

# Paul A Sieving

## List of Publications by Year in descending order

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157  
papers

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docs citations

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#	ARTICLE	IF	CITATIONS
1	Of men and mice: Human X-linked retinoschisis and fidelity in mouse modeling. <i>Progress in Retinal and Eye Research</i> , 2022, 87, 100999.	15.5	12
2	<i>Rs1h<sup>Δ</sup>/y</i> exon 3-del rat model of X-linked retinoschisis with early onset and rapid phenotype is rescued by RS1 supplementation. <i>Gene Therapy</i> , 2022, 29, 431-440.	4.5	8
3	X-Linked Retinoschisis. <i>Ophthalmology</i> , 2022, 129, 191-202.	5.2	29
4	Comprehensive variant spectrum of the <i>CNGA3</i> gene in patients affected by achromatopsia. <i>Human Mutation</i> , 2022, 43, 832-858.	2.5	8
5	Predominant Founder Effect among Recurrent Pathogenic Variants for an X-Linked Disorder. <i>Genes</i> , 2022, 13, 675.	2.4	2
6	Genetic Rescue of X-Linked Retinoschisis Mouse ( <i>Rs1<sup>Δ</sup>/y</i> ) Retina Induces Quiescence of the Retinal Microglial Inflammatory State Following AAV8- <i>RS1</i> Gene Transfer and Identifies Gene Networks Underlying Retinal Recovery. <i>Human Gene Therapy</i> , 2021, 32, 667-681.	2.7	21
7	X-linked Retinoschisis and Gene Therapy. <i>International Ophthalmology Clinics</i> , 2021, 61, 173-184.	0.7	2
8	MASSIVE ADVANCING NONEXUDATIVE TYPE 1 CHOROIDAL NEOVASCULARIZATION IN CTRP5 LATE-ONSET RETINAL DEGENERATION. <i>Retina</i> , 2021, 41, 2236-2245.	1.7	4
9	Immune function in X-linked retinoschisis subjects in an AAV8-RS1 phase I/IIa gene therapy trial. <i>Molecular Therapy</i> , 2021, 29, 2030-2040.	8.2	17
10	Host Immune Responses after Suprachoroidal Delivery of AAV8 in Nonhuman Primate Eyes. <i>Human Gene Therapy</i> , 2021, 32, 682-693.	2.7	27
11	Deciphering the genetic architecture and ethnographic distribution of IRD in three ethnic populations by whole genome sequence analysis. <i>PLoS Genetics</i> , 2021, 17, e1009848.	3.5	13
12	AMPK modulation ameliorates dominant disease phenotypes of CTRP5 variant in retinal degeneration. <i>Communications Biology</i> , 2021, 4, 1360.	4.4	19
13	Improved Ocular Tissue Models and Eye-On-A-Chip Technologies Will Facilitate Ophthalmic Drug Development. <i>Journal of Ocular Pharmacology and Therapeutics</i> , 2020, 36, 25-29.	1.4	10
14	“There Are Hills and Valleys”: Experiences of Parenting a Son With X-Linked Retinoschisis. <i>American Journal of Ophthalmology</i> , 2020, 212, 98-104.	3.3	3
15	AAVrh-10 transduces outer retinal cells in rodents and rabbits following intravitreal administration. <i>Gene Therapy</i> , 2019, 26, 386-398.	4.5	7
16	Hunting for the Adhesion Molecule, Retinoschisin, in Retina using CEMOVIS. <i>Microscopy and Microanalysis</i> , 2019, 25, 1308-1309.	0.4	1
17	The NIH Blueprint for Neuroscience Research Seeks Community Input on Future Neuroscience Investments. <i>Journal of Neuroscience</i> , 2019, 39, 774-775.	3.6	1
18	Motivations and Decision Making Processes of Men With X-linked Retinoschisis Considering Participation in an Ocular Gene Therapy Trial. <i>American Journal of Ophthalmology</i> , 2019, 204, 90-96.	3.3	7

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19	Cryo-EM of retinoschisin branched networks suggests an intercellular adhesive scaffold in the retina. <i>Journal of Cell Biology</i> , 2019, 218, 1027-1038.	5.2	17
20	Neuroethics for the National Institutes of Health BRAIN Initiative. <i>Journal of Neuroscience</i> , 2018, 38, 10583-10585.	3.6	20
21	Mutation in the intracellular chloride channel CLCC1 associated with autosomal recessive retinitis pigmentosa. <i>PLoS Genetics</i> , 2018, 14, e1007504.	3.5	25
22	Investigation of the effect of dietary docosahexaenoic acid (DHA) supplementation on macular function in subjects with autosomal recessive Stargardt macular dystrophy. <i>Ophthalmic Genetics</i> , 2018, 39, 477-486.	1.2	7
23	Retinal AAV8-RS1 Gene Therapy for X-Linked Retinoschisis: Initial Findings from a Phase I/IIa Trial by Intravitreal Delivery. <i>Molecular Therapy</i> , 2018, 26, 2282-2294.	8.2	173
24	IFT88 mutations identified in individuals with non-syndromic recessive retinal degeneration result in abnormal ciliogenesis. <i>Human Genetics</i> , 2018, 137, 447-458.	3.8	11
25	Analysis of Anatomic and Functional Measures in X-Linked Retinoschisis. , 2018, 59, 2841.		24
26	3-D retina organoids. <i>Cell Medicine</i> , 2018, 10, 215517901877375.	5.0	11
27	Accessory heterozygous mutations in cone photoreceptor CNGA3 exacerbate CNG channel-associated retinopathy. <i>Journal of Clinical Investigation</i> , 2018, 128, 5663-5675.	8.2	25
28	Neuroprotection for glaucoma: Requirements for clinical translation. <i>Experimental Eye Research</i> , 2017, 157, 34-37.	2.6	48
29	Galactose Induces Formation of Chains of the Retinal Adhesion Protein, Retinoschisin. <i>Microscopy and Microanalysis</i> , 2017, 23, 1112-1113.	0.4	0
30	Homozygosity Mapping and Genetic Analysis of Autosomal Recessive Retinal Dystrophies in 144 Consanguineous Pakistani Families. , 2017, 58, 2218.		34
31	Retinal Structure and Gene Therapy Outcome in Retinoschisin-Deficient Mice Assessed by Spectral-Domain Optical Coherence Tomography. , 2016, 57, OCT277.		32
32	Establishing the involvement of the novel gene <i>AGBL5</i> in retinitis pigmentosa by whole genome sequencing. <i>Physiological Genomics</i> , 2016, 48, 922-927.	2.3	29
33	Ocular and systemic safety of a recombinant AAV8 vector for X-linked retinoschisis gene therapy: GLP studies in rabbits and Rs1-KO mice. <i>Molecular Therapy - Methods and Clinical Development</i> , 2016, 3, 16011.	4.1	29
34	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. <i>Human Gene Therapy</i> , 2016, 27, 376-389.	2.7	60
35	Paired octamer rings of retinoschisin suggest a junctional model for cell-cell adhesion in the retina. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, 5287-5292.	7.1	49
36	Retinoschisin at 4 Å... Resolution from cryo-EM: A Junctional Model of Back-to-Back Octamers for Adhesion in the Retina. <i>Biophysical Journal</i> , 2016, 110, 348a.	0.5	0

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37	A long-term efficacy study of gene replacement therapy for RPGR-associated retinal degeneration. <i>Human Molecular Genetics</i> , 2015, 24, 3956-3970.	2.9	63
38	Biology and therapy of inherited retinal degenerative disease: insights from mouse models. <i>DMM Disease Models and Mechanisms</i> , 2015, 8, 109-129.	2.4	207
39	Convergence of Human Genetics and Animal Studies: Gene Therapy for X-Linked Retinoschisis. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2015, 5, a017368.	6.2	17
40	Synaptic pathology and therapeutic repair in adult retinoschisis mouse by AAV-RS1 transfer. <i>Journal of Clinical Investigation</i> , 2015, 125, 2891-2903.	8.2	84
41	Preclinical Safety Evaluation of a Recombinant AAV8 Vector for X-Linked Retinoschisis After Intravitreal Administration in Rabbits. <i>Human Gene Therapy Clinical Development</i> , 2014, 25, 202-211.	3.1	28
42	Sorting out Co-occurrence of Rare Monogenic Retinopathies: Stargardt Disease Co-existing with Congenital Stationary Night Blindness. <i>Ophthalmic Genetics</i> , 2014, 35, 51-56.	1.2	8
43	AIP1L1 implicated in the pathogenesis of two cases of autosomal recessive retinal degeneration. <i>Molecular Vision</i> , 2014, 20, 1-14.	1.1	6
44	Molecular modeling indicates distinct classes of missense variants with mild and severe XLRS phenotypes. <i>Human Molecular Genetics</i> , 2013, 22, 4756-4767.	2.9	32
45	Novel mutations in RPE65 identified in consanguineous Pakistani families with retinal dystrophy. <i>Molecular Vision</i> , 2013, 19, 1554-64.	1.1	5
46	Characterization of novel RS1 exonic deletions in juvenile X-linked retinoschisis. <i>Molecular Vision</i> , 2013, 19, 2209-16.	1.1	4
47	Mutations in the small nuclear riboprotein 200 kDa gene (SNRNP200) cause 1.6% of autosomal dominant retinitis pigmentosa. <i>Molecular Vision</i> , 2013, 19, 2407-17.	1.1	20
48	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. <i>Journal of Neuroscience</i> , 2012, 32, 13010-13021.	3.6	15
49	Exome Analysis Identified a Novel Mutation in the RBP4 Gene in a Consanguineous Pedigree with Retinal Dystrophy and Developmental Abnormalities. <i>PLoS ONE</i> , 2012, 7, e50205.	2.5	53
50	X-Linked Retinoschisis: <i>RS1</i> Mutation Severity and Age Affect the ERG Phenotype in a Cohort of 68 Affected Male Subjects. , 2011, 52, 9250.		54
51	Mutations in RLBPI associated with fundus albipunctatus in consanguineous Pakistani families. <i>British Journal of Ophthalmology</i> , 2011, 95, 1019-1024.	3.9	35
52	Mutations in the $\beta$ -subunit of rod phosphodiesterase identified in consanguineous Pakistani families with autosomal recessive retinitis pigmentosa. <i>Molecular Vision</i> , 2011, 17, 1373-80.	1.1	19
53	A Splice-Site Mutation in a Retina-Specific Exon of BBS8 Causes Nonsyndromic Retinitis Pigmentosa. <i>American Journal of Human Genetics</i> , 2010, 86, 805-812.	6.2	109
54	A Mutation in ZNF513, a Putative Regulator of Photoreceptor Development, Causes Autosomal-Recessive Retinitis Pigmentosa. <i>American Journal of Human Genetics</i> , 2010, 87, 400-409.	6.2	44

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55	A Mutation in SLC24A1 Implicated in Autosomal-Recessive Congenital Stationary Night Blindness. American Journal of Human Genetics, 2010, 87, 523-531.	6.2	67
56	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
57	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
58	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
59	Retinoschisin (RS1) Interacts with Negatively Charged Lipid Bilayers in the Presence of Ca <sup>2+</sup> : An Atomic Force Microscopy Study. Biochemistry, 2010, 49, 7023-7032.	2.5	19
60	At the Frontier of Vision Research: The National Eye Institute Celebrates 40 Years. American Journal of Ophthalmology, 2010, 149, 179-181.	3.3	1
61	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
62	Long-term 12 year follow-up of X-linked congenital retinoschisis. Ophthalmic Genetics, 2010, 31, 114-125.	1.2	37
63	Null Retinoschisin-Protein Expression from an <i>RS1</i> <sup>c354del1-ins18</sup> Mutation Causing Progressive and Severe XLRS in a Cross-Sectional Family Study. , 2009, 50, 5375.		14
64	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
65	Night Blindness and the Mechanism of Constitutive Signaling of Mutant G90D Rhodopsin. Journal of Neuroscience, 2008, 28, 11662-11672.	3.6	40
66	Synaptic Pathology in Retinoschisis Knockout ( <i>Rs1</i> <sup>Δ<sup>1</sup></sup> ) Mouse Retina and Modification by rAAV- <i>Rs1</i> Gene Delivery. , 2008, 49, 3677.		81
67	Photopic ERG Negative Response from Amacrine Cell Signaling in RCS Rat Retinal Degeneration. , 2008, 49, 442.		59
68	Genomics in the Era of Molecular Ophthalmology. JAMA Ophthalmology, 2008, 126, 424.	2.4	18
69	Double homozygous waltzer and Ames waltzer mice provide no evidence of retinal degeneration. Molecular Vision, 2008, 14, 2227-36.	1.1	11
70	Retinoschisin Gene Therapy and Natural History in the <i>Rs1h</i> -KO Mouse: Long-term Rescue from Retinal Degeneration. , 2007, 48, 3837.		109
71	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
72	Genetic Ophthalmology and the Era of Clinical Care. JAMA - Journal of the American Medical Association, 2007, 297, 733.	7.4	19

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73	<i>CNGB3</i> Achromatopsia with Progressive Loss of Residual Cone Function and Impaired Rod-Mediated Function. , 2007, 48, 3864.		90
74	Eyeing a New Network. <i>Science</i> , 2007, 318, 1068-1068.	12.6	2
75	Retinoschisin Is a Peripheral Membrane Protein with Affinity for Anionic Phospholipids and Affected by Divalent Cations. , 2007, 48, 991.		52
76	Severe retinitis pigmentosa mapped to 4p15 and associated with a novel mutation in the PROM1 gene. <i>Human Genetics</i> , 2007, 122, 293-299.	3.8	97
77	Preventing blindness in retinal disease: ciliary neurotrophic factor intraocular implants. <i>Canadian Journal of Ophthalmology</i> , 2007, 42, 399-402.	0.7	21
78	Identification and characterization of two mature isoforms of retinoschisin in murine retina. <i>Biochemical and Biophysical Research Communications</i> , 2006, 349, 99-105.	2.1	11
79	Haploinsufficiency Is Not the Key Mechanism of Pathogenesis in a Heterozygous <i>Elovl4</i> Knockout Mouse Model of STGD3 Disease. , 2006, 47, 3603.		45
80	Ames Waltzer Deaf Mice Have Reduced Electroretinogram Amplitudes and Complex Alternative Splicing of <i>Pcdh15</i> Transcripts. , 2006, 47, 3074.		38
81	CONGENITAL X-LINKED RETINOSCHISIS CLASSIFICATION SYSTEM. <i>Retina</i> , 2006, 26, S61-S64.	1.7	84
82	RanBP2 Modulates Cox11 and Hexokinase I Activities and Haploinsufficiency of RanBP2 Causes Deficits in Glucose Metabolism. <i>PLoS Genetics</i> , 2006, 2, e177.	3.5	86
83	Regulation of Rod Phototransduction Machinery by Ciliary Neurotrophic Factor. <i>Journal of Neuroscience</i> , 2006, 26, 13523-13530.	3.6	83
84	<i>Elovl4</i> Deletion Knock-in Mice Develop Progressive Photoreceptor Degeneration. , 2006, 47, 4558.		58
85	Ciliary neurotrophic factor (CNTF) for human retinal degeneration: Phase I trial of CNTF delivered by encapsulated cell intraocular implants. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2006, 103, 3896-3901.	7.1	548
86	Response to: "Rescuing the NIH before it is too late". <i>Journal of Clinical Investigation</i> , 2006, 116, 1462-1463.	8.2	0
87	Retinoschisin expression and localization in rodent and human pineal and consequences of mouse <i>RS1</i> gene knockout. <i>Molecular Vision</i> , 2006, 12, 1108-16.	1.1	37
88	Mutations in the gene encoding the alpha-subunit of rod phosphodiesterase in consanguineous Pakistani families. <i>Molecular Vision</i> , 2006, 12, 1283-91.	1.1	24
89	Fostering Partnerships: The National Institutes of Health (NIH) Mission. <i>Retina</i> , 2005, 25, S84-S85.	1.7	0
90	The National Eye Institute: Translational Clinical Research Initiatives on Inherited and Orphan Retinal Diseases: Personal Observations. <i>Retina</i> , 2005, 25, S8-S9.	1.7	2

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91	CNGB3 mutations account for 50% of all cases with autosomal recessive achromatopsia. European Journal of Human Genetics, 2005, 13, 302-308.	2.8	216
92	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
93	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
94	Sequencing Arrays for Screening Multiple Genes Associated with Early-Onset Human Retinal Degenerations on a High-Throughput Platform. , 2005, 46, 3355.		54
95	Autosomal Recessive Retinitis Pigmentosa Is Associated with Mutations inRP1in Three Consanguineous Pakistani Families. , 2005, 46, 2264.		50
96	Late-Onset Macular Degeneration and Long Anterior Lens Zonules Result from aCTRP5Gene Mutation. , 2005, 46, 3363.		119
97	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
98	Loss of Circadian Photoentrainment and Abnormal Retinal Electrophysiology inMath5Mutant Mice. , 2005, 46, 2540.		56
99	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
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	Encapsulated Cell-Based Intraocular Delivery of Ciliary Neurotrophic Factor in Normal Rabbit: Dose-Dependent Effects on ERG and Retinal Histology. , 2004, 45, 2420.		80
102	A Retinal Neuronal Developmental Wave of Retinoschisin Expression Begins in Ganglion Cells during Layer Formation. , 2004, 45, 3302.		93
103	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
104	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
105	Limits to growth: why neuroscience needs large-scale science. Nature Neuroscience, 2004, 7, 426-427.	14.8	34
106	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
107	Characterization of mouse orthologue of ELOVL4: genomic organization and spatial and temporal expression. Genomics, 2004, 83, 626-635.	2.9	86
108	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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109	Clinical diagnoses that overlap with choroideremia. Canadian Journal of Ophthalmology, 2003, 38, 364-372.	0.7	23
110	Long anterior zonules and pigment dispersion. American Journal of Ophthalmology, 2003, 136, 1176-1178.	3.3	50
111	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
112	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
113	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
114	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
115	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
116	X-Linked Recessive Atrophic Macular Degeneration from RPGR Mutation. Genomics, 2002, 80, 166-171.	2.9	124
117	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
118	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
119	Post-photoreceptor 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
120	Analysis of photoreceptor function and inner retinal activity in juvenile X-linked retinoschisis. Vision Research, 2001, 41, 3931-3942.	1.4	71
121	Evaluation of the ELOVL4 gene in patients with age-related macular degeneration. Ophthalmic Genetics, 2001, 22, 233-239.	1.2	53
122	Constitutive "Light" 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
123	Comparative structural and functional analysis of photoreceptor neurons of Rho <sup>-/-</sup> mice reveal increased survival on C57BL/6j in comparison to 129Sv genetic background. Visual Neuroscience, 2001, 18, 437-443.	1.0	31
124	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
125	Nrl is required for rod photoreceptor development. Nature Genetics, 2001, 29, 447-452.	21.4	795
126	Spectrum of color gene deletions and phenotype in patients with blue cone monochromacy. Human Genetics, 2000, 107, 75-82.	3.8	7



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127	Quantitative Relationship of the Scotopic and Photopic ERG to Photoreceptor Cell Loss in Light Damaged Rats. <i>Experimental Eye Research</i> , 2000, 70, 693-705.	2.6	46
128	Remapping of the RP15 Locus for X-Linked Cone-Rod Degeneration to Xp11.4-p21.1, and Identification of a De Novo Insertion in the RPGR Exon ORF15. <i>American Journal of Human Genetics</i> , 2000, 67, 1000-1003.	6.2	61
129	Spectrum of color gene deletions and phenotype in patients with blue cone monochromacy. <i>Human Genetics</i> , 2000, 107, 75-82.	3.8	33
130	Novel mutations in XLR51 causing retinoschisis, including first evidence of putative leader sequence change. <i>Human Mutation</i> , 1999, 14, 423-427.	2.5	40
131	Protein-Truncation Mutations in the RP2 Gene in a North American Cohort of Families with X-Linked Retinitis Pigmentosa. <i>American Journal of Human Genetics</i> , 1999, 64, 897-900.	6.2	78
132	Juvenile X-linked retinoschisis from XLR51 Arg213Trp mutation with preservation of the electroretinogram scotopic b-wave. <i>American Journal of Ophthalmology</i> , 1999, 128, 179-184.	3.3	81
133	Identification and Characterization of the Human Homologue (RAI2) of a Mouse Retinoic Acid-Induced Gene in Xp22. <i>Genomics</i> , 1999, 55, 275-283.	2.9	21
134	Bestrophin Gene Mutations in Patients with Best Vitelliform Macular Dystrophy. <i>Genomics</i> , 1999, 58, 98-101.	2.9	89
135	The electroretinogram of the rhodopsin knockout mouse. <i>Visual Neuroscience</i> , 1999, 16, 391-398.	1.0	83
136	Human bZIP Transcription Factor Gene NRL: Structure, Genomic Sequence, and Fine Linkage Mapping at 14q11.2 and Negative Mutation Analysis in Patients with Retinal Degeneration. <i>Genomics</i> , 1997, 45, 395-401.	2.9	28
137	Mutations in the Cone-Rod Homeobox Gene Are Associated with the Cone-Rod Dystrophy Photoreceptor Degeneration. <i>Neuron</i> , 1997, 19, 1329-1336.	8.1	250
138	Increased phase lag of the fundamental harmonic component of the 30 Hz flicker ERG in Schubert-Bornschein complete type CSNB. <i>Vision Research</i> , 1997, 37, 2471-2475.	1.4	13
139	Spectrum of Mutations in the RPGR Gene That Are Identified in 20% of Families with X-Linked Retinitis Pigmentosa. <i>American Journal of Human Genetics</i> , 1997, 61, 1287-1292.	6.2	93
140	Analysis of the RPGR Gene in 11 Pedigrees with the Retinitis Pigmentosa Type 3 Genotype: Paucity of Mutations in the Coding Region but Splice Defects in Two Families. <i>American Journal of Human Genetics</i> , 1997, 61, 571-580.	6.2	61
141	Retinopathy induced in mice by targeted disruption of the rhodopsin gene. <i>Nature Genetics</i> , 1997, 15, 216-219.	21.4	552
142	Summation of Rod and S Cone Signals at Threshold in Human Observers. <i>Vision Research</i> , 1996, 36, 2681-2688.	1.4	16
143	Linkage Study of Best's Vitelliform Macular Dystrophy (VMD2) in a Large North American Family. <i>Human Heredity</i> , 1996, 46, 211-220.	0.8	10
144	X-Linked Juvenile Retinoschisis: Localization between DXS1195, DXS418 and AFM291wf on a Single YAC. <i>Human Heredity</i> , 1996, 46, 329-335.	0.8	11

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145	Diagnostic Issues with Inherited Retinal and Macular Dystrophies. <i>Seminars in Ophthalmology</i> , 1995, 10, 279-294.	1.6	11
146	Refined Genetic Mapping of Juvenile X-Linked Retinoschisis. <i>Human Heredity</i> , 1995, 45, 206-210.	0.8	9
147	Evidence for two sites of adaptation affecting the dark-adapted ERG of cats and primates. <i>Vision Research</i> , 1995, 35, 435-442.	1.4	43
148	Push-pull model of the primate photopic electroretinogram: A role for hyperpolarizing neurons in shaping the b-wave. <i>Visual Neuroscience</i> , 1994, 11, 519-532.	1.0	354
149	Assessment of foveal cone photoreceptors in Stargardt's macular dystrophy using a small dot detection task. <i>Vision Research</i> , 1993, 33, 1509-1524.	1.4	57
150	Chapter 6 Negative components of the electroretinogram from proximal retina and photoreceptor. <i>Progress in Retinal and Eye Research</i> , 1991, 10, 121-160.	0.8	35
151	The scotopic threshold response of the cat erg is suppressed selectively by GABA and glycine. <i>Vision Research</i> , 1991, 31, 1-15.	1.4	86
152	Fenestrated Sheen Macular Dystrophy. <i>American Journal of Ophthalmology</i> , 1991, 112, 1-7.	3.3	14
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155	Contributions to the electroretinogram of currents originating in proximal retina. <i>Visual Neuroscience</i> , 1988, 1, 307-315.	1.0	37
156	Electroretinographic Findings in Selected Pedigrees With Choroideremia. <i>American Journal of Ophthalmology</i> , 1986, 101, 361-367.	3.3	51
157	Cone and rod ERGs in degenerations of central retina. <i>Survey of Ophthalmology</i> , 1984, 29, 235-236.	4.0	0