Shoji Tsuji

List of Publications by Year
in descending order

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314 papers	16,346 citations	19657 61 h-index	19190 118 g-index
327 all docs	327 docs citations	327 times ranked	18597 citing authors

#	Article	IF	Citations
1	Quantitative Evaluation of Cerebellar Function in Multiple System Atrophy with Transcranial Magnetic Stimulation. Cerebellum, 2022, 21, 219-224.	2.5	3
2	Multiple system atrophy variant with severe hippocampal pathology. Brain Pathology, 2022, 32, e13002.	4.1	18
3	ChÃ©diakâ€“Higashi syndrome presenting as a hereditary spastic paraplegia. Journal of Human Genetics, 2022, 67, 119-121.	2.3	2
4	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.	0.7	2
5	An immigrant family with Kii amyotrophic lateral sclerosis/parkinsonismâ€“dementia complex. Neurological Sciences, 2022, 43, 1423-1425.	1.9	4
6	Idiopathic Late Onset Cerebellar Ataxia (ILOCA), and Cerebellar Plus Syndrome. , 2022, , 2433-2440.		0
7	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.	0.4	5
8	CAG repeat-binding small molecule improves motor coordination impairment in a mouse model of Dentatorubralâ€“pallidoluysian atrophy. Neurobiology of Disease, 2022, 163, 105604.	4.4	11
9	Muscle Transcriptomics Shows Overexpression of Cadherin 1 in Inclusion Body Myositis. Annals of Neurology, 2022, 91, 317-328.	5.3	9
10	Elderly patients with suspected Charcot-Marie-Tooth disease should be tested for the TTR gene for effective treatments. Journal of Human Genetics, 2022, , .	2.3	4
11	An NEFH founder mutation causes broad phenotypic spectrum in multiple Japanese families. Journal of Human Genetics, 2022, 67, 399-403.	2.3	5
12	A clinical and genetic study of SPG31 in Japan. Journal of Human Genetics, 2022, , .	2.3	1
13	Complex hereditary peripheral neuropathies caused by novel variants in mitochondrial-related nuclear genes. Journal of Neurology, 2022, 269, 4129-4140.	3.6	2
14	DMD exon 2 duplication due to a complex genomic rearrangement is associated with a somatic mosaicism. Neuromuscular Disorders, 2022, 32, 263-269.	0.6	5
15	The Movement Disorder Society Criteria for the Diagnosis of Multiple System Atrophy. Movement Disorders, 2022, 37, 1131-1148.	3.9	222
16	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.	3.7	7
17	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.	3.2	12
18	Juvenile amyotrophic lateral sclerosis with complex phenotypes associated with novel SYNE1 mutations. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2021, 22, 576-578.	1.7	9

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19	Low Prevalence of NOTCH2NLC GGC Repeat Expansion in White Patients with Movement Disorders. Movement Disorders, 2021, 36, 251-255.	3.9	23
20	Genetic spectrum of <scp>Charcotâ€“Marieâ€“Tooth</scp> disease associated with myelin protein zero gene variants in Japan. Clinical Genetics, 2021, 99, 359-375.	2.0	18
21	Premature saccades: A detailed physiological analysis. Clinical Neurophysiology, 2021, 132, 63-76.	1.5	1
22	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.	2.3	4
23	Splice-site mutations in KIF5A in the Japanese case series of amyotrophic lateral sclerosis. Neurogenetics, 2021, 22, 11-17.	1.4	6
24	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.	2.3	12
25	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.	3.3	19
26	SPG9A with the new occurrence of an ALDH18A1 mutation in a CMT1A family with PMP22 duplication: case report. BMC Neurology, 2021, 21, 64.	1.8	2
27	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
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.	3.9	5
29	Clinicopathologic Features of Oculopharyngodistal Myopathy WithLRP12CGG Repeat Expansions Compared With Other Oculopharyngodistal Myopathy Subtypes. JAMA Neurology, 2021, 78, 853.	9.0	30
30	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.	0.4	0
31	COQ2 V393A confers high risk susceptibility for multiple system atrophy in East Asian population. Journal of the Neurological Sciences, 2021, 429, 117623.	0.6	17
32	Reliability and validity of Japanese version of Unified Multiple System Atrophy Rating Scale. Neurology and Clinical Neuroscience, 2021, 9, 171-180.	0.4	5
33	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.	2.3	0
34	Frequency of FMR1 Premutation Alleles in Patients with Undiagnosed Cerebellar Ataxia and Multiple System Atrophy in the Japanese Population. Cerebellum, 2021, , 1.	2.5	3
35	Candesartan prevents arteriopathy progression in cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy model. Journal of Clinical Investigation, 2021, 131, .	8.2	12
36	CNV analysis using whole exome sequencing identified biallelic CNVs of VPS13B in siblings with intellectual disability. European Journal of Medical Genetics, 2020, 63, 103610.	1.3	14

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37	VPS13Dâ€related disorders presenting as a pure and complicated form of hereditary spastic paraplegia. Molecular Genetics & Genomic Medicine, 2020, 8, e1108.	1.2	29
38	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.	0.6	1
39	A Novel de novo KIF1A Mutation in a Patient with Autism, Hyperactivity, Epilepsy, Sensory Disturbance, and Spastic Paraplegia. Internal Medicine, 2020, 59, 839-842.	0.7	13
40	Familial dementia with Lewy bodies with VPS13C mutations. Parkinsonism and Related Disorders, 2020, 81, 31-33.	2.2	3
41	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.	2.8	3
42	Identification of a novel mutation in ATP13A2 associated with a complicated form of hereditary spastic paraplegia. Neurology: Genetics, 2020, 6, e514.	1.9	15
43	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.	0.6	2
44	Advances in repeat expansion diseases and a new concept of repeat motifâ€“phenotype correlation. Current Opinion in Genetics and Development, 2020, 65, 176-185.	3.3	30
45	Clinical efficacy of haematopoietic stem cell transplantation for adult adrenoleukodystrophy. Brain Communications, 2020, 2, fcz048.	3.3	14
46	Do eye movements â€œageâ€•earlier in progeria?. Clinical Neurophysiology, 2020, 131, 1835-1836.	1.5	0
47	Comprehensive investigation of RNF213 nonsynonymous variants associated with intracranial artery stenosis. Scientific Reports, 2020, 10, 11942.	3.3	11
48	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, .	3.2	17
49	Clinical Characteristics of Neuronal Intranuclear Inclusion Disease-Related Retinopathy With CGG Repeat Expansions in the NOTCH2NLC Gene. , 2020, 61, 27.		19
50	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.	0.6	3
51	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.	1.2	1
52	A novel mutation in ABCD1 gene in a Filipino patient with adultâ€onset Xâ€linked ALD. Neurology and Clinical Neuroscience, 2020, 8, 329-331.	0.4	0
53	Clinical features of inherited neuropathy with BSCL2 mutations in Japan. Journal of the Peripheral Nervous System, 2020, 25, 125-131.	3.1	7
54	A novel mutation in the GBA2 gene in a Japanese patient with SPG46: A case report. ENeurologicalSci, 2020, 19, 100238.	1.3	6

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55	Neuron-specific analysis of histone modifications with post-mortem brains. Scientific Reports, 2020, 10, 3767.	3.3	10
56	Isolated seizure as initial presentation of GABAA receptor antibody-associated encephalitis. Journal of the Neurological Sciences, 2020, 410, 116666.	0.6	5
57	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.	2.2	15
58	DNA sequencing and other methods of exonic and genomic analyses. , 2020, , 109-120.		0
59	Prominent Spasticity and Hyperreflexia of the Legs in a Nepalese Patient with Friedreich Ataxia. Internal Medicine, 2019, 58, 2865-2869.	0.7	0
60	Noncoding CGG repeat expansions in neuronal intranuclear inclusion disease, oculopharyngodistal myopathy and an overlapping disease. Nature Genetics, 2019, 51, 1222-1232.	21.4	265
61	Increased facilitation of the primary motor cortex in de novo Parkinson's disease. Parkinsonism and Related Disorders, 2019, 66, 125-129.	2.2	20
62	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	12.8	99
63	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.	0.7	4
64	UBAP1 mutations cause juvenile-onset hereditary spastic paraplegias (SPG80) and impair UBAP1 targeting to endosomes. Journal of Human Genetics, 2019, 64, 1055-1065.	2.3	15
65	Oxygen consumption rate for evaluation of COQ2 variants associated with multiple system atrophy. Neurogenetics, 2019, 20, 51-52.	1.4	3
66	Ataxic phenotype with altered CaV3.1 channel property in a mouse model for spinocerebellar ataxia 42. Neurobiology of Disease, 2019, 130, 104516.	4.4	20
67	A critique of the second consensus criteria for multiple system atrophy. Movement Disorders, 2019, 34, 975-984.	3.9	73
68	Functional evaluation of PDGFB-variants in idiopathic basal ganglia calcification, using patient-derived iPS cells. Scientific Reports, 2019, 9, 5698.	3.3	8
69	Chronic cerebral hypoperfusion shifts the equilibrium of amyloid Î² oligomers to aggregation-prone species with higher molecular weight. Scientific Reports, 2019, 9, 2827.	3.3	27
70	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.	1.4	5
71	Neuroimaging, genetic, and enzymatic study in a Japanese family with a GBA gross deletion. Parkinsonism and Related Disorders, 2019, 61, 57-63.	2.2	6
72	The novel de novo mutation of KIF1A gene as the cause for Spastic paraplegia 30 in a Japanese case. ENeurologicalSci, 2019, 14, 34-37.	1.3	18

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73	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.	2.3	4
74	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.	1.9	28
75	PLA2G6-associated neurodegeneration presenting as a complicated form of hereditary spastic paraplegia. Journal of Human Genetics, 2019, 64, 55-59.	2.3	17
76	ExpansionsÂofÂintronic TTTCA and TTTTA repeats in benign adult familial myoclonic epilepsy. Nature Genetics, 2018, 50, 581-590.	21.4	238
77	Safety and efficacy of eculizumab in Guillain-BarrÃ© syndrome: a multicentre, double-blind, randomised phase 2 trial. Lancet Neurology, The, 2018, 17, 519-529.	10.2	111
78	No novel, high penetrant gene might remain to be found in Japanese patients with unknown MODY. Journal of Human Genetics, 2018, 63, 821-829.	2.3	3
79	Amnionless-mediated glycosylation is crucial for cell surface targeting of cubilin in renal and intestinal cells. Scientific Reports, 2018, 8, 2351.	3.3	27
80	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.	3.1	17
81	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.	2.1	11
82	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.	3.1	15
83	Mutations in COA7 cause spinocerebellar ataxia with axonal neuropathy. Brain, 2018, 141, 1622-1636.	7.6	38
84	A Homozygous LAMA2 Mutation of c.818G>A Caused Partial Merosin Deficiency in a Japanese Patient. Internal Medicine, 2018, 57, 877-882.	0.7	6
85	Authors' reply to the Drs. Finsterer and Zarrouk-Mahjoub's comments for our case report. International Journal of Cardiology, 2018, 254, 262.	1.7	1
86	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.	3.1	37
87	Novel De Novo KCND3 Mutation in a Japanese Patient with Intellectual Disability, Cerebellar Ataxia, Myoclonus, and Dystonia. Cerebellum, 2018, 17, 237-242.	2.5	21
88	The pathogenesis linked to coenzyme Q10 insufficiency in iPSC-derived neurons from patients with multiple-system atrophy. Scientific Reports, 2018, 8, 14215.	3.3	50
89	Effect of subthalamic nucleus deep brain stimulation on visual scanning. Clinical Neurophysiology, 2018, 129, 2421-2432.	1.5	9
90	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.	1.4	8

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91	HIV Dementia with a Decreased Cardiac ¹²³I-metaiodobenzylguanidine Uptake Masquerading as Dementia with Lewy Bodies. Internal Medicine, 2018, 57, 3007-3010.	0.7	3
92	Host MICA Polymorphism as a Potential Predictive Marker in Response to Chemotherapy for Colorectal Liver Metastases. Digestive Diseases, 2018, 36, 437-445.	1.9	2
93	Intronic pentanucleotide TTTCA repeat insertion in the SAMD12 gene causes familial cortical myoclonic tremor with epilepsy type 1. Brain, 2018, 141, 2280-2288.	7.6	73
94	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.	2.1	15
95	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.	1.4	1
96	JASPAC: Japan Spastic Paraplegia Research Consortium. Brain Sciences, 2018, 8, 153.	2.3	41
97	An Autopsy Case of Familial Neuronal Intranuclear Inclusion Disease with Dementia and Neuropathy. Internal Medicine, 2018, 57, 3459-3462.	0.7	21
98	Degeneration of the Substantia Nigra Following Ipsilateral Striatal Infarction. Internal Medicine, 2018, 57, 767-768.	0.7	0
99	Methylation changes and aberrant expression of FGFR3 in Lewy body disease neurons. Brain Research, 2018, 1697, 59-66.	2.2	7
100	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.	2.3	18
101	SCA42 mutation analysis in a case series of Japanese patients with spinocerebellar ataxia. Journal of Human Genetics, 2017, 62, 857-859.	2.3	25
102	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.	1.7	17
103	Tacrolimus-Induced Reversible Cerebral Vasoconstriction Syndrome with Delayed Multi-Segmental Vasoconstriction. Journal of Stroke and Cerebrovascular Diseases, 2017, 26, e75-e77.	1.6	16
104	Authors' response to â€œCompound heterozygous Fukutin mutation-related non-compactionâ€•by Finsterer and Zarrouk-Mahjoub. International Journal of Cardiology, 2017, 233, 102.	1.7	0
105	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.	0.6	12
106	Slowly progressive d-bifunctional protein deficiency with survival to adulthood diagnosed by whole-exome sequencing. Journal of the Neurological Sciences, 2017, 372, 6-10.	0.6	8
107	Altered expression of Crb2 in podocytes expands a variation of CRB2 mutations in steroid-resistant nephrotic syndrome. Pediatric Nephrology, 2017, 32, 801-809.	1.7	12
108	TBCD may be a causal gene in progressive neurodegenerative encephalopathy with atypical infantile spinal muscular atrophy. Journal of Human Genetics, 2017, 62, 473-480.	2.3	15

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109	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.	7.1	72
110	Development of a novel system to quantify the spatialâ€“temporal parameters for crutch-assisted quadrupedal gait. Advanced Robotics, 2017, 31, 80-87.	1.8	2
111	Structural Basis and Genotypeâ€“Phenotype Correlations of INSR Mutations Causing Severe Insulin Resistance. Diabetes, 2017, 66, 2713-2723.	0.6	28
112	Clinicopathologic features of myositis patients with CD8-MHC-1 complex pathology. Neurology, 2017, 89, 1060-1068.	1.1	29
113	Partial duplication of <scp>DHH</scp> causes minifascicular neuropathy. Annals of Clinical and Translational Neurology, 2017, 4, 415-421.	3.7	14
114	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.	2.5	35
115	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.	3.1	31
116	Successful management of chronic myeloid leukemia with a complication of anti-SRP antibody-associated myopathy. Leukemia and Lymphoma, 2017, 58, 1242-1245.	1.3	1
117	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.	1.3	6
118	Plasma Coenzyme Q10 Levels in Patients With Multiple System Atrophy. JAMA Neurology, 2016, 73, 977.	9.0	42
119	Cancer association as a risk factor for anti-HMGCR antibody-positive myopathy. Neurology: Neuroimmunology and NeuroInflammation, 2016, 3, e290.	6.0	71
120	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.	1.4	11
121	Tubular aggregate myopathy caused by a novel mutation in the cytoplasmic domain of STIM1. Neurology: Genetics, 2016, 2, e50.	1.9	27
122	Atypical parkinsonism caused by Pro105Leu mutation of prion protein. Neurology: Genetics, 2016, 2, e48.	1.9	12
123	BIN1 regulates BACE1 intracellular trafficking and amyloid-Î² production. Human Molecular Genetics, 2016, 25, ddw146.	2.9	67
124	AgIn: measuring the landscape of CpG methylation of individual repetitive elements. Bioinformatics, 2016, 32, 2911-2919.	4.1	29
125	Fukutin gene mutations that cause left ventricular noncompaction. International Journal of Cardiology, 2016, 222, 727-729.	1.7	14
126	Mutations in MME cause an autosomalâ€recessive Charcotâ€“Marieâ€“Tooth disease type 2. Annals of Neurology, 2016, 79, 659-672.	5.3	82

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127	Novel COL6A2 mutation in a case of limb girdle muscular dystrophy phenotype with autosomal recessive inheritance. Neurology and Clinical Neuroscience, 2016, 4, 189-191.	0.4	0
128	Human genetic variation database, a reference database of genetic variations in the Japanese population. Journal of Human Genetics, 2016, 61, 547-553.	2.3	270
129	Plasma Coenzyme Q10 Levels and Multiple System Atrophyâ€"Reply. JAMA Neurology, 2016, 73, 1499.	9.0	4
130	Modeling neurological diseases with induced pluripotent cells reprogrammed from immortalized lymphoblastoid cell lines. Molecular Brain, 2016, 9, 88.	2.6	21
131	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. Nature Communications, 2016, 7, 11253.	12.8	174
132	Assessing Cell-to-Cell DNA Methylation Variability on Individual Long Reads. Scientific Reports, 2016, 6, 21317.	3.3	11
133	Next-generation sequencing of 28 ALS-related genes in a Japanese ALS cohort. Neurobiology of Aging, 2016, 39, 219.e1-219.e8.	3.1	49
134	Genomic Landscape of Esophageal Squamous Cell Carcinoma inÂa Japanese Population. Gastroenterology, 2016, 150, 1171-1182.	1.3	265
135	Persistent pain as a non-motor symptom in corticobasal syndrome. Journal of Clinical Neuroscience, 2016, 29, 35-37.	1.5	2
136	Recovered recall memory after decompression of the fornix by surgical removal of pineal tumor. Neurology, 2016, 86, 790-791.	1.1	5
137	Adult onset ictal aphasia with epileptic discharges in Brocaâ€™s and Wernickeâ€™s areas. Clinical Neurophysiology, 2016, 127, 1754-1756.	1.5	1
138	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.	1.9	33
139	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.	1.0	18
140	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.	0.4	0
141	Late-onset spastic ataxia phenotype in a patient with a homozygous DDHD2 mutation. Scientific Reports, 2015, 4, 7132.	3.3	29
142	The 3-Second Rule in Hereditary Pure Cerebellar Ataxia: A Synchronized Tapping Study. PLoS ONE, 2015, 10, e0118592.	2.5	17
143	DNA Sequencing and Other Methods of Exonic and Genomic Analyses. , 2015, , 77-85.		0
144	Modulation of error-sensitivity during a prism adaptation task in people with cerebellar degeneration. Journal of Neurophysiology, 2015, 114, 2460-2471.	1.8	43

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145	Novel mutations in the PNPLA6 gene in Boucher-NeuhÃuser syndrome. Journal of Human Genetics, 2015, 60, 217-220.	2.3	15
146	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.	2.0	5
147	A Novel Mutation inELOVL4Leading to Spinocerebellar Ataxia (SCA) With the Hot Cross Bun Sign but Lacking Erythrokeratodermia. JAMA Neurology, 2015, 72, 797.	9.0	79
148	Variants associated with Gaucher disease in multiple system atrophy. Annals of Clinical and Translational Neurology, 2015, 2, 417-426.	3.7	90
149	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.	2.2	8
150	Epidemiology and molecular mechanism of frontotemporal lobar degeneration/amyotrophic lateral sclerosis with repeat expansion mutation inC9orf72. Journal of Neurogenetics, 2015, 29, 85-94.	1.4	17
151	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.	4.4	7
152	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
153	Mutant COQ2 in Multiple-System Atrophy. New England Journal of Medicine, 2014, 371, 80-83.	27.0	81
154	Treatable chorea associated with polycythemia vera. Neurology and Clinical Neuroscience, 2014, 2, 90-91.	0.4	0
155	Sensorimotor neuropathy in lateâ€onset <scp>K</scp>rabbe disease progressing over 40Âyears after onset. Neurology and Clinical Neuroscience, 2014, 2, 114-116.	0.4	0
156	Rapid detection of expanded short tandem repeats in personal genomics using hybrid sequencing. Bioinformatics, 2014, 30, 815-822.	4.1	61
157	Evaluation of SLC20A2 mutations that cause idiopathic basal ganglia calcification in Japan. Neurology, 2014, 82, 705-712.	1.1	71
158	Unilateral Opercular Infarction Presenting with Foix-Chavany-Marie Syndrome. Journal of Stroke and Cerebrovascular Diseases, 2014, 23, 179-181.	1.6	16
159	Triad-conditioning Transcranial Magnetic Stimulation in Parkinson's Disease. Brain Stimulation, 2014, 7, 74-79.	1.6	8
160	Exome sequencing shows a novel de novo mutation in <scp>ATL</scp>1. Neurology and Clinical Neuroscience, 2014, 2, 1-4.	0.4	2
161	Modulation of the age at onset in spinocerebellar ataxia by CAG tracts in various genes. Brain, 2014, 137, 2444-2455.	7.6	144
162	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.	2.3	53

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163	Genomic aspects of sporadic neurodegenerative diseases. Biochemical and Biophysical Research Communications, 2014, 452, 221-225.	2.1	20
164	A 3-year cohort study of the natural history of spinocerebellar ataxia type 6 in Japan. Orphanet Journal of Rare Diseases, 2014, 9, 118.	2.7	35
165	Autosomal-recessive complicated spastic paraplegia with a novel lysosomal trafficking regulator gene mutation. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 1024-1028.	1.9	27
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