Hiroyuki Ishiura

List of Publications by Citations

115 papers	2,201 citations	24 h-index	44 g-index
135 ext. papers	2,874 ext. citations	4.6 avg, IF	4.28 L-index

#	Paper	IF	Citations
115	Human genetic variation database, a reference database of genetic variations in the Japanese population. *Journal of Human Genetics*, **2016**, 61, 547-53	4.3	212
114	Expansions of intronic TTTCA and TTTTA repeats in benign adult familial myoclonic epilepsy. *Nature Genetics*, **2018**, 50, 581-590	36.3	152
113	Noncoding CGG repeat expansions in neuronal intranuclear inclusion disease, oculopharyngodistal myopathy and an overlapping disease. *Nature Genetics*, **2019**, 51, 1222-1232	36.3	132
112	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. *Nature Communications*, **2016**, 7, 11253	17.4	126
111	ERBB4 mutations that disrupt the neuregulin-ErbB4 pathway cause amyotrophic lateral sclerosis type 19. *American Journal of Human Genetics*, **2013**, 93, 900-5	11	95
110	C9ORF72 repeat expansion in amyotrophic lateral sclerosis in the Kii peninsula of Japan. *Archives of Neurology*, **2012**, 69, 1154-8		77
109	Variants associated with Gaucher disease in multiple system atrophy. *Annals of Clinical and Translational Neurology*, **2015**, 2, 417-26	5.3	76
108	The TRK-fused gene is mutated in hereditary motor and sensory neuropathy with proximal dominant involvement. *American Journal of Human Genetics*, **2012**, 91, 320-9	11	76
107	Evaluation of SLC20A2 mutations that cause idiopathic basal ganglia calcification in Japan. *Neurology*, **2014**, 82, 705-12	6.5	61
106	Identification of ATP1A3 mutations by exome sequencing as the cause of alternating hemiplegia of childhood in Japanese patients. *PLoS ONE*, **2013**, 8, e56120	3.7	61
105	A Novel Mutation in ELOVL4 Leading to Spinocerebellar Ataxia (SCA) With the Hot Cross Bun Sign but Lacking Erythrokeratodermia: A Broadened Spectrum of SCA34. *JAMA Neurology*, **2015**, 72, 797-805	17.2	53
104	Mutations in MME cause an autosomal-recessive Charcot-Marie-Tooth disease type 2. *Annals of Neurology*, **2016**, 79, 659-72	9.4	53
103	Rapid detection of expanded short tandem repeats in personal genomics using hybrid sequencing. *Bioinformatics*, **2014**, 30, 815-22	7.2	50
102	A recurrent de novo FAM111A mutation causes Kenny-Caffey syndrome type 2. *Journal of Bone and Mineral Research*, **2014**, 29, 992-8	6.3	49
101	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. *Nature Communications*, **2019**, 10, 4920	17.4	48
100	Intronic pentanucleotide TTTCA repeat insertion in the SAMD12 gene causes familial cortical myoclonic tremor with epilepsy type 1. *Brain*, **2018**, 141, 2280-2288	11.2	45
99	Molecular epidemiology and clinical spectrum of hereditary spastic paraplegia in the Japanese population based on comprehensive mutational analyses. *Journal of Human Genetics*, **2014**, 59, 163-72	4.3	38

98	Development of a high-throughput microarray-based resequencing system for neurological disorders and its application to molecular genetics of amyotrophic lateral sclerosis. *Archives of Neurology*, **2008**, 65, 1326-32		37
97	Posterior column ataxia with retinitis pigmentosa in a Japanese family with a novel mutation in FLVCR1. *Neurogenetics*, **2011**, 12, 117-21	3	31
96	The pathogenesis linked to coenzyme Q10 insufficiency in iPSC-derived neurons from patients with multiple-system atrophy. *Scientific Reports*, **2018**, 8, 14215	4.9	30
95	CSF1R mutations identified in three families with autosomal dominantly inherited leukoencephalopathy. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2012**, 159B, 951-7	3.5	29
94	Lymphomatoid granulomatosis involving central nervous system successfully treated with rituximab alone. *Archives of Neurology*, **2008**, 65, 662-5		29
93	Mutations in COA7 cause spinocerebellar ataxia with axonal neuropathy. *Brain*, **2018**, 141, 1622-1636	11.2	27
92	Hypertrophic pachymeningitis and tracheobronchial stenosis in IgG4-related disease: case presentation and literature review. *Internal Medicine*, **2012**, 51, 935-41	1.1	27
91	Three-Year Follow-Up of High-Dose Ubiquinol Supplementation in a Case of Familial Multiple System Atrophy with Compound Heterozygous COQ2 Mutations. *Cerebellum*, **2017**, 16, 664-672	4.3	24
90	Plasma Coenzyme Q10 Levels in Patients With Multiple System Atrophy. *JAMA Neurology*, **2016**, 73, 977-80	17.2	24
89	JASPAC: Japan Spastic Paraplegia Research Consortium. *Brain Sciences*, **2018**, 8,	3.4	23
88	Molecular epidemiological study of familial amyotrophic lateral sclerosis in Japanese population by whole-exome sequencing and identification of novel HNRNPA1 mutation. *Neurobiology of Aging*, **2018**, 61, 255.e9-255.e16	5.6	22
87	AgIn: measuring the landscape of CpG methylation of individual repetitive elements. *Bioinformatics*, **2016**, 32, 2911-9	7.2	22
86	Clinical and genetic diversities of Charcot-Marie-Tooth disease with MFN2 mutations in a large case study. *Journal of the Peripheral Nervous System*, **2017**, 22, 191-199	4.7	19
85	Autosomal-recessive complicated spastic paraplegia with a novel lysosomal trafficking regulator gene mutation. *Journal of Neurology, Neurosurgery and Psychiatry*, **2014**, 85, 1024-8	5.5	17
84	Ataxic phenotype with altered Ca3.1 channel property in a mouse model for spinocerebellar ataxia 42. *Neurobiology of Disease*, **2019**, 130, 104516	7.5	15
83	Modeling neurological diseases with induced pluripotent cells reprogrammed from immortalized lymphoblastoid cell lines. *Molecular Brain*, **2016**, 9, 88	4.5	15
82	Burden of rare variants in causative genes for amyotrophic lateral sclerosis (ALS) accelerates age at onset of ALS. *Journal of Neurology, Neurosurgery and Psychiatry*, **2019**, 90, 537-542	5.5	15
81	Epidemiology and molecular mechanism of frontotemporal lobar degeneration/amyotrophic lateral sclerosis with repeat expansion mutation in C9orf72. *Journal of Neurogenetics*, **2015**, 29, 85-94	1.6	14

80	Structural Basis and Genotype-Phenotype Correlations of INSR Mutations Causing Severe Insulin Resistance. *Diabetes*, **2017**, 66, 2713-2723	0.9	14
79	Clinicopathologic features of myositis patients with CD8-MHC-1 complex pathology. *Neurology*, **2017**, 89, 1060-1068	6.5	14
78	VPS13D-related disorders presenting as a pure and complicated form of hereditary spastic paraplegia. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1108	2.3	14
77	The novel mutation of gene as the cause for Spastic paraplegia 30 in a Japanese case. *ENeurologicalSci*, **2019**, 14, 34-37	2.1	14
76	Identification of candidate genes involved in the etiology of sporadic Tourette syndrome by exome sequencing. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2017**, 174, 712-723	3.5	13
75	An Autopsy Case of Familial Neuronal Intranuclear Inclusion Disease with Dementia and Neuropathy. *Internal Medicine*, **2018**, 57, 3459-3462	1.1	13
74	Clinical features and haplotype analysis of newly identified Japanese patients with gelsolin-related familial amyloidosis of Finnish type. *Neurogenetics*, **2012**, 13, 237-43	3	13
73	Fukutin gene mutations that cause left ventricular noncompaction. *International Journal of Cardiology*, **2016**, 222, 727-729	3.2	13
72	Novel mutations in the ALDH18A1 gene in complicated hereditary spastic paraplegia with cerebellar ataxia and cognitive impairment. *Journal of Human Genetics*, **2018**, 63, 1009-1013	4.3	12
71	Novel mutations in the PNPLA6 gene in Boucher-Neuh□ser syndrome. *Journal of Human Genetics*, **2015**, 60, 217-20	4.3	12
70	TBCD may be a causal gene in progressive neurodegenerative encephalopathy with atypical infantile spinal muscular atrophy. *Journal of Human Genetics*, **2017**, 62, 473-480	4.3	11
69	Novel De Novo KCND3 Mutation in a Japanese Patient with Intellectual Disability, Cerebellar Ataxia, Myoclonus, and Dystonia. *Cerebellum*, **2018**, 17, 237-242	4.3	11
68	Advances in repeat expansion diseases and a new concept of repeat motif-phenotype correlation. *Current Opinion in Genetics and Development*, **2020**, 65, 176-185	4.9	11
67	PLA2G6-associated neurodegeneration presenting as a complicated form of hereditary spastic paraplegia. *Journal of Human Genetics*, **2019**, 64, 55-59	4.3	11
66	Low Prevalence of NOTCH2NLC GGC Repeat Expansion in White Patients with Movement Disorders. *Movement Disorders*, **2021**, 36, 251-255	7	11
65	Partial duplication of causes minifascicular neuropathy: A novel mutation detection of. *Annals of Clinical and Translational Neurology*, **2017**, 4, 415-421	5.3	10
64	Atypical parkinsonism caused by Pro105Leu mutation of prion protein: A broad clinical spectrum. *Neurology: Genetics*, **2016**, 2, e48	3.8	10
63	UBAP1 mutations cause juvenile-onset hereditary spastic paraplegias (SPG80) and impair UBAP1 targeting to endosomes. *Journal of Human Genetics*, **2019**, 64, 1055-1065	4.3	9

62	Slowly progressive d-bifunctional protein deficiency with survival to adulthood diagnosed by whole-exome sequencing. *Journal of the Neurological Sciences*, **2017**, 372, 6-10	3.2	8
61	Clinical and genetic features of Charcot-Marie-Tooth disease 2F and hereditary motor neuropathy 2B in Japan. *Journal of the Peripheral Nervous System*, **2018**, 23, 40-48	4.7	8
60	Frequency and characteristics of the TBK1 gene variants in Japanese patients with sporadic amyotrophic lateral sclerosis. *Neurobiology of Aging*, **2018**, 64, 158.e15-158.e19	5.6	8
59	A Novel de novo KIF1A Mutation in a Patient with Autism, Hyperactivity, Epilepsy, Sensory Disturbance, and Spastic Paraplegia. *Internal Medicine*, **2020**, 59, 839-842	1.1	8
58	Clinical efficacy of haematopoietic stem cell transplantation for adult adrenoleukodystrophy. *Brain Communications*, **2020**, 2, fcz048	4.5	8
57	Ataxic form of autosomal recessive PEX10-related peroxisome biogenesis disorders with a novel compound heterozygous gene mutation and characteristic clinical phenotype. *Journal of the Neurological Sciences*, **2017**, 375, 424-429	3.2	7
56	Adult-onset neuronal intranuclear inclusion disease mimicking Fragile X-associated tremor-ataxia syndrome in ethnic Chinese patients. *Parkinsonism and Related Disorders*, **2020**, 74, 25-27	3.6	7
55	Identification of a novel mutation in associated with a complicated form of hereditary spastic paraplegia. *Neurology: Genetics*, **2020**, 6, e514	3.8	7
54	Clinical Characteristics of Neuronal Intranuclear Inclusion Disease-Related Retinopathy With CGG Repeat Expansions in the NOTCH2NLC Gene **2020**, 61, 27		7
53	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. *Brain*, **2021**, 144, 1422-1434	11.2	7
52	A Homozygous LAMA2 Mutation of c.818G>A Caused Partial Merosin Deficiency in a Japanese Patient. *Internal Medicine*, **2018**, 57, 877-882	1.1	6
51	Genetic spectrum of Charcot-Marie-Tooth disease associated with myelin protein zero gene variants in Japan. *Clinical Genetics*, **2021**, 99, 359-375	4	6
50	Functional evaluation of PDGFB-variants in idiopathic basal ganglia calcification, using patient-derived iPS cells. *Scientific Reports*, **2019**, 9, 5698	4.9	5
49	Comprehensive investigation of RNF213 nonsynonymous variants associated with intracranial artery stenosis. *Scientific Reports*, **2020**, 10, 11942	4.9	5
48	Randomized phase 2 study of perampanel for sporadic amyotrophic lateral sclerosis. *Journal of Neurology*, **2021**, 1	5.5	5
47	Exome sequencing reveals a novel missense mutation in the KIAA0196 gene in a Japanese patient with SPG8. *Clinical Neurology and Neurosurgery*, **2016**, 144, 36-8	2	5
46	Juvenile amyotrophic lateral sclerosis with complex phenotypes associated with novel mutations. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2021**, 22, 576-578	3.6	4
45	Neuroimaging, genetic, and enzymatic study in a Japanese family with a GBA gross deletion. *Parkinsonism and Related Disorders*, **2019**, 61, 57-63	3.6	4

44	Cranial Nerve Involvement and Dysautonomia in Post-COVID-19 Guillain-Barr Syndrome. *Internal Medicine*, **2021**, 60, 3477-3480	1.1	4
43	A novel mutation in the gene in a Japanese patient with SPG46: A case report. *ENeurologicalSci*, **2020**, 19, 100238	2.1	3
42	Familial dementia with Lewy bodies with VPS13C mutations. *Parkinsonism and Related Disorders*, **2020**, 81, 31-33	3.6	3
41	Selective impairment of On-reading (Chinese-style pronunciation) in alexia with agraphia for kanji due to subcortical hemorrhage in the left posterior middle temporal gyrus. *Neurocase*, **2020**, 26, 220-226	0.8	3
40	A novel homozygous mutation of the TFG gene in a patient with early onset spastic paraplegia and later onset sensorimotor polyneuropathy. *Journal of Human Genetics*, **2019**, 64, 171-176	4.3	3
39	Loss-of-function variants in NEK1 are associated with an increased risk of sporadic ALS in the Japanese population. *Journal of Human Genetics*, **2021**, 66, 237-241	4.3	3
38	HIV Dementia with a Decreased Cardiac I-metaiodobenzylguanidine Uptake Masquerading as Dementia with Lewy Bodies. *Internal Medicine*, **2018**, 57, 3007-3010	1.1	3
37	Exome sequencing shows a novel de novo mutation in ATL1. *Neurology and Clinical Neuroscience*, **2014**, 2, 1-4	0.3	2
36	Exome analysis reveals a Japanese family with spinocerebellar ataxia, autosomal recessive 1. *Journal of the Neurological Sciences*, **2013**, 331, 158-60	3.2	2
35	A Japanese family with primary familial brain calcification presenting with paroxysmal kinesigenic dyskinesia - A comprehensive mutational analysis. *Journal of the Neurological Sciences*, **2020**, 418, 117091	3.2	2
34	Clinical Impact of Copy Number Variation on the Genetic Diagnosis of Syndromic Aortopathies. *Circulation Genomic and Precision Medicine*, **2021**, 14, e003458	5.2	2
33	COQ2 V393A confers high risk susceptibility for multiple system atrophy in East Asian population. *Journal of the Neurological Sciences*, **2021**, 429, 117623	3.2	2
32	Association of ATXN2 intermediate-length CAG repeats with amyotrophic lateral sclerosis correlates with the distributions of normal CAG repeat alleles among individual ethnic populations. *Neurogenetics*, **2019**, 20, 65-71	3	1
31	Clinical features of inherited neuropathy with BSCL2 mutations in Japan. *Journal of the Peripheral Nervous System*, **2020**, 25, 125-131	4.7	1
30	No novel, high penetrant gene might remain to be found in Japanese patients with unknown MODY. *Journal of Human Genetics*, **2018**, 63, 821-829	4.3	1
29	Frequency of FMR1 Premutation Alleles in Patients with Undiagnosed Cerebellar Ataxia and Multiple System Atrophy in the Japanese Population. *Cerebellum*, **2021**, 1	4.3	1
28	A Role of Aging in the Progression of Cortical Excitability in Benign Adult Familial Myoclonus Epilepsy type 1 Patients. *Movement Disorders*, **2021**, 36, 2446-2448	7	1
27	Splice-site mutations in KIF5A in the Japanese case series of amyotrophic lateral sclerosis. *Neurogenetics*, **2021**, 22, 11-17	3	1

26	An autopsy case of G gangliosidosis type II in a patient who survived a long duration with artificial respiratory support. *Neuropathology*, **2020**, 40, 379-388	2	0
25	A case of late-onset Krabbe disease which showed subacute progression of spastic paresis with bilateral spinal cord lesions. *Neurology and Clinical Neuroscience*, **2018**, 6, 104-106	0.3	0
24	An immigrant family with Kii amyotrophic lateral sclerosis/parkinsonism-dementia complex. *Neurological Sciences*, **2021**, 43, 1423	3.5	0
23	Adrenoleukodystrophy siblings with a novel ABCD1 missense variant presenting with phenotypic differences: a case report and literature review. *Journal of Human Genetics*, **2021**, 66, 535-537	4.3	0
22	SPG9A with the new occurrence of an ALDH18A1 mutation in a CMT1A family with PMP22 duplication: case report. *BMC Neurology*, **2021**, 21, 64	3.1	0
21	Reliability and validity of Japanese version of Unified Multiple System Atrophy Rating Scale. *Neurology and Clinical Neuroscience*, **2021**, 9, 171-180	0.3	0
20	DMD exon 2 duplication due to a complex genomic rearrangement is associated with a somatic mosaicism.. *Neuromuscular Disorders*, **2021**,	2.9	0
19	Nonsyndromic arteriopathy and aortopathy and vascular Ehlers-Danlos syndrome causing COL3A1 variants.. *American Journal of Medical Genetics, Part A*, **2022**,	2.5	0
18	TDP-43 Proteinopathy Presenting with Typical Symptoms of Parkinson Disease.. *Movement Disorders*, **2022**,	7	0
17	Authors response to "Compound heterozygous Fukutin mutation-related non-compaction" by Finsterer and Zarrouk-Mahjoub. *International Journal of Cardiology*, **2017**, 233, 102	3.2	
16	Novel variant of CSF1R in sporadic case with early-onset cognitive impairment. *Neurology and Clinical Neuroscience*, **2020**, 8, 430-432	0.3	
15	A novel mutation in ABCD1 gene in a Filipino patient with adult-onset X-linked ALD. *Neurology and Clinical Neuroscience*, **2020**, 8, 329-331	0.3	
14	Authors reply to the Drs. Finsterer and Zarrouk-Mahjoub comments for our case report. *International Journal of Cardiology*, **2018**, 254, 262	3.2	
13	Prominent Spasticity and Hyperreflexia of the Legs in a Nepalese Patient with Friedreich Ataxia. *Internal Medicine*, **2019**, 58, 2865-2869	1.1	
12	Sporadic progressive myoclonic epilepsy with early-onset dementia caused by a de novo mutation in PSEN1. *Neurology and Clinical Neuroscience*, **2019**, 7, 294-296	0.3	
11	Novel mutation in the membrane metalloendopeptidase gene in a patient with the autosomal recessive form of Charcot-Marie-Tooth disease. *Neurology and Clinical Neuroscience*, **2017**, 5, 124-126	0.3	
10	Family with congenital contractural arachnodactyly due to a novel multiexon deletion of the gene.. *Clinical Case Reports (discontinued)*, **2022**, 10, e05335	0.7	
9	DNA sequencing and other methods of exonic and genomic analyses **2020**, 109-120		

8	A novel multi-exon deletion in the dysferlin gene of a limb-girdle muscular dystrophy type 2B Filipino patient. *Neurology and Clinical Neuroscience*, **2020**, 8, 419-421	0.3
7	Do eye movements "age" earlier in progeria?. *Clinical Neurophysiology*, **2020**, 131, 1835-1836	4.3
6	A Case of Irreversible Corneal Edema Associated with Dentatorubropallidoluysian Atrophy Following Corneal Endothelial Transplantation. *SN Comprehensive Clinical Medicine*, **2021**, 3, 2029-2032	2.7
5	Novel COL6A2 mutation in a case of limb girdle muscular dystrophy phenotype with autosomal recessive inheritance. *Neurology and Clinical Neuroscience*, **2016**, 4, 189-191	0.3
4	Severe dilated cardiomyopathy and ventricular arrhythmia in a patient with Emery-Dreifuss muscular dystrophy harboring a novel frameshift mutation in EMD. *Neurology and Clinical Neuroscience*, **2021**, 9, 490	0.3
3	Cerebellar Ataxia as a Common Clinical Presentation Associated with DNMT1 p.Y511H and a Review of the Literature. *Journal of Molecular Neuroscience*, **2021**, 71, 1796-1801	3.3
2	Complex hereditary peripheral neuropathies caused by novel variants in mitochondrial-related nuclear genes.. *Journal of Neurology*, **2022**, 1	5.5
1	Efficacy of canakinumab on AA amyloidosis in late-onset NLRP3-associated autoinflammatory disease with an I574F somatic mosaic mutation.. *Clinical Rheumatology*, **2022**, 1	3.9