

Hiroyuki Ishiura

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.

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	Family with congenital contractural arachnodactyly due to a novel multiexon deletion of the gene.. <i>Clinical Case Reports (discontinued)</i> , 2022 , 10, e05335	0.7	
114	Complex hereditary peripheral neuropathies caused by novel variants in mitochondrial-related nuclear genes.. <i>Journal of Neurology</i> , 2022 , 1	5.5	
113	Efficacy of canakinumab on AA amyloidosis in late-onset NLRP3-associated autoinflammatory disease with an I574F somatic mosaic mutation.. <i>Clinical Rheumatology</i> , 2022 , 1	3.9	
112	Nonsyndromic arteriopathy and aortopathy and vascular Ehlers-Danlos syndrome causing COL3A1 variants.. <i>American Journal of Medical Genetics, Part A</i> , 2022 ,	2.5	0
111	TDP-43 Proteinopathy Presenting with Typical Symptoms of Parkinson Disease.. <i>Movement Disorders</i> , 2022 ,	7	0
110	An immigrant family with Kii amyotrophic lateral sclerosis/parkinsonism-dementia complex. <i>Neurological Sciences</i> , 2021 , 43, 1423	3.5	0
109	Frequency of FMR1 Premutation Alleles in Patients with Undiagnosed Cerebellar Ataxia and Multiple System Atrophy in the Japanese Population. <i>Cerebellum</i> , 2021 , 1	4.3	1
108	Juvenile amyotrophic lateral sclerosis with complex phenotypes associated with novel mutations. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2021 , 22, 576-578	3.6	4
107	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. <i>Brain</i> , 2021 , 144, 1422-1434	11.2	7
106	Randomized phase 2 study of perampanel for sporadic amyotrophic lateral sclerosis. <i>Journal of Neurology</i> , 2021 , 1	5.5	5
105	A Case of Irreversible Corneal Edema Associated with Dentatorubropallidoluysian Atrophy Following Corneal Endothelial Transplantation. <i>SN Comprehensive Clinical Medicine</i> , 2021 , 3, 2029-2032	2.7	
104	A Role of Aging in the Progression of Cortical Excitability in Benign Adult Familial Myoclonus Epilepsy type 1 Patients. <i>Movement Disorders</i> , 2021 , 36, 2446-2448	7	1
103	Low Prevalence of NOTCH2NLC GGC Repeat Expansion in White Patients with Movement Disorders. <i>Movement Disorders</i> , 2021 , 36, 251-255	7	11
102	Genetic spectrum of Charcot-Marie-Tooth disease associated with myelin protein zero gene variants in Japan. <i>Clinical Genetics</i> , 2021 , 99, 359-375	4	6
101	Adrenoleukodystrophy siblings with a novel ABCD1 missense variant presenting with phenotypic differences: a case report and literature review. <i>Journal of Human Genetics</i> , 2021 , 66, 535-537	4.3	0
100	Splice-site mutations in KIF5A in the Japanese case series of amyotrophic lateral sclerosis. <i>Neurogenetics</i> , 2021 , 22, 11-17	3	1
99	Loss-of-function variants in NEK1 are associated with an increased risk of sporadic ALS in the Japanese population. <i>Journal of Human Genetics</i> , 2021 , 66, 237-241	4.3	3

98	Cranial Nerve Involvement and Dysautonomia in Post-COVID-19 Guillain-Barré Syndrome. <i>Internal Medicine</i> , 2021 , 60, 3477-3480	1.1	4
97	SPG9A with the new occurrence of an ALDH18A1 mutation in a CMT1A family with PMP22 duplication: case report. <i>BMC Neurology</i> , 2021 , 21, 64	3.1	0
96	Clinical Impact of Copy Number Variation on the Genetic Diagnosis of Syndromic Aortopathies. <i>Circulation Genomic and Precision Medicine</i> , 2021 , 14, e003458	5.2	2
95	Severe dilated cardiomyopathy and ventricular arrhythmia in a patient with Emery-Dreifuss muscular dystrophy harboring a novel frameshift mutation in EMD. <i>Neurology and Clinical Neuroscience</i> , 2021 , 9, 490	0.3	
94	COQ2 V393A confers high risk susceptibility for multiple system atrophy in East Asian population. <i>Journal of the Neurological Sciences</i> , 2021 , 429, 117623	3.2	2
93	Reliability and validity of Japanese version of Unified Multiple System Atrophy Rating Scale. <i>Neurology and Clinical Neuroscience</i> , 2021 , 9, 171-180	0.3	0
92	Cerebellar Ataxia as a Common Clinical Presentation Associated with DNMT1 p.Y511H and a Review of the Literature. <i>Journal of Molecular Neuroscience</i> , 2021 , 71, 1796-1801	3.3	
91	DMD exon 2 duplication due to a complex genomic rearrangement is associated with a somatic mosaicism.. <i>Neuromuscular Disorders</i> , 2021 ,	2.9	0
90	Novel variant of CSF1R in sporadic case with early-onset cognitive impairment. <i>Neurology and Clinical Neuroscience</i> , 2020 , 8, 430-432	0.3	
89	An autopsy case of G gangliosidosis type II in a patient who survived a long duration with artificial respiratory support. <i>Neuropathology</i> , 2020 , 40, 379-388	2	0
88	A novel mutation in ABCD1 gene in a Filipino patient with adult-onset X-linked ALD. <i>Neurology and Clinical Neuroscience</i> , 2020 , 8, 329-331	0.3	
87	Clinical features of inherited neuropathy with BSCL2 mutations in Japan. <i>Journal of the Peripheral Nervous System</i> , 2020 , 25, 125-131	4.7	1
86	A novel mutation in the gene in a Japanese patient with SPG46: A case report. <i>ENeurologicalSci</i> , 2020 , 19, 100238	2.1	3
85	Adult-onset neuronal intranuclear inclusion disease mimicking Fragile X-associated tremor-ataxia syndrome in ethnic Chinese patients. <i>Parkinsonism and Related Disorders</i> , 2020 , 74, 25-27	3.6	7
84	DNA sequencing and other methods of exonic and genomic analyses 2020 , 109-120		
83	VPS13D-related disorders presenting as a pure and complicated form of hereditary spastic paraplegia. <i>Molecular Genetics & Genomic Medicine</i> , 2020 , 8, e1108	2.3	14
82	A Novel de novo KIF1A Mutation in a Patient with Autism, Hyperactivity, Epilepsy, Sensory Disturbance, and Spastic Paraplegia. <i>Internal Medicine</i> , 2020 , 59, 839-842	1.1	8
81	Familial dementia with Lewy bodies with VPS13C mutations. <i>Parkinsonism and Related Disorders</i> , 2020 , 81, 31-33	3.6	3

80	A novel multi-exon deletion in the dysferlin gene of a limb-girdle muscular dystrophy type 2B Filipino patient. <i>Neurology and Clinical Neuroscience</i> , 2020 , 8, 419-421	0.3	
79	Identification of a novel mutation in associated with a complicated form of hereditary spastic paraplegia. <i>Neurology: Genetics</i> , 2020 , 6, e514	3.8	7
78	A Japanese family with primary familial brain calcification presenting with paroxysmal kinesigenic dyskinesia - A comprehensive mutational analysis. <i>Journal of the Neurological Sciences</i> , 2020 , 418, 117091	3.2	2
77	Advances in repeat expansion diseases and a new concept of repeat motif-phenotype correlation. <i>Current Opinion in Genetics and Development</i> , 2020 , 65, 176-185	4.9	11
76	Clinical efficacy of haematopoietic stem cell transplantation for adult adrenoleukodystrophy. <i>Brain Communications</i> , 2020 , 2, fcz048	4.5	8
75	Do eye movements "age" earlier in progeria?. <i>Clinical Neurophysiology</i> , 2020 , 131, 1835-1836	4.3	
74	Comprehensive investigation of RNF213 nonsynonymous variants associated with intracranial artery stenosis. <i>Scientific Reports</i> , 2020 , 10, 11942	4.9	5
73	Clinical Characteristics of Neuronal Intranuclear Inclusion Disease-Related Retinopathy With CGG Repeat Expansions in the NOTCH2NLC Gene 2020 , 61, 27		7
72	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. <i>Neurocase</i> , 2020 , 26, 220-226	0.8	3
71	UBAP1 mutations cause juvenile-onset hereditary spastic paraplegias (SPG80) and impair UBAP1 targeting to endosomes. <i>Journal of Human Genetics</i> , 2019 , 64, 1055-1065	4.3	9
70	Ataxic phenotype with altered Ca _v 3.1 channel property in a mouse model for spinocerebellar ataxia 42. <i>Neurobiology of Disease</i> , 2019 , 130, 104516	7.5	15
69	Functional evaluation of PDGFB-variants in idiopathic basal ganglia calcification, using patient-derived iPSC cells. <i>Scientific Reports</i> , 2019 , 9, 5698	4.9	5
68	Association of ATXN2 intermediate-length CAG repeats with amyotrophic lateral sclerosis correlates with the distributions of normal CAG repeat alleles among individual ethnic populations. <i>Neurogenetics</i> , 2019 , 20, 65-71	3	1
67	Prominent Spasticity and Hyperreflexia of the Legs in a Nepalese Patient with Friedreich Ataxia. <i>Internal Medicine</i> , 2019 , 58, 2865-2869	1.1	
66	Noncoding CGG repeat expansions in neuronal intranuclear inclusion disease, oculopharyngodistal myopathy and an overlapping disease. <i>Nature Genetics</i> , 2019 , 51, 1222-1232	36.3	132
65	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. <i>Nature Communications</i> , 2019 , 10, 4920	17.4	48
64	Sporadic progressive myoclonic epilepsy with early-onset dementia caused by a de novo mutation in PSEN1. <i>Neurology and Clinical Neuroscience</i> , 2019 , 7, 294-296	0.3	
63	Neuroimaging, genetic, and enzymatic study in a Japanese family with a GBA gross deletion. <i>Parkinsonism and Related Disorders</i> , 2019 , 61, 57-63	3.6	4

62	The novel mutation of gene as the cause for Spastic paraplegia 30 in a Japanese case. <i>ENeurologicalSci</i> , 2019 , 14, 34-37	2.1	14
61	A novel homozygous mutation of the TFG gene in a patient with early onset spastic paraplegia and later onset sensorimotor polyneuropathy. <i>Journal of Human Genetics</i> , 2019 , 64, 171-176	4.3	3
60	Burden of rare variants in causative genes for amyotrophic lateral sclerosis (ALS) accelerates age at onset of ALS. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2019 , 90, 537-542	5.5	15
59	PLA2G6-associated neurodegeneration presenting as a complicated form of hereditary spastic paraplegia. <i>Journal of Human Genetics</i> , 2019 , 64, 55-59	4.3	11
58	Expansions of intronic TTTCA and TTTTA repeats in benign adult familial myoclonic epilepsy. <i>Nature Genetics</i> , 2018 , 50, 581-590	36.3	152
57	No novel, high penetrant gene might remain to be found in Japanese patients with unknown MODY. <i>Journal of Human Genetics</i> , 2018 , 63, 821-829	4.3	1
56	Clinical and genetic features of Charcot-Marie-Tooth disease 2F and hereditary motor neuropathy 2B in Japan. <i>Journal of the Peripheral Nervous System</i> , 2018 , 23, 40-48	4.7	8
55	Frequency and characteristics of the TBK1 gene variants in Japanese patients with sporadic amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2018 , 64, 158.e15-158.e19	5.6	8
54	Mutations in COA7 cause spinocerebellar ataxia with axonal neuropathy. <i>Brain</i> , 2018 , 141, 1622-1636	11.2	27
53	A Homozygous LAMA2 Mutation of c.818G>A Caused Partial Merosin Deficiency in a Japanese Patient. <i>Internal Medicine</i> , 2018 , 57, 877-882	1.1	6
52	Authors Reply to the Drs. Finsterer and Zarrouk-Mahjoub comments for our case report. <i>International Journal of Cardiology</i> , 2018 , 254, 262	3.2	
51	Molecular epidemiological study of familial amyotrophic lateral sclerosis in Japanese population by whole-exome sequencing and identification of novel HNRNPA1 mutation. <i>Neurobiology of Aging</i> , 2018 , 61, 255.e9-255.e16	5.6	22
50	Novel De Novo KCND3 Mutation in a Japanese Patient with Intellectual Disability, Cerebellar Ataxia, Myoclonus, and Dystonia. <i>Cerebellum</i> , 2018 , 17, 237-242	4.3	11
49	Intronic pentanucleotide TTTCA repeat insertion in the SAMD12 gene causes familial cortical myoclonic tremor with epilepsy type 1. <i>Brain</i> , 2018 , 141, 2280-2288	11.2	45
48	A case of late-onset Krabbe disease which showed subacute progression of spastic paresis with bilateral spinal cord lesions. <i>Neurology and Clinical Neuroscience</i> , 2018 , 6, 104-106	0.3	0
47	JASPAC: Japan Spastic Paraplegia Research Consortium. <i>Brain Sciences</i> , 2018 , 8,	3.4	23
46	An Autopsy Case of Familial Neuronal Intranuclear Inclusion Disease with Dementia and Neuropathy. <i>Internal Medicine</i> , 2018 , 57, 3459-3462	1.1	13
45	Novel mutations in the ALDH18A1 gene in complicated hereditary spastic paraplegia with cerebellar ataxia and cognitive impairment. <i>Journal of Human Genetics</i> , 2018 , 63, 1009-1013	4.3	12

44	The pathogenesis linked to coenzyme Q10 insufficiency in iPSC-derived neurons from patients with multiple-system atrophy. <i>Scientific Reports</i> , 2018 , 8, 14215	4.9	30
43	HIV Dementia with a Decreased Cardiac I-metaiodobenzylguanidine Uptake Masquerading as Dementia with Lewy Bodies. <i>Internal Medicine</i> , 2018 , 57, 3007-3010	1.1	3
42	Identification of candidate genes involved in the etiology of sporadic Tourette syndrome by exome sequencing. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2017 , 174, 712-723	3.5	13
41	Authors' response to "Compound heterozygous Fukutin mutation-related non-compactation" by Finsterer and Zarrouk-Mahjoub. <i>International Journal of Cardiology</i> , 2017 , 233, 102	3.2	
40	Ataxic form of autosomal recessive PEX10-related peroxisome biogenesis disorders with a novel compound heterozygous gene mutation and characteristic clinical phenotype. <i>Journal of the Neurological Sciences</i> , 2017 , 375, 424-429	3.2	7
39	Slowly progressive d-bifunctional protein deficiency with survival to adulthood diagnosed by whole-exome sequencing. <i>Journal of the Neurological Sciences</i> , 2017 , 372, 6-10	3.2	8
38	TBCD may be a causal gene in progressive neurodegenerative encephalopathy with atypical infantile spinal muscular atrophy. <i>Journal of Human Genetics</i> , 2017 , 62, 473-480	4.3	11
37	Novel mutation in the membrane metalloendopeptidase gene in a patient with the autosomal recessive form of Charcot-Marie-Tooth disease. <i>Neurology and Clinical Neuroscience</i> , 2017 , 5, 124-126	0.3	
36	Structural Basis and Genotype-Phenotype Correlations of INSR Mutations Causing Severe Insulin Resistance. <i>Diabetes</i> , 2017 , 66, 2713-2723	0.9	14
35	Clinicopathologic features of myositis patients with CD8-MHC-1 complex pathology. <i>Neurology</i> , 2017 , 89, 1060-1068	6.5	14
34	Partial duplication of causes minifascicular neuropathy: A novel mutation detection of. <i>Annals of Clinical and Translational Neurology</i> , 2017 , 4, 415-421	5.3	10
33	Three-Year Follow-Up of High-Dose Ubiquinol Supplementation in a Case of Familial Multiple System Atrophy with Compound Heterozygous COQ2 Mutations. <i>Cerebellum</i> , 2017 , 16, 664-672	4.3	24
32	Clinical and genetic diversities of Charcot-Marie-Tooth disease with MFN2 mutations in a large case study. <i>Journal of the Peripheral Nervous System</i> , 2017 , 22, 191-199	4.7	19
31	Human genetic variation database, a reference database of genetic variations in the Japanese population. <i>Journal of Human Genetics</i> , 2016 , 61, 547-53	4.3	212
30	Modeling neurological diseases with induced pluripotent cells reprogrammed from immortalized lymphoblastoid cell lines. <i>Molecular Brain</i> , 2016 , 9, 88	4.5	15
29	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. <i>Nature Communications</i> , 2016 , 7, 11253	17.4	126
28	Plasma Coenzyme Q10 Levels in Patients With Multiple System Atrophy. <i>JAMA Neurology</i> , 2016 , 73, 977-802	8.2	24
27	Exome sequencing reveals a novel missense mutation in the KIAA0196 gene in a Japanese patient with SPG8. <i>Clinical Neurology and Neurosurgery</i> , 2016 , 144, 36-8	2	5

26	Atypical parkinsonism caused by Pro105Leu mutation of prion protein: A broad clinical spectrum. <i>Neurology: Genetics</i> , 2016 , 2, e48	3.8	10
25	AgIn: measuring the landscape of CpG methylation of individual repetitive elements. <i>Bioinformatics</i> , 2016 , 32, 2911-9	7.2	22
24	Fukutin gene mutations that cause left ventricular noncompaction. <i>International Journal of Cardiology</i> , 2016 , 222, 727-729	3.2	13
23	Mutations in MME cause an autosomal-recessive Charcot-Marie-Tooth disease type 2. <i>Annals of Neurology</i> , 2016 , 79, 659-72	9.4	53
22	Novel COL6A2 mutation in a case of limb girdle muscular dystrophy phenotype with autosomal recessive inheritance. <i>Neurology and Clinical Neuroscience</i> , 2016 , 4, 189-191	0.3	
21	A Novel Mutation in ELOVL4 Leading to Spinocerebellar Ataxia (SCA) With the Hot Cross Bun Sign but Lacking Erythrokeratodermia: A Broadened Spectrum of SCA34. <i>JAMA Neurology</i> , 2015 , 72, 797-805	17.2	53
20	Variants associated with Gaucher disease in multiple system atrophy. <i>Annals of Clinical and Translational Neurology</i> , 2015 , 2, 417-26	5.3	76
19	Epidemiology and molecular mechanism of frontotemporal lobar degeneration/amyotrophic lateral sclerosis with repeat expansion mutation in C9orf72. <i>Journal of Neurogenetics</i> , 2015 , 29, 85-94	1.6	14
18	Novel mutations in the PNPLA6 gene in Boucher-Neuhuser syndrome. <i>Journal of Human Genetics</i> , 2015 , 60, 217-20	4.3	12
17	Exome sequencing shows a novel de novo mutation in ATL1. <i>Neurology and Clinical Neuroscience</i> , 2014 , 2, 1-4	0.3	2
16	Molecular epidemiology and clinical spectrum of hereditary spastic paraplegia in the Japanese population based on comprehensive mutational analyses. <i>Journal of Human Genetics</i> , 2014 , 59, 163-72	4.3	38
15	Autosomal-recessive complicated spastic paraplegia with a novel lysosomal trafficking regulator gene mutation. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014 , 85, 1024-8	5.5	17
14	Rapid detection of expanded short tandem repeats in personal genomics using hybrid sequencing. <i>Bioinformatics</i> , 2014 , 30, 815-22	7.2	50
13	Evaluation of SLC20A2 mutations that cause idiopathic basal ganglia calcification in Japan. <i>Neurology</i> , 2014 , 82, 705-12	6.5	61
12	A recurrent de novo FAM111A mutation causes Kenny-Caffey syndrome type 2. <i>Journal of Bone and Mineral Research</i> , 2014 , 29, 992-8	6.3	49
11	Exome analysis reveals a Japanese family with spinocerebellar ataxia, autosomal recessive 1. <i>Journal of the Neurological Sciences</i> , 2013 , 331, 158-60	3.2	2
10	ERBB4 mutations that disrupt the neuregulin-ErbB4 pathway cause amyotrophic lateral sclerosis type 19. <i>American Journal of Human Genetics</i> , 2013 , 93, 900-5	11	95
9	Identification of ATP1A3 mutations by exome sequencing as the cause of alternating hemiplegia of childhood in Japanese patients. <i>PLoS ONE</i> , 2013 , 8, e56120	3.7	61

8	CSF1R mutations identified in three families with autosomal dominantly inherited leukoencephalopathy. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2012 , 159B, 951-7	3.5	29
7	Clinical features and haplotype analysis of newly identified Japanese patients with gelsolin-related familial amyloidosis of Finnish type. <i>Neurogenetics</i> , 2012 , 13, 237-43	3	13
6	The TRK-fused gene is mutated in hereditary motor and sensory neuropathy with proximal dominant involvement. <i>American Journal of Human Genetics</i> , 2012 , 91, 320-9	11	76
5	C9ORF72 repeat expansion in amyotrophic lateral sclerosis in the Kii peninsula of Japan. <i>Archives of Neurology</i> , 2012 , 69, 1154-8		77
4	Hypertrophic pachymeningitis and tracheobronchial stenosis in IgG4-related disease: case presentation and literature review. <i>Internal Medicine</i> , 2012 , 51, 935-41	1.1	27
3	Posterior column ataxia with retinitis pigmentosa in a Japanese family with a novel mutation in FLVCR1. <i>Neurogenetics</i> , 2011 , 12, 117-21	3	31
2	Lymphomatoid granulomatosis involving central nervous system successfully treated with rituximab alone. <i>Archives of Neurology</i> , 2008 , 65, 662-5		29
1	Development of a high-throughput microarray-based resequencing system for neurological disorders and its application to molecular genetics of amyotrophic lateral sclerosis. <i>Archives of Neurology</i> , 2008 , 65, 1326-32		37