Takaaki Hayashi

List of Publications by Citations

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	Position of a ‘green-red’ hybrid gene in the visual pigment array determines colour-vision phenotype. *Nature Genetics*, **1999**, 22, 90-3	36.3	86
105	RINX(VSX1), a novel homeobox gene expressed in the inner nuclear layer of the adult retina. *Genomics*, **2000**, 67, 128-39	4.3	74
104	Subfoveal choroidal thickness in multiple evanescent white dot syndrome. *Australasian journal of optometry, The*, **2012**, 95, 212-7	2.7	68
103	X-linked high myopia associated with cone dysfunction. *JAMA Ophthalmology*, **2004**, 122, 897-908		62
102	The molecular basis of dichromatic color vision in males with multiple red and green visual pigment genes. *Human Molecular Genetics*, **2002**, 11, 23-32	5.6	47
101	Whole exome analysis identifies frequent CNGA1 mutations in Japanese population with autosomal recessive retinitis pigmentosa. *PLoS ONE*, **2014**, 9, e108721	3.7	44
100	Novel RP1L1 Variants and Genotype-Photoreceptor Microstructural Phenotype Associations in Cohort of Japanese Patients With Occult Macular Dystrophy **2016**, 57, 4837-46		43
99	A novel homozygous GRK1 mutation (P391H) in 2 siblings with Oguchi disease with markedly reduced cone responses. *Ophthalmology*, **2007**, 114, 134-41	7.3	35
98	CYP4V2 mutations in two Japanese patients with Bietti’s crystalline dystrophy. *Ophthalmic Research*, **2005**, 37, 262-9	2.9	35
97	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
96	Foveal Hypoplasia in Patients with Stickler Syndrome. *Ophthalmology*, **2017**, 124, 896-902	7.3	32
95	Novel NR2E3 mutations (R104Q, R334G) associated with a mild form of enhanced S-cone syndrome demonstrate compound heterozygosity. *Ophthalmology*, **2005**, 112, 2115	7.3	31
94	Autosomal recessive cone-rod dystrophy associated with compound heterozygous mutations in the EYS gene. *Documenta Ophthalmologica*, **2014**, 128, 211-7	2.2	30
93	Compound heterozygous CNGA3 mutations (R436W, L633P) in a Japanese patient with congenital achromatopsia. *Visual Neuroscience*, **2006**, 23, 395-402	1.7	30
92	Whole-exome sequencing identifies a novel ALMS1 mutation (p.Q2051X) in two Japanese brothers with Alström syndrome. *Molecular Vision*, **2013**, 19, 2393-406	2.3	30
91	Autosomal dominant occult macular dystrophy with an RP1L1 mutation (R45W). *Optometry and Vision Science*, **2012**, 89, 684-91	2.1	24
90	Improvement of central visual function following steroid pulse therapy in acute zonal occult outer retinopathy. *Documenta Ophthalmologica*, **2012**, 124, 249-54	2.2	23

89	RPE65 Mutations in Two Japanese Families with Leber Congenital Amaurosis. *Ophthalmic Genetics*, **2016**, 37, 161-9	1.2	22
88	Four Japanese male patients with juvenile retinoschisis: only three have mutations in the RS1 gene. *American Journal of Ophthalmology*, **2004**, 138, 788-98	4.9	22
87	Molecular Diagnosis of 34 Japanese Families with Leber Congenital Amaurosis Using Targeted Next Generation Sequencing. *Scientific Reports*, **2018**, 8, 8279	4.9	21
86	Enhanced S-cone syndrome in a Japanese family with a nonsense NR2E3 mutation (Q350X). *Acta Ophthalmologica*, **2004**, 82, 616-22		19
85	Mutation analysis of BEST1 in Japanese patients with Best vitelliform macular dystrophy. *British Journal of Ophthalmology*, **2015**, 99, 1577-82	5.5	16
84	Clinical Stages of Occult Macular Dystrophy Based on Optical Coherence Tomographic Findings **2019**, 60, 4691-4700		16
83	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	15
82	Macular Dysfunction in Oguchi Disease with the Frequent Mutation 1147delA in the SAG Gene. *Ophthalmic Research*, **2011**, 46, 175-80	2.9	15
81	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	1.4	15
80	X-linked Retinitis Pigmentosa in Japan: Clinical and Genetic Findings in Male Patients and Female Carriers. *International Journal of Molecular Sciences*, **2019**, 20,	6.3	14
79	RHO Mutations (p.W126L and p.A346P) in Two Japanese Families with Autosomal Dominant Retinitis Pigmentosa. *Journal of Ophthalmology*, **2014**, 2014, 210947	2	14
78	OAT mutations and clinical features in two Japanese brothers with gyrate atrophy of the choroid and retina. *Documenta Ophthalmologica*, **2014**, 128, 137-48	2.2	14
77	Elderly case of pseudo-unilateral occult macular dystrophy with Arg45Trp mutation in RP1L1 gene. *Documenta Ophthalmologica*, **2013**, 127, 141-6	2.2	14
76	Novel biallelic loss-of-function KCNV2 variants in cone dystrophy with supernormal rod responses. *Documenta Ophthalmologica*, **2019**, 138, 229-239	2.2	13
75	Novel C8orf37 Mutations in Patients with Early-onset Retinal Dystrophy, Macular Atrophy, Cataracts, and High Myopia. *Ophthalmic Genetics*, **2016**, 37, 68-75	1.2	13
74	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, 1642-8	5.5	13
73	Compound heterozygous RDH5 mutations in familial fleck retina with night blindness. *Acta Ophthalmologica*, **2006**, 84, 254-8		13
72	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	2.6	13

71	RDH5-Related Fundus Albipunctatus in a Large Japanese Cohort **2020**, 61, 53		13
70	Electroretinographic abnormalities associated with pregabalin: a case report. *Documenta Ophthalmologica*, **2020**, 140, 279-287	2.2	12
69	Novel homozygous CLN3 missense variant in isolated retinal dystrophy: A case report and electron microscopic findings. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1308	2.3	12
68	CLINICAL COURSE OF PARANEOPLASTIC RETINOPATHY WITH ANTI-TRPM1 AUTOANTIBODY IN JAPANESE COHORT. *Retina*, **2019**, 39, 2410-2418	3.6	11
67	Phenotypical Characteristics of POC1B-Associated Retinopathy in Japanese Cohort: Cone Dystrophy With Normal Funduscopic Appearance **2019**, 60, 3432-3446		11
66	Early onset flecked retinal dystrophy associated with new compound heterozygous variants. *Molecular Vision*, **2018**, 24, 286-296	2.3	11
65	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	4.9	10
64	Congenital Achromatopsia and Macular Atrophy Caused by a Novel Recessive PDE6C Mutation (p.E591K). *Ophthalmic Genetics*, **2015**, 36, 137-44	1.2	10
63	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	10
62	Heterozygous GGC repeat expansion of in a patient with neuronal intranuclear inclusion disease and progressive retinal dystrophy. *Ophthalmic Genetics*, **2020**, 41, 93-95	1.2	9
61	Dominant optic atrophy caused by a novel OPA1 splice site mutation (IVS20+1G-->A) associated with intron retention. *Ophthalmic Research*, **2005**, 37, 214-24	2.9	9
60	Molecular analysis of human red/green visual pigment gene locus: relationship to color vision. *Methods in Enzymology*, **2000**, 316, 651-70	1.7	9
59	New truncation mutation of the NR2E3 gene in a Japanese patient with enhanced S-cone syndrome. *Japanese Journal of Ophthalmology*, **2016**, 60, 476-485	2.6	9
58	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, 201-8	3.8	8
57	Clinical heterogeneity between two Japanese siblings with congenital achromatopsia. *Visual Neuroscience*, **2004**, 21, 413-20	1.7	8
56	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. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2020**, 184, 656-674	3.1	8
55	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	3.2	7
54	Novel form of a single X-linked visual pigment gene in a unique dichromatic color-vision defect. *Visual Neuroscience*, **2006**, 23, 411-7	1.7	7

53	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	1.3	7
52	KCNV2-Associated Retinopathy: Genetics, Electrophysiology, and Clinical Course-KCNV2 Study Group Report 1. *American Journal of Ophthalmology*, **2021**, 225, 95-107	4.9	7
51	Somatic instability of expanded CAG repeats of ATXN7 in Japanese patients with spinocerebellar ataxia type 7. *Documenta Ophthalmologica*, **2015**, 130, 189-95	2.2	6
50	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	4.9	6
49	Long-term observation of a Japanese mucolipidosis IV patient with a novel homozygous p.F313del variant of MCOLN1. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1500-1505	2.5	6
48	Novel mutations in enhanced S-cone syndrome. *Ophthalmology*, **2013**, 120, 431.e1-6	7.3	6
47	Compound heterozygous splice site variants in the SCLT1 gene highlight an additional candidate locus for Senior-L□en syndrome. *Scientific Reports*, **2018**, 8, 16733	4.9	6
46	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	5
45	Genotype determination of the OPN1LW/OPN1MW genes: novel disease-causing mechanisms in Japanese patients with blue cone monochromacy. *Scientific Reports*, **2018**, 8, 11507	4.9	5
44	Differences in ocular findings in two siblings: one with complete and other with incomplete achromatopsia. *Documenta Ophthalmologica*, **2017**, 134, 141-147	2.2	5
43	Heterozygous deletion of the OPA1 gene in patients with dominant optic atrophy. *Japanese Journal of Ophthalmology*, **2017**, 61, 395-401	2.6	5
42	Improvement in S-cone-mediated visual fields and rod function after correction of vitamin A deficiency. *European Journal of Ophthalmology*, **2011**, 21, 657-60	1.9	5
41	Electroretinograms of eyes with Stickler syndrome. *Documenta Ophthalmologica*, **2020**, 140, 233-243	2.2	5
40	Characterization of GUCA1A-associated dominant cone/cone-rod dystrophy: low prevalence among Japanese patients with inherited retinal dystrophies. *Scientific Reports*, **2019**, 9, 16851	4.9	5
39	Novel biallelic TRPM1 variants in an elderly patient with complete congenital stationary night blindness. *Documenta Ophthalmologica*, **2021**, 142, 265-273	2.2	5
38	Clinical findings of end-stage retinitis pigmentosa with a homozygous variant (p.R653X). *American Journal of Ophthalmology Case Reports*, **2019**, 13, 110-115	1.3	4
37	Novel biallelic splice-site BBS1 variants in Bardet-Biedle syndrome: a case report of the first Japanese patient. *Documenta Ophthalmologica*, **2020**, 141, 77-88	2.2	3
36	Gene Analysis in 20 Japanese Patients with Angioid Streaks Revealing Four Frequent and Two Novel Variants and Pseudodominant Inheritance. *Journal of Ophthalmology*, **2017**, 2017, 1079687	2	3

35	Multifocal electroretinographic evaluation of macular function in acute posterior multifocal placoid pigment epitheliopathy. *Documenta Ophthalmologica*, **2013**, 126, 253-8	2.2	3
34	Number and arrangement of the red and green visual pigment genes in color-normal Japanese males. *Color Research and Application*, **2001**, 26, S84-S88	1.3	3
33	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	2.3	3
32	Spontaneous Resolution of Large Macular Retinoschisis in Enhanced S-Cone Syndrome. *Ophthalmic Surgery Lasers and Imaging Retina*, **2016**, 47, 187-90	1.4	3
31	Clinical findings in eyes with BEST1-related retinopathy complicated by choroidal neovascularization. *Graefe's Archive for Clinical and Experimental Ophthalmology*, **2021**, 1	3.8	3
30	Genetic defects of CHM and visual acuity outcome in 24 choroideremia patients from 16 Japanese families. *Scientific Reports*, **2020**, 10, 15883	4.9	3
29	Complete congenital stationary night blindness associated with a novel variant (p.Asn216Lys) in middle-aged and older adult patients. *Ophthalmic Genetics*, **2021**, 42, 412-419	1.2	3
28	New variants and in silico analyses in GRK1 associated Oguchi disease. *Human Mutation*, **2021**, 42, 164-176	4.7	3
27	A novel homozygous CYP4V2 variant (p.S121Y) associated with a choroideremia-like phenotype. *Ophthalmic Genetics*, **2017**, 38, 286-287	1.2	2
26	Clinical features of a toddler with bilateral bullous retinoschisis with a novel mutation. *American Journal of Ophthalmology Case Reports*, **2017**, 5, 76-80	1.3	2
25	Unique and progressive retinal degeneration in a patient with cancer associated retinopathy. *American Journal of Ophthalmology Case Reports*, **2020**, 20, 100908	1.3	2
24	Improvement of reduced electroretinographic responses in thymoma-associated retinopathy: a case report and literature review. *Documenta Ophthalmologica*, **2020**, 141, 195-204	2.2	2
23	Reconstruction of Photoreceptor Outer Layers after Steroid Therapy in Solar Retinopathy. *Case Reports in Ophthalmological Medicine*, **2018**, 2018, 7850467	0.7	2
22	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	1.4	2
21	Genotype-Phenotype Correlations in -Associated Retinal Dystrophies: A Multi-Center Cohort Study in JAPAN. *Journal of Clinical Medicine*, **2021**, 10,	5.1	2
20	Progress of macular atrophy during 30 months' follow-up in a patient with spinocerebellar ataxia type1 (SCA1). *Documenta Ophthalmologica*, **2021**, 142, 87-98	2.2	2
19	Genotype and Long-term Clinical Course of Bietti Crystalline Dystrophy in Korean and Japanese Patients. *Ophthalmology Retina*, **2021**, 5, 1269-1279	3.8	2
18	A new PDE6A missense variant p.Arg544Gln in rod-cone dystrophy. *Documenta Ophthalmologica*, **2021**, 143, 107-114	2.2	2

17	Novel use of Finesse Flex loop for macular hole retinal detachment. *American Journal of Ophthalmology Case Reports*, **2020**, 18, 100703	1.3	1
16	Clinical Course and Electron Microscopic Findings in Lymphocytes of Patients with -Associated Retinopathy. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	1
15	Closure of a full-thickness macular hole without vitrectomy in choroideraemia. *Australasian journal of optometry, The*, **2017**, 100, 294-295	2.7	1
14	The second Japanese family with Malattia Leventinese/Doyne honeycomb retinal dystrophy. *Documenta Ophthalmologica*, **2021**, 1	2.2	1
13	The first Japanese family of CDH3-related hypotrichosis with juvenile macular dystrophy. *Molecular Genetics & Genomic Medicine*, **2021**, 9, e1688	2.3	1
12	A novel heterozygous splice site OPA1 mutation causes exon 10 skipping in Japanese patients with dominant optic atrophy. *Ophthalmic Genetics*, **2016**, 37, 354-6	1.2	1
11	Vitamin A deficiency after prolonged intake of an unbalanced diet in a Japanese hemodialysis patient. *Documenta Ophthalmologica*, **2021**, 143, 85-91	2.2	1
10	Homozygous single nucleotide duplication of SLC38A8 in autosomal recessive foveal hypoplasia: The first Japanese case report. *Documenta Ophthalmologica*, **2021**, 143, 323-330	2.2	0
9	Spontaneous Improvement of Visual Acuity in a 13-Year-Old Boy with Neuromyelitis Optica Spectrum Disorder. *Neuro-Ophthalmology*, **2019**, 43, 114-119	0.9	
8	Color vision in an elderly patient with protanopic genotype and successfully treated unilateral age-related macular degeneration. *International Ophthalmology*, **2011**, 31, 471-5	2.2	
7	Cone Dysfunction Syndrome in the Japanese Population. *Essentials in Ophthalmology*, **2017**, 129-135	0.2	
6	OPA1??????????????????????????. *Japanese Orthoptic Journal*, **2010**, 39, 117-122	0	
5	??????????????????????????. *Japanese Orthoptic Journal*, **2010**, 39, 123-128	0	
4	Transient electroretinographic abnormalities that mimic those of KCNV2 retinopathy: a case report. *Documenta Ophthalmologica*, **2021**, 143, 221-228	2.2	
3	Vision Improvement after Osimertinib Treatment in Paraneoplastic Optic Neuropathy Associated with Lung Adenocarcinoma. *Case Reports in Ophthalmological Medicine*, **2021**, 2021, 2832021	0.7	
2	Polypoidal choroidal vasculopathy in a patient with DMPK-associated myotonic dystrophy.. *Documenta Ophthalmologica*, **2022**, 1	2.2	
1	Clinical characterization of autosomal dominant retinitis pigmentosa with NRL mutation in a three-generation Japanese family. *Documenta Ophthalmologica*,	2.2	