Hiroyuki Ishiura

List of Publications by Year
in descending order

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125 papers	3,398 citations	172207 29 h-index	174990 52 g-index
135 all docs	135 docs citations	135 times ranked	5950 citing authors

#	Article	IF	Citations
1	Human genetic variation database, a reference database of genetic variations in the Japanese population. Journal of Human Genetics, 2016, 61, 547-553.	1.1	270
2	Noncoding CGG repeat expansions in neuronal intranuclear inclusion disease, oculopharyngodistal myopathy and an overlapping disease. Nature Genetics, 2019, 51, 1222-1232.	9.4	265
3	ExpansionsÂofÂintronic TTTCA and TTTTA repeats in benign adult familial myoclonic epilepsy. Nature Genetics, 2018, 50, 581-590.	9.4	238
4	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. Nature Communications, 2016, 7, 11253.	5.8	174
5	ERBB4 Mutations that Disrupt the Neuregulin-ErbB4 Pathway Cause Amyotrophic Lateral Sclerosis Type 19. American Journal of Human Genetics, 2013, 93, 900-905.	2.6	123
6	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	5.8	99
7	The TRK-Fused Gene Is Mutated in Hereditary Motor and Sensory Neuropathy with Proximal Dominant Involvement. American Journal of Human Genetics, 2012, 91, 320-329.	2.6	98
8	Variants associated with Gaucher disease in multiple system atrophy. Annals of Clinical and Translational Neurology, 2015, 2, 417-426.	1.7	90
9	C9ORF72 Repeat Expansion in Amyotrophic Lateral Sclerosis in the Kii Peninsula of Japan. Archives of Neurology, 2012, 69, 1154-8.	4.9	88
10	Mutations in MME cause an autosomalâ€recessive Charcotâ€“Marieâ€“Tooth disease type 2. Annals of Neurology, 2016, 79, 659-672.	2.8	82
11	A Novel Mutation inELOVL4Leading to Spinocerebellar Ataxia (SCA) With the Hot Cross Bun Sign but Lacking Erythrokeratodermia. JAMA Neurology, 2015, 72, 797.	4.5	79
12	Identification of ATP1A3 Mutations by Exome Sequencing as the Cause of Alternating Hemiplegia of Childhood in Japanese Patients. PLoS ONE, 2013, 8, e56120.	1.1	79
13	Intronic pentanucleotide TTTCA repeat insertion in the SAMD12 gene causes familial cortical myoclonic tremor with epilepsy type 1. Brain, 2018, 141, 2280-2288.	3.7	73
14	Evaluation of SLC20A2 mutations that cause idiopathic basal ganglia calcification in Japan. Neurology, 2014, 82, 705-712.	1.5	71
15	A recurrent de novo FAM111A mutation causes kennyâ€caffey syndrome type 2. Journal of Bone and Mineral Research, 2014, 29, 992-998.	3.1	68
16	Rapid detection of expanded short tandem repeats in personal genomics using hybrid sequencing. Bioinformatics, 2014, 30, 815-822.	1.8	61
17	Molecular epidemiology and clinical spectrum of hereditary spastic paraplegia in the Japanese population based on comprehensive mutational analyses. Journal of Human Genetics, 2014, 59, 163-172.	1.1	53
18	The pathogenesis linked to coenzyme Q10 insufficiency in iPSC-derived neurons from patients with multiple-system atrophy. Scientific Reports, 2018, 8, 14215.	1.6	50

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19	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.	4.9	44
20	Plasma Coenzyme Q10 Levels in Patients With Multiple System Atrophy. JAMA Neurology, 2016, 73, 977.	4.5	42
21	JASPAC: Japan Spastic Paraplegia Research Consortium. Brain Sciences, 2018, 8, 153.	1.1	41
22	Posterior column ataxia with retinitis pigmentosa in a Japanese family with a novel mutation in FLVCR1. Neurogenetics, 2011, 12, 117-121.	0.7	38
23	Mutations in COA7 cause spinocerebellar ataxia with axonal neuropathy. Brain, 2018, 141, 1622-1636.	3.7	38
24	Lymphomatoid Granulomatosis Involving Central Nervous System Successfully Treated With Rituximab Alone. Archives of Neurology, 2008, 65, 662-5.	4.9	37
25	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.	1.5	37
26	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.	1.4	35
27	CSF1R mutations identified in three families with autosomal dominantly inherited leukoencephalopathy. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2012, 159B, 951-957.	1.1	34
28	Hypertrophic Pachymeningitis and Tracheobronchial Stenosis in IgG4-related Disease: Case Presentation and Literature Review. Internal Medicine, 2012, 51, 935-941.	0.3	31
29	Clinical and genetic diversities of Charcotâ€Marieâ€Tooth disease with MFN2 mutations in a large case study. Journal of the Peripheral Nervous System, 2017, 22, 191-199.	1.4	31
30	Advances in repeat expansion diseases and a new concept of repeat motifâ€“phenotype correlation. Current Opinion in Genetics and Development, 2020, 65, 176-185.	1.5	30
31	AgIn: measuring the landscape of CpG methylation of individual repetitive elements. Bioinformatics, 2016, 32, 2911-2919.	1.8	29
32	Clinicopathologic features of myositis patients with CD8-MHC-1 complex pathology. Neurology, 2017, 89, 1060-1068.	1.5	29
33	VPS13Dâ€related disorders presenting as a pure and complicated form of hereditary spastic paraplegia. Molecular Genetics & Genomic Medicine, 2020, 8, e1108.	0.6	29
34	Structural Basis and Genotypeâ€“Phenotype Correlations of INSR Mutations Causing Severe Insulin Resistance. Diabetes, 2017, 66, 2713-2723.	0.3	28
35	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.	0.9	28
36	Autosomal-recessive complicated spastic paraplegia with a novel lysosomal trafficking regulator gene mutation. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 1024-1028.	0.9	27

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37	Low Prevalence of NOTCH2NLC GGC Repeat Expansion in White Patients with Movement Disorders. Movement Disorders, 2021, 36, 251-255.	2.2	23
38	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	3.7	22
39	Modeling neurological diseases with induced pluripotent cells reprogrammed from immortalized lymphoblastoid cell lines. Molecular Brain, 2016, 9, 88.	1.3	21
40	Novel De Novo KCND3 Mutation in a Japanese Patient with Intellectual Disability, Cerebellar Ataxia, Myoclonus, and Dystonia. Cerebellum, 2018, 17, 237-242.	1.4	21
41	An Autopsy Case of Familial Neuronal Intranuclear Inclusion Disease with Dementia and Neuropathy. Internal Medicine, 2018, 57, 3459-3462.	0.3	21
42	Ataxic phenotype with altered CaV3.1 channel property in a mouse model for spinocerebellar ataxia 42. Neurobiology of Disease, 2019, 130, 104516.	2.1	20
43	Clinical Characteristics of Neuronal Intranuclear Inclusion Disease-Related Retinopathy With CGG Repeat Expansions in the NOTCH2NLC Gene. , 2020, 61, 27.		19
44	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.	1.1	18
45	The novel de novo mutation of KIF1A gene as the cause for Spastic paraplegia 30 in a Japanese case. ENeurologicalSci, 2019, 14, 34-37.	0.5	18
46	Genetic spectrum of <scp>Charcotâ€“Marieâ€“Tooth</scp> disease associated with myelin protein zero gene variants in Japan. Clinical Genetics, 2021, 99, 359-375.	1.0	18
47	Epidemiology and molecular mechanism of frontotemporal lobar degeneration/amyotrophic lateral sclerosis with repeat expansion mutation inC9orf72. Journal of Neurogenetics, 2015, 29, 85-94.	0.6	17
48	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.	1.1	17
49	Clinical and genetic features of Charcotâ€Marieâ€Tooth disease 2F and hereditary motor neuropathy 2B in Japan. Journal of the Peripheral Nervous System, 2018, 23, 40-48.	1.4	17
50	PLA2G6-associated neurodegeneration presenting as a complicated form of hereditary spastic paraplegia. Journal of Human Genetics, 2019, 64, 55-59.	1.1	17
51	COQ2 V393A confers high risk susceptibility for multiple system atrophy in East Asian population. Journal of the Neurological Sciences, 2021, 429, 117623.	0.3	17
52	Clinical features and haplotype analysis of newly identified Japanese patients with gelsolin-related familial amyloidosis of Finnish type. Neurogenetics, 2012, 13, 237-243.	0.7	15
53	Novel mutations in the PNPLA6 gene in Boucher-NeuhÃuser syndrome. Journal of Human Genetics, 2015, 60, 217-220.	1.1	15
54	TBCD may be a causal gene in progressive neurodegenerative encephalopathy with atypical infantile spinal muscular atrophy. Journal of Human Genetics, 2017, 62, 473-480.	1.1	15

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55	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.	1.5	15
56	UBAP1 mutations cause juvenile-onset hereditary spastic paraplegias (SPG80) and impair UBAP1 targeting to endosomes. Journal of Human Genetics, 2019, 64, 1055-1065.	1.1	15
57	Identification of a novel mutation in ATP13A2 associated with a complicated form of hereditary spastic paraplegia. Neurology: Genetics, 2020, 6, e514.	0.9	15
58	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.	1.1	15
59	Randomized phase 2 study of perampanel for sporadic amyotrophic lateral sclerosis. Journal of Neurology, 2022, 269, 885-896.	1.8	15
60	Fukutin gene mutations that cause left ventricular noncompaction. International Journal of Cardiology, 2016, 222, 727-729.	0.8	14
61	Partial duplication of <scp>DHH</scp> causes minifascicular neuropathy. Annals of Clinical and Translational Neurology, 2017, 4, 415-421.	1.7	14
62	Clinical efficacy of haematopoietic stem cell transplantation for adult adrenoleukodystrophy. Brain Communications, 2020, 2, fcz048.	1.5	14
63	A Novel de novo KIF1A Mutation in a Patient with Autism, Hyperactivity, Epilepsy, Sensory Disturbance, and Spastic Paraplegia. Internal Medicine, 2020, 59, 839-842.	0.3	13
64	Atypical parkinsonism caused by Pro105Leu mutation of prion protein. Neurology: Genetics, 2016, 2, e48.	0.9	12
65	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.	0.3	12
66	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.	1.1	12
67	Exome sequencing reveals a novel missense mutation in the KIAA0196 gene in a Japanese patient with SPG8. Clinical Neurology and Neurosurgery, 2016, 144, 36-38.	0.6	11
68	Comprehensive investigation of RNF213 nonsynonymous variants associated with intracranial artery stenosis. Scientific Reports, 2020, 10, 11942.	1.6	11
69	<scp>TDP</scp>â€43 Proteinopathy Presenting with Typical Symptoms of Parkinson's Disease. Movement Disorders, 2022, 37, 1561-1563.	2.2	10
70	Juvenile amyotrophic lateral sclerosis with complex phenotypes associated with novel SYNE1 mutations. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2021, 22, 576-578.	1.1	9
71	Muscle Transcriptomics Shows Overexpression of Cadherin 1 in Inclusion Body Myositis. Annals of Neurology, 2022, 91, 317-328.	2.8	9
72	Slowly progressive d-bifunctional protein deficiency with survival to adulthood diagnosed by whole-exome sequencing. Journal of the Neurological Sciences, 2017, 372, 6-10.	0.3	8

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73	Functional evaluation of PDGFB-variants in idiopathic basal ganglia calcification, using patient-derived iPS cells. Scientific Reports, 2019, 9, 5698.	1.6	8
74	Cranial Nerve Involvement and Dysautonomia in Post-COVID-19 Guillain-BarrÃ© Syndrome. Internal Medicine, 2021, 60, 3477-3480.	0.3	8
75	Clinical features of inherited neuropathy with BSCL2 mutations in Japan. Journal of the Peripheral Nervous System, 2020, 25, 125-131.	1.4	7
76	Clinical Impact of Copy Number Variation on the Genetic Diagnosis of Syndromic Aortopathies. Circulation Genomic and Precision Medicine, 2021, 14, e003458.	1.6	7
77	Isolated Paravermal Hyperintensities in Neuronal Intranuclear Inclusion Disease. Neurology, 2022, 98, 938-939.	1.5	7
78	Novel de novo <scp>POLR3B</scp> mutations responsible for demyelinating Charcotâ€“Marieâ€“Tooth disease in Japan. Annals of Clinical and Translational Neurology, 2022, 9, 747-755.	1.7	7
79	A Homozygous LAMA2 Mutation of c.818G>A Caused Partial Merosin Deficiency in a Japanese Patient. Internal Medicine, 2018, 57, 877-882.	0.3	6
80	Neuroimaging, genetic, and enzymatic study in a Japanese family with a GBA gross deletion. Parkinsonism and Related Disorders, 2019, 61, 57-63.	1.1	6
81	A novel mutation in the GBA2 gene in a Japanese patient with SPG46: A case report. ENeurologicalSci, 2020, 19, 100238.	0.5	6
82	Splice-site mutations in KIF5A in the Japanese case series of amyotrophic lateral sclerosis. Neurogenetics, 2021, 22, 11-17.	0.7	6
83	Exome analysis reveals a Japanese family with spinocerebellar ataxia, autosomal recessive 1. Journal of the Neurological Sciences, 2013, 331, 158-160.	0.3	5
84	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.	0.7	5
85	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.	2.2	5
86	Reliability and validity of Japanese version of Unified Multiple System Atrophy Rating Scale. Neurology and Clinical Neuroscience, 2021, 9, 171-180.	0.2	5
87	Randomized, doubleâ€blind, placeboâ€controlled phase 1 study to evaluate the safety and pharmacokinetics of high doses of ubiquinol in healthy adults. Neurology and Clinical Neuroscience, 2022, 10, 14-24.	0.2	5
88	An NEFH founder mutation causes broad phenotypic spectrum in multiple Japanese families. Journal of Human Genetics, 2022, 67, 399-403.	1.1	5
89	DMD exon 2 duplication due to a complex genomic rearrangement is associated with a somatic mosaicism. Neuromuscular Disorders, 2022, 32, 263-269.	0.3	5
90	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.	1.1	4

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91	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.	1.1	4
92	An immigrant family with Kii amyotrophic lateral sclerosis/parkinsonismâ€“dementia complex. Neurological Sciences, 2022, 43, 1423-1425.	0.9	4
93	Elderly patients with suspected Charcot-Marie-Tooth disease should be tested for the TTR gene for effective treatments. Journal of Human Genetics, 2022, , .	1.1	4
94	No novel, high penetrant gene might remain to be found in Japanese patients with unknown MODY. Journal of Human Genetics, 2018, 63, 821-829.	1.1	3
95	HIV Dementia with a Decreased Cardiac ¹²³I-metaiodobenzylguanidine Uptake Masquerading as Dementia with Lewy Bodies. Internal Medicine, 2018, 57, 3007-3010.	0.3	3
96	Familial dementia with Lewy bodies with VPS13C mutations. Parkinsonism and Related Disorders, 2020, 81, 31-33.	1.1	3
97	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.	0.2	3
98	Frequency of FMR1 Premutation Alleles in Patients with Undiagnosed Cerebellar Ataxia and Multiple System Atrophy in the Japanese Population. Cerebellum, 2021, , 1.	1.4	3
99	Exome sequencing shows a novel de novo mutation in <scp>ATL</scp>1. Neurology and Clinical Neuroscience, 2014, 2, 1-4.	0.2	2
100	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.	0.2	2
101	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.	0.3	2
102	SPG9A with the new occurrence of an ALDH18A1 mutation in a CMT1A family with PMP22 duplication: case report. BMC Neurology, 2021, 21, 64.	0.8	2
103	ChÃ©diakâ€“Higashi syndrome presenting as a hereditary spastic paraplegia. Journal of Human Genetics, 2022, 67, 119-121.	1.1	2
104	Diagnostic Values of Venous Peak Lactate, Lactate-to-pyruvate Ratio, and Fold Increase in Lactate from Baseline in Aerobic Exercise Tests in Patients with Mitochondrial Diseases. Internal Medicine, 2022, 61, 1939-1946.	0.3	2
105	Complex hereditary peripheral neuropathies caused by novel variants in mitochondrial-related nuclear genes. Journal of Neurology, 2022, 269, 4129-4140.	1.8	2
106	Efficacy of canakinumab on AA amyloidosis in late-onset NLRP3-associated autoinflammatory disease with an I574F somatic mosaic mutation. Clinical Rheumatology, 2022, 41, 2233-2237.	1.0	2
107	Nonsyndromic arteriopathy and aortopathy and vascular Ehlersâ€“Danlos syndrome <scp>causing COL3A1</scp> variants. American Journal of Medical Genetics, Part A, 2022, 188, 2777-2782.	0.7	2
108	Authors' reply to the Drs. Finsterer and Zarrouk-Mahjoub's comments for our case report. International Journal of Cardiology, 2018, 254, 262.	0.8	1

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109	An autopsy case of G M1 gangliosidosis type II in a patient who survived a long duration with artificial respiratory support. Neuropathology, 2020, 40, 379-388.	0.7	1
110	A clinical and genetic study of SPG31 in Japan. Journal of Human Genetics, 2022, , .	1.1	1
111	Novel COL6A2 mutation in a case of limb girdle muscular dystrophy phenotype with autosomal recessive inheritance. Neurology and Clinical Neuroscience, 2016, 4, 189-191.	0.2	0
112	Authors' response to â€œCompound heterozygous Fukutin mutation-related non-compactionâ€•by Finsterer and Zarrouk-Mahjoub. International Journal of Cardiology, 2017, 233, 102.	0.8	0
113	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.	0.2	0
114	Prominent Spasticity and Hyperreflexia of the Legs in a Nepalese Patient with Friedreich Ataxia. Internal Medicine, 2019, 58, 2865-2869.	0.3	0
115	Sporadic progressive myoclonic epilepsy with earlyâ€onset dementia caused by a de novo mutation in PSEN1. Neurology and Clinical Neuroscience, 2019, 7, 294-296.	0.2	0
116	A novel multiâ€exon deletion in the dysferlin gene of a limbâ€girdle muscular dystrophy type 2B Filipino patient. Neurology and Clinical Neuroscience, 2020, 8, 419-421.	0.2	0
117	Do eye movements â€œageâ€•earlier in progeria?. Clinical Neurophysiology, 2020, 131, 1835-1836.	0.7	0
118	Novel variant of CSF1R in sporadic case with earlyâ€onset cognitive impairment. Neurology and Clinical Neuroscience, 2020, 8, 430-432.	0.2	0
119	A novel mutation in ABCD1 gene in a Filipino patient with adultâ€onset Xâ€linked ALD. Neurology and Clinical Neuroscience, 2020, 8, 329-331.	0.2	0
120	A Case of Irreversible Corneal Edema Associated with Dentatorubropallidoluysian Atrophy Following Corneal Endothelial Transplantation. SN Comprehensive Clinical Medicine, 2021, 3, 2029-2032.	0.3	0
121	Severe dilated cardiomyopathy and ventricular arrhythmia in a patient with Emeryâ€“Dreifuss muscular dystrophy harboring a novel frameshift mutation in EMD. Neurology and Clinical Neuroscience, 2021, 9, 490.	0.2	0
122	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.	1.1	0
123	DNA sequencing and other methods of exonic and genomic analyses. , 2020, , 109-120.		0
124	Family with congenital contractural arachnodactyly due to a novel multiexon deletion of the FBN2 gene. Clinical Case Reports (discontinued), 2022, 10, e05335.	0.2	0
125	Expression profile analysis in cells overexpressing <scp>DRPLA cDNA</scp> to explore the roles of <scp>DRPLAp</scp> as a transcriptional coregulator. Neurology and Clinical Neuroscience, 0, , .	0.2	0