

Takaaki Hayashi

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

106
papers

1,329
citations

20
h-index

31
g-index

110
ext. papers

1,529
ext. citations

3.1
avg, IF

4.2
L-index

#	Paper	IF	Citations
106	Polypoidal choroidal vasculopathy in a patient with DMPK-associated myotonic dystrophy.. <i>Documenta Ophthalmologica</i> , 2022 , 1	2.2	
105	The second Japanese family with Malattia Leventinese/Doyme honeycomb retinal dystrophy. <i>Documenta Ophthalmologica</i> , 2021 , 1	2.2	1
104	Clinical findings in eyes with BEST1-related retinopathy complicated by choroidal neovascularization. <i>Graefes Archive for Clinical and Experimental Ophthalmology</i> , 2021 , 1	3.8	3
103	Transient electroretinographic abnormalities that mimic those of KCNV2 retinopathy: a case report. <i>Documenta Ophthalmologica</i> , 2021 , 143, 221-228	2.2	
102	Complete congenital stationary night blindness associated with a novel variant (p.Asn216Lys) in middle-aged and older adult patients. <i>Ophthalmic Genetics</i> , 2021 , 42, 412-419	1.2	3
101	The first Japanese family of CDH3-related hypotrichosis with juvenile macular dystrophy. <i>Molecular Genetics & Genomic Medicine</i> , 2021 , 9, e1688	2.3	1
100	KCNV2-Associated Retinopathy: Genetics, Electrophysiology, and Clinical Course-KCNV2 Study Group Report 1. <i>American Journal of Ophthalmology</i> , 2021 , 225, 95-107	4.9	7
99	Genotype-Phenotype Correlations in -Associated Retinal Dystrophies: A Multi-Center Cohort Study in JAPAN. <i>Journal of Clinical Medicine</i> , 2021 , 10,	5.1	2
98	Homozygous single nucleotide duplication of SLC38A8 in autosomal recessive foveal hypoplasia: The first Japanese case report. <i>Documenta Ophthalmologica</i> , 2021 , 143, 323-330	2.2	0
97	Vision Improvement after Osimertinib Treatment in Paraneoplastic Optic Neuropathy Associated with Lung Adenocarcinoma. <i>Case Reports in Ophthalmological Medicine</i> , 2021 , 2021, 2832021	0.7	
96	Progress of macular atrophy during 30 months follow-up in a patient with spinocerebellar ataxia type1 (SCA1). <i>Documenta Ophthalmologica</i> , 2021 , 142, 87-98	2.2	2
95	New variants and in silico analyses in GRK1 associated Oguchi disease. <i>Human Mutation</i> , 2021 , 42, 164-176	4.7	3
94	Novel biallelic TRPM1 variants in an elderly patient with complete congenital stationary night blindness. <i>Documenta Ophthalmologica</i> , 2021 , 142, 265-273	2.2	5
93	Genotype and Long-term Clinical Course of Bietti Crystalline Dystrophy in Korean and Japanese Patients. <i>Ophthalmology Retina</i> , 2021 , 5, 1269-1279	3.8	2
92	Vitamin A deficiency after prolonged intake of an unbalanced diet in a Japanese hemodialysis patient. <i>Documenta Ophthalmologica</i> , 2021 , 143, 85-91	2.2	1
91	A new PDE6A missense variant p.Arg544Gln in rod-cone dystrophy. <i>Documenta Ophthalmologica</i> , 2021 , 143, 107-114	2.2	2
90	Unique and progressive retinal degeneration in a patient with cancer associated retinopathy. <i>American Journal of Ophthalmology Case Reports</i> , 2020 , 20, 100908	1.3	2

89	Novel use of Finesse Flex loop for macular hole retinal detachment. <i>American Journal of Ophthalmology Case Reports</i> , 2020 , 18, 100703	1.3	1
88	Clinical and Genetic Characteristics of 18 Patients from 13 Japanese Families with CRX-associated retinal disorder: Identification of Genotype-phenotype Association. <i>Scientific Reports</i> , 2020 , 10, 9531	4.9	6
87	Novel biallelic splice-site BBS1 variants in Bardet-Biedle syndrome: a case report of the first Japanese patient. <i>Documenta Ophthalmologica</i> , 2020 , 141, 77-88	2.2	3
86	Clinical Course and Electron Microscopic Findings in Lymphocytes of Patients with -Associated Retinopathy. <i>International Journal of Molecular Sciences</i> , 2020 , 21,	6.3	1
85	Heterozygous GGC repeat expansion of in a patient with neuronal intranuclear inclusion disease and progressive retinal dystrophy. <i>Ophthalmic Genetics</i> , 2020 , 41, 93-95	1.2	9
84	Improvement of reduced electroretinographic responses in thymoma-associated retinopathy: a case report and literature review. <i>Documenta Ophthalmologica</i> , 2020 , 141, 195-204	2.2	2
83	Genetic Spectrum of EYS-associated Retinal Disease in a Large Japanese Cohort: Identification of Disease-associated Variants with Relatively High Allele Frequency. <i>Scientific Reports</i> , 2020 , 10, 5497	4.9	10
82	Long-term observation of a Japanese mucopolidosis IV patient with a novel homozygous p.F313del variant of MCOLN1. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 1500-1505	2.5	6
81	Electroretinographic abnormalities associated with pregabalin: a case report. <i>Documenta Ophthalmologica</i> , 2020 , 140, 279-287	2.2	12
80	Electroretinograms of eyes with Stickler syndrome. <i>Documenta Ophthalmologica</i> , 2020 , 140, 233-243	2.2	5
79	Genetic defects of CHM and visual acuity outcome in 24 choroideremia patients from 16 Japanese families. <i>Scientific Reports</i> , 2020 , 10, 15883	4.9	3
78	Clinical and genetic characteristics of 10 Japanese patients with PROM1-associated retinal disorder: A report of the phenotype spectrum and a literature review in the Japanese population. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2020 , 184, 656-674	3.1	8
77	Coexistence of GNAT1 and ABCA4 variants associated with Nougaret-type congenital stationary night blindness and childhood-onset cone-rod dystrophy. <i>Documenta Ophthalmologica</i> , 2020 , 140, 147-157	2.2	10
76	RDH5-Related Fundus Albipunctatus in a Large Japanese Cohort 2020 , 61, 53		13
75	Novel homozygous CLN3 missense variant in isolated retinal dystrophy: A case report and electron microscopic findings. <i>Molecular Genetics & Genomic Medicine</i> , 2020 , 8, e1308	2.3	12
74	CLINICAL COURSE OF PARANEOPlastic RETINOPATHY WITH ANTI-TRPM1 AUTOANTIBODY IN JAPANESE COHORT. <i>Retina</i> , 2019 , 39, 2410-2418	3.6	11
73	Novel biallelic loss-of-function KCNV2 variants in cone dystrophy with supernormal rod responses. <i>Documenta Ophthalmologica</i> , 2019 , 138, 229-239	2.2	13
72	X-linked Retinitis Pigmentosa in Japan: Clinical and Genetic Findings in Male Patients and Female Carriers. <i>International Journal of Molecular Sciences</i> , 2019 , 20,	6.3	14

71	Spontaneous Improvement of Visual Acuity in a 13-Year-Old Boy with Neuromyelitis Optica Spectrum Disorder. <i>Neuro-Ophthalmology</i> , 2019 , 43, 114-119	0.9	
70	Phenotypical Characteristics of POC1B-Associated Retinopathy in Japanese Cohort: Cone Dystrophy With Normal Fundusoscopic Appearance 2019 , 60, 3432-3446		11
69	Clinical Stages of Occult Macular Dystrophy Based on Optical Coherence Tomographic Findings 2019 , 60, 4691-4700		16
68	Autosomal dominant optic atrophy with gene mutations accompanied by auditory neuropathy and other systemic complications in a Japanese cohort. <i>Molecular Vision</i> , 2019 , 25, 559-573	2.3	3
67	High-Resolution Retinal Imaging Reveals Preserved Cone Photoreceptor Density and Choroidal Thickness in Female Carriers of Choroideremia. <i>Ophthalmic Surgery Lasers and Imaging Retina</i> , 2019 , 50, 76-85	1.4	2
66	Characterization of GUCA1A-associated dominant cone/cone-rod dystrophy: low prevalence among Japanese patients with inherited retinal dystrophies. <i>Scientific Reports</i> , 2019 , 9, 16851	4.9	5
65	Clinical findings of end-stage retinitis pigmentosa with a homozygous variant (p.R653X). <i>American Journal of Ophthalmology Case Reports</i> , 2019 , 13, 110-115	1.3	4
64	Autosomal dominant retinitis pigmentosa with macular involvement associated with a disease haplotype that included a novel PRPH2 variant (p.Cys250Gly). <i>Ophthalmic Genetics</i> , 2018 , 39, 357-365	1.2	5
63	Neuronal intranuclear hyaline inclusion disease presenting with childhood-onset night blindness associated with progressive retinal dystrophy. <i>Journal of the Neurological Sciences</i> , 2018 , 388, 84-86	3.2	7
62	Reconstruction of Photoreceptor Outer Layers after Steroid Therapy in Solar Retinopathy. <i>Case Reports in Ophthalmological Medicine</i> , 2018 , 2018, 7850467	0.7	2
61	Genotype determination of the OPN1LW/OPN1MW genes: novel disease-causing mechanisms in Japanese patients with blue cone monochromacy. <i>Scientific Reports</i> , 2018 , 8, 11507	4.9	5
60	Molecular Diagnosis of 34 Japanese Families with Leber Congenital Amaurosis Using Targeted Next Generation Sequencing. <i>Scientific Reports</i> , 2018 , 8, 8279	4.9	21
59	Early onset flecked retinal dystrophy associated with new compound heterozygous variants. <i>Molecular Vision</i> , 2018 , 24, 286-296	2.3	11
58	Compound heterozygous splice site variants in the SCLT1 gene highlight an additional candidate locus for Senior-Lkjen syndrome. <i>Scientific Reports</i> , 2018 , 8, 16733	4.9	6
57	A novel homozygous CYP4V2 variant (p.S121Y) associated with a choroideremia-like phenotype. <i>Ophthalmic Genetics</i> , 2017 , 38, 286-287	1.2	2
56	Clinical features of a toddler with bilateral bullous retinoschisis with a novel mutation. <i>American Journal of Ophthalmology Case Reports</i> , 2017 , 5, 76-80	1.3	2
55	Foveal Hypoplasia in Patients with Stickler Syndrome. <i>Ophthalmology</i> , 2017 , 124, 896-902	7.3	32
54	Gene Analysis in 20 Japanese Patients with Angioid Streaks Revealing Four Frequent and Two Novel Variants and Pseudodominant Inheritance. <i>Journal of Ophthalmology</i> , 2017 , 2017, 1079687	2	3

53	Differences in ocular findings in two siblings: one with complete and other with incomplete achromatopsia. <i>Documenta Ophthalmologica</i> , 2017 , 134, 141-147	2.2	5
52	Heterozygous deletion of the OPA1 gene in patients with dominant optic atrophy. <i>Japanese Journal of Ophthalmology</i> , 2017 , 61, 395-401	2.6	5
51	Closure of a full-thickness macular hole without vitrectomy in choroideraemia. <i>Australasian journal of optometry, The</i> , 2017 , 100, 294-295	2.7	1
50	Cone Dysfunction Syndrome in the Japanese Population. <i>Essentials in Ophthalmology</i> , 2017 , 129-135	0.2	
49	RPE65 Mutations in Two Japanese Families with Leber Congenital Amaurosis. <i>Ophthalmic Genetics</i> , 2016 , 37, 161-9	1.2	22
48	Novel C8orf37 Mutations in Patients with Early-onset Retinal Dystrophy, Macular Atrophy, Cataracts, and High Myopia. <i>Ophthalmic Genetics</i> , 2016 , 37, 68-75	1.2	13
47	Spontaneous Resolution of Large Macular Retinoschisis in Enhanced S-Cone Syndrome. <i>Ophthalmic Surgery Lasers and Imaging Retina</i> , 2016 , 47, 187-90	1.4	3
46	High-Resolution Adaptive Optics Retinal Image Analysis at Early Stage Central Areolar Choroidal Dystrophy With PRPH2 Mutation. <i>Ophthalmic Surgery Lasers and Imaging Retina</i> , 2016 , 47, 1115-1126	1.4	15
45	Novel RP1L1 Variants and Genotype-Photoreceptor Microstructural Phenotype Associations in Cohort of Japanese Patients With Occult Macular Dystrophy 2016 , 57, 4837-46		43
44	A novel heterozygous splice site OPA1 mutation causes exon 10 skipping in Japanese patients with dominant optic atrophy. <i>Ophthalmic Genetics</i> , 2016 , 37, 354-6	1.2	1
43	New truncation mutation of the NR2E3 gene in a Japanese patient with enhanced S-cone syndrome. <i>Japanese Journal of Ophthalmology</i> , 2016 , 60, 476-485	2.6	9
42	Somatic instability of expanded CAG repeats of ATXN7 in Japanese patients with spinocerebellar ataxia type 7. <i>Documenta Ophthalmologica</i> , 2015 , 130, 189-95	2.2	6
41	Mutation analysis of BEST1 in Japanese patients with Best@ vitelliform macular dystrophy. <i>British Journal of Ophthalmology</i> , 2015 , 99, 1577-82	5.5	16
40	Congenital Achromatopsia and Macular Atrophy Caused by a Novel Recessive PDE6C Mutation (p.E591K). <i>Ophthalmic Genetics</i> , 2015 , 36, 137-44	1.2	10
39	Autosomal recessive cone-rod dystrophy associated with compound heterozygous mutations in the EYS gene. <i>Documenta Ophthalmologica</i> , 2014 , 128, 211-7	2.2	30
38	Occult macular dystrophy with bilateral chronic subfoveal serous retinal detachment associated with a novel RP1L1 mutation (p.S1199P). <i>Documenta Ophthalmologica</i> , 2014 , 129, 49-56	2.2	15
37	RHO Mutations (p.W126L and p.A346P) in Two Japanese Families with Autosomal Dominant Retinitis Pigmentosa. <i>Journal of Ophthalmology</i> , 2014 , 2014, 210947	2	14
36	Whole exome analysis identifies frequent CNGA1 mutations in Japanese population with autosomal recessive retinitis pigmentosa. <i>PLoS ONE</i> , 2014 , 9, e108721	3.7	44

35	Three-year visual outcome of photodynamic therapy plus intravitreal bevacizumab with or without subtenon triamcinolone acetonide injections for polypoidal choroidal vasculopathy. <i>British Journal of Ophthalmology</i> , 2014 , 98, 1642-8	5.5	13
34	OAT mutations and clinical features in two Japanese brothers with gyrate atrophy of the choroid and retina. <i>Documenta Ophthalmologica</i> , 2014 , 128, 137-48	2.2	14
33	Novel mutations in enhanced S-cone syndrome. <i>Ophthalmology</i> , 2013 , 120, 431.e1-6	7.3	6
32	Multifocal electroretinographic evaluation of macular function in acute posterior multifocal placoid pigment epitheliopathy. <i>Documenta Ophthalmologica</i> , 2013 , 126, 253-8	2.2	3
31	Elderly case of pseudo-unilateral occult macular dystrophy with Arg45Trp mutation in RP1L1 gene. <i>Documenta Ophthalmologica</i> , 2013 , 127, 141-6	2.2	14
30	Whole-exome sequencing identifies a novel ALMS1 mutation (p.Q2051X) in two Japanese brothers with Alström syndrome. <i>Molecular Vision</i> , 2013 , 19, 2393-406	2.3	30
29	Subfoveal choroidal thickness in multiple evanescent white dot syndrome. <i>Australasian journal of optometry, The</i> , 2012 , 95, 212-7	2.7	68
28	Improvement of central visual function following steroid pulse therapy in acute zonal occult outer retinopathy. <i>Documenta Ophthalmologica</i> , 2012 , 124, 249-54	2.2	23
27	Autosomal dominant occult macular dystrophy with an RP1L1 mutation (R45W). <i>Optometry and Vision Science</i> , 2012 , 89, 684-91	2.1	24
26	Improvement in S-cone-mediated visual fields and rod function after correction of vitamin A deficiency. <i>European Journal of Ophthalmology</i> , 2011 , 21, 657-60	1.9	5
25	Color vision in an elderly patient with protanopic genotype and successfully treated unilateral age-related macular degeneration. <i>International Ophthalmology</i> , 2011 , 31, 471-5	2.2	
24	A novel mutation (Cys83Tyr) in the second zinc finger of NR2E3 in enhanced S-cone syndrome. <i>Graefes Archive for Clinical and Experimental Ophthalmology</i> , 2011 , 249, 201-8	3.8	8
23	Macular Dysfunction in Oguchi Disease with the Frequent Mutation 1147delA in the SAG Gene. <i>Ophthalmic Research</i> , 2011 , 46, 175-80	2.9	15
22	A novel haplotype with the R345W mutation in the EFEMP1 gene associated with autosomal dominant drusen in a Japanese family 2010 , 51, 1643-50		34
21	OPA1?????????????????????????????. <i>Japanese Orthoptic Journal</i> , 2010 , 39, 117-122	0	
20	?????????????????????????????. <i>Japanese Orthoptic Journal</i> , 2010 , 39, 123-128	0	
19	A novel homozygous GRK1 mutation (P391H) in 2 siblings with Oguchi disease with markedly reduced cone responses. <i>Ophthalmology</i> , 2007 , 114, 134-41	7.3	35
18	Novel form of a single X-linked visual pigment gene in a unique dichromatic color-vision defect. <i>Visual Neuroscience</i> , 2006 , 23, 411-7	1.7	7

17	Compound heterozygous CNGA3 mutations (R436W, L633P) in a Japanese patient with congenital achromatopsia. <i>Visual Neuroscience</i> , 2006 , 23, 395-402	1.7	30
16	Compound heterozygous RDH5 mutations in familial fleck retina with night blindness. <i>Acta Ophthalmologica</i> , 2006 , 84, 254-8		13
15	Dominant optic atrophy caused by a novel OPA1 splice site mutation (IVS20+1G-->A) associated with intron retention. <i>Ophthalmic Research</i> , 2005 , 37, 214-24	2.9	9
14	CYP4V2 mutations in two Japanese patients with Bietti's crystalline dystrophy. <i>Ophthalmic Research</i> , 2005 , 37, 262-9	2.9	35
13	Novel NR2E3 mutations (R104Q, R334G) associated with a mild form of enhanced S-cone syndrome demonstrate compound heterozygosity. <i>Ophthalmology</i> , 2005 , 112, 2115	7.3	31
12	Expression of <i>rinx/vsx1</i> during postnatal eye development in cone-bipolar, differentiating ganglion, and lens fiber cells. <i>Japanese Journal of Ophthalmology</i> , 2005 , 49, 93-105	2.6	13
11	Clinical heterogeneity between two Japanese siblings with congenital achromatopsia. <i>Visual Neuroscience</i> , 2004 , 21, 413-20	1.7	8
10	Enhanced S-cone syndrome in a Japanese family with a nonsense NR2E3 mutation (Q350X). <i>Acta Ophthalmologica</i> , 2004 , 82, 616-22		19
9	Four Japanese male patients with juvenile retinoschisis: only three have mutations in the RS1 gene. <i>American Journal of Ophthalmology</i> , 2004 , 138, 788-98	4.9	22
8	X-linked high myopia associated with cone dysfunction. <i>JAMA Ophthalmology</i> , 2004 , 122, 897-908		62
7	The molecular basis of dichromatic color vision in males with multiple red and green visual pigment genes. <i>Human Molecular Genetics</i> , 2002 , 11, 23-32	5.6	47
6	The importance of gene order in expression of the red and green visual pigment genes and in color vision. <i>Color Research and Application</i> , 2001 , 26, S79-S83	1.3	7
5	Number and arrangement of the red and green visual pigment genes in color-normal Japanese males. <i>Color Research and Application</i> , 2001 , 26, S84-S88	1.3	3
4	Molecular analysis of human red/green visual pigment gene locus: relationship to color vision. <i>Methods in Enzymology</i> , 2000 , 316, 651-70	1.7	9
3	RINX(VSX1), a novel homeobox gene expressed in the inner nuclear layer of the adult retina. <i>Genomics</i> , 2000 , 67, 128-39	4.3	74
2	Position of a green-red hybrid gene in the visual pigment array determines colour-vision phenotype. <i>Nature Genetics</i> , 1999 , 22, 90-3	36.3	86
1	Clinical characterization of autosomal dominant retinitis pigmentosa with NRL mutation in a three-generation Japanese family. <i>Documenta Ophthalmologica</i> ,	2.2	