Kaoru Fujinami

List of Publications by Year
in descending order

76 papers	2,365 citations	279798 23 h-index	265206 42 g-index
80 all docs	80 docs citations	80 times ranked	1709 citing authors

#	Article	IF	Citations
1	Longitudinal Changes of Fixation Stability and Location Within 24 Months in Stargardt Disease: ProgStar Report No. 16. American Journal of Ophthalmology, 2022, 233, 78-89.	3.3	5
2	Clinical course of a Japanese girl with Leber congenital amaurosis associated with a novel nonsense pathogenic variant in NMNAT1: a case report and mini review. Ophthalmic Genetics, 2022, , 1-9.	1.2	1
3	Visual Field Characteristics in East Asian Patients With Occult Macular Dystrophy (Miyake Disease): EAOMD Report No. 3. , 2022, 63, 12.		3
4	Stargardt Macular Dystrophy. , 2022, , 151-168.		1
5	X-Linked Retinoschisis. Ophthalmology, 2022, 129, 542-551.	5.2	19
6	A recurrent variant in LIM2 causes an isolated congenital sutural/lamellar cataract in a Japanese family. Ophthalmic Genetics, 2022, 43, 622-626.	1.2	4
7	Sector Retinitis Pigmentosa: Extending the Molecular Genetics Basis and Elucidating the Natural History. American Journal of Ophthalmology, 2021, 221, 299-310.	3.3	20
8	Spatial Functional Characteristics of East Asian Patients With Occult Macular Dystrophy (Miyake) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	3.3	10
9	New variants and in silico analyses in GRK1 associated Oguchi disease. Human Mutation, 2021, 42, 164-176.	2.5	7
10	Genotype and Long-term Clinical Course of Bietti Crystalline Dystrophy in Korean and Japanese Patients. Ophthalmology Retina, 2021, 5, 1269-1279.	2.4	6
11	Inherited retinal diseases: Therapeutics, clinical trials and end pointsâ€"A review. Clinical and Experimental Ophthalmology, 2021, 49, 270-288.	2.6	68
12	KCNV2-Associated Retinopathy: Detailed Retinal Phenotype and Structural Endpointsâ€"KCNV2 Study Group Report 2. American Journal of Ophthalmology, 2021, 230, 1-11.	3.3	11
13	Prediction of causative genes in inherited retinal disorder from fundus photography and autofluorescence imaging using deep learning techniques. British Journal of Ophthalmology, 2021, 105, 1272-1279.	3.9	12
14	The Progression of Stargardt Disease Using Volumetric Hill of Vision Analyses Over 24 Months: ProgStar Report No.15. American Journal of Ophthalmology, 2021, 230, 123-133.	3.3	10
15	KCNV2-Associated Retinopathy: Genetics, Electrophysiology, and Clinical Courseâ€"KCNV2 Study Group Report 1. American Journal of Ophthalmology, 2021, 225, 95-107.	3.3	17
16	Machine Learning Algorithms to Detect Subclinical Keratoconus: Systematic Review. JMIR Medical Informatics, 2021, 9, e27363.	2.6	12
17	Long-term follow-up of a Chinese patient with KCNV2-retinopathy. Ophthalmic Genetics, 2021, 42, 144-149.	1.2	0
18	Genetic and Phenotypic Landscape of PRPH2-Associated Retinal Dystrophy in Japan. Genes, 2021, 12, 1817.	2.4	9

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19	A Japanese boy with double diagnoses of 2p15p16.1 microdeletion syndrome and RP2-associated retinal disorder. Human Genome Variation, 2021, 8, 46.	0.7	0
20	GUCY2D-Associated Leber Congenital Amaurosis: A Retrospective Natural History Study in Preparation for Trials of Novel Therapies. American Journal of Ophthalmology, 2020, 210, 59-70.	3.3	39
21	Prospective Cohort Study of Childhood-Onset Stargardt Disease: Fundus Autofluorescence Imaging, Progression, Comparison with Adult-Onset Disease, and Disease Symmetry. American Journal of Ophthalmology, 2020, 211, 159-175.	3.3	41
22	Retinal imaging in inherited retinal diseases. Annals of Eye Science, 2020, 5, 25-25.	2.1	17
23	Clinical and genetic characteristics of 10 Japanese patients with PROM1 â€associated retinal disorder: A report of the phenotype spectrum and a literature review in the Japanese population. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 656-674.	1.6	21
24	RP2 â€associated retinal disorder in a Japanese cohort: Report of novel variants and a literature review, identifying a genotypeâ€"phenotype association. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 675-693.	1.6	5
25	Clinical and genetic characteristics of Stargardt disease in a large Western China cohort: Report 1. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 694-707.	1.6	7
26	Resolving the dark matter of ABCA4 for 1054 Stargardt disease probands through integrated genomics and transcriptomics. Genetics in Medicine, 2020, 22, 1235-1246.	2.4	92
27	Clinical and Genetic Characteristics of 18 Patients from 13 Japanese Families with CRX-associated retinal disorder: Identification of Genotype-phenotype Association. Scientific Reports, 2020, 10, 9531.	3.3	24
28	Clinical and Genetic Characteristics of 15 Affected Patients From 12 Japanese Families with GUCY2D-Associated Retinal Disorder. Translational Vision Science and Technology, 2020, 9, 2.	2.2	15
29	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.	3.3	20
30	Genetic Spectrum of EYS-associated Retinal Disease in a Large Japanese Cohort: Identification of Disease-associated Variants with Relatively High Allele Frequency. Scientific Reports, 2020, 10, 5497.	3.3	21
31	Inherited cataracts: molecular genetics, clinical features, disease mechanisms and novel therapeutic approaches. British Journal of Ophthalmology, 2020, 104, 1331-1337.	3.9	49
32	RDH5-Related Fundus Albipunctatus in a Large Japanese Cohort. , 2020, 61, 53.		20
33	Phenogenon: Gene to phenotype associations for rare genetic diseases. PLoS ONE, 2020, 15, e0230587.	2.5	6
34	Detailed genetic characteristics of an international large cohort of patients with Stargardt disease: ProgStar study report 8. British Journal of Ophthalmology, 2019, 103, 390-397.	3.9	45
35	Phenotypical Characteristics of POC1B-Associated Retinopathy in Japanese Cohort: Cone Dystrophy With Normal Funduscopic Appearance. , 2019, 60, 3432.		18
36	Clinical and Genetic Characteristics of East Asian Patients with Occult Macular Dystrophy (Miyake) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	5.2	28

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37	Clinical Stages of Occult Macular Dystrophy Based on Optical Coherence Tomographic Findings. , 2019, 60, 4691.		25
38	Clinical and genetic characteristics of 14 patients from 13 Japanese families with RPGR-associated retinal disorder: report of eight novel variants. Human Genome Variation, 2019, 6, 34.	0.7	15
39	Prediction of Causative Genes in Inherited Retinal Disorders from Spectral-Domain Optical Coherence Tomography Utilizing Deep Learning Techniques. Journal of Ophthalmology, 2019, 2019, 1-7.	1.3	32
40	Cross-Sectional and Longitudinal Assessment of the Ellipsoid Zone in Childhood-Onset Stargardt Disease. Translational Vision Science and Technology, 2019, 8, 1.	2.2	40
41	Novel mutations in the RS1 gene in Japanese patients with X-linked congenital retinoschisis. Human Genome Variation, 2019, 6, 3.	0.7	18
42	Characterization of GUCA1A-associated dominant cone/cone-rod dystrophy: low prevalence among Japanese patients with inherited retinal dystrophies. Scientific Reports, 2019, 9, 16851.	3.3	7
43	Stargardt Disease in Asian Population. Essentials in Ophthalmology, 2019, , 279-295.	0.1	2
44	Autosomal dominant optic atrophy with gene mutations accompanied by auditory neuropathy and other systemic complications in a Japanese cohort. Molecular Vision, 2019, 25, 559-573.	1.1	5
45	Case of cone dystrophy with normal fundus appearance associated with biallelic POC1B variants. Ophthalmic Genetics, 2018, 39, 255-262.	1.2	17
46	Early Patterns of Macular Degeneration in ABCA4-Associated Retinopathy. Ophthalmology, 2018, 125, 735-746.	5.2	55
47	Missense variants in the X-linked genePRPS1cause retinal degeneration in females. Human Mutation, 2018, 39, 80-91.	2.5	23
48	A Cross-Sectional and Longitudinal Study of Retinal Sensitivity inRPE65-Associated Leber Congenital Amaurosis. , 2018, 59, 3330.		19
49	Retinal findings in a patient with mutations in ABCC6 and ABCA4. Eye, 2018, 32, 1542-1543.	2.1	5
50	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.	3.3	24
51	Visual Acuity Change Over 24 Months and Its Association With Foveal Phenotype and Genotype in Individuals With Stargardt Disease. JAMA Ophthalmology, 2018, 136, 920.	2.5	44
52	Stargardt disease: clinical features, molecular genetics, animal models and therapeutic options. British Journal of Ophthalmology, 2017, 101, 25-30.	3.9	265
53	Parafoveal Photoreceptor Abnormalities in Asymptomatic Patients With RP1L1 Mutations in Families With Occult Macular Dystrophy. , 2017, 58, 6020.		21
54	One-year outcome of intravitreal aflibercept injection for age-related macular degeneration resistant to ranibizumab: rapid morphologic recovery and subsequent visual improvement. Clinical Ophthalmology, 2016, 10, 969.	1.8	13

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55	Phenotype and Progression of Retinal Degeneration Associated With Nullizigosity of ABCA4. , 2016, 57, 4668.		40
56	The Effect on Retinal Structure and Function of 15 Specific ABCA4 Mutations: A Detailed Examination of 82 Hemizygous Patients. , 2016, 57, 5963.		41
57	NovelRP1L1Variants and Genotypeâ€"Photoreceptor Microstructural Phenotype Associations in Cohort of Japanese Patients With Occult Macular Dystrophy. , 2016, 57, 4837.		54
58	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
59	Clinical and Genetic Findings of Autosomal Recessive Bestrophinopathy in Japanese Cohort. American Journal of Ophthalmology, 2016, 168, 86-94.	3.3	33
60	Internal Limiting Membrane Peeling to Prevent Post-vitrectomy Epiretinal Membrane Development in Retinal Detachment. American Journal of Ophthalmology, 2016, 171, 1-10.	3.3	53
61	Association of Retinal Artery and Other Inner Retinal Structures With Distribution of Tapetal-like Reflex in Oguchi's Disease. , 2015, 56, 2162.		5
62	Macular Hole Associated with Vogt-Koyanagi-Harada Disease at the Acute Uveitic Stage. Case Reports in Ophthalmology, 2015, 6, 328-332.	0.7	10
63	Clinical and Molecular Characteristics ofÂChildhood-Onset Stargardt Disease. Ophthalmology, 2015, 122, 326-334.	5.2	146
64	Clinical course of focal choroidal excavation in Vogt–Koyanagi–Harada disease. Clinical Ophthalmology, 2014, 8, 2461.	1.8	18
65	Longitudinal followâ€up of siblings with a discordant Stargardt disease phenotype. Acta Ophthalmologica, 2014, 92, e331-2.	1.1	9
66	Fine central macular dots associated with childhoodâ€onset Stargardt Disease. Acta Ophthalmologica, 2014, 92, e157-9.	1.1	17
67	Haptic Breakage after Transscleral Fixation of a Single-Piece Acrylic Intraocular Lens. Case Reports in Ophthalmology, 2014, 5, 212-216.	0.7	5
68	The Clinical Effect of Homozygous ABCA4 Alleles in 18 Patients. Ophthalmology, 2013, 120, 2324-2331.	5.2	56
69	Clinical and Molecular Analysis of Stargardt Disease With Preserved Foveal Structure and Function. American Journal of Ophthalmology, 2013, 156, 487-501.e1.	3.3	100
70	A Longitudinal Study of Stargardt Disease: Clinical and Electrophysiologic Assessment, Progression, and Genotype Correlations. American Journal of Ophthalmology, 2013, 155, 1075-1088.e13.	3.3	121
71	ABCA4Gene Screening by Next-Generation Sequencing in a British Cohort. , 2013, 54, 6662.		47
72	A Longitudinal Study of Stargardt Disease: Quantitative Assessment of Fundus Autofluorescence, Progression, and Genotype Correlations. , 2013, 54, 8181.		119

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73	Molecular characteristics of four Japanese cases with KCNV2 retinopathy: report of novel disease-causing variants. Molecular Vision, 2013, 19, 1580-90.	1.1	14
74	CLINICAL CHARACTERISTICS OF OCCULT MACULAR DYSTROPHY IN FAMILY WITH MUTATION OF RP1L1 GENE. Retina, 2012, 32, 1135-1147.	1.7	48
75	Stargardt Disease with Preserved Central Vision: identification of a putative novel mutation in ATPâ€binding cassette transporter gene. Acta Ophthalmologica, 2011, 89, e297-8.	1.1	14
76	Fundus Autofluorescence in Autosomal Dominant Occult Macular Dystrophy. JAMA Ophthalmology, 2011, 129, 597.	2.4	38