Eric A Pierce

List of Publications by Year
in descending order

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104	10,581	66343	42399
papers	citations	42	92
		h-index	g-index

119	119	119	10886
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Safety and Efficacy of Gene Transfer for Leber's Congenital Amaurosis. New England Journal of Medicine, 2008, 358, 2240-2248.	27.0	1,941
2	Age-dependent effects of RPE65 gene therapy for Leber's congenital amaurosis: a phase 1 dose-escalation trial. Lancet, The, 2009, 374, 1597-1605.	13.7	774
3	Gene Therapy for Leber's Congenital Amaurosis is Safe and Effective Through 1.5 Years After Vector Administration. Molecular Therapy, 2010, 18, 643-650.	8.2	503
4	Regulation of Vascular Endothelial Growth Factor by Oxygen in a Model of Retinopathy of Prematurity. JAMA Ophthalmology, 1996, 114, 1219.	2.4	435
5	Safety and durability of effect of contralateral-eye administration of AAV2 gene therapy in patients with childhood-onset blindness caused by RPE65 mutations: a follow-on phase 1 trial. Lancet, The, 2016, 388, 661-672.	13.7	377
6	AAV2 Gene Therapy Readministration in Three Adults with Congenital Blindness. Science Translational Medicine, 2012, 4, 120ra15.	12.4	340
7	TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum. Nature Genetics, 2011, 43, 189-196.	21.4	326
8	The Proteome of the Mouse Photoreceptor Sensory Cilium Complex. Molecular and Cellular Proteomics, 2007, 6, 1299-1317.	3.8	310
9	Three-Year Follow-up after Unilateral Subretinal Delivery of Adeno-Associated Virus in Patients with Leber Congenital Amaurosis Type 2. Ophthalmology, 2013, 120, 1283-1291.	5.2	301
10	Comparative analysis of RNA-Seq alignment algorithms and the RNA-Seq unified mapper (RUM). Bioinformatics, 2011, 27, 2518-2528.	4.1	298
11	Candidate exome capture identifies mutation of SDCCAG8 as the cause of a retinal-renal ciliopathy. Nature Genetics, 2010, 42, 840-850.	21.4	295
12	Panel-based genetic diagnostic testing for inherited eye diseases is highly accurate and reproducible, and more sensitive for variant detection, than exome sequencing. Genetics in Medicine, 2015, 17, 253-261.	2.4	216
13	Mutations in a gene encoding a new oxygen-regulated photoreceptor protein cause dominant retinitis pigmentosa. Nature Genetics, 1999, 22, 248-254.	21.4	174
14	NMNAT1 mutations cause Leber congenital amaurosis. Nature Genetics, 2012, 44, 1040-1045.	21.4	171
15	Inherited Retinal Degenerations: Current Landscape and Knowledge Gaps. Translational Vision Science and Technology, 2018, 7, 6.	2.2	168
16	Mutations in TSPAN12 Cause Autosomal-Dominant Familial Exudative Vitreoretinopathy. American Journal of Human Genetics, 2010, 86, 248-253.	6.2	161
17	Photoreceptor Cilia and Retinal Ciliopathies. Cold Spring Harbor Perspectives in Biology, 2017, 9, a028274.	5.5	154
18	Transcriptome analyses of the human retina identify unprecedented transcript diversity and 3.5 Mb of novel transcribed sequence via significant alternative splicing and novel genes. BMC Genomics, 2013, 14, 486.	2.8	151

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19	The Retinitis Pigmentosa 1 Protein Is a Photoreceptor Microtubule-Associated Protein. Journal of Neuroscience, 2004, 24, 6427-6436.	3.6	140
20	Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2013, 93, 482-495.	6.2	138
21	Mutations in IFT172 cause isolated retinal degeneration and Bardetâ€“Biedl syndrome. Human Molecular Genetics, 2015, 24, 230-242.	2.9	136
22	The Status ofRPE65Gene Therapy Trials: Safety and Efficacy. Cold Spring Harbor Perspectives in Medicine, 2015, 5, a017285.	6.2	130
23	The R345W mutation in EFEMP1 is pathogenic and causes AMD-like deposits in mice. Human Molecular Genetics, 2007, 16, 2411-2422.	2.9	129
24	RP1 Is Required for the Correct Stacking of Outer Segment Discs. , 2003, 44, 4171.		122
25	Progressive photoreceptor degeneration, outer segment dysplasia, and rhodopsin mislocalization in mice with targeted disruption of the retinitis pigmentosa-1 (Rp1) gene. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 5698-5703.	7.1	113
26	Allele-Specific CRISPR-Cas9 Genome Editing of the Single-Base P23H Mutation for Rhodopsin-Associated Dominant Retinitis Pigmentosa. CRISPR Journal, 2018, 1, 55-64.	2.9	96
27	Identification and subcellular localization of the RP1 protein in human and mouse photoreceptors. Investigative Ophthalmology and Visual Science, 2002, 43, 22-32.	3.3	89
28	Pathways to photoreceptor cell death in inherited retinal degenerations. BioEssays, 2001, 23, 605-618.	2.5	88
29	Generation ofCreTransgenic Mice with Postnatal RPE-Specific Ocular Expression. , 2011, 52, 1378.		85
30	Isolation, culture and characterization of primary mouse RPE cells. Nature Protocols, 2016, 11, 1206-1218.	12.0	79
31	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
32	Three Gene-Targeted Mouse Models of RNA Splicing Factor RP Show Late-Onset RPE and Retinal Degeneration. , 2011, 52, 190.		70
33	Analysis of Kinesin-2 Function in Photoreceptor Cells Using SynchronousCre-loxP Knockout ofKif3awithRHO-Cre. , 2006, 47, 5039.		68
34	Quantification of the cytoplasmic spaces of living cells with EGFP reveals arrestin-EGFP to be in disequilibrium in dark adapted rod photoreceptors. Journal of Cell Science, 2004, 117, 3049-3059.	2.0	66
35	Mutations in Pre-mRNA Processing Factors 3, 8, and 31 Cause Dysfunction of the Retinal Pigment Epithelium. American Journal of Pathology, 2014, 184, 2641-2652.	3.8	62
36	A local complement response by RPE causes early-stage macular degeneration. Human Molecular Genetics, 2015, 24, 5555-5569.	2.9	62

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37	Mouse Models of NMNAT1-Leber Congenital Amaurosis (LCA9) Recapitulate Key Features of the Human Disease. American Journal of Pathology, 2016, 186, 1925-1938.	3.8	61
38	Copy-number variation contributes 9% of pathogenicity in the inherited retinal degenerations. Genetics in Medicine, 2020, 22, 1079-1087.	2.4	59
39	Changes in extracellular matrix cause RPE cells to make basal deposits and activate the alternative complement pathway. Human Molecular Genetics, 2018, 27, 147-159.	2.9	58
40	Advancing Clinical Trials for Inherited Retinal Diseases: Recommendations from the Second Monaciano Symposium. Translational Vision Science and Technology, 2020, 9, 2.	2.2	56
41	Why Do Mutations in the Ubiquitously Expressed Housekeeping GeneIMPDH1Cause Retina-Specific Photoreceptor Degeneration?. , 2006, 47, 3754.		55
42	Retinal expression, regulation, and functional bioactivity of prostacyclin-stimulating factor. Journal of Clinical Investigation, 2000, 106, 541-550.	8.2	53
43	Retinal Degeneration and Failure of Photoreceptor Outer Segment Formation in Mice with Targeted Deletion of the Joubert Syndrome Gene, Ahi1. Journal of Neuroscience, 2010, 30, 8759-8768.	3.6	52
44	Copy-number variation is an important contributor to the genetic causality of inherited retinal degenerations. Genetics in Medicine, 2017, 19, 643-651.	2.4	51
45	Retinopathy of Prematurity. Molecular Diagnosis and Therapy, 2003, 3, 261-277.	3.3	48
46	Mouse genetics and proteomic analyses demonstrate a critical role for complement in a model of DHRD/ML, an inherited macular degeneration. Human Molecular Genetics, 2014, 23, 52-68.	2.9	47
47	Mitochondrial disease genetic diagnostics: optimized whole-exome analysis for all MitoCarta nuclear genes and the mitochondrial genome. Discovery Medicine, 2012, 14, 389-99.	0.5	47
48	USMG5 Ashkenazi Jewish founder mutation impairs mitochondrial complex V dimerization and ATP synthesis. Human Molecular Genetics, 2018, 27, 3305-3312.	2.9	45
49	Systemic Diseases Associated with Retinal Dystrophies. Seminars in Ophthalmology, 2014, 29, 319-328.	1.6	44
50	Extracellular Matrix Alterations and Deposit Formation in AMD. Advances in Experimental Medicine and Biology, 2016, 854, 53-58.	1.6	43
51	Temporal and Tissue Specific Regulation of RP-Associated Splicing Factor Genes PRPF3, PRPF31 and PRPC8â€"Implications in the Pathogenesis of RP. PLoS ONE, 2011, 6, e15860.	2.5	42
52	A novelHSD17B10mutation impairing the activities of the mitochondrial RNase P complex causes X-linked intractable epilepsy and neurodevelopmental regression. RNA Biology, 2016, 13, 477-485.	3.1	42
53	C3a triggers formation of sub-retinal pigment epithelium deposits via the ubiquitin proteasome pathway. Scientific Reports, 2018, 8, 9679.	3.3	42
54	CRB1: One Gene, Many Phenotypes. Seminars in Ophthalmology, 2013, 28, 397-405.	1.6	37

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55	Exome-Based Mapping and Variant Prioritization for Inherited Mendelian Disorders. American Journal of Human Genetics, 2014, 94, 373-384.	6.2	37
56	Application of Next-Generation Sequencing to Identify Genes and Mutations Causing Autosomal Dominant Retinitis Pigmentosa (adRP). Advances in Experimental Medicine and Biology, 2014, 801, 123-129.	1.6	37
57	Targeted High-Throughput DNA Sequencing for Gene Discovery in Retinitis Pigmentosa. Advances in Experimental Medicine and Biology, 2010, 664, 325-331.	1.6	35
58	The Genetic Basis of Pericentral Retinitis Pigmentosaâ€"A Form of Mild Retinitis Pigmentosa. Genes, 2017, 8, 256.	2.4	34
59	Photoreceptor Sensory Cilia and Inherited Retinal Degeneration. Advances in Experimental Medicine and Biology, 2010, 664, 223-232.	1.6	34
60	Expression and Activation of STAT3 in Ischemia-Induced Retinopathy. , 2005, 46, 4409.		31
61	Decreased Levels of the RNA Splicing Factor Prpf3 in Mice and Zebrafish Do Not Cause Photoreceptor Degeneration. , 2008, 49, 3830.		31
62	Prognosis for splicing factor PRPF8 retinitis pigmentosa, novel mutations and correlation between human and yeast phenotypes. Human Mutation, 2010, 31, E1361-E1376.	2.5	31
63	Mutations in the Kinesin-2 Motor KIF3B Cause an Autosomal-Dominant Ciliopathy. American Journal of Human Genetics, 2020, 106, 893-904.	6.2	29
64	AAV-Mediated Gene Augmentation Therapy Restores Critical Functions in Mutant PRPF31+/â^' iPSC-Derived RPE Cells. Molecular Therapy - Methods and Clinical Development, 2019, 15, 392-402.	4.1	28
65	Contribution of noncoding pathogenic variants to RPGRIP1-mediated inherited retinal degeneration. Genetics in Medicine, 2019, 21, 694-704.	2.4	27
66	Genetic Testing for Inherited Eye Disease. JAMA Ophthalmology, 2013, 131, 1265.	2.5	26
67	In Vivo Assessment of Potential Therapeutic Approaches for USH2A-Associated Diseases. Advances in Experimental Medicine and Biology, 2019, 1185, 91-96.	1.6	26
68	Characterization of RP1L1, a highly polymorphic paralog of the retinitis pigmentosa 1 (RP1) gene. Molecular Vision, 2003, 9, 129-37.	1.1	26
69	Targeted Exon Sequencing in Usher Syndrome Type I. Investigative Ophthalmology and Visual Science, 2014, 55, 8488-8496.	3.3	24
70	RNA-Seq: Improving Our Understanding of Retinal Biology and Disease. Cold Spring Harbor Perspectives in Medicine, 2015, 5, a017152.	6.2	23
71	Expression of Wild-Type Rp1 Protein in Rp1 Knock-in Mice Rescues the Retinal Degeneration Phenotype. PLoS ONE, 2012, 7, e43251.	2.5	22
72	Seeing the Light. Science Translational Medicine, 2013, 5, 175fs8.	12.4	22

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73	Course of Ocular Function inPRPF31Retinitis Pigmentosa. Seminars in Ophthalmology, 2016, 31, 49-52.	1.6	22
74	Characterizing variants of unknown significance in rhodopsin: A functional genomics approach. Human Mutation, 2019, 40, 1127-1144.	2.5	22
75	Knockdown ofttc26disrupts ciliogenesis of the photoreceptor cells and the pronephros in zebrafish. Molecular Biology of the Cell, 2012, 23, 3069-3078.	2.1	21
76	Ift172 conditional knock-out mice exhibit rapid retinal degeneration and protein trafficking defects. Human Molecular Genetics, 2018, 27, 2012-2024.	2.9	21
77	The Severity of Retinal Degeneration inRp1hGene-Targeted Mice Is Dependent on Genetic Background. , 2009, 50, 1566.		20
78	A Murine Rp1 Missense Mutation Causes Protein Mislocalization and Slowly Progressive Photoreceptor Degeneration. American Journal of Pathology, 2014, 184, 2721-2729.	3.8	18
79	Characterization of retinal inosine monophosphate dehydrogenase 1 in several mammalian species. Molecular Vision, 2007, 13, 1866-72.	1.1	17
80	Expanding the phenotypic spectrum in RDH12-associated retinal disease. Journal of Physical Education and Sports Management, 2020, 6, a004754.	1.2	16
81	Parthanatos as aÂCell Death Pathway Underlying Retinal Disease. Advances in Experimental Medicine and Biology, 2019, 1185, 323-327.	1.6	16
82	Gene Therapy Preserves Retinal Structure and Function in a Mouse Model of NMNAT1-Associated Retinal Degeneration. Molecular Therapy - Methods and Clinical Development, 2020, 18, 582-594.	4.1	15
83	Mutant Nmnat1 leads to a retina-specific decrease of NAD+ accompanied by increased poly(ADP-ribose) in a mouse model of NMNAT1-associated retinal degeneration. Human Molecular Genetics, 2021, 30, 644-657.	2.9	14
84	The importance of genetic testing as demonstrated by two cases of -associated retinal generation misdiagnosed as LCA. Molecular Vision, 2017, 23, 695-706.	1.1	13
85	A radiometric immunosorbent assay for the detection of anti-hormone-binding protein antibodies. Analytical Biochemistry, 1986, 153, 67-74.	2.4	12
86	A combined RNA-seq and whole genome sequencing approach for identification of non-coding pathogenic variants in single families. Human Molecular Genetics, 2020, 29, 967-979.	2.9	12
87	Ocular Toxoplasmosis: Pathogenesis, Diagnosis, and Management. Seminars in Ophthalmology, 1993, 8, 40-52.	1.6	9
88	Controversies in the Management of Retinopathy of Prematurity. International Ophthalmology Clinics, 1994, 34, 121-148.	0.7	9
89	Complement C5 is not critical for the formation of sub-RPE deposits in Efemp1 mutant mice. Scientific Reports, 2021, 11, 10416.	3.3	8
90	Broadening INPP5E phenotypic spectrum: detection of rare variants in syndromic and non-syndromic IRD. Npj Genomic Medicine, 2021, 6, 53.	3.8	8

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91	Reduced nuclear NAD+ drives DNA damage and subsequent immune activation in the retina. Human Molecular Genetics, 2022, 31, 1370-1388.	2.9	8
92	<scp>WDR34</scp>, a candidate gene for nonâ€syndromic rodâ€cone dystrophy. Clinical Genetics, 2021, 99, 298-302.	2.0	7
93	Paradigm Shifts in Ophthalmic Diagnostics. Transactions of the American Ophthalmological Society, 2016, 114, WP1.	1.4	7
94	Beyond Sector Retinitis Pigmentosa: Expanding the Phenotype and Natural History of the Rhodopsin Gene Codon 106 Mutation (Gly-to-Arg) in Autosomal Dominant Retinitis Pigmentosa. Genes, 2021, 12, 1853.	2.4	6
95	Detection of Large Structural Variants Causing Inherited Retinal Diseases. Advances in Experimental Medicine and Biology, 2019, 1185, 197-202.	1.6	4
96	Mitochondrial tRNA-serine (AGY) m.C12264T mutation causes severe multisystem disease with cataracts. Discovery Medicine, 2012, 13, 143-50.	0.5	4
97	Biallelic -associated retinal dystrophies: Expanding the mutational and clinical spectrum. Molecular Vision, 2020, 26, 423-433.	1.1	4
98	Novel RCBTB1 variants causing later-onset non-syndromic retinal dystrophy with macular chorioretinal atrophy. Ophthalmic Genetics, 2022, , 1-8.	1.2	2
99	Gene editing technology: Towards precision medicine in inherited retinal diseases. Seminars in Ophthalmology, 2021, 36, 176-184.	1.6	1
100	The RP1 Gene and Protein in Photoreceptor Biology. , 2004, , 223-257.		1
101	Retinitis pigmentosa and related disorders. , 2010, , 579-589.		0
102	RP1 Mutation Analysis. , 2001, , 55-61.		0
103	The 1,25-Dihydroxycholecalciferol Receptor. , 1987, , 319-337.		0
104	A Hidden Structural Variation in a Known IRD Gene: A Cautionary Tale of Two New Disease Candidate Genes. Journal of Physical Education and Sports Management, 2021, , mcs.a006131.	1.2	0