Samuel G Jacobson

List of Publications by Year
in descending order

285 papers	27,556 citations	4103 90 h-index	9118 149 g-index
289 all docs	289 docs citations	289 times ranked	13815 citing authors

#	Article	IF	Citations
1	Full-field stimulus testing: Role in the clinic and as an outcome measure in clinical trials of severe childhood retinal disease. Progress in Retinal and Eye Research, 2022, 87, 101000.	7.3	28
2	Comprehensive variant spectrum of the CNGA3 gene in patients affected by achromatopsia. Human Mutation, 2022, 43, 832-858.	1.1	8
3	Intravitreal antisense oligonucleotide sepofarsen in Leber congenital amaurosis type 10: a phase 1b/2 trial. Nature Medicine, 2022, 28, 1014-1021.	15.2	46
4	Restoration of Cone Sensitivity to Individuals with Congenital Photoreceptor Blindness within the Phase 1/2 Sepofarsen Trial. Ophthalmology Science, 2022, 2, 100133.	1.0	5
5	Mobility test to assess functional vision in dark-adapted patients with Leber congenital amaurosis. BMC Ophthalmology, 2022, 22, .	0.6	4
6	The landscape of submicroscopic structural variants at the OPN1LW/OPN1MW gene cluster on Xq28 underlying blue cone monochromacy. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	3.3	2
7	Leber Congenital Amaurosis Due to GUCY2D Mutations: Longitudinal Analysis of Retinal Structure and Visual Function. International Journal of Molecular Sciences, 2021, 22, 2031.	1.8	7
8	Durable vision improvement after a single treatment with antisense oligonucleotide sepofarsen: a case report. Nature Medicine, 2021, 27, 785-789.	15.2	41
9	Safety and improved efficacy signals following gene therapy in childhood blindness caused by GUCY2D mutations. IScience, 2021, 24, 102409.	1.9	22
10	A Novel ARL3 Gene Mutation Associated With Autosomal Dominant Retinal Degeneration. Frontiers in Cell and Developmental Biology, 2021, 9, 720782.	1.8	13
11	Gene therapy reforms photoreceptor structure and restores vision in NPHP5-associated Leber congenital amaurosis. Molecular Therapy, 2021, 29, 2456-2468.	3.7	18
12	Macular Rod Function in Retinitis Pigmentosa Measured With Scotopic Microperimetry. Translational Vision Science and Technology, 2021, 10, 3.	1.1	5
13	Measures of Function and Structure to Determine Phenotypic Features, Natural History, and Treatment Outcomes in Inherited Retinal Diseases. Annual Review of Vision Science, 2021, 7, 747-772.	2.3	14
14	Childhood-onset genetic cone-rod photoreceptor diseases and underlying pathobiology. EBioMedicine, 2021, 63, 103200.	2.7	4
15	RPGR isoform imbalance causes ciliary defects due to exon ORF15 mutations in X-linked retinitis pigmentosa (XLRP). Human Molecular Genetics, 2021, 29, 3706-3716.	1.4	16
16	Comparative Natural History of Visual Function From Patients With Biallelic Variants inBBS1andBBS10. , 2021, 62, 26.		11
17	Deepâ€intronic variants in CNGB3 cause achromatopsia by pseudoexon activation. Human Mutation, 2020, 41, 255-264.	1.1	26
18	Long-Term Structural Outcomes of Late-Stage RPE65 Gene Therapy. Molecular Therapy, 2020, 28, 266-278.	3.7	56

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19	Toxicity and Efficacy Evaluation of an Adeno-Associated Virus Vector Expressing Codon-Optimized RPGR Delivered by Subretinal Injection in a Canine Model of X-linked Retinitis Pigmentosa. Human Gene Therapy, 2020, 31, 253-267.	1.4	22
20	Progress in treating inherited retinal diseases: Early subretinal gene therapy clinical trials and candidates for future initiatives. Progress in Retinal and Eye Research, 2020, 77, 100827.	7.3	133
21	Rod function deficit in retained photoreceptors of patients with class B Rhodopsin mutations. Scientific Reports, 2020, 10, 12552.	1.6	10
22	Foveal Therapy in Blue Cone Monochromacy: Predictions of Visual Potential From Artificial Intelligence. Frontiers in Neuroscience, 2020, 14, 800.	1.4	7
23	Dose Range Finding Studies with Two RPGR Transgenes in a Canine Model of X-Linked Retinitis Pigmentosa Treated with Subretinal Gene Therapy. Human Gene Therapy, 2020, 31, 743-755.	1.4	15
24	Faster Sensitivity Loss around Dense Scotomas than for Overall Macular Sensitivity in Stargardt Disease: ProgStar Report No. 14. American Journal of Ophthalmology, 2020, 216, 219-225.	1.7	20
25	Transient pupillary light reflex in CEP290- or NPHP5-associated Leber congenital amaurosis: Latency as a potential outcome measure of cone function. Vision Research, 2020, 168, 53-63.	0.7	14
26	Reading Performance in Blue Cone Monochromacy: Defining an Outcome Measure for a Clinical Trial. Translational Vision Science and Technology, 2020, 9, 13.	1.1	5
27	Treatment Potential for Macular Cone Vision in Leber Congenital Amaurosis Due toCEP290orNPHP5Mutations: Predictions From Artificial Intelligence. , 2019, 60, 2551.		27
28	Short-Wavelength Sensitive Cone (S-cone) Testing as an Outcome Measure for NR2E3 Clinical Treatment Trials. International Journal of Molecular Sciences, 2019, 20, 2497.	1.8	13
29	Mutation spectrum and clinical investigation of achromatopsia patients with mutations in the GNAT2 gene. Human Mutation, 2019, 40, 1145-1155.	1.1	15
30	Leber Congenital Amaurosis (LCA): Potential for Improvement of Vision. , 2019, 60, 1680.		50
31	Autosomal Dominant Retinitis Pigmentosa Due to Class B Rhodopsin Mutations: An Objective Outcome for Future Treatment Trials. International Journal of Molecular Sciences, 2019, 20, 5344.	1.8	11
32	Effect of an intravitreal antisense oligonucleotide on vision in Leber congenital amaurosis due to a photoreceptor cilium defect. Nature Medicine, 2019, 25, 225-228.	15.2	177
33	A G86R mutation in the calcium-sensor protein GCAP1 alters regulation of retinal guanylyl cyclase and causes dominant cone-rod degeneration. Journal of Biological Chemistry, 2019, 294, 3476-3488.	1.6	29
34	BEST1 gene therapy corrects a diffuse retina-wide microdetachment modulated by light exposure. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E2839-E2848.	3.3	62
35	Nonsyndromic Retinitis Pigmentosa in the Ashkenazi Jewish Population. Ophthalmology, 2018, 125, 725-734.	2.5	30
36	Expanded Retinal Disease Spectrum Associated With Autosomal Recessive Mutations in GUCY2D. American Journal of Ophthalmology, 2018, 190, 58-68.	1.7	20

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37	Blue Cone Monochromacy Caused by the C203R Missense Mutation or Large Deletion Mutations. , 2018, 59, 5762.		21
38	Translational Retinal Research and Therapies. Translational Vision Science and Technology, 2018, 7, 8.	1.1	11
39	Progression in X-linked Retinitis Pigmentosa Due toORF15-RPGRMutations: Assessment of Localized Vision Changes Over 2 Years. , 2018, 59, 4558.		17
40	Efficacy Outcome Measures for Clinical Trials of USH2A Caused by the Common c.2299delG Mutation. American Journal of Ophthalmology, 2018, 193, 114-129.	1.7	19
41	Cone Vision Changes in the Enhanced S-Cone Syndrome Caused byNR2E3Gene Mutations. , 2018, 59, 3209.		19
42	Mutation-independent rhodopsin gene therapy by knockdown and replacement with a single AAV vector. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E8547-E8556.	3.3	114
43	Longitudinal Changes of Fixation Location and Stability Within 12 Months in Stargardt Disease: ProgStar Report No. 12. American Journal of Ophthalmology, 2018, 193, 54-61.	1.7	24
44	Dysfunction of cGMP signalling in photoreceptors by a macular dystrophy-related mutation in the calcium sensor GCAP1. Human Molecular Genetics, 2017, 26, ddw374.	1.4	34
45	Defining Outcomes for Clinical Trials of Leber Congenital Amaurosis Caused by GUCY2D Mutations. American Journal of Ophthalmology, 2017, 177, 44-57.	1.7	29
46	Optimization of Retinal Gene Therapy for X-Linked Retinitis Pigmentosa Due to RPGR Mutations. Molecular Therapy, 2017, 25, 1866-1880.	3.7	60
47	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.	1.4	23
48	Novel pathogenic mutations in C1QTNF5 support a dominant negative disease mechanism in late-onset retinal degeneration. Scientific Reports, 2017, 7, 12147.	1.6	30
49	CDHR1 mutations in retinal dystrophies. Scientific Reports, 2017, 7, 6992.	1.6	49
50	InÂVitro Modeling Using Ciliopathy-Patient-Derived Cells Reveals Distinct Cilia Dysfunctions Caused by CEP290 Mutations. Cell Reports, 2017, 20, 384-396.	2.9	120
51	EYS Mutations Causing Autosomal Recessive Retinitis Pigmentosa: Changes of Retinal Structure and Function with Disease Progression. Genes, 2017, 8, 178.	1.0	35
52	Outcome Measures for Clinical Trials of Leber Congenital Amaurosis Caused by the Intronic Mutation in the CEP290 Gene. , 2017, 58, 2609.		46
53	Pupillary Light Reflexes in Severe Photoreceptor Blindness Isolate the Melanopic Component of Intrinsically Photosensitive Retinal Ganglion Cells. , 2017, 58, 3215.		13
54	Imaging Lenticular Autofluorescence in Older Subjects. , 2017, 58, 4940.		9

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55	Postretinal Structure and Function in Severe Congenital Photoreceptor Blindness Caused by Mutations in the GUCY2D Gene. , 2017, 58, 959.		16
56	Complexity of the Class B Phenotype in Autosomal Dominant Retinitis Pigmentosa Due to Rhodopsin Mutations. , 2016, 57, 4847.		30
57	Visual Function and Central Retinal Structure in Choroideremia. , 2016, 57, OCT377.		65
58	Developing an Outcome Measure With High Luminance for Optogenetics Treatment of Severe Retinal Degenerations and for Gene Therapy of Cone Diseases. , 2016, 57, 3211.		18
59	Outer Retinal Changes Including the Ellipsoid Zone Band in Usher Syndrome 1B due toMYO7AMutations. , 2016, 57, OCT253.		26
60	Retinal Structure Measurements as Inclusion Criteria for Stem Cellâ€“Based Therapies of Retinal Degenerations. , 2016, 57, ORSFn1.		13
61	Automated Light- and Dark-Adapted Perimetry for Evaluating Retinitis Pigmentosa: Filling a Need to Accommodate Multicenter Clinical Trials. , 2016, 57, 3118.		40
62	Mutation Detection in Patients with Retinal Dystrophies Using Targeted Next Generation Sequencing. PLoS ONE, 2016, 11, e0145951.	1.1	91
63	Patterns of Individual Variation in Visual Pathway Structure and Function in the Sighted and Blind. PLoS ONE, 2016, 11, e0164677.	1.1	38
64	Visual Acuity Loss and Associated Risk Factors in the Retrospective Progression of Stargardt Disease Study (ProgStar Report No. 2). Ophthalmology, 2016, 123, 1887-1897.	2.5	59
65	DNA Methylation Profiling of Uniparental Disomy Subjects Provides a Map of Parental Epigenetic Bias in the Human Genome. American Journal of Human Genetics, 2016, 99, 555-566.	2.6	66
66	Overlap of abnormal photoreceptor development and progressive degeneration in Leber congenital amaurosis caused byNPHP5mutation. Human Molecular Genetics, 2016, 25, 4211-4226.	1.4	35
67	Variegated yet non-random rod and cone photoreceptor disease patterns inRPGR-ORF15-associated retinal degeneration. Human Molecular Genetics, 2016, 25, 5444-5459.	1.4	35
68	SPATA7: Evolving phenotype from cone-rod dystrophy to retinitis pigmentosa. Ophthalmic Genetics, 2016, 37, 333-338.	0.5	17
69	Leber Congenital Amaurosis: Genotypes and Retinal Structure Phenotypes. Advances in Experimental Medicine and Biology, 2016, 854, 169-175.	0.8	27
70	Functional Rescue of Retinal Degeneration-Associated Mutant RPE65 Proteins. Advances in Experimental Medicine and Biology, 2016, 854, 525-532.	0.8	5
71	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.	1.6	66
72	Autofluorescence Imaging With Near-Infrared Excitation: Normalization by Reflectance to Reduce Signal From Choroidal Fluorophores. , 2015, 56, 3393.		48

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73	Molecular Heterogeneity Within the Clinical Diagnosis of Pericentral Retinal Degeneration. , 2015, 56, 6007.		20
74	Predicting Progression ofABCA4-Associated Retinal Degenerations Based on Longitudinal Measurements of the Leading Disease Front. , 2015, 56, 5946.		36
75	Temperature-sensitive retinoid isomerase activity of RPE65 mutants associated with Leber Congenital Amaurosis. Journal of Biochemistry, 2015, 158, 115-125.	0.9	10
76	Pseudo-Fovea Formation After Gene Therapy for RPE65-LCA. Investigative Ophthalmology and Visual Science, 2015, 56, 526-537.	3.3	39
77	Genetics and Disease Expression in the CNGA3 Form of Achromatopsia. Ophthalmology, 2015, 122, 997-1007.	2.5	61
78	Protein misfolding and the pathogenesis of ABCA4-associated retinal degenerations. Human Molecular Genetics, 2015, 24, 3220-3237.	1.4	69
79	Improvement in vision: a new goal for treatment of hereditary retinal degenerations. Expert Opinion on Orphan Drugs, 2015, 3, 563-575.	0.5	23
80	Improvement and Decline in Vision with Gene Therapy in Childhood Blindness. New England Journal of Medicine, 2015, 372, 1920-1926.	13.9	333
81	Outcome measure for the treatment of cone photoreceptor diseases: orientation to a scene with cone-only contrast. BMC Ophthalmology, 2015, 15, 98.	0.6	4
82	Successful arrest of photoreceptor and vision loss expands the therapeutic window of retinal gene therapy to later stages of disease. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, E5844-53.	3.3	75
83	Gene Augmentation for X-Linked Retinitis Pigmentosa Caused by Mutations in RPGR. Cold Spring Harbor Perspectives in Medicine, 2015, 5, a017392-a017392.	2.9	19
84	Drusen and Photoreceptor Abnormalities in African-Americans with Intermediate Non-neovascular Age-related Macular Degeneration. Current Eye Research, 2015, 40, 398-406.	0.7	15
85	Blue Cone Monochromacy: Visual Function and Efficacy Outcome Measures for Clinical Trials. PLoS ONE, 2015, 10, e0125700.	1.1	29
86	Canine Retina Has a Primate Fovea-Like Bouquet of Cone Photoreceptors Which Is Affected by Inherited Macular Degenerations. PLoS ONE, 2014, 9, e90390.	1.1	100
87	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.	1.1	23
88	TULP1Mutations Causing Early-Onset Retinal Degeneration: Preserved but Insensitive Macular Cones. , 2014, 55, 5354.		47
89	Late-Onset Retinal Degeneration Caused byC1QTNF5Mutation. JAMA Ophthalmology, 2014, 132, 1252.	1.4	21
90	Inner and Outer Retinal Changes in Retinal Degenerations Associated WithABCA4Mutations. , 2014, 55, 1810.		48

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91	Rare and common variants in extracellular matrix gene Fibrillin 2 (FBN2) are associated with macular degeneration. Human Molecular Genetics, 2014, 23, 5827-5837.	1.4	52
92	Rescue of Enzymatic Function for Disease-associated RPE65 Proteins Containing Various Missense Mutations in Non-active Sites. Journal of Biological Chemistry, 2014, 289, 18943-18956.	1.6	42
93	Human retinal gene therapy for Leber congenital amaurosis shows advancing retinal degeneration despite enduring visual improvement. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, E517-25.	3.3	401
94	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.	2.6	120
95	Expanded Clinical Spectrum of Enhanced S-Cone Syndrome. JAMA Ophthalmology, 2013, 131, 1324.	1.4	59
96	Non-exomic and synonymous variants in ABCA4 are an important cause of Stargardt disease. Human Molecular Genetics, 2013, 22, 5136-5145.	1.4	159
97	Reply to Townes-Anderson: RPE65 gene therapy does not alter the natural history of retinal degeneration. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, E1706.	3.3	4
98	Determining consequences of retinal membrane guanylyl cyclase (RetGC1) deficiency in human Leber congenital amaurosis en route to therapy: residual cone-photoreceptor vision correlates with biochemical properties of the mutants. Human Molecular Genetics, 2013, 22, 168-183.	1.4	89
99	Human Cone Visual Pigment Deletions Spare Sufficient Photoreceptors to Warrant Gene Therapy. Human Gene Therapy, 2013, 24, 993-1006.	1.4	97
100	Comprehensive molecular diagnosis of 179 Leber congenital amaurosis and juvenile retinitis pigmentosa patients by targeted next generation sequencing. Journal of Medical Genetics, 2013, 50, 674-688.	1.5	139
101	AAV-Mediated Gene Therapy in the Guanylate Cyclase (RetGC1/RetGC2) Double Knockout Mouse Model of Leber Congenital Amaurosis. Human Gene Therapy, 2013, 24, 189-202.	1.4	60
102	Visual Acuity Changes in Patients With Leber Congenital Amaurosis and Mutations in CEP290. JAMA Ophthalmology, 2013, 131, 178.	1.4	44
103	Intervisit Variability of Visual Parameters in Leber Congenital Amaurosis Caused byRPE65Mutations. , 2013, 54, 1378.		24
104	Abnormal Thickening as well as Thinning of the Photoreceptor Layer in Intermediate Age-Related Macular Degeneration. , 2013, 54, 1603.		77
105	Gene Therapy for Retinitis Pigmentosa Caused by MFRP Mutations: Human Phenotype and Preliminary Proof of Concept. Human Gene Therapy, 2012, 23, 367-376.	1.4	35
106	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, 2132-2137.	3.3	237
107	Gene Therapy for Leber Congenital Amaurosis Caused by RPE65 Mutations. JAMA Ophthalmology, 2012, 130, 9.	2.6	580
108	Mutations inRPGRandRP2Account for 15% of Males with Simplex Retinal Degenerative Disease. , 2012, 53, 8232.		108

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109	Macular Function in Macular Degenerations: Repeatability of Microperimetry as a Potential Outcome Measure forABCA4-Associated Retinopathy Trials. , 2012, 53, 841.		105
110	RPGR-Associated Retinal Degeneration in Human X-Linked RP and a Murine Model. , 2012, 53, 5594.		56
111	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.	2.6	121
112	Combining Cep290 and Mkks ciliopathy alleles in mice rescues sensory defects and restores ciliogenesis. Journal of Clinical Investigation, 2012, 122, 1233-1245.	3.9	75
113	Combining Cep290 and Mkks ciliopathy alleles in mice rescues sensory defects and restores ciliogenesis. Journal of Clinical Investigation, 2012, 122, 3025-3025.	3.9	0
114	Retinal Disease Course in Usher Syndrome 1B Due toMYO7AMutations. , 2011, 52, 7924.		68
115	Autosomal Recessive Retinitis Pigmentosa Caused by Mutations in theMAKGene. , 2011, 52, 9665.		39
116	MpdzNull Allele in an Avian Model of Retinal Degeneration and Mutations in Human Leber Congenital Amaurosis and Retinitis Pigmentosa. , 2011, 52, 7432.		24
117	IQCB1Mutations in Patients with Leber Congenital Amaurosis. , 2011, 52, 834.		107
118	Variations in NPHP5 in Patients With Nonsyndromic Leber Congenital Amaurosis and Senior-Loken Syndrome. JAMA Ophthalmology, 2011, 129, 81.	2.6	62
119	A Missense Mutation in DHDDS, Encoding Dehydrodolichyl Diphosphate Synthase, Is Associated with Autosomal-Recessive Retinitis Pigmentosa in Ashkenazi Jews. American Journal of Human Genetics, 2011, 88, 207-215.	2.6	120
120	BBS genotype-phenotype assessment of a multiethnic patient cohort calls for a revision of the disease definition. Human Mutation, 2011, 32, 610-619.	1.1	100
121	Human Retinal Disease fromAIPL1Gene Mutations: Foveal Cone Loss with Minimal Macular Photoreceptors and Rod Function Remaining. , 2011, 52, 70.		59
122	HumanCRB1-Associated Retinal Degeneration: Comparison with therd8 Crb1-Mutant Mouse Model. , 2011, 52, 6898.		98
123	Probing Mechanisms of Photoreceptor Degeneration in a New Mouse Model of the Common Form of Autosomal Dominant Retinitis Pigmentosa due to P23H Opsin Mutations. Journal of Biological Chemistry, 2011, 286, 10551-10567.	1.6	221
124	The Usher 1B protein, MYO7A, is required for normal localization and function of the visual retinoid cycle enzyme, RPE65. Human Molecular Genetics, 2011, 20, 2560-2570.	1.4	56
125	Exome sequencing and analysis of induced pluripotent stem cells identify the cilia-related gene male germ cell-associated kinase (MAK) as a cause of retinitis pigmentosa. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, E569-76.	3.3	186
126	Defective photoreceptor phagocytosis in a mouse model of enhanced Sâ€cone syndrome causes progressive retinal degeneration. FASEB Journal, 2011, 25, 3157-3176.	0.2	76

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127	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, 1411-1423.	1.4	115
128	Characterisation of a C1qtnf5 Ser163Arg Knock-In Mouse Model of Late-Onset Retinal Macular Degeneration. PLoS ONE, 2011, 6, e27433.	1.1	16
129	Dissecting the pathogenic mechanisms of mutations in the pore region of the human cone photoreceptor cyclic nucleotide-gated channel. Human Mutation, 2010, 31, 830-839.	1.1	20
130	Normal Central Retinal Function and Structure Preserved in Retinitis Pigmentosa. , 2010, 51, 1079.		81
131	Retinal Disease in Rpe65-Deficient Mice: Comparison to Human Leber Congenital Amaurosis Due toRPE65Mutations. , 2010, 51, 5304.		27
132	Molecular Anthropology Meets Genetic Medicine to Treat Blindness in the North African Jewish Population: Human Gene Therapy Initiated in Israel. Human Gene Therapy, 2010, 21, 1749-1757.	1.4	65
133	Treatment Possibilities for Retinitis Pigmentosa. New England Journal of Medicine, 2010, 363, 1669-1671.	13.9	71
134	The Genomic, Biochemical, and Cellular Responses of the Retina in Inherited Photoreceptor Degenerations and Prospects for the Treatment of These Disorders. Annual Review of Neuroscience, 2010, 33, 441-472.	5.0	143
135	Visual Acuity in Patients with Leber's Congenital Amaurosis and Early Childhood-Onset Retinitis Pigmentosa. Ophthalmology, 2010, 117, 1190-1198.	2.5	95
136	Harmonin in the Murine Retina and the Retinal Phenotypes ofUsh1c-Mutant Mice and Human USH1C. , 2009, 50, 3881.		41
137	Defining the Residual Vision in Leber Congenital Amaurosis Caused byRPE65Mutations. , 2009, 50, 2368.		109
138	Disease Boundaries in the Retina of Patients with Usher Syndrome Caused byMYO7AGene Mutations. , 2009, 50, 1886.		83
139	Retinal Pigment Epithelium Defects in Humans and Mice with Mutations inMYO7A: Imaging Melanosome-Specific Autofluorescence. , 2009, 50, 4386.		75
140	Loss of cone photoreceptors caused by chromophore depletion is partially prevented by the artificial chromophore pro-drug, 9-cis-retinyl acetate. Human Molecular Genetics, 2009, 18, 2277-2287.	1.4	77
141	ABCA4 disease progression and a proposed strategy for gene therapy. Human Molecular Genetics, 2009, 18, 931-941.	1.4	163
142	CERKLMutations Cause an Autosomal Recessive Cone-Rod Dystrophy with Inner Retinopathy. , 2009, 50, 5944.		83
143	A common allele in RPGRIP1L is a modifier of retinal degeneration in ciliopathies. Nature Genetics, 2009, 41, 739-745.	9.4	255
144	Subconjunctivally implantable hydrogels with degradable and thermoresponsive properties for sustained release of insulin to the retina. Biomaterials, 2009, 30, 6541-6547.	5.7	86

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145	Loss of the Metalloprotease ADAM9 Leads to Cone-Rod Dystrophy in Humans and Retinal Degeneration in Mice. American Journal of Human Genetics, 2009, 84, 683-691.	2.6	76
146	Missense Mutations in a Retinal Pigment Epithelium Protein, Bestrophin-1, Cause Retinitis Pigmentosa. American Journal of Human Genetics, 2009, 85, 581-592.	2.6	156
147	TRPM1 Is Mutated in Patients with Autosomal-Recessive Complete Congenital Stationary Night Blindness. American Journal of Human Genetics, 2009, 85, 720-729.	2.6	207
148	Vision 1 Year after Gene Therapy for Leber's Congenital Amaurosis. New England Journal of Medicine, 2009, 361, 725-727.	13.9	197
149	Mutation analysis in the long isoform of USH2A in American patients with Usher Syndrome type II. Journal of Human Genetics, 2009, 54, 732-738.	1.1	35
150	HumanRPE65Gene Therapy for Leber Congenital Amaurosis: Persistence of Early Visual Improvements and Safety at 1 Year. Human Gene Therapy, 2009, 20, 999-1004.	1.4	305
151	Leber congenital amaurosis caused by Lebercilin (LCA5) mutation: retained photoreceptors adjacent to retinal disorganization. Molecular Vision, 2009, 15, 1098-106.	1.1	26
152	ABCA4 gene analysis in patients with autosomal recessive cone and cone rod dystrophies. European Journal of Human Genetics, 2008, 16, 812-819.	1.4	54
153	Usher syndromes due to MYO7A, PCDH15, USH2A or GPR98 mutations share retinal disease mechanism. Human Molecular Genetics, 2008, 17, 2405-2415.	1.4	106
154	Human gene therapy for RPE65 isomerase deficiency activates the retinoid cycle of vision but with slow rod kinetics. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 15112-15117.	3.3	639
155	Treatment of Leber Congenital Amaurosis Due toRPE65Mutations by Ocular Subretinal Injection of Adeno-Associated Virus Gene Vector: Short-Term Results of a Phase I Trial. Human Gene Therapy, 2008, 19, 979-990.	1.4	880
156	Retinal Laminar Architecture in Human Retinitis Pigmentosa Caused byRhodopsinGene Mutations. , 2008, 49, 1580.		118
157	Retinal Disease in Usher Syndrome III Caused by Mutations in the Clarin-1 Gene. , 2008, 49, 2651.		75
158	Photoreceptor Layer Topography in Children with Leber Congenital Amaurosis Caused byRPE65Mutations. , 2008, 49, 4573.		86
159	Phase I Trial of Leber Congenital Amaurosis due to RPE65 Mutations by Ocular Subretinal Injection of Adeno-Associated Virus Gene Vector: Short-Term Results. Human Gene Therapy, 2008, .	1.4	13
160	Human cone photoreceptor dependence on RPE65 isomerase. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 15123-15128.	3.3	135
161	Evidence for retinal remodelling in retinitis pigmentosa caused by PDE6B mutation. British Journal of Ophthalmology, 2007, 91, 699-701.	2.1	35
162	Full-field stimulus testing (FST) to quantify visual perception in severely blind candidates for treatment trials. Physiological Measurement, 2007, 28, N51-N56.	1.2	92

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163	Leber Congenital Amaurosis Caused by an RPGRIP1 Mutation Shows Treatment Potential. Ophthalmology, 2007, 114, 895-898.	2.5	44
164	Reduced-illuminance autofluorescence imaging in ABCA4-associated retinal degenerations. Journal of the Optical Society of America A: Optics and Image Science, and Vision, 2007, 24, 1457.	0.8	131
165	Canine and Human Visual Cortex Intact and Responsive Despite Early Retinal Blindness from RPE65 Mutation. PLoS Medicine, 2007, 4, e230.	3.9	107
166	RDH12andRPE65, Visual Cycle Genes Causing Leber Congenital Amaurosis, Differ in Disease Expression. , 2007, 48, 332.		66
167	Macular Pigment and Lutein Supplementation inABCA4-Associated Retinal Degenerations. , 2007, 48, 1319.		63
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