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	Reduced nuclear NAD+ drives DNA damage and subsequent immune activation in the retina. Human Molecular Genetics, 2022, 31, 1370-1388.	2.9	8
2	Novel RCBTB1 variants causing later-onset non-syndromic retinal dystrophy with macular chorioretinal atrophy. Ophthalmic Genetics, 2022, , 1-8.	1.2	2
3	<scp>WDR34</scp>, a candidate gene for nonâ€syndromic rodâ€cone dystrophy. Clinical Genetics, 2021, 99, 298-302.	2.0	7
4	Gene editing technology: Towards precision medicine in inherited retinal diseases. Seminars in Ophthalmology, 2021, 36, 176-184.	1.6	1
5	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
6	Complement C5 is not critical for the formation of sub-RPE deposits in Efemp1 mutant mice. Scientific Reports, 2021, 11, 10416.	3.3	8
7	Broadening INPP5E phenotypic spectrum: detection of rare variants in syndromic and non-syndromic IRD. Npj Genomic Medicine, 2021, 6, 53.	3.8	8
8	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
9	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
10	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
11	Advancing Clinical Trials for Inherited Retinal Diseases: Recommendations from the Second Monaciano Symposium. Translational Vision Science and Technology, 2020, 9, 2.	2.2	56
12	Mutations in the Kinesin-2 Motor KIF3B Cause an Autosomal-Dominant Ciliopathy. American Journal of Human Genetics, 2020, 106, 893-904.	6.2	29
13	Expanding the phenotypic spectrum in RDH12-associated retinal disease. Journal of Physical Education and Sports Management, 2020, 6, a004754.	1.2	16
14	Copy-number variation contributes 9% of pathogenicity in the inherited retinal degenerations. Genetics in Medicine, 2020, 22, 1079-1087.	2.4	59
15	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
16	Biallelic -associated retinal dystrophies: Expanding the mutational and clinical spectrum. Molecular Vision, 2020, 26, 423-433.	1.1	4
17	Contribution of noncoding pathogenic variants to RPGRIP1-mediated inherited retinal degeneration. Genetics in Medicine, 2019, 21, 694-704.	2.4	27
18	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

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19	Characterizing variants of unknown significance in rhodopsin: A functional genomics approach. Human Mutation, 2019, 40, 1127-1144.	2.5	22
20	In Vivo Assessment of Potential Therapeutic Approaches for USH2A-Associated Diseases. Advances in Experimental Medicine and Biology, 2019, 1185, 91-96.	1.6	26
21	Detection of Large Structural Variants Causing Inherited Retinal Diseases. Advances in Experimental Medicine and Biology, 2019, 1185, 197-202.	1.6	4
22	Parthanatos as aÂCell Death Pathway Underlying Retinal Disease. Advances in Experimental Medicine and Biology, 2019, 1185, 323-327.	1.6	16
23	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
24	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
25	Ift172 conditional knock-out mice exhibit rapid retinal degeneration and protein trafficking defects. Human Molecular Genetics, 2018, 27, 2012-2024.	2.9	21
26	Inherited Retinal Degenerations: Current Landscape and Knowledge Gaps. Translational Vision Science and Technology, 2018, 7, 6.	2.2	168
27	C3a triggers formation of sub-retinal pigment epithelium deposits via the ubiquitin proteasome pathway. Scientific Reports, 2018, 8, 9679.	3.3	42
28	USMG5 Ashkenazi Jewish founder mutation impairs mitochondrial complex V dimerization and ATP synthesis. Human Molecular Genetics, 2018, 27, 3305-3312.	2.9	45
29	Photoreceptor Cilia and Retinal Ciliopathies. Cold Spring Harbor Perspectives in Biology, 2017, 9, a028274.	5.5	154
30	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
31	The Genetic Basis of Pericentral Retinitis Pigmentosaâ€"A Form of Mild Retinitis Pigmentosa. Genes, 2017, 8, 256.	2.4	34
32	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
33	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
34	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
35	Isolation, culture and characterization of primary mouse RPE cells. Nature Protocols, 2016, 11, 1206-1218.	12.0	79
36	Course of Ocular Function inPRPF31Retinitis Pigmentosa. Seminars in Ophthalmology, 2016, 31, 49-52.	1.6	22

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37	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
38	Extracellular Matrix Alterations and Deposit Formation in AMD. Advances in Experimental Medicine and Biology, 2016, 854, 53-58.	1.6	43
39	Paradigm Shifts in Ophthalmic Diagnostics. Transactions of the American Ophthalmological Society, 2016, 114, WP1.	1.4	7
40	RNA-Seq: Improving Our Understanding of Retinal Biology and Disease. Cold Spring Harbor Perspectives in Medicine, 2015, 5, a017152.	6.2	23
41	The Status ofRPE65Gene Therapy Trials: Safety and Efficacy. Cold Spring Harbor Perspectives in Medicine, 2015, 5, a017285.	6.2	130
42	A local complement response by RPE causes early-stage macular degeneration. Human Molecular Genetics, 2015, 24, 5555-5569.	2.9	62
43	Mutations in IFT172 cause isolated retinal degeneration and Bardetâ€“Biedl syndrome. Human Molecular Genetics, 2015, 24, 230-242.	2.9	136
44	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
45	Targeted Exon Sequencing in Usher Syndrome Type I. Investigative Ophthalmology and Visual Science, 2014, 55, 8488-8496.	3.3	24
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	Exome-Based Mapping and Variant Prioritization for Inherited Mendelian Disorders. American Journal of Human Genetics, 2014, 94, 373-384.	6.2	37
48	Systemic Diseases Associated with Retinal Dystrophies. Seminars in Ophthalmology, 2014, 29, 319-328.	1.6	44
49	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
50	A Murine Rp1 Missense Mutation Causes Protein Mislocalization and Slowly Progressive Photoreceptor Degeneration. American Journal of Pathology, 2014, 184, 2721-2729.	3.8	18
51	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
52	Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2013, 93, 482-495.	6.2	138
53	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
54	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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55	Genetic Testing for Inherited Eye Disease. JAMA Ophthalmology, 2013, 131, 1265.	2.5	26
56	Seeing the Light. Science Translational Medicine, 2013, 5, 175fs8.	12.4	22
57	CRB1: One Gene, Many Phenotypes. Seminars in Ophthalmology, 2013, 28, 397-405.	1.6	37
58	NMNAT1 mutations cause Leber congenital amaurosis. Nature Genetics, 2012, 44, 1040-1045.	21.4	171
59	Knockdown ofttc26disrupts ciliogenesis of the photoreceptor cells and the pronephros in zebrafish. Molecular Biology of the Cell, 2012, 23, 3069-3078.	2.1	21
60	Expression of Wild-Type Rp1 Protein in Rp1 Knock-in Mice Rescues the Retinal Degeneration Phenotype. PLoS ONE, 2012, 7, e43251.	2.5	22
61	AAV2 Gene Therapy Readministration in Three Adults with Congenital Blindness. Science Translational Medicine, 2012, 4, 120ra15.	12.4	340
62	Mitochondrial tRNA-serine (AGY) m.C12264T mutation causes severe multisystem disease with cataracts. Discovery Medicine, 2012, 13, 143-50.	0.5	4
63	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
64	Comparative analysis of RNA-Seq alignment algorithms and the RNA-Seq unified mapper (RUM). Bioinformatics, 2011, 27, 2518-2528.	4.1	298
65	Generation ofCreTransgenic Mice with Postnatal RPE-Specific Ocular Expression. , 2011, 52, 1378.		85
66	Three Gene-Targeted Mouse Models of RNA Splicing Factor RP Show Late-Onset RPE and Retinal Degeneration. , 2011, 52, 190.		70
67	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
68	TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum. Nature Genetics, 2011, 43, 189-196.	21.4	326
69	Mutations in TSPAN12 Cause Autosomal-Dominant Familial Exudative Vitreoretinopathy. American Journal of Human Genetics, 2010, 86, 248-253.	6.2	161
70	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
71	Candidate exome capture identifies mutation of SDCCAG8 as the cause of a retinal-renal ciliopathy. Nature Genetics, 2010, 42, 840-850.	21.4	295
72	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

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73	Retinitis pigmentosa and related disorders. , 2010, , 579-589.		0
74	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
75	Photoreceptor Sensory Cilia and Inherited Retinal Degeneration. Advances in Experimental Medicine and Biology, 2010, 664, 223-232.	1.6	34
76	Targeted High-Throughput DNA Sequencing for Gene Discovery in Retinitis Pigmentosa. Advances in Experimental Medicine and Biology, 2010, 664, 325-331.	1.6	35
77	The Severity of Retinal Degeneration inRp1hGene-Targeted Mice Is Dependent on Genetic Background. , 2009, 50, 1566.		20
78	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
79	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
80	Decreased Levels of the RNA Splicing Factor Prpf3 in Mice and Zebrafish Do Not Cause Photoreceptor Degeneration. , 2008, 49, 3830.		31
81	Safety and Efficacy of Gene Transfer for Leber's Congenital Amaurosis. New England Journal of Medicine, 2008, 358, 2240-2248.	27.0	1,941
82	The R345W mutation in EFEMP1 is pathogenic and causes AMD-like deposits in mice. Human Molecular Genetics, 2007, 16, 2411-2422.	2.9	129
83	The Proteome of the Mouse Photoreceptor Sensory Cilium Complex. Molecular and Cellular Proteomics, 2007, 6, 1299-1317.	3.8	310
84	Characterization of retinal inosine monophosphate dehydrogenase 1 in several mammalian species. Molecular Vision, 2007, 13, 1866-72.	1.1	17
85	Analysis of Kinesin-2 Function in Photoreceptor Cells Using SynchronousCre-loxP Knockout ofKif3awithRHO-Cre. , 2006, 47, 5039.		68
86	Why Do Mutations in the Ubiquitously Expressed Housekeeping GeneIMPDH1Cause Retina-Specific Photoreceptor Degeneration?. , 2006, 47, 3754.		55
87	Expression and Activation of STAT3 in Ischemia-Induced Retinopathy. , 2005, 46, 4409.		31
88	The Retinitis Pigmentosa 1 Protein Is a Photoreceptor Microtubule-Associated Protein. Journal of Neuroscience, 2004, 24, 6427-6436.	3.6	140
89	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
90	The RP1 Gene and Protein in Photoreceptor Biology. , 2004, , 223-257.		1

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91	Retinopathy of Prematurity. Molecular Diagnosis and Therapy, 2003, 3, 261-277.	3.3	48
92	RP1 Is Required for the Correct Stacking of Outer Segment Discs. , 2003, 44, 4171.		122
93	Characterization of RP1L1, a highly polymorphic paralog of the retinitis pigmentosa 1 (RP1) gene. Molecular Vision, 2003, 9, 129-37.	1.1	26
94	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
95	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
96	Pathways to photoreceptor cell death in inherited retinal degenerations. BioEssays, 2001, 23, 605-618.	2.5	88
97	RP1 Mutation Analysis. , 2001, , 55-61.		0
98	Retinal expression, regulation, and functional bioactivity of prostacyclin-stimulating factor. Journal of Clinical Investigation, 2000, 106, 541-550.	8.2	53
99	Mutations in a gene encoding a new oxygen-regulated photoreceptor protein cause dominant retinitis pigmentosa. Nature Genetics, 1999, 22, 248-254.	21.4	174
100	Regulation of Vascular Endothelial Growth Factor by Oxygen in a Model of Retinopathy of Prematurity. JAMA Ophthalmology, 1996, 114, 1219.	2.4	435
101	Controversies in the Management of Retinopathy of Prematurity. International Ophthalmology Clinics, 1994, 34, 121-148.	0.7	9
102	Ocular Toxoplasmosis: Pathogenesis, Diagnosis, and Management. Seminars in Ophthalmology, 1993, 8, 40-52.	1.6	9
103	The 1,25-Dihydroxycholecalciferol Receptor. , 1987, , 319-337.		0
104	A radiometric immunosorbent assay for the detection of anti-hormone-binding protein antibodies. Analytical Biochemistry, 1986, 153, 67-74.	2.4	12