, 42, 399-402.	0.7	21
104	Neuroethics for the National Institutes of Health BRAIN Initiative. Journal of Neuroscience, 2018, 38, 10583-10585.	3.6	20
105	Mutations in the small nuclear riboprotein 200 kDa gene (SNRNP200) cause 1.6% of autosomal dominant retinitis pigmentosa. Molecular Vision, 2013, 19, 2407-17.	1.1	20
106	Genetic Ophthalmology and the Era of Clinical Care. JAMA - Journal of the American Medical Association, 2007, 297, 733.	7.4	19
107	Retinoschisin (RS1) Interacts with Negatively Charged Lipid Bilayers in the Presence of Ca²⁺: An Atomic Force Microscopy Study. Biochemistry, 2010, 49, 7023-7032.	2.5	19
108	Mutations in the Î²-subunit of rod phosphodiesterase identified in consanguineous Pakistani families with autosomal recessive retinitis pigmentosa. Molecular Vision, 2011, 17, 1373-80.	1.1	19

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109	AMPK modulation ameliorates dominant disease phenotypes of CTRP5 variant in retinal degeneration. Communications Biology, 2021, 4, 1360.	4.4	19
110	Genomics in the Era of Molecular Ophthalmology. JAMA Ophthalmology, 2008, 126, 424.	2.4	18
111	Convergence of Human Genetics and Animal Studies: Gene Therapy for X-Linked Retinoschisis. Cold Spring Harbor Perspectives in Medicine, 2015, 5, a017368.	6.2	17
112	Cryo-EM of retinoschisin branched networks suggests an intercellular adhesive scaffold in the retina. Journal of Cell Biology, 2019, 218, 1027-1038.	5.2	17
113	Immune function in X-linked retinoschisis subjects in an AAV8-RS1 phase I/IIa gene therapy trial. Molecular Therapy, 2021, 29, 2030-2040.	8.2	17
114	Summation of Rod and S Cone Signals at Threshold in Human Observers. Vision Research, 1996, 36, 2681-2688.	1.4	16
115	Loss of Retinoschisin (RS1) Cell Surface Protein in Maturing Mouse Rod Photoreceptors Elevates the Luminance Threshold for Light-Driven Translocation of Transducin But Not Arrestin. Journal of Neuroscience, 2012, 32, 13010-13021.	3.6	15
116	Fenestrated Sheen Macular Dystrophy. American Journal of Ophthalmology, 1991, 112, 1-7.	3.3	14
117	Null Retinoschisin-Protein Expression from anRS1c354del1-ins18 Mutation Causing Progressive and Severe XLRS in a Cross-Sectional Family Study. , 2009, 50, 5375.		14
118	Increased phase lag of the fundamental harmonic component of the 30 Hz flicker ERG in Schubert-Bornschein complete type CSNB. Vision Research, 1997, 37, 2471-2475.	1.4	13
119	Deciphering the genetic architecture and ethnographic distribution of IRD in three ethnic populations by whole genome sequence analysis. PLoS Genetics, 2021, 17, e1009848.	3.5	13
120	A Novel Locus for Autosomal Recessive Retinitis Pigmentosa in a Consanguineous Pakistani Family Maps to Chromosome 2p. American Journal of Ophthalmology, 2010, 149, 861-866.	3.3	12
121	Of men and mice: Human X-linked retinoschisis and fidelity in mouse modeling. Progress in Retinal and Eye Research, 2022, 87, 100999.	15.5	12
122	Diagnostic Issues with Inherited Retinal and Macular Dystrophies. Seminars in Ophthalmology, 1995, 10, 279-294.	1.6	11
123	X-Linked Juvenile Retinoschisis: Localization between (DXS1195, DXS418) and AFM291wf5 on a Single YAC. Human Heredity, 1996, 46, 329-335.	0.8	11
124	Identification and characterization of two mature isoforms of retinoschisin in murine retina. Biochemical and Biophysical Research Communications, 2006, 349, 99-105.	2.1	11
125	IFT88 mutations identified in individuals with non-syndromic recessive retinal degeneration result in abnormal ciliogenesis. Human Genetics, 2018, 137, 447-458.	3.8	11
126	3-D retina organoids. Cell Medicine, 2018, 10, 215517901877375.	5.0	11

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127	Double homozygous waltzer and Ames waltzer mice provide no evidence of retinal degeneration. Molecular Vision, 2008, 14, 2227-36.	1.1	11
128	Leber's Congenital Amaurosis With Marbelized Fundus and Juvenile Nephronophthisis. American Journal of Ophthalmology, 1989, 107, 426-428.	3.3	10
129	Linkage Study of Best‘s Vitelliform Macular Dystrophy (VMD2) in a Large North American Family. Human Heredity, 1996, 46, 211-220.	0.8	10
130	Improved Ocular Tissue Models and Eye-On-A-Chip Technologies Will Facilitate Ophthalmic Drug Development. Journal of Ocular Pharmacology and Therapeutics, 2020, 36, 25-29.	1.4	10
131	Refined Genetic Mapping of Juvenile X-Linked Retinoschisis. Human Heredity, 1995, 45, 206-210.	0.8	9
132	Sorting out Co-occurrence of Rare Monogenic Retinopathies: Stargardt Disease Co-existing with Congenital Stationary Night Blindness. Ophthalmic Genetics, 2014, 35, 51-56.	1.2	8
133	Rs1hâ^'/y exon 3-del rat model of X-linked retinoschisis with early onset and rapid phenotype is rescued by RS1 supplementation. Gene Therapy, 2022, 29, 431-440.	4.5	8
134	Mutation screening of patients with Leber Congenital Amaurosis or the enhanced S-Cone Syndrome reveals a lack of sequence variations in the NRL gene. Molecular Vision, 2003, 9, 14-7.	1.1	8
135	Comprehensive variant spectrum of the CNGA3 gene in patients affected by achromatopsia. Human Mutation, 2022, 43, 832-858.	2.5	8
136	Spectrum of color gene deletions and phenotype in patients with blue cone monochromacy. Human Genetics, 2000, 107, 75-82.	3.8	7
137	Investigation of the effect of dietary docosahexaenoic acid (DHA) supplementation on macular function in subjects with autosomal recessive Stargardt macular dystrophy. Ophthalmic Genetics, 2018, 39, 477-486.	1.2	7
138	AAVrh-10 transduces outer retinal cells in rodents and rabbits following intravitreal administration. Gene Therapy, 2019, 26, 386-398.	4.5	7
139	Motivations and Decision Making Processes of Men With X-linked Retinoschisis Considering Participation in an Ocular Gene Therapy Trial. American Journal of Ophthalmology, 2019, 204, 90-96.	3.3	7
140	AIPL1 implicated in the pathogenesis of two cases of autosomal recessive retinal degeneration. Molecular Vision, 2014, 20, 1-14.	1.1	6
141	Novel mutations in RPE65 identified in consanguineous Pakistani families with retinal dystrophy. Molecular Vision, 2013, 19, 1554-64.	1.1	5
142	Evaluation of the ELOVL4 gene in patients with autosomal recessive retinitis pigmentosa and Leber congenital amaurosis. Molecular Vision, 2003, 9, 49-51.	1.1	5
143	MASSIVE ADVANCING NONEXUDATIVE TYPE 1 CHOROIDAL NEOVASCULARIZATION IN CTRP5 LATE-ONSET RETINAL DEGENERATION. Retina, 2021, 41, 2236-2245.	1.7	4
144	Characterization of novel RS1 exonic deletions in juvenile X-linked retinoschisis. Molecular Vision, 2013, 19, 2209-16.	1.1	4

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145	â€œThere Are Hills and Valleysâ€• Experiences of Parenting a Son With X-Linked Retinoschisis. American Journal of Ophthalmology, 2020, 212, 98-104.	3.3	3
146	The National Eye Institute: Translational Clinical Research Initiatives on Inherited and Orphan Retinal Diseases: Personal Observations. Retina, 2005, 25, S8-S9.	1.7	2
147	Eyeing a New Network. Science, 2007, 318, 1068-1068.	12.6	2
148	X-linked Retinoschisis and Gene Therapy. International Ophthalmology Clinics, 2021, 61, 173-184.	0.7	2
149	Predominant Founder Effect among Recurrent Pathogenic Variants for an X-Linked Disorder. Genes, 2022, 13, 675.	2.4	2
150	At the Frontier of Vision Research: The National Eye Institute Celebrates 40 Years. American Journal of Ophthalmology, 2010, 149, 179-181.	3.3	1
151	Hunting for the Adhesion Molecule, Retinoschisin, in Retina using CEMOVIS. Microscopy and Microanalysis, 2019, 25, 1308-1309.	0.4	1
152	The NIH Blueprint for Neuroscience Research Seeks Community Input on Future Neuroscience Investments. Journal of Neuroscience, 2019, 39, 774-775.	3.6	1
153	Cone and rod ERGs in degenerations of central retina. Survey of Ophthalmology, 1984, 29, 235-236.	4.0	0
154	Fostering Partnerships: The National Institutes of Health (NIH) Mission. Retina, 2005, 25, S84-S85.	1.7	0
155	Retinoschisin at 4 Ã... Resolution from cryo-EM: A Junctional Model of Back-to-Back Octamers for Adhesion in the Retina. Biophysical Journal, 2016, 110, 348a.	0.5	0
156	Galactose Induces Formation of Chains of the Retinal Adhesion Protein, Retinoschisin. Microscopy and Microanalysis, 2017, 23, 1112-1113.	0.4	0
157	Response to: "Rescuing the NIH before it is too late". Journal of Clinical Investigation, 2006, 116, 1462-1463.	8.2	0