Takaaki Hayashi

List of Publications by Year
in descending order

108 papers	1,747 citations	331670 21 h-index	377865 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. Nature Genetics, 1999, 22, 90-93.	21.4	104
2	RINX(VSX1), a Novel Homeobox Gene Expressed in the Inner Nuclear Layer of the Adult Retina. Genomics, 2000, 67, 128-139.	2.9	78
3	X-Linked High Myopia Associated With Cone Dysfunction. JAMA Ophthalmology, 2004, 122, 897.	2.4	74
4	Subfoveal choroidal thickness in multiple evanescent white dot syndrome. Australasian journal of optometry, The, 2012, 95, 212-217.	1.3	74
5	The molecular basis of dichromatic color vision in males with multiple red and green visual pigment genes. Human Molecular Genetics, 2002, 11, 23-32.	2.9	57
6	Whole Exome Analysis Identifies Frequent CNGA1 Mutations in Japanese Population with Autosomal Recessive Retinitis Pigmentosa. PLoS ONE, 2014, 9, e108721.	2.5	56
7	NovelRP1L1Variants and Genotypeâ€"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. Scientific Reports, 2018, 8, 8279.	3.3	48
9	A Novel Homozygous GRK1 Mutation (P391H) in 2 Siblings with Oguchi Disease with Markedly Reduced Cone Responses. Ophthalmology, 2007, 114, 134-141.e1.	5.2	45
10	Foveal Hypoplasia in Patients with Stickler Syndrome. Ophthalmology, 2017, 124, 896-902.	5.2	39
11	Novel NR2E3 Mutations (R104Q, R334G) Associated with a Mild Form of Enhanced S-Cone Syndrome Demonstrate Compound Heterozygosity. Ophthalmology, 2005, 112, 2115.e1-2115.e10.	5.2	37
12	A Novel Haplotype with the R345W Mutation in theEFEMP1Gene Associated with Autosomal Dominant Drusen in a Japanese Family. , 2010, 51, 1643.		37
13	CYP4V2 Mutations in Two Japanese Patients with Biettiâ€™s Crystalline Dystrophy. Ophthalmic Research, 2005, 37, 262-269.	1.9	36
14	Compound heterozygous CNGA3 mutations (R436W, L633P) in a Japanese patient with congenital achromatopsia. Visual Neuroscience, 2006, 23, 395-402.	1.0	34
15	Autosomal recessive coneâ€"rod dystrophy associated with compound heterozygous mutations in the EYS gene. Documenta Ophthalmologica, 2014, 128, 211-217.	2.2	33
16	Autosomal Dominant Occult Macular Dystrophy with an RP1L1 Mutation (R45W). Optometry and Vision Science, 2012, 89, 684-691.	1.2	31
17	Whole-exome sequencing identifies a novel ALMS1 mutation (p.Q2051X) in two Japanese brothers with AlstrÃ¶m syndrome. Molecular Vision, 2013, 19, 2393-406.	1.1	31
18	Improvement of central visual function following steroid pulse therapy in acute zonal occult outer retinopathy. Documenta Ophthalmologica, 2012, 124, 249-254.	2.2	29

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19	X-linked Retinitis Pigmentosa in Japan: Clinical and Genetic Findings in Male Patients and Female Carriers. International Journal of Molecular Sciences, 2019, 20, 1518.	4.1	26
20	Clinical Stages of Occult Macular Dystrophy Based on Optical Coherence Tomographic Findings. , 2019, 60, 4691.		25
21	RPE65 Mutations in Two Japanese Families with Leber Congenital Amaurosis. Ophthalmic Genetics, 2016, 37, 161-169.	1.2	24
22	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
23	Enhanced S-cone syndrome in a Japanese family with a nonsense NR2E3 mutation (Q350X). Acta Ophthalmologica, 2004, 82, 616-622.	0.3	22
24	Four Japanese male patients with juvenile retinoschisis: Only three have mutations in the RS1 gene. American Journal of Ophthalmology, 2004, 138, 788-798.	3.3	22
25	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
26	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
27	Macular Dysfunction in Oguchi Disease with the Frequent Mutation 1147delA in the <i>SAG</i> Gene. Ophthalmic Research, 2011, 46, 175-180.	1.9	20
28	Mutation analysis ofBEST1in Japanese patients with Best's vitelliform macular dystrophy. British Journal of Ophthalmology, 2015, 99, 1577-1582.	3.9	20
29	Heterozygous GGC repeat expansion of NOTCH2NLC in a patient with neuronal intranuclear inclusion disease and progressive retinal dystrophy. Ophthalmic Genetics, 2020, 41, 93-95.	1.2	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 PRPH2 Mutation. Ophthalmic Surgery Lasers and Imaging Retina, 2016, 47, 1115-1126.	0.7	20
32	Novel homozygous CLN3 missense variant in isolated retinal dystrophy: A case report and electron microscopic findings. Molecular Genetics & Genomic Medicine, 2020, 8, e1308.	1.2	19
33	Occult macular dystrophy with bilateral chronic subfoveal serous retinal detachment associated with a novel RP1L1 mutation (p.S1199P). Documenta Ophthalmologica, 2014, 129, 49-56.	2.2	18
34	Phenotypical Characteristics ofPOC1B-Associated Retinopathy in Japanese Cohort: Cone Dystrophy With Normal Funduscopic Appearance. , 2019, 60, 3432.		18
35	Expression of Rinx/Vsx1 during postnatal eye development in cone-bipolar, differentiating ganglion, and lens fiber cells. Japanese Journal of Ophthalmology, 2005, 49, 93-105.	1.9	17
36	NovelC8orf37Mutations in Patients with Early-onset Retinal Dystrophy, Macular Atrophy, Cataracts, and High Myopia. Ophthalmic Genetics, 2016, 37, 1-8.	1.2	17

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37	OAT mutations and clinical features in two Japanese brothers with gyrate atrophy of the choroid and retina. Documenta Ophthalmologica, 2014, 128, 137-148.	2.2	17
38	KCNV2-Associated Retinopathy: Genetics, Electrophysiology, and Clinical Courseâ€"KCNV2 Study Group Report 1. American Journal of Ophthalmology, 2021, 225, 95-107.	3.3	17
39	[43] Molecular analysis of human red/ green visual pigment gene locus: relationship to color vision. Methods in Enzymology, 2000, 316, 651-670.	1.0	16
40	Elderly case of pseudo-unilateral occult macular dystrophy with Arg45Trp mutation in RP1L1 gene. Documenta Ophthalmologica, 2013, 127, 141-146.	2.2	16
41	RHOMutations (p.W126L and p.A346P) in Two Japanese Families with Autosomal Dominant Retinitis Pigmentosa. Journal of Ophthalmology, 2014, 2014, 1-10.	1.3	16
42	Electroretinographic abnormalities associated with pregabalin: a case report. Documenta Ophthalmologica, 2020, 140, 279-287.	2.2	16
43	CLINICAL COURSE OF PARANEOPLASTIC RETINOPATHY WITH ANTI-TRPM1 AUTOANTIBODY IN JAPANESE COHORT. Retina, 2019, 39, 2410-2418.	1.7	15
44	Early onset flecked retinal dystrophy associated with new compound heterozygous variants. Molecular Vision, 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. British Journal of Ophthalmology, 2014, 98, 1642-1648.	3.9	14
46	Novel biallelic loss-of-function KCNV2 variants in cone dystrophy with supernormal rod responses. Documenta Ophthalmologica, 2019, 138, 229-239.	2.2	14
47	Coexistence of GNAT1 and ABCA4 variants associated with Nougaret-type congenital stationary night blindness and childhood-onset cone-rod dystrophy. Documenta Ophthalmologica, 2020, 140, 147-157.	2.2	14
48	Compound heterozygous RDH5 mutations in familial fleck retina with night blindness. Acta Ophthalmologica, 2005, 84, 254-258.	0.3	13
49	Neuronal intranuclear hyaline inclusion disease presenting with childhood-onset night blindness associated with progressive retinal dystrophy. Journal of the Neurological Sciences, 2018, 388, 84-86.	0.6	12
50	Congenital Achromatopsia and Macular Atrophy Caused by a Novel RecessivePDE6CMutation (p.E591K). Ophthalmic Genetics, 2015, 36, 137-144.	1.2	11
51	Dominant Optic Atrophy Caused by a Novel OPA1 Splice Site Mutation (IVS20+1Gâ†'A) Associated with Intron Retention. Ophthalmic Research, 2005, 37, 214-224.	1.9	10
52	A novel mutation (Cys83Tyr) in the second zinc finger of NR2E3 in enhanced S-cone syndrome. Graefe's Archive for Clinical and Experimental Ophthalmology, 2011, 249, 201-208.	1.9	10
53	New truncation mutation of the NR2E3 gene in a Japanese patient with enhanced S-cone syndrome. Japanese Journal of Ophthalmology, 2016, 60, 476-485.	1.9	10
54	Genotype determination of the OPN1LW/OPN1MW genes: novel disease-causing mechanisms in Japanese patients with blue cone monochromacy. Scientific Reports, 2018, 8, 11507.	3.3	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. Color Research and Application, 2001, 26, S79-S83.	1.6	9
56	Clinical heterogeneity between two Japanese siblings with congenital achromatopsia. Visual Neuroscience, 2004, 21, 413-420.	1.0	9
57	Compound heterozygous splice site variants in the SCLT1 gene highlight an additional candidate locus for Senior-LÃ¸ken syndrome. Scientific Reports, 2018, 8, 16733.	3.3	9
58	Genotype-Phenotype Correlations in RP1-Associated Retinal Dystrophies: A Multi-Center Cohort Study in JAPAN. Journal of Clinical Medicine, 2021, 10, 2265.	2.4	9
59	Genetic and Phenotypic Landscape of PRPH2-Associated Retinal Dystrophy in Japan. Genes, 2021, 12, 1817.	2.4	9
60	Novel form of a single X-linked visual pigment gene in a unique dichromatic color-vision defect. Visual Neuroscience, 2006, 23, 411-417.	1.0	8
61	Novel Mutations in Enhanced S-cone Syndrome. Ophthalmology, 2013, 120, 431-431.e6.	5.2	8
62	Novel biallelic TRPM1 variants in an elderly patient with complete congenital stationary night blindness. Documenta Ophthalmologica, 2021, 142, 265-273.	2.2	8
63	Somatic instability of expanded CAG repeats of ATXN7 in Japanese patients with spinocerebellar ataxia type 7. Documenta Ophthalmologica, 2015, 130, 189-195.	2.2	7
64	Differences in ocular findings in two siblings: one with complete and other with incomplete achromatopsia. Documenta Ophthalmologica, 2017, 134, 141-147.	2.2	7
65	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
66	New variants and in silico analyses in GRK1 associated Oguchi disease. Human Mutation, 2021, 42, 164-176.	2.5	7
67	High-Resolution Retinal Imaging Reveals Preserved Cone Photoreceptor Density and Choroidal Thickness in Female Carriers of Choroideremia. Ophthalmic Surgery Lasers and Imaging Retina, 2019, 50, 76-85.	0.7	7
68	Clinical findings in eyes with BEST1-related retinopathy complicated by choroidal neovascularization. Graefe's Archive for Clinical and Experimental Ophthalmology, 2022, 260, 1125-1137.	1.9	7
69	Autosomal dominant retinitis pigmentosa with macular involvement associated with a disease haplotype that included a novel PRPH2 variant (p.Cys250Gly). Ophthalmic Genetics, 2018, 39, 357-365.	1.2	6
70	Electroretinograms of eyes with Stickler syndrome. Documenta Ophthalmologica, 2020, 140, 233-243.	2.2	6
71	Longâ€term observation of a Japanese mucolipidosis <scp>IV</scp> patient with a novel homozygous p.F313del variant of <scp>MCOLN1</scp>. American Journal of Medical Genetics, Part A, 2020, 182, 1500-1505.	1.2	6
72	Genotype and Long-term Clinical Course of Bietti Crystalline Dystrophy in Korean and Japanese Patients. Ophthalmology Retina, 2021, 5, 1269-1279.	2.4	6

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73	A new PDE6A missense variant p.Arg544Gln in rodâ€"cone dystrophy. Documenta Ophthalmologica, 2021, 143, 107-114.	2.2	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.	1.3	5
75	Heterozygous deletion of the OPA1 gene in patients with dominant optic atrophy. Japanese Journal of Ophthalmology, 2017, 61, 395-401.	1.9	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.	0.7	5
77	Spontaneous Resolution of Large Macular Retinoschisis in Enhanced S-Cone Syndrome. Ophthalmic Surgery Lasers and Imaging Retina, 2016, 47, 187-190.	0.7	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	ABCC6 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.	1.3	4
80	Genetic defects of CHM and visual acuity outcome in 24 choroideremia patients from 16 Japanese families. Scientific Reports, 2020, 10, 15883.	3.3	4
81	Novel use of Finesse Flex loop for macular hole retinal detachment. American Journal of Ophthalmology Case Reports, 2020, 18, 100703.	0.7	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.	2.2	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.	1.2	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.	1.6	3
85	Multifocal electroretinographic evaluation of macular function in acute posterior multifocal placoid pigment epitheliopathy. Documenta Ophthalmologica, 2013, 126, 253-258.	2.2	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.	1.2	3
87	Reconstruction of Photoreceptor Outer Layers after Steroid Therapy in Solar Retinopathy. Case Reports in Ophthalmological Medicine, 2018, 2018, 1-5.	0.5	3
88	Unique and progressive retinal degeneration in a patient with cancer associated retinopathy. American Journal of Ophthalmology Case Reports, 2020, 20, 100908.	0.7	3
89	Clinical Course and Electron Microscopic Findings in Lymphocytes of Patients with DRAM2-Associated Retinopathy. International Journal of Molecular Sciences, 2020, 21, 1331.	4.1	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.	2.2	3

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91	The first Japanese family of CDH3â€related hypotrichosis with juvenile macular dystrophy. Molecular Genetics & Genomic Medicine, 2021, 9, e1688.	1.2	3
92	Homozygous single nucleotide duplication of SLC38A8 in autosomal recessive foveal hypoplasia: The first Japanese case report. Documenta Ophthalmologica, 2021, 143, 323-330.	2.2	3
93	Clinical characterization of autosomal dominant retinitis pigmentosa with NRL mutation in a three-generation Japanese family. Documenta Ophthalmologica, 0, , .	2.2	3
94	A novel homozygous CYP4V2 variant (p.S121Y) associated with a choroideremia-like phenotype. Ophthalmic Genetics, 2017, 38, 286-287.	1.2	2
95	Clinical features of a toddler with bilateral bullous retinoschisis with a novel RS1 mutation. American Journal of Ophthalmology Case Reports, 2017, 5, 76-80.	0.7	2
96	Closure of a fullâ€thickness macular hole without vitrectomy in choroideraemia. Australasian journal of optometry, The, 2017, 100, 294-295.	1.3	2
97	Improvement of reduced electroretinographic responses in thymoma-associated retinopathy: a case report and literature review. Documenta Ophthalmologica, 2020, 141, 195-204.	2.2	2
98	Vitamin A deficiency after prolonged intake of an unbalanced diet in a Japanese hemodialysis patient. Documenta Ophthalmologica, 2021, 143, 85-91.	2.2	2
99	The second Japanese family with Malattia Leventinese/Doyne honeycomb retinal dystrophy. Documenta Ophthalmologica, 2021, , 1.	2.2	2
100	Transient electroretinographic abnormalities that mimic those of KCNV2 retinopathy: a case report. Documenta Ophthalmologica, 2021, 143, 221-228.	2.2	1
101	Polypoidal choroidal vasculopathy in a patient with DMPK-associated myotonic dystrophy. Documenta Ophthalmologica, 2022, 144, 217-226.	2.2	1
102	Color vision in an elderly patient with protanopic genotype and successfully treated unilateral age-related macular degeneration. International Ophthalmology, 2011, 31, 471-475.	1.4	0
103	Spontaneous Improvement of Visual Acuity in a 13-Year-Old Boy with Neuromyelitis Optica Spectrum Disorder. Neuro-Ophthalmology, 2019, 43, 114-119.	1.0	0
104	Vision Improvement after Osimertinib Treatment in Paraneoplastic Optic Neuropathy Associated with Lung Adenocarcinoma. Case Reports in Ophthalmological Medicine, 2021, 2021, 1-7.	0.5	0
105	OPA1é°ä¼åå‰ç•°ã,'æœ‰ã•™ã,‹å„æŸ“è‰²ä½“å„ªæ€§è¦–ç¥žçµŒèŽ,®ã®é»„æ–‘æ©Ÿèƒ½. Japanese Orthoptic Journal, 2010, 39, 11	0.1	0
106	é»„æ–‘éƒ¨éŒä½“æ©Ÿèƒ½ä½Žä¸‹ã«ã,ˆã,‹å¾ŒŒå©é•é»„è‰²è¦šç•°å„ã,'åˆä½µã•—ãŸåºƒç—…. Japanese Orthoptic Journal, 2010, 39, 12	0.1	0
107	Cone Dysfunction Syndrome in the Japanese Population. Essentials in Ophthalmology, 2017, , 129-135.	0.1	0
108	Multimodal Imaging of Subfoveal Pachydrusen Containing a Blood Flow Signal. Case Reports in Ophthalmological Medicine, 2022, 2022, 1-6.	0.5	0