Shoji Tsuji

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/288217/publications.pdf

Version: 2024-02-01

314	16,346	19608 61	19136 118
papers	citations	h-index	g-index

327	327	327	18597
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Frequency of the C9orf72 hexanucleotide repeat expansion in patients with amyotrophic lateral sclerosis and frontotemporal dementia: a cross-sectional study. Lancet Neurology, The, 2012, 11, 323-330.	4.9	1,039
2	Interference by Huntingtin and Atrophin-1 with CBP-Mediated Transcription Leading to Cellular Toxicity. Science, 2001, 291, 2423-2428.	6.0	1,035
3	Hereditary progressive dystonia with marked diurnal fluctuation caused by mutations in the GTP cyclohydrolase I gene. Nature Genetics, 1994, 8, 236-242.	9.4	800
4	Î±-Synuclein immunoreactivity in glial cytoplasmic inclusions in multiple system atrophy. Neuroscience Letters, 1998, 249, 180-182.	1.0	581
5	SCA17, a novel autosomal dominant cerebellar ataxia caused by an expanded polyglutamine in TATA-binding protein. Human Molecular Genetics, 2001, 10, 1441-1448.	1.4	569
6	Association of HTRA1 Mutations and Familial Ischemic Cerebral Small-Vessel Disease. New England Journal of Medicine, 2009, 360, 1729-1739.	13.9	407
7	Expanded polyglutamine stretches interact with TAFII130, interfering with CREB-dependent transcription. Nature Genetics, 2000, 26, 29-36.	9.4	388
8	Early-onset ataxia with ocular motor apraxia and hypoalbuminemia is caused by mutations in a new HIT superfamily gene. Nature Genetics, 2001, 29, 184-188.	9.4	376
9	Suppression of aggregate formation and apoptosis by transglutaminase inhibitors in cells expressing truncated DRPLA protein with an expanded polyglutamine stretch. Nature Genetics, 1998, 18, 111-117.	9.4	372
10	Mis-sense mutation Val→Ile in exon 17 of amyloid precursor protein gene in Japanese familial Alzheimer's disease. Lancet, The, 1991, 337, 978-979.	6.3	311
11	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
12	Genomic Landscape of Esophageal Squamous Cell Carcinoma inÂa Japanese Population. Gastroenterology, 2016, 150, 1171-1182.	0.6	265
13	Noncoding CGG repeat expansions in neuronal intranuclear inclusion disease, oculopharyngodistal myopathy and an overlapping disease. Nature Genetics, 2019, 51, 1222-1232.	9.4	265
14	Spinocerebellar Ataxia Type 31 Is Associated with â€œInsertedâ€•Penta-Nucleotide Repeats Containing (TGGAA)n. American Journal of Human Genetics, 2009, 85, 544-557.	2.6	260
15	Synphilin-1 is present in Lewy bodies in Parkinson's disease. Annals of Neurology, 2000, 47, 521-523.	2.8	246
16	ExpansionsÂofÂintronic TTTCA and TTTTA repeats in benign adult familial myoclonic epilepsy. Nature Genetics, 2018, 50, 581-590.	9.4	238
17	The Movement Disorder Society Criteria for the Diagnosis of Multiple System Atrophy. Movement Disorders, 2022, 37, 1131-1148.	2.2	222
18	An autopsy case of autosomal-recessive juvenile parkinsonism with a homozygous exon 4 deletion in theparkin gene. Movement Disorders, 2000, 15, 884-888.	2.2	220

#	Article	IF	Citations
19	Mutations for Gaucher Disease Confer High Susceptibility to Parkinson Disease. Archives of Neurology, 2009, 66, 571-6.	4.9	183
20	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. Nature Communications, 2016, 7, 11253.	5.8	174
21	Efficacy and safety of leuprorelin in patients with spinal and bulbar muscular atrophy (JASMITT study): a multicentre, randomised, double-blind, placebo-controlled trial. Lancet Neurology, The, 2010, 9, 875-884.	4.9	170
22	Genetic association of the very low density lipoprotein (VLDL) receptor gene with sporadic Alzheimer's disease. Nature Genetics, 1995, 11, 207-209.	9.4	169
23	Dentatorubral-pallidoluysian atrophy: Clinical features are closely related to unstable expansions of trinucleotide (CAG) repeat. Annals of Neurology, 1995, 37, 769-775.	2.8	154
24	ApoEâ€“Îµ4 and earlyâ€“onset Alzheimer's. Nature Genetics, 1994, 7, 10-11.	9.4	145
25	Modulation of the age at onset in spinocerebellar ataxia by CAG tracts in various genes. Brain, 2014, 137, 2444-2455.	3.7	144
26	A novel locus for dominant cerebellar ataxia (SCA14) maps to a 10.2-cM interval flanked by D19S206 and D19S605 on chromosome 19q13.4-qter. Annals of Neurology, 2000, 48, 156-163.	2.8	136
27	Sporadic ataxias in Japan â€“ a population-based epidemiological study. Cerebellum, 2008, 7, 189-197.	1.4	131
28	Multiplex Families With Multiple System Atrophy. Archives of Neurology, 2007, 64, 545.	4.9	127
29	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
30	Safety and efficacy of eculizumab in Guillain-BarrÃ© syndrome: a multicentre, double-blind, randomised phase 2 trial. Lancet Neurology, The, 2018, 17, 519-529.	4.9	111
31	Novel mutations, pseudo-dominant inheritance, and possible familial affects in patients with autosomal recessive juvenile parkinsonism. Annals of Neurology, 2000, 48, 245-250.	2.8	102
32	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	5.8	99
33	Age at onset influences on wide-ranged clinical features of sporadic amyotrophic lateral sclerosis. Journal of the Neurological Sciences, 2009, 276, 163-169.	0.3	98
34	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
35	Gene locus for autosomal recessive distal myopathy with rimmed vacuoles maps to chromosome 9. Annals of Neurology, 1997, 41, 432-437.	2.8	97
36	Trinucleotide repeat length and rate of progression of Huntington's disease. Annals of Neurology, 1994, 36, 630-635.	2.8	95

#	Article	IF	Citations
37	Analysis of the expression level of Î±-synuclein mRNA using postmortem brain samples from pathologically confirmed cases of multiple system atrophy. Acta Neuropathologica, 2001, 102, 188-190.	3.9	95
38	Widespread occurrence of intranuclear atrophin-1 accumulation in the central nervous system neurons of patients with dentatorubral-pallidoluysian atrophy. Annals of Neurology, 2001, 49, 14-23.	2.8	94
39	Variants associated with Gaucher disease in multiple system atrophy. Annals of Clinical and Translational Neurology, 2015, 2, 417-426.	1.7	90
40	Hereditary dentatorubral-pallidoluysian atrophy: detection of widespread ubiquitinated neuronal and glial intranuclear inclusions in the brain. Acta Neuropathologica, 1998, 96, 547-552.	3.9	88
41	C9ORF72 Repeat Expansion in Amyotrophic Lateral Sclerosis in the Kii Peninsula of Japan. Archives of Neurology, 2012, 69, 1154-8.	4.9	88
42	Mechanisms of Genomic Instabilities Underlying Two Common Fragile-Site-Associated Loci, PARK2 and DMD, in Germ Cell and Cancer Cell Lines. American Journal of Human Genetics, 2010, 87, 75-89.	2.6	85
43	Mutations in MME cause an autosomalâ€recessive Charcotâ€“Marieâ€“Tooth disease type 2. Annals of Neurology, 2016, 79, 659-672.	2.8	82
44	Mutant COQ2 in Multiple-System Atrophy. New England Journal of Medicine, 2014, 371, 80-83.	13.9	81
45	Exome Sequencing Reveals a Homozygous SYT14 Mutation in Adult-Onset, Autosomal-Recessive Spinocerebellar Ataxia with Psychomotor Retardation. American Journal of Human Genetics, 2011, 89, 320-327.	2.6	79
46	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
47	Aprataxin, the causative protein for EAOH is a nuclear protein with a potential role as a DNA repair protein. Annals of Neurology, 2004, 55, 241-249.	2.8	76
48	A homozygous mutation ofC12orf65causes spastic paraplegia with optic atrophy and neuropathy (SPG55). Journal of Medical Genetics, 2012, 49, 777-784.	1.5	76
49	Pathology of CAG repeat diseases. Neuropathology, 2000, 20, 319-325.	0.7	74
50	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
51	A critique of the second consensus criteria for multiple system atrophy. Movement Disorders, 2019, 34, 975-984.	2.2	73
52	Quantitative evaluation of the pyramidal tract segmented by diffusion tensor tractography: feasibility study in patients with amyotrophic lateral sclerosis. Radiation Medicine, 2005, 23, 195-9.	0.8	73
53	Genetics of neurodegenerative diseases: insights from high-throughput resequencing. Human Molecular Genetics, 2010, 19, R65-R70.	1.4	72
54	Neuron-specific methylome analysis reveals epigenetic regulation and tau-related dysfunction of BRCA1 in Alzheimerâ€™s disease. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E9645-E9654.	3.3	72

#	Article	IF	Citations
55	Evaluation of SLC20A2 mutations that cause idiopathic basal ganglia calcification in Japan. Neurology, 2014, 82, 705-712.	1.5	71
56	Cancer association as a risk factor for anti-HMGCR antibody-positive myopathy. Neurology: Neuroimmunology and NeuroInflammation, 2016, 3, e290.	3.1	71
57	Progressive atrophy of cerebellum and brainstem as a function of age and the size of the expanded CAG repeats in theMJD1 gene in Machado-Joseph disease. Annals of Neurology, 1998, 43, 288-296.	2.8	70
58	Linkage of autosomal recessive hereditary spastic paraplegia with mental impairment and thin corpus callosum to chromosome 15q13-15. Annals of Neurology, 2000, 48, 108-112.	2.8	70
59	BIN1 regulates BACE1 intracellular trafficking and amyloid-Î² production. Human Molecular Genetics, 2016, 25, ddw146.	1.4	67
60	Pathology of CAG repeat diseases. Neuropathology, 2000, 20, 319-325.	0.7	67
61	No mutation in the entire coding region of the Î±-synuclein gene in pathologically confirmed cases of multiple system atrophy. Neuroscience Letters, 1999, 270, 110-112.	1.0	66
62	A novel monoclonal antibody reveals a conformational alteration shared by amyotrophic lateral sclerosisâ€linked SOD1 mutants. Annals of Neurology, 2012, 72, 739-749.	2.8	65
63	A novel ferritin light chain gene mutation in a Japanese family with neuroferritinopathy: Description of clinical features and implications for genotypeâ€“phenotype correlations. Movement Disorders, 2009, 24, 441-445.	2.2	64
64	Dentatorubralâ€“pallidoluysian atrophy. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2012, 103, 587-594.	1.0	64
65	Rapid detection of expanded short tandem repeats in personal genomics using hybrid sequencing. Bioinformatics, 2014, 30, 815-822.	1.8	61
66	Adult onset globoid cell leukodystrophy (Krabbe disease): analysis of galactosylceramidase cDNA from four Japanese patients. Human Genetics, 1997, 100, 450-456.	1.8	60
67	14-3-3 protein ? chain gene (YWHAH) polymorphism and its genetic association with schizophrenia. American Journal of Medical Genetics Part A, 1999, 88, 164-167.	2.4	60
68	Aprataxin, causative gene product for EAOH/AOA1, repairs DNA single-strand breaks with damaged 3â€²-phosphate and 3â€²-phosphoglycolate ends. Nucleic Acids Research, 2007, 35, 3797-3809.	6.5	60
69	Fatty Acid Elongation Activity in Fibroblasts from Patients with Adrenoleukodystrophy (ALD)1. Journal of Biochemistry, 1984, 96, 1241-1247.	0.9	58
70	Autosomal dominant cerebellar ataxia (SCA6): clinical, genetic and neuropathological study in a family. Acta Neuropathologica, 1998, 95, 333-337.	3.9	58
71	Mutational Analysis and Genotype-Phenotype Correlation of 29 Unrelated Japanese Patients With X-linked Adrenoleukodystrophy. Archives of Neurology, 1999, 56, 295.	4.9	58
72	The FHA domain of aprataxin interacts with the C-terminal region of XRCC1. Biochemical and Biophysical Research Communications, 2004, 325, 1279-1285.	1.0	56

#	Article	IF	Citations
73	A functional variant in ZNF512B is associated with susceptibility to amyotrophic lateral sclerosis in Japanese. Human Molecular Genetics, 2011, 20, 3684-3692.	1.4	53
74	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
75	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
76	Dentatorubral-pallidoluysian atrophy (DRPLA): Close correlation of CAG repeat expansions with the wide spectrum of clinical presentations and prominent anticipation. Seminars in Cell Biology, 1995, 6, 37-44.	3.5	49
77	Next-generation sequencing of 28 ALS-related genes in a Japanese ALS cohort. Neurobiology of Aging, 2016, 39, 219.e1-219.e8.	1.5	49
78	Epidemiology of X-linked adrenoleukodystrophy in Japan. Journal of Human Genetics, 2002, 47, 0590-0593.	1.1	45
79	Toxicity of expanded polyglutamine-domain proteins in Escherichia coli. FEBS Letters, 1996, 399, 135-139.	1.3	44
80	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
81	Modulation of error-sensitivity during a prism adaptation task in people with cerebellar degeneration. Journal of Neurophysiology, 2015, 114, 2460-2471.	0.9	43
82	TDP-43 M337V Mutation in Familial Amyotrophic Lateral Sclerosis in Japan. Internal Medicine, 2010, 49, 331-334.	0.3	42
83	Plasma Coenzyme Q10 Levels in Patients With Multiple System Atrophy. JAMA Neurology, 2016, 73, 977.	4.5	42
84	Hereditary dentatorubral-pallidoluysian atrophy: ubiquitinated filamentous inclusions in the cerebellar dentate nucleus neurons. Acta Neuropathologica, 1998, 95, 479-482.	3.9	41
85	JASPAC: Japan Spastic Paraplegia Research Consortium. Brain Sciences, 2018, 8, 153.	1.1	41
86	MITOCHONDRIAL DNA MUTATION IN FAMILY WITH LEBER'S HEREDITARY OPTIC NEUROPATHY. Lancet, The, 1989, 333, 1076-1077.	6.3	40
87	Appropriate data cleaning methods for genome-wide association study. Journal of Human Genetics, 2008, 53, 886-893.	1.1	40
88	Apolipoprotein E Îµ4 allele and progression of cortical Lewy body pathology in Parkinson's disease. Acta Neuropathologica, 1998, 95, 450-454.	3.9	38
89	Autosomal dominant diffuse Lewy body disease. Acta Neuropathologica, 1998, 96, 207-210.	3.9	38
90	Severe neurological phenotypes of Q129 DRPLA transgenic mice serendipitously created by en masse expansion of CAG repeats in Q76 DRPLA mice. Human Molecular Genetics, 2009, 18, 723-736.	1.4	38

#	Article	IF	Citations
91	Posterior column ataxia with retinitis pigmentosa in a Japanese family with a novel mutation in FLVCR1. Neurogenetics, 2011, 12, 117-121.	0.7	38
92	Mutations in COA7 cause spinocerebellar ataxia with axonal neuropathy. Brain, 2018, 141, 1622-1636.	3.7	38
93	Lack of association of very low density lipoprotein receptor gene polymorphism with caucasian Alzheimer's disease. Annals of Neurology, 1996, 40, 251-254.	2.8	37
94	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
95	Interaction of expanded polyglutamine stretches with nuclear transcription factors leads to aberrant transcriptional regulation in polyglutamine diseases. Neuropathology, 2000, 20, 326-333.	0.7	36
96	Mitochondrial ND3 as the novel causative gene for Leber hereditary optic neuropathy and dystonia. Neurogenetics, 2009, 10, 337-345.	0.7	35
97	A 3-year cohort study of the natural history of spinocerebellar ataxia type 6 in Japan. Orphanet Journal of Rare Diseases, 2014, 9, 118.	1.2	35
98	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
99	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
100	X-linked nonprogressive congenital cerebellar hypoplasia: Clinical description and mapping to chromosome Xq. Annals of Neurology, 1996, 40, 75-83.	2.8	33
101	A rapid functional decline type of amyotrophic lateral sclerosis is linked to low expression ofTTN. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 851-858.	0.9	33
102	Effect of intravenous immunoglobulin in Lambert-Eaton myasthenic syndrome with small-cell lung cancer: Correlation with the titer of anti-voltage-gateo calcium channel antibody. Muscle and Nerve, 1994, 17, 1073-1075.	1.0	32
103	Adult-onset leukoencephalopathies with vanishing white matter with novel missense mutations in EIF2B2, EIF2B3, and EIF2B5. Neurogenetics, 2011, 12, 259-261.	0.7	32
104	TRPM7 is not associated with amyotrophic lateral sclerosisâ€parkinsonism dementia complex in the Kii peninsula of Japan. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 310-313.	1.1	31
105	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
106	Thalamic form of Creutzfeldt-Jakob disease or fatal insomnia? Report of a sporadic case with normal prion protein genotype. Acta Neuropathologica, 1997, 93, 317-322.	3.9	30
107	Phosphorylation in the C-terminal domain of Aquaporin-4 is required for Golgi transition in primary cultured astrocytes. Biochemical and Biophysical Research Communications, 2008, 377, 463-468.	1.0	30
108	Diffusion tensor tract-specific analysis of the uncinate fasciculus in patients with amyotrophic lateral sclerosis. Neuroradiology, 2010, 52, 729-733.	1.1	30

#	Article	IF	Citations
109	Genotypeâ€“phenotype correlations in early onset ataxia with ocular motor apraxia and hypoalbuminaemia. Brain, 2011, 134, 1387-1399.	3.7	30
110	Increased gene dosage of myelin protein zero causes Charcotâ€Marieâ€Tooth disease. Annals of Neurology, 2012, 71, 84-92.	2.8	30
111	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
112	Clinicopathologic Features of Oculopharyngodistal Myopathy WithLRP12CGG Repeat Expansions Compared With Other Oculopharyngodistal Myopathy Subtypes. JAMA Neurology, 2021, 78, 853.	4.5	30
113	Identification of novel SNPs of ABCD1, ABCD2, ABCD3, and ABCD4 genes in patients with X-linked adrenoleukodystrophy (ALD) based on comprehensive resequencing and association studies with ALD phenotypes. Neurogenetics, 2011, 12, 41-50.	0.7	29
114	Late-onset spastic ataxia phenotype in a patient with a homozygous DDHD2 mutation. Scientific Reports, 2015, 4, 7132.	1.6	29
115	AgIn: measuring the landscape of CpG methylation of individual repetitive elements. Bioinformatics, 2016, 32, 2911-2919.	1.8	29
116	Clinicopathologic features of myositis patients with CD8-MHC-1 complex pathology. Neurology, 2017, 89, 1060-1068.	1.5	29
117	VPS13Dâ€related disorders presenting as a pure and complicated form of hereditary spastic paraplegia. Molecular Genetics & Genomic Medicine, 2020, 8, e1108.	0.6	29
118	SNP HiTLink: a high-throughput linkage analysis system employing dense SNP data. BMC Bioinformatics, 2009, 10, 121.	1.2	28
119	Structural Basis and Genotypeâ€“Phenotype Correlations of INSR Mutations Causing Severe Insulin Resistance. Diabetes, 2017, 66, 2713-2723.	0.3	28
120	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
121	Assignment of the human ST2 gene to chromosome 2 at q11.2. Human Genetics, 1996, 97, 561-563.	1.8	27
122	Decrease in benzodiazepine receptor binding in a patient with Angelman syndrome detected by iodine-123 iomazenil and single-photon emission tomography. European Journal of Nuclear Medicine and Molecular Imaging, 1996, 23, 598-604.	2.2	27
123	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
124	Tubular aggregate myopathy caused by a novel mutation in the cytoplasmic domain of STIM1. Neurology: Genetics, 2016, 2, e50.	0.9	27
125	Amnionless-mediated glycosylation is crucial for cell surface targeting of cubilin in renal and intestinal cells. Scientific Reports, 2018, 8, 2351.	1.6	27
126	Chronic cerebral hypoperfusion shifts the equilibrium of amyloid Î² oligomers to aggregation-prone species with higher molecular weight. Scientific Reports, 2019, 9, 2827.	1.6	27

#	Article	IF	Citations
127	Lack of association between dopamine D2 receptor gene Cys311 variant and schizophrenia. , 1996, 67, 208-211.		26
128	Transcortical sensory aphasia following left frontal infarction. Journal of Neurology, 1998, 245, 69-76.	1.8	26
129	SCA42 mutation analysis in a case series of Japanese patients with spinocerebellar ataxia. Journal of Human Genetics, 2017, 62, 857-859.	1.1	25
130	Neuronal atrophy and synaptic alteration in a mouse model of dentatorubral-pallidoluysian atrophy. Brain, 2006, 129, 2353-2362.	3.7	24
131	DRPLA transgenic mouse substrains carrying single copy of full-length mutant human DRPLA gene with variable sizes of expanded CAG repeats exhibit CAG repeat length- and age-dependent changes in behavioral abnormalities and gene expression profiles. Neurobiology of Disease, 2012, 46, 336-350.	2.1	23
132	Low Prevalence of NOTCH2NLC GGC Repeat Expansion in White Patients with Movement Disorders. Movement Disorders, 2021, 36, 251-255.	2.2	23
133	Quantitation of heteroplasmy of mitochondrial trnaLeu(UUR) gene using PCR-SSCP. Muscle and Nerve, 1995, 18, 1390-1397.	1.0	22
134	Ataxia with isolated vitamin E deficiency and retinitis pigmentosa. Annals of Neurology, 1998, 43, 273-273.	2.8	22
135	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	3.7	22
136	Molecular genetic evidence of clinical heterogeneity in Fukuyama-type congenital muscular dystrophy. Human Genetics, 1997, 99, 427-432.	1.8	21
137	Modeling neurological diseases with induced pluripotent cells reprogrammed from immortalized lymphoblastoid cell lines. Molecular Brain, 2016, 9, 88.	1.3	21
138	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
139	An Autopsy Case of Familial Neuronal Intranuclear Inclusion Disease with Dementia and Neuropathy. Internal Medicine, 2018, 57, 3459-3462.	0.3	21
140	Genomic aspects of sporadic neurodegenerative diseases. Biochemical and Biophysical Research Communications, 2014, 452, 221-225.	1.0	20
141	Increased facilitation of the primary motor cortex in de novo Parkinson's disease. Parkinsonism and Related Disorders, 2019, 66, 125-129.	1.1	20
142	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
143	Lack of association between dopamine D4 receptor gene and schizophrenia. American Journal of Medical Genetics Part A, 1995, 60, 580-582.	2.4	19
144	Unstable expansion of triplet repeats as a new disease mechanism for neurodegenerative diseases. Japanese Journal of Human Genetics, 1996, 41, 279-290.	0.8	19

#	Article	IF	Citations
145	Mutational analysis of familial and sporadic amyotrophic lateral sclerosis withOPTNmutations in Japanese population. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 562-566.	2.3	19
146	The Neurogenomics View of Neurological Diseases. JAMA Neurology, 2013, 70, 689.	4.5	19
147	Clinical Characteristics of Neuronal Intranuclear Inclusion Disease-Related Retinopathy With CGG Repeat Expansions in the NOTCH2NLC Gene. , 2020, 61, 27.		19
148	HLA genotype-clinical phenotype correlations in multiple sclerosis and neuromyelitis optica spectrum disorders based on Japan MS/NMOSD Biobank data. Scientific Reports, 2021, 11, 607.	1.6	19
149	Association study between schizophrenia and dopamine D3 receptor gene polymorphism. , 1996, 67, 366-368.		18
150	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
151	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
152	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
153	Multiple system atrophy variant with severe hippocampal pathology. Brain Pathology, 2022, 32, e13002.	2.1	18
154	Differential Effect of HDAC3 on Cytoplasmic and Nuclear Huntingtin Aggregates. PLoS ONE, 2014, 9, e111277.	1.1	18
155	A Prospective, Multicenter, Randomized Phase II Study to Evaluate the Efficacy and Safety of Eculizumab in Patients with Guillain-BarrÃ© Syndrome (GBS): Protocol of Japanese Eculizumab Trial for GBS (JET-GBS). JMIR Research Protocols, 2016, 5, e210.	0.5	18
156	The 3-Second Rule in Hereditary Pure Cerebellar Ataxia: A Synchronized Tapping Study. PLoS ONE, 2015, 10, e0118592.	1.1	17
157	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
158	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
159	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
160	PLA2G6-associated neurodegeneration presenting as a complicated form of hereditary spastic paraplegia. Journal of Human Genetics, 2019, 64, 55-59.	1.1	17
161	First Report of Multidrug-Resistant Carbapenemase-Producing Bacteria Coharboring mcr-9 Associated with Respiratory Disease Complex in Pets: Potential of Animal-Human Transmission. Antimicrobial Agents and Chemotherapy, 2020, 65, .	1.4	17
162	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

#	Article	IF	Citations
163	Unilateral Opercular Infarction Presenting with Foix-Chavany-Marie Syndrome. Journal of Stroke and Cerebrovascular Diseases, 2014, 23, 179-181.	0.7	16
164	Tacrolimus-Induced Reversible Cerebral Vasoconstriction Syndrome with Delayed Multi-Segmental Vasoconstriction. Journal of Stroke and Cerebrovascular Diseases, 2017, 26, e75-e77.	0.7	16
165	Human leukocyte antigens in Fisher's syundrome. Annals of Neurology, 1993, 33, 655-657.	2.8	15
166	Familial cases presenting very early onset autosomal dominant Alzheimerâ€™s disease with I143T in presenilin-1 gene: implication for genotypeâ€“phenotype correlation. Neurogenetics, 2008, 9, 65-67.	0.7	15
167	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
168	Novel mutations in the PNPLA6 gene in Boucher-NeuhÃuser syndrome. Journal of Human Genetics, 2015, 60, 217-220.	1.1	15
169	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
170	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
171	Does the Clock Tick Slower or Faster in Parkinsonâ€™s Disease? â€“ Insights Gained From the Synchronized Tapping Task. Frontiers in Psychology, 2018, 9, 1178.	1.1	15
172	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
173	Identification of a novel mutation in ATP13A2 associated with a complicated form of hereditary spastic paraplegia. Neurology: Genetics, 2020, 6, e514.	0.9	15
174	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
175	Mutational analysis of the anion exchanger 3 gene in familial paroxysmal dystonic choreoathetosis linked to chromosome 2q. , 1999, 88, 733-737.		14
176	Fukutin gene mutations that cause left ventricular noncompaction. International Journal of Cardiology, 2016, 222, 727-729.	0.8	14
177	Partial duplication of <scp>DHH</scp> causes minifascicular neuropathy. Annals of Clinical and Translational Neurology, 2017, 4, 415-421.	1.7	14
178	CNV analysis using whole exome sequencing identified biallelic CNVs of VPS13B in siblings with intellectual disability. European Journal of Medical Genetics, 2020, 63, 103610.	0.7	14
179	Clinical efficacy of haematopoietic stem cell transplantation for adult adrenoleukodystrophy. Brain Communications, 2020, 2, fcz048.	1.5	14
180	Reduced morning cortisol secretion in patients with multiple system atrophy. Clinical Autonomic Research, 2001, 11, 271-272.	1.4	13

#	Article	IF	Citations
181	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
182	Strong linkage disequilibrium and haplotype analysis in Japanese pedigrees with Machado-Joseph disease. , 1996, 67, 437-444.		12
183	Molecular Genetics of Triplet Repeats: Unstable Expansion of Triplet Repeats as a New Mechanism for Neurodegenerative Diseases.. Internal Medicine, 1997, 36, 3-8.	0.3	12
184	Atypical parkinsonism caused by Pro105Leu mutation of prion protein. Neurology: Genetics, 2016, 2, e48.	0.9	12
185	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
186	Altered expression of Crb2 in podocytes expands a variation of CRB2 mutations in steroid-resistant nephrotic syndrome. Pediatric Nephrology, 2017, 32, 801-809.	0.9	12
187	Targeted deep sequencing of DNA from multiple tissue types improves the diagnostic rate and reveals a highly diverse phenotype of mosaic neurofibromatosis type 2. Journal of Medical Genetics, 2021, 58, 701-711.	1.5	12
188	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
189	Candesartan prevents arteriopathy progression in cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy model. Journal of Clinical Investigation, 2021, 131, .	3.9	12
190	Stereotyped hand clasping: An unusual tardive movement disorder. Movement Disorders, 1993, 8, 230-231.	2.2	11
191	Two Japanese cases with aspartylglycosaminuria: clinical and morphological features. Clinical Genetics, 1991, 40, 318-325.	1.0	11
192	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
193	Assessing Cell-to-Cell DNA Methylation Variability on Individual Long Reads. Scientific Reports, 2016, 6, 21317.	1.6	11
194	Cilostazol alleviates white matter degeneration caused by chronic cerebral hypoperfusion in mice: Implication of its mechanism from gene expression analysis. Neuroscience Letters, 2018, 662, 247-252.	1.0	11
195	Comprehensive investigation of RNF213 nonsynonymous variants associated with intracranial artery stenosis. Scientific Reports, 2020, 10, 11942.	1.6	11
196	CAG repeat-binding small molecule improves motor coordination impairment in a mouse model of Dentatorubralâ€“pallidoluysian atrophy. Neurobiology of Disease, 2022, 163, 105604.	2.1	11
197	Letters to the editor. Muscle and Nerve, 1993, 16, 217-222.	1.0	10
198	Shy-Drager Syndrome with Abnormal Circadian Rhythm of Plasma Antidiuretic Hormone Secretion and Urinary Excretion.. Internal Medicine, 1993, 32, 225-227.	0.3	10

#	Article	IF	Citations
199	A de novo splice donor site mutation causes in-frame deletion of 14 amino acids in the proteolipid protein in Pelizaeus-Merzbacher disease. Annals of Neurology, 1999, 46, 112-115.	2.8	10
200	An open trial of longâ€term testosterone suppression in spinal and bulbar muscular atrophy. Muscle and Nerve, 2013, 47, 816-822.	1.0	10
201	Neuron-specific analysis of histone modifications with post-mortem brains. Scientific Reports, 2020, 10, 3767.	1.6	10
202	Partial deletions of putative adrenoleukodystrophy (ALD) gene in Japanese ALD patients. Human Mutation, 1995, 6, 263-267.	1.1	9
203	Pick's disease: selective occurrence of apolipoprotein E-immunoreactive Pick bodies in the limbic system. Acta Neuropathologica, 1998, 95, 1-4.	3.9	9
204	Efficacy of Early Plasmapheresis in Bickerstaff's Encephalitis.. Internal Medicine, 1998, 37, 986-989.	0.3	9
205	Mutational Analysis of X-Linked Adrenoleukodystrophy Gene. Cell Biochemistry and Biophysics, 2000, 32, 177-185.	0.9	9
206	Effect of subthalamic nucleus deep brain stimulation on visual scanning. Clinical Neurophysiology, 2018, 129, 2421-2432.	0.7	9
207	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
208	Familial amyotrophic lateral sclerosis with a mutation in the Cu/Zn superoxide dismutase gene. Acta Neuropathologica, 1994, 88, 185-188.	3.9	9
209	Top-Down but Not Bottom-Up Visual Scanning is Affected in Hereditary Pure Cerebellar Ataxia. PLoS ONE, 2014, 9, e116181.	1.1	9
210	Muscle Transcriptomics Shows Overexpression of Cadherin 1 in Inclusion Body Myositis. Annals of Neurology, 2022, 91, 317-328.	2.8	9
211	Familial amyotrophic lateral sclerosis with novel A4DSOD1mutation with late age at onset and rapid progressive course. Neurology and Clinical Neuroscience, 2013, 1, 45-47.	0.2	8
212	Triad-conditioning Transcranial Magnetic Stimulation in Parkinson's Disease. Brain Stimulation, 2014, 7, 74-79.	0.7	8
213	The first Japanese familial case of spinocerebellar ataxia 23 with a novel mutation in the PDYN gene. Parkinsonism and Related Disorders, 2015, 21, 332-334.	1.1	8
214	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
215	Simultaneous detection of reduced and oxidized forms of coenzyme Q10 in human cerebral spinal fluid as a potential marker of oxidative stress. Journal of Clinical Biochemistry and Nutrition, 2018, 63, 205-210.	0.6	8
216	Functional evaluation of PDGFB-variants in idiopathic basal ganglia calcification, using patient-derived iPS cells. Scientific Reports, 2019, 9, 5698.	1.6	8

#	Article	IF	Citations
217	Recurrent cerebral aneurysm formation and rupture within a short period due to invasive aspergillosis of the nasal sinus; pathological analysis of the catastrophic clinical course. International Journal of Clinical and Experimental Pathology, 2015, 8, 13510-22.	0.5	8
218	Recurrent K3E mutation in Cu/Zn superoxide dismutase gene associated with amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 608-614.	1.1	7
219	Monitoring daily life activity shows less activity among female dementia patients. Neurology and Clinical Neuroscience, 2013, 1, 91-95.	0.2	7
220	A systematic immunoprecipitation approach reinforces the concept of common conformational alterations in amyotrophic lateral sclerosis-linked SOD1 mutants. Neurobiology of Disease, 2015, 82, 478-486.	2.1	7
221	Methylation changes and aberrant expression of FGFR3 in Lewy body disease neurons. Brain Research, 2018, 1697, 59-66.	1.1	7
222	Clinical features of inherited neuropathy with BSCL2 mutations in Japan. Journal of the Peripheral Nervous System, 2020, 25, 125-131.	1.4	7
223	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
224	Elevated Serum Uric Acid Levels Are Related to Cognitive Deterioration in an Elderly Japanese Population. Dementia and Geriatric Cognitive Disorders Extra, 2017, 6, 580-588.	0.6	6
225	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
226	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
227	A novel mutation in the GBA2 gene in a Japanese patient with SPG46: A case report. ENeurologicalSci, 2020, 19, 100238.	0.5	6
228	Splice-site mutations in KIF5A in the Japanese case series of amyotrophic lateral sclerosis. Neurogenetics, 2021, 22, 11-17.	0.7	6
229	Dentatorubral-pallidoluysian atrophy: clinical aspects and molecular genetics. Advances in Neurology, 2002, 89, 231-9.	0.8	6
230	Cerebellar Ataxia and Peripheral Neuropathy due to Chronic Bromvalerylurea Poisoning.. Internal Medicine, 1997, 36, 742-746.	0.3	5
231	Adultâ€onset vanishing white matter disease with novel missense mutations in a subunit of translational regulator, <scp>EIF2B4</scp>. Clinical Genetics, 2015, 88, 401-403.	1.0	5
232	Recovered recall memory after decompression of the fornix by surgical removal of pineal tumor. Neurology, 2016, 86, 790-791.	1.5	5
233	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
234	Isolated seizure as initial presentation of GABAA receptor antibody-associated encephalitis. Journal of the Neurological Sciences, 2020, 410, 116666.	0.3	5

#	Article	IF	Citations
235	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
236	Reliability and validity of Japanese version of Unified Multiple System Atrophy Rating Scale. Neurology and Clinical Neuroscience, 2021, 9, 171-180.	0.2	5
237	Synphilin-1 is present in Lewy bodies in Parkinson's disease. , 2000, 47, 521.		5
238	SNP Haplotype Mapping in a Small ALS Family. PLoS ONE, 2009, 4, e5687.	1.1	5
239	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
240	An NEFH founder mutation causes broad phenotypic spectrum in multiple Japanese families. Journal of Human Genetics, 2022, 67, 399-403.	1.1	5
241	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
242	Genetic polymorphism of the Tau gene and neurodegenerative diseases with Tau pathology among Japanese. Annals of Neurology, 1998, 44, 707-707.	2.8	4
243	Studies of the candidate genes in X-linked congenital cerebellar hypoplasia. Journal of Neurology, 1999, 246, 1177-1180.	1.8	4
244	Plasma Coenzyme Q10 Levels and Multiple System Atrophyâ€”Reply. JAMA Neurology, 2016, 73, 1499.	4.5	4
245	Novel SLC20A2 variant in a Japanese patient with idiopathic basal ganglia calcification-1 (IBGC1) associated with dopa-responsive parkinsonism. Human Genome Variation, 2019, 6, 44.	0.4	4
246	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
247	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
248	Linkage of autosomal recessive hereditary spastic paraplegia with mental impairment and thin corpus callosum to chromosome 15q13â€“15. , 2000, 48, 108.		4
249	An immigrant family with Kii amyotrophic lateral sclerosis/parkinsonismâ€“dementia complex. Neurological Sciences, 2022, 43, 1423-1425.	0.9	4
250	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
251	Effects of acidic phospholipids on antiganglioside antibodies in Guillain-BarrÃ© syndrome: Role of the disialosyl residue. Clinical and Experimental Neuroimmunology, 2013, 4, 70-74.	0.5	3
252	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

#	Article	IF	Citations
253	HIV Dementia with a Decreased Cardiac ¹²³I-metaiodobenzylguanidine Uptake Masquerading as Dementia with Lewy Bodies. Internal Medicine, 2018, 57, 3007-3010.	0.3	3
254	Oxygen consumption rate for evaluation of COQ2 variants associated with multiple system atrophy. Neurogenetics, 2019, 20, 51-52.	0.7	3
255	Familial dementia with Lewy bodies with VPS13C mutations. Parkinsonism and Related Disorders, 2020, 81, 31-33.	1.1	3
256	Clinical usefulness of multigene screening with phenotype-driven bioinformatics analysis for the diagnosis of patients with monogenic diabetes or severe insulin resistance. Diabetes Research and Clinical Practice, 2020, 169, 108461.	1.1	3
257	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
258	Quantitative Evaluation of Cerebellar Function in Multiple System Atrophy with Transcranial Magnetic Stimulation. Cerebellum, 2022, 21, 219-224.	1.4	3
259	Assignment of the human ST2 gene to chromosome 2 at q11.2. Human Genetics, 1996, 97, 561-563.	1.8	3
260	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
261	Letters to the editor. Muscle and Nerve, 1993, 16, 109-117.	1.0	2
262	Letters to the editor. Muscle and Nerve, 1994, 17, 112-120.	1.0	2
263	Gene Abnormalities in Medical Diseases. Neurodegenerative Diseases: Molecular Mechanisms of Spinocerebellar Ataxia.. Internal Medicine, 1997, 36, 154-156.	0.3	2
264	Alzheimer's disease as a polygenic disease. Neuropathology, 1998, 18, 111-115.	0.7	2
265	Exome sequencing shows a novel de novo mutation in <scp>ATL</scp>1. Neurology and Clinical Neuroscience, 2014, 2, 1-4.	0.2	2
266	Persistent pain as a non-motor symptom in corticobasal syndrome. Journal of Clinical Neuroscience, 2016, 29, 35-37.	0.8	2
267	Development of a novel system to quantify the spatialâ€“temporal parameters for crutch-assisted quadrupedal gait. Advanced Robotics, 2017, 31, 80-87.	1.1	2
268	Host MICA Polymorphism as a Potential Predictive Marker in Response to Chemotherapy for Colorectal Liver Metastases. Digestive Diseases, 2018, 36, 437-445.	0.8	2
269	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
270	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

#	Article	IF	Citations
271	ChÃ©diakâ€“Higashi syndrome presenting as a hereditary spastic paraplegia. Journal of Human Genetics, 2022, 67, 119-121.	1.1	2
272	Sporadic ataxias in Japan - a population-based epidemiological study. Cerebellum, 2008, 7, 1-9.	1.4	2
273	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
274	Complex hereditary peripheral neuropathies caused by novel variants in mitochondrial-related nuclear genes. Journal of Neurology, 2022, 269, 4129-4140.	1.8	2
275	Adrenoleukodystrophy with involvement of the cerebral cortex. Neuropathology, 1997, 17, 106-111.	0.7	1
276	MERRF/MELAS overlap syndrome associated with 3243 tRNALeu(UUR)mutation of mitochondrial DNA. Neuropathology, 1998, 18, 321-327.	0.7	1
277	O4-06-05: SORL1 is genetically associated with late-onset Alzheimer's disease in Japanese, Koreans and Caucasians. , 2013, 9, P693-P694.		1
278	P1-078: BIN1 REGULATES BACE1 TRAFFICKING AND AB PRODUCTION. , 2014, 10, P331-P332.		1
279	Adult onset ictal aphasia with epileptic discharges in Brocaâ€™s and Wernickeâ€™s areas. Clinical Neurophysiology, 2016, 127, 1754-1756.	0.7	1
280	Successful management of chronic myeloid leukemia with a complication of anti-SRP antibody-associated myopathy. Leukemia and Lymphoma, 2017, 58, 1242-1245.	0.6	1
281	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
282	Optic neuropathy and decorticate-like posture as presenting symptoms of Bickerstaffâ€™s brainstem encephalitis: A case report and literature review. Clinical Neurology and Neurosurgery, 2018, 173, 159-162.	0.6	1
283	Rituximab improves not only back stiffness but also â€œstiff eyesâ€•in stiff person syndrome: Implications for immune-mediated treatment. Journal of the Neurological Sciences, 2020, 408, 116506.	0.3	1
284	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
285	Premature saccades: A detailed physiological analysis. Clinical Neurophysiology, 2021, 132, 63-76.	0.7	1
286	Lack of association between dopamine D2 receptor gene Cys311 variant and schizophrenia. American Journal of Medical Genetics Part A, 1996, 67, 208-211.	2.4	1
287	Synphilin-1 is present in Lewy bodies in Parkinson's disease. , 2000, 47, 521.		1
288	Widespread occurrence of intranuclear atrophin-1 accumulation in the central nervous system neurons of patients with dentatorubral-pallidoluysian atrophy. , 2001, 49, 14.		1

#	Article	IF	Citations
289	An autopsy case of autosomal-recessive juvenile parkinsonism with a homozygous exon 4 deletion in the parkin gene. , 2000, 15, 884.		1
290	Polymyositis associated with focal mesangial proliferative glomerulonephritis with depositions of immune complexes. Clinical Rheumatology, 2007, 26, 792.	1.0	1
291	A clinical and genetic study of SPG31 in Japan. Journal of Human Genetics, 2022, , .	1.1	1
292	3P-212 Mercury Chloride Decreases the Water Permeability of Aquaporin-4-Reconstituted Proteoliposomes(The 46th Annual Meeting of the Biophysical Society of Japan). Seibutsu Butsuri, 2008, 48, S160.	0.0	0
293	HTLV-I-like sequence in MS. Acta Neurologica Scandinavica, 2009, 91, 516-517.	1.0	0
294	Aberrant expression of myogenin in inclusion body myositis: Immunohistochemical studies of transcription factors regulating myogenesis in inflammatory myopathies. Clinical and Experimental Neuroimmunology, 2012, 3, 129-137.	0.5	0
295	Treatable chorea associated with polycythemia vera. Neurology and Clinical Neuroscience, 2014, 2, 90-91.	0.2	0
296	Sensorimotor neuropathy in lateâ€onset <scp>K</scp>rabbe disease progressing over 40Âyears after onset. Neurology and Clinical Neuroscience, 2014, 2, 114-116.	0.2	0
297	Abadie's sign in adrenomyeloneuropathy. Journal of the Neurological Sciences, 2014, 340, 245-246.	0.3	0
298	Accumulation of transportin 1 in the fused in sarcomaâ€positive neuronal inclusions in sporadic amyotrophic lateral sclerosis without <scp>FUS</scp> mutation. Neurology and Clinical Neuroscience, 2015, 3, 194-196.	0.2	0
299	DNA Sequencing and Other Methods of Exonic and Genomic Analyses. , 2015, , 77-85.		0
300	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
301	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
302	Degeneration of the Substantia Nigra Following Ipsilateral Striatal Infarction. Internal Medicine, 2018, 57, 767-768.	0.3	0
303	Prominent Spasticity and Hyperreflexia of the Legs in a Nepalese Patient with Friedreich Ataxia. Internal Medicine, 2019, 58, 2865-2869.	0.3	0
304	Do eye movements â€œageâ€•earlier in progeria?. Clinical Neurophysiology, 2020, 131, 1835-1836.	0.7	0
305	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
306	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

#	Article	IF	Citations
307	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
308	Dentatorubral-pallidoluysian atrophy. , 2001, , 481-490.		0
309	Idiopathic Late Onset Cerebellar Ataxia (ILOCA), and Cerebellar plus Syndrome. , 2013, , 2143-2150.		0
310	Diagnosis and Management of Senile Dementia.. Journal of the Japanese Association of Rural Medicine, 1996, 44, 790-794.	0.0	0
311	Molecular Genetics of Neurologic Diseases. Japanese Journal of Neurosurgery, 1999, 8, 26-32.	0.0	0
312	DNA sequencing and other methods of exonic and genomic analyses. , 2020, , 109-120.		0
313	Idiopathic Late Onset Cerebellar Ataxia (ILOCA), and Cerebellar Plus Syndrome. , 2022, , 2433-2440.		0
314	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