

Takaaki Hayashi

List of Publications by Year in descending order

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Version: 2024-02-01

108
papers

1,747
citations

331259

21
h-index

377514

34
g-index

110
all docs

110
docs citations

110
times ranked

1663
citing authors

#	ARTICLE	IF	CITATIONS
1	Position of a 'green-red' hybrid gene in the visual pigment array determines colour-vision phenotype. <i>Nature Genetics</i> , 1999, 22, 90-93.	9.4	104
2	RINX(VSX1), a Novel Homeobox Gene Expressed in the Inner Nuclear Layer of the Adult Retina. <i>Genomics</i> , 2000, 67, 128-139.	1.3	78
3	X-Linked High Myopia Associated With Cone Dysfunction. <i>JAMA Ophthalmology</i> , 2004, 122, 897.	2.6	74
4	Subfoveal choroidal thickness in multiple evanescent white dot syndrome. <i>Australasian journal of optometry</i> , 2012, 95, 212-217.	0.6	74
5	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.	1.4	57
6	Whole Exome Analysis Identifies Frequent CNGA1 Mutations in Japanese Population with Autosomal Recessive Retinitis Pigmentosa. <i>PLoS ONE</i> , 2014, 9, e108721.	1.1	56
7	Novel <i>RP1L1</i> Variants and Genotype-Photoreceptor Microstructural Phenotype Associations in Cohort of Japanese Patients With Occult Macular Dystrophy. , 2016, 57, 4837.		54
8	Molecular Diagnosis of 34 Japanese Families with Leber Congenital Amaurosis Using Targeted Next Generation Sequencing. <i>Scientific Reports</i> , 2018, 8, 8279.	1.6	48
9	A Novel Homozygous GRK1 Mutation (P391H) in 2 Siblings with Oguchi Disease with Markedly Reduced Cone Responses. <i>Ophthalmology</i> , 2007, 114, 134-141.e1.	2.5	45
10	Foveal Hypoplasia in Patients with Stickler Syndrome. <i>Ophthalmology</i> , 2017, 124, 896-902.	2.5	39
11	Novel NR2E3 Mutations (R104Q, R334G) Associated with a Mild Form of Enhanced S-Cone Syndrome Demonstrate Compound Heterozygosity. <i>Ophthalmology</i> , 2005, 112, 2115.e1-2115.e10.	2.5	37
12	A Novel Haplotype with the R345W Mutation in the <i>EFEMP1</i> Gene Associated with Autosomal Dominant Drusen in a Japanese Family. , 2010, 51, 1643.		37
13	<i>CYP4V2</i> Mutations in Two Japanese Patients with Bietti's Crystalline Dystrophy. <i>Ophthalmic Research</i> , 2005, 37, 262-269.	1.0	36
14	Compound heterozygous CNGA3 mutations (R436W, L633P) in a Japanese patient with congenital achromatopsia. <i>Visual Neuroscience</i> , 2006, 23, 395-402.	0.5	34
15	Autosomal recessive cone-rod dystrophy associated with compound heterozygous mutations in the EYS gene. <i>Documenta Ophthalmologica</i> , 2014, 128, 211-217.	1.0	33
16	Autosomal Dominant Occult Macular Dystrophy with an <i>RP1L1</i> Mutation (R45W). <i>Optometry and Vision Science</i> , 2012, 89, 684-691.	0.6	31
17	Whole-exome sequencing identifies a novel <i>ALMS1</i> mutation (p.Q2051X) in two Japanese brothers with Alström syndrome. <i>Molecular Vision</i> , 2013, 19, 2393-406.	1.1	31
18	Improvement of central visual function following steroid pulse therapy in acute zonal occult outer retinopathy. <i>Documenta Ophthalmologica</i> , 2012, 124, 249-254.	1.0	29

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19	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, 1518.	1.8	26
20	Clinical Stages of Occult Macular Dystrophy Based on Optical Coherence Tomographic Findings. , 2019, 60, 4691.		25
21	<i>RPE65</i> Mutations in Two Japanese Families with Leber Congenital Amaurosis. <i>Ophthalmic Genetics</i> , 2016, 37, 161-169.	0.5	24
22	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.	1.6	24
23	Enhanced S-cone syndrome in a Japanese family with a nonsense NR2E3 mutation (Q350X). <i>Acta Ophthalmologica</i> , 2004, 82, 616-622.	0.4	22
24	Four Japanese male patients with juvenile retinoschisis: Only three have mutations in the RS1 gene. <i>American Journal of Ophthalmology</i> , 2004, 138, 788-798.	1.7	22
25	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.	0.7	21
26	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.	1.6	21
27	Macular Dysfunction in Oguchi Disease with the Frequent Mutation 1147delA in the <i>SAG</i> Gene. <i>Ophthalmic Research</i> , 2011, 46, 175-180.	1.0	20
28	Mutation analysis of <i>BEST1</i> in Japanese patients with Best's vitelliform macular dystrophy. <i>British Journal of Ophthalmology</i> , 2015, 99, 1577-1582.	2.1	20
29	Heterozygous GGC repeat expansion of <i>NOTCH2NLC</i> in a patient with neuronal intranuclear inclusion disease and progressive retinal dystrophy. <i>Ophthalmic Genetics</i> , 2020, 41, 93-95.	0.5	20
30	RDH5-Related Fundus Albipunctatus in a Large Japanese Cohort. , 2020, 61, 53.		20
31	High-Resolution Adaptive Optics Retinal Image Analysis at Early Stage Central Areolar Choroidal Dystrophy With <i>PRPH2</i> Mutation. <i>Ophthalmic Surgery Lasers and Imaging Retina</i> , 2016, 47, 1115-1126.	0.4	20
32	Novel homozygous <i>CLN3</i> missense variant in isolated retinal dystrophy: A case report and electron microscopic findings. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1308.	0.6	19
33	Occult macular dystrophy with bilateral chronic subfoveal serous retinal detachment associated with a novel <i>RP1L1</i> mutation (p.S1199P). <i>Documenta Ophthalmologica</i> , 2014, 129, 49-56.	1.0	18
34	Phenotypical Characteristics of <i>POC1B</i> -Associated Retinopathy in Japanese Cohort: Cone Dystrophy With Normal Fundusoscopic Appearance. , 2019, 60, 3432.		18
35	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.	0.9	17
36	Novel <i>C8orf37</i> Mutations in Patients with Early-onset Retinal Dystrophy, Macular Atrophy, Cataracts, and High Myopia. <i>Ophthalmic Genetics</i> , 2016, 37, 1-8.	0.5	17

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37	OAT mutations and clinical features in two Japanese brothers with gyrate atrophy of the choroid and retina. <i>Documenta Ophthalmologica</i> , 2014, 128, 137-148.	1.0	17
38	KCNV2-Associated Retinopathy: Genetics, Electrophysiology, and Clinical Course—KCNV2 Study Group Report 1. <i>American Journal of Ophthalmology</i> , 2021, 225, 95-107.	1.7	17
39	[43] Molecular analysis of human red/ green visual pigment gene locus: relationship to color vision. <i>Methods in Enzymology</i> , 2000, 316, 651-670.	0.4	16
40	Elderly case of pseudo-unilateral occult macular dystrophy with Arg45Trp mutation in RP1L1 gene. <i>Documenta Ophthalmologica</i> , 2013, 127, 141-146.	1.0	16
41	RHOMutations (p.W126L and p.A346P) in Two Japanese Families with Autosomal Dominant Retinitis Pigmentosa. <i>Journal of Ophthalmology</i> , 2014, 2014, 1-10.	0.6	16
42	Electroretinographic abnormalities associated with pregabalin: a case report. <i>Documenta Ophthalmologica</i> , 2020, 140, 279-287.	1.0	16
43	CLINICAL COURSE OF PARANEOPLASTIC RETINOPATHY WITH ANTI-TRPM1 AUTOANTIBODY IN JAPANESE COHORT. <i>Retina</i> , 2019, 39, 2410-2418.	1.0	15
44	Early onset flecked retinal dystrophy associated with new compound heterozygous variants. <i>Molecular Vision</i> , 2018, 24, 286-296.	1.1	15
45	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, 1642-1648.	2.1	14
46	Novel biallelic loss-of-function KCNV2 variants in cone dystrophy with supernormal rod responses. <i>Documenta Ophthalmologica</i> , 2019, 138, 229-239.	1.0	14
47	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.	1.0	14
48	Compound heterozygous RDH5 mutations in familial fleck retina with night blindness. <i>Acta Ophthalmologica</i> , 2005, 84, 254-258.	0.4	13
49	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.	0.3	12
50	Congenital Achromatopsia and Macular Atrophy Caused by a Novel Recessive PDE6C Mutation (p.E591K). <i>Ophthalmic Genetics</i> , 2015, 36, 137-144.	0.5	11
51	Dominant Optic Atrophy Caused by a Novel OPA1 Splice Site Mutation (IVS20+1G→A) Associated with Intron Retention. <i>Ophthalmic Research</i> , 2005, 37, 214-224.	1.0	10
52	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, 201-208.	1.0	10
53	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.	0.9	10
54	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.	1.6	10

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55	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.	0.8	9
56	Clinical heterogeneity between two Japanese siblings with congenital achromatopsia. <i>Visual Neuroscience</i> , 2004, 21, 413-420.	0.5	9
57	Compound heterozygous splice site variants in the SCLT1 gene highlight an additional candidate locus for Senior-LÅkken syndrome. <i>Scientific Reports</i> , 2018, 8, 16733.	1.6	9
58	Genotype-Phenotype Correlations in RP1-Associated Retinal Dystrophies: A Multi-Center Cohort Study in JAPAN. <i>Journal of Clinical Medicine</i> , 2021, 10, 2265.	1.0	9
59	Genetic and Phenotypic Landscape of PRPH2-Associated Retinal Dystrophy in Japan. <i>Genes</i> , 2021, 12, 1817.	1.0	9
60	Novel form of a single X-linked visual pigment gene in a unique dichromatic color-vision defect. <i>Visual Neuroscience</i> , 2006, 23, 411-417.	0.5	8
61	Novel Mutations in Enhanced S-cone Syndrome. <i>Ophthalmology</i> , 2013, 120, 431-431.e6.	2.5	8
62	Novel biallelic TRPM1 variants in an elderly patient with complete congenital stationary night blindness. <i>Documenta Ophthalmologica</i> , 2021, 142, 265-273.	1.0	8
63	Somatic instability of expanded CAG repeats of ATXN7 in Japanese patients with spinocerebellar ataxia type 7. <i>Documenta Ophthalmologica</i> , 2015, 130, 189-195.	1.0	7
64	Differences in ocular findings in two siblings: one with complete and other with incomplete achromatopsia. <i>Documenta Ophthalmologica</i> , 2017, 134, 141-147.	1.0	7
65	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.	1.6	7
66	New variants and in silico analyses in GRK1 associated Oguchi disease. <i>Human Mutation</i> , 2021, 42, 164-176.	1.1	7
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.	0.4	7
68	Clinical findings in eyes with BEST1-related retinopathy complicated by choroidal neovascularization. <i>Graefe's Archive for Clinical and Experimental Ophthalmology</i> , 2022, 260, 1125-1137.	1.0	7
69	Autosomal dominant retinitis pigmentosa with macular involvement associated with a disease haplotype that included a novel <i>PRPH2</i> variant (p.Cys250Gly). <i>Ophthalmic Genetics</i> , 2018, 39, 357-365.	0.5	6
70	Electroretinograms of eyes with Stickler syndrome. <i>Documenta Ophthalmologica</i> , 2020, 140, 233-243.	1.0	6
71	Long-term observation of a Japanese mucopolidosis IV patient with a novel homozygous p.F313del variant of <i>MCOLN1</i> . <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1500-1505.	0.7	6
72	Genotype and Long-term Clinical Course of Bietti Crystalline Dystrophy in Korean and Japanese Patients. <i>Ophthalmology Retina</i> , 2021, 5, 1269-1279.	1.2	6

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73	A new PDE6A missense variant p.Arg544Gln in rodâ€™cone dystrophy. Documenta Ophthalmologica, 2021, 143, 107-114.	1.0	6
74	Improvement in S-coneâ€™mediated Visual Fields and Rod Function after Correction of Vitamin a Deficiency. European Journal of Ophthalmology, 2011, 21, 657-660.	0.7	5
75	Heterozygous deletion of the OPA1 gene in patients with dominant optic atrophy. Japanese Journal of Ophthalmology, 2017, 61, 395-401.	0.9	5
76	Clinical findings of end-stage retinitis pigmentosa with a homozygous PDE6A variant (p.R653X). American Journal of Ophthalmology Case Reports, 2019, 13, 110-115.	0.4	5
77	Spontaneous Resolution of Large Macular Retinoschisis in Enhanced S-Cone Syndrome. Ophthalmic Surgery Lasers and Imaging Retina, 2016, 47, 187-190.	0.4	5
78	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
79	<i>ABCC6</i> Gene Analysis in 20 Japanese Patients with Angioid Streaks Revealing Four Frequent and Two Novel Variants and Pseudodominant Inheritance. Journal of Ophthalmology, 2017, 2017, 1-7.	0.6	4
80	Genetic defects of CHM and visual acuity outcome in 24 choroideremia patients from 16 Japanese families. Scientific Reports, 2020, 10, 15883.	1.6	4
81	Novel use of Finesse Flex loop for macular hole retinal detachment. American Journal of Ophthalmology Case Reports, 2020, 18, 100703.	0.4	4
82	Novel biallelic splice-site BBS1 variants in Bardetâ€™Biedle syndrome: a case report of the first Japanese patient. Documenta Ophthalmologica, 2020, 141, 77-88.	1.0	4
83	Complete congenital stationary night blindness associated with a novel NYX variant (p.Asn216Lys) in middle-aged and older adult patients. Ophthalmic Genetics, 2021, 42, 412-419.	0.5	4
84	Number and arrangement of the red and green visual pigment genes in color-normal Japanese males. Color Research and Application, 2001, 26, S84-S88.	0.8	3
85	Multifocal electroretinographic evaluation of macular function in acute posterior multifocal placoid pigment epitheliopathy. Documenta Ophthalmologica, 2013, 126, 253-258.	1.0	3
86	A novel heterozygous splice site OPA1 mutation causes exon 10 skipping in Japanese patients with dominant optic atrophy. Ophthalmic Genetics, 2016, 37, 354-356.	0.5	3
87	Reconstruction of Photoreceptor Outer Layers after Steroid Therapy in Solar Retinopathy. Case Reports in Ophthalmological Medicine, 2018, 2018, 1-5.	0.3	3
88	Unique and progressive retinal degeneration in a patient with cancer associated retinopathy. American Journal of Ophthalmology Case Reports, 2020, 20, 100908.	0.4	3
89	Clinical Course and Electron Microscopic Findings in Lymphocytes of Patients with DRAM2-Associated Retinopathy. International Journal of Molecular Sciences, 2020, 21, 1331.	1.8	3
90	Progress of macular atrophy during 30 monthsâ€™ follow-up in a patient with spinocerebellar ataxia type1 (SCA1). Documenta Ophthalmologica, 2021, 142, 87-98.	1.0	3

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91	The first Japanese family of <i>CDH3</i> -related hypotrichosis with juvenile macular dystrophy. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1688.	0.6	3
92	Homozygous single nucleotide duplication of <i>SLC38A8</i> in autosomal recessive foveal hypoplasia: The first Japanese case report. <i>Documenta Ophthalmologica</i> , 2021, 143, 323-330.	1.0	3
93	Clinical characterization of autosomal dominant retinitis pigmentosa with <i>NRL</i> mutation in a three-generation Japanese family. <i>Documenta Ophthalmologica</i> , 0, , .	1.0	3
94	A novel homozygous <i>CYP4V2</i> variant (p.S121Y) associated with a choroideremia-like phenotype. <i>Ophthalmic Genetics</i> , 2017, 38, 286-287.	0.5	2
95	Clinical features of a toddler with bilateral bullous retinoschisis with a novel <i>RS1</i> mutation. <i>American Journal of Ophthalmology Case Reports</i> , 2017, 5, 76-80.	0.4	2
96	Closure of a full-thickness macular hole without vitrectomy in choroideraemia. <i>Australasian journal of optometry, The</i> , 2017, 100, 294-295.	0.6	2
97	Improvement of reduced electroretinographic responses in thymoma-associated retinopathy: a case report and literature review. <i>Documenta Ophthalmologica</i> , 2020, 141, 195-204.	1.0	2
98	Vitamin A deficiency after prolonged intake of an unbalanced diet in a Japanese hemodialysis patient. <i>Documenta Ophthalmologica</i> , 2021, 143, 85-91.	1.0	2
99	The second Japanese family with Malattia Leventinese/Doyne honeycomb retinal dystrophy. <i>Documenta Ophthalmologica</i> , 2021, , 1.	1.0	2
100	Transient electroretinographic abnormalities that mimic those of <i>KCNV2</i> retinopathy: a case report. <i>Documenta Ophthalmologica</i> , 2021, 143, 221-228.	1.0	1
101	Polypoidal choroidal vasculopathy in a patient with <i>DMPK</i> -associated myotonic dystrophy. <i>Documenta Ophthalmologica</i> , 2022, 144, 217-226.	1.0	1
102	Color vision in an elderly patient with protanopic genotype and successfully treated unilateral age-related macular degeneration. <i>International Ophthalmology</i> , 2011, 31, 471-475.	0.6	0
103	Spontaneous Improvement of Visual Acuity in a 13-Year-Old Boy with Neuromyelitis Optica Spectrum Disorder. <i>Neuro-Ophthalmology</i> , 2019, 43, 114-119.	0.4	0
104	Vision Improvement after Osimertinib Treatment in Paraneoplastic Optic Neuropathy Associated with Lung Adenocarcinoma. <i>Case Reports in Ophthalmological Medicine</i> , 2021, 2021, 1-7.	0.3	0
105	OPA1-related macular dystrophy in a Japanese family. <i>Japanese Orthoptic Journal</i> , 2010, 39, 11-15.		
106	OPA1-related macular dystrophy in a Japanese family. <i>Japanese Orthoptic Journal</i> , 2010, 39, 11-15.		
107	Cone Dysfunction Syndrome in the Japanese Population. <i>Essentials in Ophthalmology</i> , 2017, , 129-135.	0.0	0
108	Multimodal Imaging of Subfoveal Pachydrusen Containing a Blood Flow Signal. <i>Case Reports in Ophthalmological Medicine</i> , 2022, 2022, 1-6.	0.3	0