Samuel G Jacobson

List of Publications by Year
in descending order

285 papers	27,556 citations	3531 90 h-index	7950 149 g-index
289 all docs	289 docs citations	289 times ranked	12692 citing authors

#	Article	IF	Citations
1	Gene therapy restores vision in a canine model of childhood blindness. Nature Genetics, 2001, 28, 92-95.	21.4	1,130
2	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.	2.7	880
3	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.	7.1	639
4	Mutations in MERTK, the human orthologue of the RCS rat retinal dystrophy gene, cause retinitis pigmentosa. Nature Genetics, 2000, 26, 270-271.	21.4	622
5	Gene Therapy for Leber Congenital Amaurosis Caused by RPE65 Mutations. JAMA Ophthalmology, 2012, 130, 9.	2.4	580
6	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
7	Mutation of a nuclear receptor gene, NR2E3, causes enhanced S cone syndrome, a disorder of retinal cell fate. Nature Genetics, 2000, 24, 127-131.	21.4	439
8	Long-Term Restoration of Rod and Cone Vision by Single Dose rAAV-Mediated Gene Transfer to the Retina in a Canine Model of Childhood Blindness. Molecular Therapy, 2005, 12, 1072-1082.	8.2	421
9	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.	7.1	401
10	Improvement and Decline in Vision with Gene Therapy in Childhood Blindness. New England Journal of Medicine, 2015, 372, 1920-1926.	27.0	333
11	Identification of the gene (BBS1) most commonly involved in Bardet-Biedl syndrome, a complex human obesity syndrome. Nature Genetics, 2002, 31, 435-438.	21.4	327
12	Total colourblindness is caused by mutations in the gene encoding the Î±-subunit of the cone photoreceptor cGMP-gated cation channel. Nature Genetics, 1998, 19, 257-259.	21.4	321
13	Mutations in NYX, encoding the leucine-rich proteoglycan nyctalopin, cause X-linked complete congenital stationary night blindness. Nature Genetics, 2000, 26, 319-323.	21.4	309
14	HumanRPE65Gene Therapy for Leber Congenital Amaurosis: Persistence of Early Visual Improvements and Safety at 1 Year. Human Gene Therapy, 2009, 20, 999-1004.	2.7	305
15	CNGA3 Mutations in Hereditary Cone Photoreceptor Disorders. American Journal of Human Genetics, 2001, 69, 722-737.	6.2	294
16	De novo mutations in the CRX homeobox gene associated with Leber congenital amaurosis. Nature Genetics, 1998, 18, 311-312.	21.4	276
17	A common allele in RPGRIP1L is a modifier of retinal degeneration in ciliopathies. Nature Genetics, 2009, 41, 739-745.	21.4	255
18	Identification of the gene that, when mutated, causes the human obesity syndrome BBS4. Nature Genetics, 2001, 28, 188-191.	21.4	254

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19	Mutations in the Cone-Rod Homeobox Gene Are Associated with the Cone-Rod Dystrophy Photoreceptor Degeneration. Neuron, 1997, 19, 1329-1336.	8.1	250
20	Identifying photoreceptors in blind eyes caused by RPE65 mutations: Prerequisite for human gene therapy success. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 6177-6182.	7.1	249
21	Genetically engineered large animal model for studying cone photoreceptor survival and degeneration in retinitis pigmentosa. Nature Biotechnology, 1997, 15, 965-970.	17.5	247
22	Mutations in the Cone Photoreceptor G-Protein Î±-Subunit Gene GNAT2 in Patients with Achromatopsia. American Journal of Human Genetics, 2002, 71, 422-425.	6.2	245
23	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.	7.1	237
24	Automated Light- and Dark- Adapted Perimetry for Evaluating Retinitis Pigmentosa. Ophthalmology, 1986, 93, 1604-1611.	5.2	232
25	Mutations in ABCA4 result in accumulation of lipofuscin before slowing of the retinoid cycle: a reappraisal of the human disease sequence. Human Molecular Genetics, 2004, 13, 525-534.	2.9	231
26	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
27	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.	3.4	221
28	The nuclear receptor NR2E3 plays a role in human retinal photoreceptor differentiation and degeneration. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 473-478.	7.1	218
29	CNGB3 mutations account for 50% of all cases with autosomal recessive achromatopsia. European Journal of Human Genetics, 2005, 13, 302-308.	2.8	216
30	Retinal Function and Rhodopsin Levels in Autosomal Dominant Retinitis Pigmentosa With Rhodopsin Mutations. American Journal of Ophthalmology, 1991, 112, 256-271.	3.3	213
31	Long-Term Protection of Retinal Structure but Not Function Using RAAV.CNTF in Animal Models of Retinitis Pigmentosa. Molecular Therapy, 2001, 4, 461-472.	8.2	209
32	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
33	Crumbs homolog 1 (CRB1) mutations result in a thick human retina with abnormal lamination. Human Molecular Genetics, 2003, 12, 1073-1078.	2.9	205
34	Autosomal Dominant Retinitis Pigmentosa Caused by the Threonine-17-Methionine Rhodopsin Mutation: Retinal Histopathology and Immunocytochemistry. Experimental Eye Research, 1994, 58, 397-408.	2.6	197
35	Night blindness in Sorsby's fundus dystrophy reversed by vitamin A. Nature Genetics, 1995, 11, 27-32.	21.4	197
36	Vision 1 Year after Gene Therapy for Leber's Congenital Amaurosis. New England Journal of Medicine, 2009, 361, 725-727.	27.0	197

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37	Safety of Recombinant Adeno-Associated Virus Type 2â€“RPE65 Vector Delivered by Ocular Subretinal Injection. Molecular Therapy, 2006, 13, 1074-1084.	8.2	196
38	In Utero Gene Therapy Rescues Vision in a Murine Model of Congenital Blindness. Molecular Therapy, 2004, 9, 182-188.	8.2	191
39	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.	7.1	186
40	Diagnostic Clinical Findings of a New Syndrome with Night Blindness, Maculopathy, and Enhanced S Cone Sensitivity. American Journal of Ophthalmology, 1990, 110, 124-134.	3.3	183
41	In vivo dynamics of retinal injury and repair in the rhodopsin mutant dog model of human retinitis pigmentosa. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 5233-5238.	7.1	183
42	Effect of an intravitreal antisense oligonucleotide on vision in Leber congenital amaurosis due to a photoreceptor cilium defect. Nature Medicine, 2019, 25, 225-228.	30.7	177
43	Mutation in a short-chain collagen gene, CTRP5 , results in extracellular deposit formation in late-onset retinal degeneration: a genetic model for age-related macular degeneration. Human Molecular Genetics, 2003, 12, 2657-2667.	2.9	172
44	ABCA4 disease progression and a proposed strategy for gene therapy. Human Molecular Genetics, 2009, 18, 931-941.	2.9	163
45	Non-exomic and synonymous variants in ABCA4 are an important cause of Stargardt disease. Human Molecular Genetics, 2013, 22, 5136-5145.	2.9	159
46	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
47	Remodeling of the Human Retina in Choroideremia: Rab Escort Protein 1 (REP-1) Mutations. , 2006, 47, 4113.		156
48	Missense Mutations in a Retinal Pigment Epithelium Protein, Bestrophin-1, Cause Retinitis Pigmentosa. American Journal of Human Genetics, 2009, 85, 581-592.	6.2	156
49	TULP1 mutation in two extended Dominican kindreds with autosomal recessive Retinitis pigmentosa. Nature Genetics, 1998, 18, 177-179.	21.4	151
50	Early age-related maculopathy and self-reported visual difficulty in daily life2 2The authors have no commercial interests in any device or product mentioned in this article.. Ophthalmology, 2002, 109, 1235-1242.	5.2	151
51	Naturally occurring rhodopsin mutation in the dog causes retinal dysfunction and degeneration mimicking human dominant retinitis pigmentosa. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 6328-6333.	7.1	150
52	Centrosomal-ciliary geneCEP290/NPHP6 mutations result in blindness with unexpected sparing of photoreceptors and visual brain: implications for therapy of Leber congenital amaurosis. Human Mutation, 2007, 28, 1074-1083.	2.5	148
53	Quantifying rod photoreceptor-mediated vision in retinal degenerations: dark-adapted thresholds as outcome measures. Experimental Eye Research, 2005, 80, 259-272.	2.6	145
54	Retinal Dystrophy Due to Paternal Isodisomy for Chromosome 1 or Chromosome 2, with Homoallelism for Mutations in RPE65 or MERTK, Respectively. American Journal of Human Genetics, 2002, 70, 224-229.	6.2	144

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55	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.	10.7	143
56	Safety in Nonhuman Primates of Ocular AAV2-RPE65, a Candidate Treatment for Blindness in Leber Congenital Amaurosis. Human Gene Therapy, 2006, 17, 845-858.	2.7	142
57	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.	3.2	139
58	Human cone photoreceptor dependence on RPE65 isomerase. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 15123-15128.	7.1	135
59	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.	15.5	133
60	Title is missing!. Nature Genetics, 2001, 28, 92-95.	21.4	132
61	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.	1.5	131
62	Identification of a locus, distinct from RDS-peripherin, for autosomal recessive retinitis pigmentosa on chromosome 6p. Human Molecular Genetics, 1994, 3, 1401-1403.	2.9	129
63	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
64	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.	6.2	120
65	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
66	InÂVitro Modeling Using Ciliopathy-Patient-Derived Cells Reveals Distinct Cilia Dysfunctions Caused by CEP290 Mutations. Cell Reports, 2017, 20, 384-396.	6.4	120
67	Pattern of Retinal Dysfunction in Acute Zonal Occult Outer Retinopathy. Ophthalmology, 1995, 102, 1187-1198.	5.2	119
68	Mutations in the PDE6B Gene in Autosomal Recessive Retinitis Pigmentosa. Genomics, 1995, 30, 1-7.	2.9	118
69	Retinal Laminar Architecture in Human Retinitis Pigmentosa Caused byRhodopsinGene Mutations. , 2008, 49, 1580.		118
70	Evaluation of Complex Inheritance Involving the Most Common Bardet-Biedl Syndrome Locus (BBS1). American Journal of Human Genetics, 2003, 72, 429-437.	6.2	117
71	ABCA4-Associated Retinal Degenerations Spare Structure and Function of the Human Parapapillary Retina. , 2005, 46, 4739.		117
72	An Alternative Phototransduction Model for Human Rod and Cone ERG a -waves: Normal Parameters and Variation with Age. Vision Research, 1996, 36, 2609-2621.	1.4	116

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73	Characterization of Usher syndrome type I gene mutations in an Usher syndrome patient population. Human Genetics, 2005, 116, 292-299.	3.8	116
74	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.	2.9	115
75	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.	7.1	114
76	In vivo function of the orphan nuclear receptor NR2E3 in establishing photoreceptor identity during mammalian retinal development. Human Molecular Genetics, 2006, 15, 2588-2602.	2.9	113
77	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
78	Defining the Residual Vision in Leber Congenital Amaurosis Caused byRPE65Mutations. , 2009, 50, 2368.		109
79	Mutations inRPGRandRP2Account for 15% of Males with Simplex Retinal Degenerative Disease. , 2012, 53, 8232.		108
80	Canine and Human Visual Cortex Intact and Responsive Despite Early Retinal Blindness from RPE65 Mutation. PLoS Medicine, 2007, 4, e230.	8.4	107
81	Inner Retinal Abnormalities in X-linked Retinitis Pigmentosa withRPGRMutations. , 2007, 48, 4759.		107
82	IQCB1Mutations in Patients with Leber Congenital Amaurosis. , 2011, 52, 834.		107
83	Lifespan and mitochondrial control of neurodegeneration. Nature Genetics, 2004, 36, 1153-1158.	21.4	106
84	Usher syndromes due to MYO7A, PCDH15, USH2A or GPR98 mutations share retinal disease mechanism. Human Molecular Genetics, 2008, 17, 2405-2415.	2.9	106
85	Macular Function in Macular Degenerations: Repeatability of Microperimetry as a Potential Outcome Measure forABCA4-Associated Retinopathy Trials. , 2012, 53, 841.		105
86	BBS genotype-phenotype assessment of a multiethnic patient cohort calls for a revision of the disease definition. Human Mutation, 2011, 32, 610-619.	2.5	100
87	Canine Retina Has a Primate Fovea-Like Bouquet of Cone Photoreceptors Which Is Affected by Inherited Macular Degenerations. PLoS ONE, 2014, 9, e90390.	2.5	100
88	Rod and cone visual cycle consequences of a null mutation in the 11-cis-retinol dehydrogenase gene in man. Visual Neuroscience, 2000, 17, 667-678.	1.0	99
89	Interocular asymmetry of visual function in heterozygotes of X-linked retinitis pigmentosa. Experimental Eye Research, 1989, 48, 679-691.	2.6	98
90	Enhanced S cone syndrome: Evidence for an abnormally large number of S cones. Vision Research, 1995, 35, 1473-1481.	1.4	98

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91	HumanCRB1-Associated Retinal Degeneration: Comparison with therd8 Crb1-Mutant Mouse Model. , 2011, 52, 6898.		98
92	Human Cone Visual Pigment Deletions Spare Sufficient Photoreceptors to Warrant Gene Therapy. Human Gene Therapy, 2013, 24, 993-1006.	2.7	97
93	Macular Pigment and Lutein Supplementation in Choroideremia. Experimental Eye Research, 2002, 74, 371-381.	2.6	96
94	Visual Acuity in Patients with Leber's Congenital Amaurosis and Early Childhood-Onset Retinitis Pigmentosa. Ophthalmology, 2010, 117, 1190-1198.	5.2	95
95	Spinocerebellar Ataxia Type 7 (SCA7) Shows a Coneâ€"Rod Dystrophy Phenotype. Experimental Eye Research, 2002, 74, 737-745.	2.6	94
96	Nuclear receptor NR2E3 gene mutations distort human retinal laminar architecture and cause an unusual degeneration. Human Molecular Genetics, 2004, 13, 1893-1902.	2.9	94
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	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
99	Impairment of the Transient Pupillary Light Reflex inRpe65^{â^'/â^'}Mice and Humans with Leber Congenital Amaurosis. , 2004, 45, 1259.		92
100	Mutation analysis ofNR2E3 andNRL genes in Enhanced S Cone Syndrome. Human Mutation, 2004, 24, 439-439.	2.5	92
101	Full-field stimulus testing (FST) to quantify visual perception in severely blind candidates for treatment trials. Physiological Measurement, 2007, 28, N51-N56.	2.1	92
102	Augmented rod bipolar cell function in partial receptor loss: an ERG study in P23H rhodopsin transgenic and aging normal rats. Vision Research, 2001, 41, 2779-2797.	1.4	91
103	Mutation Detection in Patients with Retinal Dystrophies Using Targeted Next Generation Sequencing. PLoS ONE, 2016, 11, e0145951.	2.5	91
104	Relatively Enhanced S Cone Function in the Goldmann-Favre Syndrome. American Journal of Ophthalmology, 1991, 111, 446-453.	3.3	90
105	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.	2.9	89
106	Photoreceptor Layer Topography in Children with Leber Congenital Amaurosis Caused byRPE65Mutations. , 2008, 49, 4573.		86
107	Subconjunctivally implantable hydrogels with degradable and thermoresponsive properties for sustained release of insulin to the retina. Biomaterials, 2009, 30, 6541-6547.	11.4	86
108	Visual function and rhodopsin levels in humans with vitamin A deficiency. Experimental Eye Research, 1988, 46, 185-197.	2.6	84

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109	Dominant late-onset retinal degeneration with regional variation of sub-retinal pigment epithelium deposits, retinal function, and photoreceptor degeneration. Ophthalmology, 2000, 107, 2256-2266.	5.2	83
110	Usher Syndrome Type III: Revised Genomic Structure of the USH3 Gene and Identification of Novel Mutations. American Journal of Human Genetics, 2002, 71, 607-617.	6.2	83
111	Retinal Disease Expression in Bardet-Biedl Syndrome-1 (BBS1) Is a Spectrum from Maculopathy to Retina-Wide Degeneration. , 2006, 47, 5004.		83
112	Disease Boundaries in the Retina of Patients with Usher Syndrome Caused byMYO7AGene Mutations. , 2009, 50, 1886.		83
113	CERKLMutations Cause an Autosomal Recessive Cone-Rod Dystrophy with Inner Retinopathy. , 2009, 50, 5944.		83
114	Normal Central Retinal Function and Structure Preserved in Retinitis Pigmentosa. , 2010, 51, 1079.		81
115	Lentiviral Expression of Retinal Guanylate Cyclase-1 (RetGC1) Restores Vision in an Avian Model of Childhood Blindness. PLoS Medicine, 2006, 3, e201.	8.4	80
116	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
117	Foveal Cone Electroretinograms in Retinitis Pigmentosa and Juvenile Macular Degeneration. American Journal of Ophthalmology, 1979, 88, 702-707.	3.3	77
118	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.	2.9	77
119	Abnormal Thickening as well as Thinning of the Photoreceptor Layer in Intermediate Age-Related Macular Degeneration. , 2013, 54, 1603.		77
120	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.	6.2	76
121	Defective photoreceptor phagocytosis in a mouse model of enhanced Sâ€cone syndrome causes progressive retinal degeneration. FASEB Journal, 2011, 25, 3157-3176.	0.5	76
122	Retinal Rod Photoreceptorâ€“Specific Gene Mutation Perturbs Cone Pathway Development. Neuron, 1999, 23, 549-557.	8.1	75
123	Retinal Disease in Usher Syndrome III Caused by Mutations in the Clarin-1 Gene. , 2008, 49, 2651.		75
124	Retinal Pigment Epithelium Defects in Humans and Mice with Mutations inMYO7A: Imaging Melanosome-Specific Autofluorescence. , 2009, 50, 4386.		75
125	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.	7.1	75
126	Combining Cep290 and Mkks ciliopathy alleles in mice rescues sensory defects and restores ciliogenesis. Journal of Clinical Investigation, 2012, 122, 1233-1245.	8.2	75

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127	QRX, a novel homeobox gene, modulates photoreceptor gene expression. Human Molecular Genetics, 2004, 13, 1025-1040.	2.9	73
128	Pitfalls in Homozygosity Mapping. American Journal of Human Genetics, 2000, 67, 1348-1351.	6.2	72
129	Treatment Possibilities for Retinitis Pigmentosa. New England Journal of Medicine, 2010, 363, 1669-1671.	27.0	71
130	Abnormal Rod Dark Adaptation in Autosomal Dominant Retinitis Pigmentosa With Proline-23-Histidine Rhodopsin Mutation. American Journal of Ophthalmology, 1992, 113, 165-174.	3.3	69
131	Protein misfolding and the pathogenesis of ABCA4-associated retinal degenerations. Human Molecular Genetics, 2015, 24, 3220-3237.	2.9	69
132	In vivo micropathology of Best macular dystrophy with optical coherence tomography. Experimental Eye Research, 2003, 76, 203-211.	2.6	68
133	Retinal Disease Course in Usher Syndrome 1B Due toMYO7AMutations. , 2011, 52, 7924.		68
134	RDH12andRPE65, Visual Cycle Genes Causing Leber Congenital Amaurosis, Differ in Disease Expression. , 2007, 48, 332.		66
135	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.	3.3	66
136	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.	6.2	66
137	A Novel Locus (RP24) for X-linked Retinitis Pigmentosa Maps to Xq26-27. American Journal of Human Genetics, 1998, 63, 1439-1447.	6.2	65
138	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.	2.7	65
139	Visual Function and Central Retinal Structure in Choroideremia. , 2016, 57, OCT377.		65
140	Nonhuman Primate Models for Diabetic Ocular Neovascularization Using AAV2-Mediated Overexpression of Vascular Endothelial Growth Factor. Diabetes, 2005, 54, 1141-1149.	0.6	64
141	Photoreceptor Rosettes with Blue Cone Opsin Immunoreactivity in Retinitis Pigmentosa. Ophthalmology, 1990, 97, 1620-1631.	5.2	63
142	Genetic Heterogeneity of Usher Syndrome: Analysis of 151 Families with Usher Type I. American Journal of Human Genetics, 2000, 67, 1569-1574.	6.2	63
143	Macular Pigment and Lutein Supplementation inABCA4-Associated Retinal Degenerations. , 2007, 48, 1319.		63
144	Human Rod Monochromacy: Linkage Analysis and Mapping of a Cone Photoreceptor Expressed Candidate Gene on Chromosome 2q11. Genomics, 1998, 51, 325-331.	2.9	62

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145	Variations in NPHP5 in Patients With Nonsyndromic Leber Congenital Amaurosis and Senior-Loken Syndrome. JAMA Ophthalmology, 2011, 129, 81.	2.4	62
146	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.	7.1	62
147	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
148	Genetics and Disease Expression in the CNGA3 Form of Achromatopsia. Ophthalmology, 2015, 122, 997-1007.	5.2	61
149	A Peripherin/Retinal Degeneration Slow Mutation (Pro-210-Arg) Associated with Macular and Peripheral Retinal Degeneration. Ophthalmology, 1995, 102, 246-255.	5.2	60
150	Disease Expression in Usher Syndrome Caused byVLGR1Gene Mutation (USH2C) and Comparison withUSH2APhenotype. , 2005, 46, 734.		60
151	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.	2.7	60
152	Optimization of Retinal Gene Therapy for X-Linked Retinitis Pigmentosa Due to RPGR Mutations. Molecular Therapy, 2017, 25, 1866-1880.	8.2	60
153	Human Retinal Disease fromAIPL1Gene Mutations: Foveal Cone Loss with Minimal Macular Photoreceptors and Rod Function Remaining. , 2011, 52, 70.		59
154	Expanded Clinical Spectrum of Enhanced S-Cone Syndrome. JAMA Ophthalmology, 2013, 131, 1324.	2.5	59
155	Visual Acuity Loss and Associated Risk Factors in the Retrospective Progression of Stargardt Disease Study (ProgStar Report No. 2). Ophthalmology, 2016, 123, 1887-1897.	5.2	59
156	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.	2.9	56
157	RPGR-Associated Retinal Degeneration in Human X-Linked RP and a Murine Model. , 2012, 53, 5594.		56
158	Long-Term Structural Outcomes of Late-Stage RPE65 Gene Therapy. Molecular Therapy, 2020, 28, 266-278.	8.2	56
159	ABCA4 gene analysis in patients with autosomal recessive cone and cone rod dystrophies. European Journal of Human Genetics, 2008, 16, 812-819.	2.8	54
160	Four novel mutations in the RPE65 gene in patients with Leber congenital amaurosis. Human Mutation, 2001, 18, 164-164.	2.5	52
161	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
162	Clinicopathologic effects of mutant GUCY2D in Leber congenital amaurosis. Ophthalmology, 2003, 110, 549-558.	5.2	50

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163	Leber Congenital Amaurosis (LCA): Potential for Improvement of Vision. , 2019, 60, 1680.		50
164	CDHR1 mutations in retinal dystrophies. Scientific Reports, 2017, 7, 6992.	3.3	49
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