

# Kari E Branham

## List of Publications by Year in descending order

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Version: 2024-02-01

86  
papers

9,710  
citations

147801

31  
h-index

88630

70  
g-index

90  
all docs

90  
docs citations

90  
times ranked

19421  
citing authors

#	ARTICLE	IF	CITATIONS
1	Real-world outcomes of voretigene neparvovec treatment in pediatric patients with RPE65-associated Leber congenital amaurosis. <i>Graefe's Archive for Clinical and Experimental Ophthalmology</i> , 2022, 260, 1543-1550.	1.9	27
2	Tissue-specific genotype-phenotype correlations among USH2A-related disorders in the RUSH2A study. <i>Human Mutation</i> , 2022, 43, 613-624.	2.5	10
3	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
4	Adherence and satisfaction in Argus II prosthesis users: a self determination theory model. <i>Ophthalmic Genetics</i> , 2022, 43, 462-469.	1.2	0
5	The landscape of submicroscopic structural variants at the <i>OPN1LW/OPN1MW</i> gene cluster on Xq28 underlying blue cone monochromacy. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, .	7.1	2
6	Clinical trial design for neuroprotection in RHO autosomal dominant retinitis pigmentosa; outcome measure considerations. <i>Ophthalmic Genetics</i> , 2021, 42, 170-177.	1.2	2
7	A novel think tank program to promote innovation and strategic planning in ophthalmic surgery. <i>Perioperative Care and Operating Room Management</i> , 2021, 22, 100147.	0.3	1
8	Association of No-Cost Genetic Testing Program Implementation and Patient Characteristics With Access to Genetic Testing for Inherited Retinal Degenerations. <i>JAMA Ophthalmology</i> , 2021, 139, 449.	2.5	6
9	Characterization of the Spectrum of Ophthalmic Changes in Patients With Alagille Syndrome. , 2021, 62, 27.		11
10	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
11	Association of Smoking, Alcohol Consumption, Blood Pressure, Body Mass Index, and Glycemic Risk Factors With Age-Related Macular Degeneration. <i>JAMA Ophthalmology</i> , 2021, 139, 1299.	2.5	29
12	Deep intronic variants in <i>CNGB3</i> cause achromatopsia by pseudoexon activation. <i>Human Mutation</i> , 2020, 41, 255-264.	2.5	26
13	Advancing Clinical Trials for Inherited Retinal Diseases: Recommendations from the Second Monaciano Symposium. <i>Translational Vision Science and Technology</i> , 2020, 9, 2.	2.2	56
14	Genetic testing for inherited retinal degenerations: Triumphs and tribulations. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2020, 184, 571-577.	1.6	10
15	Surgical outcomes of Glaucoma associated with Axenfeld-Rieger syndrome. <i>BMC Ophthalmology</i> , 2020, 20, 172.	1.4	21
16	Family-based exome sequencing identifies rare coding variants in age-related macular degeneration. <i>Human Molecular Genetics</i> , 2020, 29, 2022-2034.	2.9	26
17	Contribution of noncoding pathogenic variants to RGRIP1-mediated inherited retinal degeneration. <i>Genetics in Medicine</i> , 2019, 21, 694-704.	2.4	27
18	Peripheral Pigmented Retinal Lesions in Stargardt Disease. <i>American Journal of Ophthalmology</i> , 2018, 188, 104-110.	3.3	12

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19	Identification of Novel Deletions as the Underlying Cause of Retinal Degeneration in Two Pedigrees. <i>Advances in Experimental Medicine and Biology</i> , 2018, 1074, 229-236.	1.6	2
20	Molecular Findings in Families with an Initial Diagnose of Autosomal Dominant Retinitis Pigmentosa (adRP). <i>Advances in Experimental Medicine and Biology</i> , 2018, 1074, 237-245.	1.6	7
21	Double hyperautofluorescent ring on fundus autofluorescence in <i>ABCA4</i> . <i>Ophthalmic Genetics</i> , 2018, 39, 87-91.	1.2	2
22	Reliability of kinetic visual field testing in children with mutation-proven retinal dystrophies: Implications for therapeutic clinical trials. <i>Ophthalmic Genetics</i> , 2018, 39, 22-28.	1.2	8
23	Mutations in the gene <i>PDE6C</i> encoding the catalytic subunit of the cone photoreceptor phosphodiesterase in patients with achromatopsia. <i>Human Mutation</i> , 2018, 39, 1366-1371.	2.5	18
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	<i>ABCA4</i> . , 2018, , 1-5.		0
26	<i>CLN3</i> . , 2018, , 59-60.		0
27	<i>CNGA3</i> . , 2018, , 65-66.		0
28	<i>CNGB3</i> . , 2018, , 71-74.		0
29	<i>KLHL7</i> . , 2018, , 129-131.		0
30	<i>RP2</i> . , 2018, , 229-231.		0
31	<i>RPGR</i> . , 2018, , 237-242.		1
32	<i>SAG</i> . , 2018, , 251-251.		3
33	Prevalence of Antiretinal Antibodies in Acute Zonal Occult Outer Retinopathy: A Comprehensive Review of 25 Cases. <i>American Journal of Ophthalmology</i> , 2017, 176, 210-218.	3.3	29
34	Cystoid macular changes on optical coherence tomography in a patient with maternally inherited diabetes and deafness (MIDD)-associated macular dystrophy. <i>Ophthalmic Genetics</i> , 2017, 38, 467-472.	1.2	9
35	Genetic analysis of 10 pedigrees with inherited retinal degeneration by exome sequencing and phenotype-genotype association. <i>Physiological Genomics</i> , 2017, 49, 216-229.	2.3	23
36	Peripheral Visual Fields in <i>ABCA4</i> Stargardt Disease and Correlation With Disease Extent on Ultra-widefield Fundus Autofluorescence. <i>American Journal of Ophthalmology</i> , 2017, 184, 181-188.	3.3	12

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37	<i>C2orf71</i> Mutations as a Frequent Cause of Autosomal-Recessive Retinitis Pigmentosa: Clinical Analysis and Presentation of 8 Novel Mutations. , 2017, 58, 3840.		13
38	A Novel Dominant Mutation in <i>SAG</i>, the Arrestin-1 Gene, Is a Common Cause of Retinitis Pigmentosa in Hispanic Families in the Southwestern United States. , 2017, 58, 2774.		31
39	Expansion of Severely Constricted Visual Field Using Google Glass. Ophthalmic Surgery Lasers and Imaging Retina, 2016, 47, 486-489.	0.7	16
40	Establishing the involvement of the novel gene<i>AGBL5</i>in retinitis pigmentosa by whole genome sequencing. Physiological Genomics, 2016, 48, 922-927.	2.3	29
41	A reference panel of 64,976 haplotypes for genotype imputation. Nature Genetics, 2016, 48, 1279-1283.	21.4	2,421
42	Worldwide Argus II implantation: recommendations to optimize patient outcomes. BMC Ophthalmology, 2016, 16, 52.	1.4	39
43	A large genome-wide association study of age-related macular degeneration highlights contributions of rare and common variants. Nature Genetics, 2016, 48, 134-143.	21.4	1,167
44	Multimodal Imaging in Wagner Syndrome. Ophthalmic Surgery Lasers and Imaging Retina, 2016, 47, 574-579.	0.7	9
45	Differential DNA methylation identified in the blood and retina of AMD patients. Epigenetics, 2015, 10, 698-707.	2.7	62
46	Advancing Therapeutic Strategies for Inherited Retinal Degeneration: Recommendations From the Monaciano Symposium. Investigative Ophthalmology and Visual Science, 2015, 56, 918-931.	3.3	92
47	Genome-wide association study and meta-analysis of intraocular pressure. Human Genetics, 2014, 133, 41-57.	3.8	93
48	The Ophthalmic Experience: Unanticipated Primary Findings in the Era of Next Generation Sequencing. Journal of Genetic Counseling, 2014, 23, 588-593.	1.6	5
49	Ancestry estimation and control of population stratification for sequence-based association studies. Nature Genetics, 2014, 46, 409-415.	21.4	136
50	Next generation sequencing-based molecular diagnosis of retinitis pigmentosa: identification of a novel genotype-phenotype correlation and clinical refinements. Human Genetics, 2014, 133, 331-345.	3.8	204
51	Expression of Thyrotropin Receptor, Thyroglobulin, Sodium-Iodide Symporter, and Thyroperoxidase by Fibrocytes Depends on AIRE. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E1236-E1244.	3.6	52
52	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
53	Hypomethylation of the IL17RC Promoter in Peripheral Blood Leukocytes Is Not A Hallmark of Age-Related Macular Degeneration. Cell Reports, 2013, 5, 1527-1535.	6.4	42
54	Diagnostic Fundus Autofluorescence Patterns in Achromatopsia. American Journal of Ophthalmology, 2013, 156, 1211-1219.e2.	3.3	38

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55	Clinical Phenotypes and Prognostic Full-Field Electroretinographic Findings in Stargardt Disease. <i>American Journal of Ophthalmology</i> , 2013, 155, 465-473.e3.	3.3	39
56	Mutations in the X-Linked Retinitis Pigmentosa Genes <i>RPGR</i> and <i>RP2</i> Found in 8.5% of Families with a Provisional Diagnosis of Autosomal Dominant Retinitis Pigmentosa. , 2013, 54, 1411.		113
57	Seven new loci associated with age-related macular degeneration. <i>Nature Genetics</i> , 2013, 45, 433-439.	21.4	687
58	Identification of a rare coding variant in complement 3 associated with age-related macular degeneration. <i>Nature Genetics</i> , 2013, 45, 1375-1379.	21.4	158
59	Providing comprehensive genetic-based ophthalmic care. <i>Clinical Genetics</i> , 2013, 84, 183-189.	2.0	12
60	Screening of a Large Cohort of Leber Congenital Amaurosis and Retinitis Pigmentosa Patients Identifies Novel <i>LCA5</i> Mutations and New Genotype-Phenotype Correlations. <i>Human Mutation</i> , 2013, 34, 1537-1546.	2.5	32
61	Phenotypic Conservation in Patients With X-Linked Retinitis Pigmentosa Caused by <i>RPGR</i> Mutations. <i>JAMA Ophthalmology</i> , 2013, 131, 1016.	2.5	31
62	Mutations in the small nuclear riboprotein 200 kDa gene ( <i>SNRNP200</i> ) cause 1.6% of autosomal dominant retinitis pigmentosa. <i>Molecular Vision</i> , 2013, 19, 2407-17.	1.1	20
63	Complement factor H genetic variant and age-related macular degeneration: effect size, modifiers and relationship to disease subtype. <i>International Journal of Epidemiology</i> , 2012, 41, 250-262.	1.9	79
64	Establishing baseline rod electroretinogram values in achromatopsia and cone dystrophy. <i>Documenta Ophthalmologica</i> , 2012, 125, 229-233.	2.2	13
65	Next-generation genetic testing for retinitis pigmentosa. <i>Human Mutation</i> , 2012, 33, 963-972.	2.5	258
66	Mutations in <i>RPGR</i> and <i>RP2</i> Account for 15% of Males with Simplex Retinal Degenerative Disease. , 2012, 53, 8232.		108
67	Complement Factor D in Age-Related Macular Degeneration. , 2011, 52, 8828.		92
68	Long-term follow-up of a family with dominant X-linked retinitis pigmentosa. <i>Eye</i> , 2010, 24, 764-774.	2.1	27
69	Loss of lysophosphatidylcholine acyltransferase 1 leads to photoreceptor degeneration in <i>rd11</i> mice. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2010, 107, 15523-15528.	7.1	55
70	E2-2 Protein and Fuchs's Corneal Dystrophy. <i>New England Journal of Medicine</i> , 2010, 363, 1016-1024.	27.0	247
71	Peripapillary Dark Choroid Ring as a Helpful Diagnostic Sign in Advanced Stargardt Disease. <i>American Journal of Ophthalmology</i> , 2010, 149, 656-660.e2.	3.3	25
72	Genetic variants near <i>TIMP3</i> and high-density lipoprotein-associated loci influence susceptibility to age-related macular degeneration. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2010, 107, 7401-7406.	7.1	475

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73	Mutations in a BTB-Kelch Protein, KLHL7, Cause Autosomal-Dominant Retinitis Pigmentosa. American Journal of Human Genetics, 2009, 84, 792-800.	6.2	89
74	Toll-like Receptor Polymorphisms and Age-Related Macular Degeneration. , 2008, 49, 1652.		79
75	Retinal Phenotype of an X-Linked Pseudo-usher Syndrome in Association with the G173R Mutation in the RPGR Gene. Advances in Experimental Medicine and Biology, 2008, 613, 221-227.	1.6	4
76	Molecular Testing for Hereditary Retinal Disease as Part of Clinical Care. JAMA Ophthalmology, 2007, 125, 252.	2.4	37
77	A variant of mitochondrial protein LOC387715/ARMS2, not HTRA1, is strongly associated with age-related macular degeneration. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 16227-16232.	7.1	398
78	High-Resolution Imaging with Adaptive Optics in Patients with Inherited Retinal Degeneration. , 2007, 48, 3283.		241
79	Inner Retinal Abnormalities in X-linked Retinitis Pigmentosa with <i>RPGR</i> Mutations. , 2007, 48, 4759.		107
80	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
81	CFH haplotypes without the Y402H coding variant show strong association with susceptibility to age-related macular degeneration. Nature Genetics, 2006, 38, 1049-1054.	21.4	318
82	Strong Association of the Y402H Variant in Complement Factor H at 1q32 with Susceptibility to Age-Related Macular Degeneration. American Journal of Human Genetics, 2005, 77, 149-153.	6.2	327
83	Toll-like receptor 4 variant D299G is associated with susceptibility to age-related macular degeneration. Human Molecular Genetics, 2005, 14, 1449-1455.	2.9	177
84	Biomarkers of Cardiovascular Disease as Risk Factors for Age-Related Macular Degeneration. Ophthalmology, 2005, 112, 2076-2080.	5.2	143
85	Association of Apolipoprotein E Alleles with Susceptibility to Age-Related Macular Degeneration in a Large Cohort from a Single Center. Investigative Ophthalmology and Visual Science, 2004, 45, 1306-1310.	3.3	129
86	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