

# Shoji Tsuji

## List of Publications by Year in descending order

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314  
papers

16,346  
citations

19657

61  
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19190

118  
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327  
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327  
docs citations

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times ranked

18597  
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. <i>Lancet Neurology</i> , The, 2012, 11, 323-330.	10.2	1,039
2	Interference by Huntingtin and Atrophin-1 with CBP-Mediated Transcription Leading to Cellular Toxicity. <i>Science</i> , 2001, 291, 2423-2428.	12.6	1,035
3	Hereditary progressive dystonia with marked diurnal fluctuation caused by mutations in the GTP cyclohydrolase I gene. <i>Nature Genetics</i> , 1994, 8, 236-242.	21.4	800
4	Î±-Synuclein immunoreactivity in glial cytoplasmic inclusions in multiple system atrophy. <i>Neuroscience Letters</i> , 1998, 249, 180-182.	2.1	581
5	SCA17, a novel autosomal dominant cerebellar ataxia caused by an expanded polyglutamine in TATA-binding protein. <i>Human Molecular Genetics</i> , 2001, 10, 1441-1448.	2.9	569
6	Association of HTRA1 Mutations and Familial Ischemic Cerebral Small-Vessel Disease. <i>New England Journal of Medicine</i> , 2009, 360, 1729-1739.	27.0	407
7	Expanded polyglutamine stretches interact with TAFII130, interfering with CREB-dependent transcription. <i>Nature Genetics</i> , 2000, 26, 29-36.	21.4	388
8	Early-onset ataxia with ocular motor apraxia and hypoalbuminemia is caused by mutations in a new HIT superfamily gene. <i>Nature Genetics</i> , 2001, 29, 184-188.	21.4	376
9	Suppression of aggregate formation and apoptosis by transglutaminase inhibitors in cells expressing truncated DRPLA protein with an expanded polyglutamine stretch. <i>Nature Genetics</i> , 1998, 18, 111-117.	21.4	372
10	Mis-sense mutation Val&arr;lle in exon 17 of amyloid precursor protein gene in Japanese familial Alzheimer's disease. <i>Lancet</i> , The, 1991, 337, 978-979.	18.7	311
11	Human genetic variation database, a reference database of genetic variations in the Japanese population. <i>Journal of Human Genetics</i> , 2016, 61, 547-553.	2.3	270
12	Genomic Landscape of Esophageal Squamous Cell Carcinoma inÂa Japanese Population. <i>Gastroenterology</i> , 2016, 150, 1171-1182.	1.3	265
13	Noncoding CGG repeat expansions in neuronal intranuclear inclusion disease, oculopharyngodistal myopathy and an overlapping disease. <i>Nature Genetics</i> , 2019, 51, 1222-1232.	21.4	265
14	Spinocerebellar Ataxia Type 31 Is Associated with â€œInsertedâ€•Penta-Nucleotide Repeats Containing (TGGAA)n. <i>American Journal of Human Genetics</i> , 2009, 85, 544-557.	6.2	260
15	Synphilin-1 is present in Lewy bodies in Parkinson's disease. <i>Annals of Neurology</i> , 2000, 47, 521-523.	5.3	246
16	Expansions ofÎintrinsic TTTCA and TTTTA repeats in benign adult familial myoclonic epilepsy. <i>Nature Genetics</i> , 2018, 50, 581-590.	21.4	238
17	The Movement Disorder Society Criteria for the Diagnosis of Multiple System Atrophy. <i>Movement Disorders</i> , 2022, 37, 1131-1148.	3.9	222
18	An autopsy case of autosomal-recessive juvenile parkinsonism with a homozygous exon 4 deletion in the parkin gene. <i>Movement Disorders</i> , 2000, 15, 884-888.	3.9	220

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19	Mutations for Gaucher Disease Confer High Susceptibility to Parkinson Disease. <i>Archives of Neurology</i> , 2009, 66, 571-6.	4.5	183
20	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. <i>Nature Communications</i> , 2016, 7, 11253.	12.8	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. <i>Lancet Neurology</i> , The, 2010, 9, 875-884.	10.2	170
22	Genetic association of the very low density lipoprotein (VLDL) receptor gene with sporadic Alzheimer's disease. <i>Nature Genetics</i> , 1995, 11, 207-209.	21.4	169
23	Dentatorubral-pallidoluysian atrophy: Clinical features are closely related to unstable expansions of trinucleotide (CAG) repeat. <i>Annals of Neurology</i> , 1995, 37, 769-775.	5.3	154
24	ApoE $\epsilon$ 4 and early-onset Alzheimer's. <i>Nature Genetics</i> , 1994, 7, 10-11.	21.4	145
25	Modulation of the age at onset in spinocerebellar ataxia by CAG tracts in various genes. <i>Brain</i> , 2014, 137, 2444-2455.	7.6	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. <i>Annals of Neurology</i> , 2000, 48, 156-163.	5.3	136
27	Sporadic ataxias in Japan – a population-based epidemiological study. <i>Cerebellum</i> , 2008, 7, 189-197.	2.5	131
28	Multiplex Families With Multiple System Atrophy. <i>Archives of Neurology</i> , 2007, 64, 545.	4.5	127
29	ERBB4 Mutations that Disrupt the Neuregulin-ErbB4 Pathway Cause Amyotrophic Lateral Sclerosis Type 19. <i>American Journal of Human Genetics</i> , 2013, 93, 900-905.	6.2	123
30	Safety and efficacy of eculizumab in Guillain-Barré syndrome: a multicentre, double-blind, randomised phase 2 trial. <i>Lancet Neurology</i> , The, 2018, 17, 519-529.	10.2	111
31	Novel mutations, pseudo-dominant inheritance, and possible familial affects in patients with autosomal recessive juvenile parkinsonism. <i>Annals of Neurology</i> , 2000, 48, 245-250.	5.3	102
32	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. <i>Nature Communications</i> , 2019, 10, 4920.	12.8	99
33	Age at onset influences on wide-ranged clinical features of sporadic amyotrophic lateral sclerosis. <i>Journal of the Neurological Sciences</i> , 2009, 276, 163-169.	0.6	98
34	The TRK-Fused Gene Is Mutated in Hereditary Motor and Sensory Neuropathy with Proximal Dominant Involvement. <i>American Journal of Human Genetics</i> , 2012, 91, 320-329.	6.2	98
35	Gene locus for autosomal recessive distal myopathy with rimmed vacuoles maps to chromosome 9. <i>Annals of Neurology</i> , 1997, 41, 432-437.	5.3	97
36	Trinucleotide repeat length and rate of progression of Huntington's disease. <i>Annals of Neurology</i> , 1994, 36, 630-635.	5.3	95

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37	Analysis of the expression level of $\alpha$ -synuclein mRNA using postmortem brain samples from pathologically confirmed cases of multiple system atrophy. <i>Acta Neuropathologica</i> , 2001, 102, 188-190.	7.7	95
38	Widespread occurrence of intranuclear atrophin-1 accumulation in the central nervous system neurons of patients with dentatorubral-pallidolusian atrophy. <i>Annals of Neurology</i> , 2001, 49, 14-23.	5.3	94
39	Variants associated with Gaucher disease in multiple system atrophy. <i>Annals of Clinical and Translational Neurology</i> , 2015, 2, 417-426.	3.7	90
40	Hereditary dentatorubral-pallidolusian atrophy: detection of widespread ubiquitinated neuronal and glial intranuclear inclusions in the brain. <i>Acta Neuropathologica</i> , 1998, 96, 547-552.	7.7	88
41	C9ORF72 Repeat Expansion in Amyotrophic Lateral Sclerosis in the Kii Peninsula of Japan. <i>Archives of Neurology</i> , 2012, 69, 1154-8.	4.5	88
42	Mechanisms of Genomic Instabilities Underlying Two Common Fragile-Site-Associated Loci, PARK2 and DMD, in Germ Cell and Cancer Cell Lines. <i>American Journal of Human Genetics</i> , 2010, 87, 75-89.	6.2	85
43	Mutations in <i>MME</i> cause an autosomal recessive Charcot-Marie-Tooth disease type 2. <i>Annals of Neurology</i> , 2016, 79, 659-672.	5.3	82
44	Mutant <i>COQ2</i> in Multiple-System Atrophy. <i>New England Journal of Medicine</i> , 2014, 371, 80-83.	27.0	81
45	Exome Sequencing Reveals a Homozygous SYT14 Mutation in Adult-Onset, Autosomal-Recessive Spinocerebellar Ataxia with Psychomotor Retardation. <i>American Journal of Human Genetics</i> , 2011, 89, 320-327.	6.2	79
46	A Novel Mutation in <i>ELOVL4</i> Leading to Spinocerebellar Ataxia (SCA) With the Hot Cross Bun Sign but Lacking Erythrokeratoderma. <i>JAMA Neurology</i> , 2015, 72, 797.	9.0	79
47	Aprataxin, the causative protein for EAOH is a nuclear protein with a potential role as a DNA repair protein. <i>Annals of Neurology</i> , 2004, 55, 241-249.	5.3	76
48	A homozygous mutation of <i>C12orf65</i> causes spastic paraplegia with optic atrophy and neuropathy (SPG55). <i>Journal of Medical Genetics</i> , 2012, 49, 777-784.	3.2	76
49	Pathology of CAG repeat diseases. <i>Neuropathology</i> , 2000, 20, 319-325.	1.2	74
50	Intronic pentanucleotide TTCA repeat insertion in the SAMD12 gene causes familial cortical myoclonic tremor with epilepsy type 1. <i>Brain</i> , 2018, 141, 2280-2288.	7.6	73
51	A critique of the second consensus criteria for multiple system atrophy. <i>Movement Disorders</i> , 2019, 34, 975-984.	3.9	73
52	Quantitative evaluation of the pyramidal tract segmented by diffusion tensor tractography: feasibility study in patients with amyotrophic lateral sclerosis. <i>Radiation Medicine</i> , 2005, 23, 195-9.	0.8	73
53	Genetics of neurodegenerative diseases: insights from high-throughput resequencing. <i>Human Molecular Genetics</i> , 2010, 19, R65-R70.	2.9	72
54	Neuron-specific methylome analysis reveals epigenetic regulation and tau-related dysfunction of BRCA1 in Alzheimer's disease. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017, 114, E9645-E9654.	7.1	72

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55	Evaluation of <i>SLC20A2</i> mutations that cause idiopathic basal ganglia calcification in Japan. <i>Neurology</i> , 2014, 82, 705-712.	1.1	71
56	Cancer association as a risk factor for anti-HMGCR antibody-positive myopathy. <i>Neurology: Neuroimmunology and NeuroInflammation</i> , 2016, 3, e290.	6.0	71
57	Progressive atrophy of cerebellum and brainstem as a function of age and the size of the expanded CAG repeats in the MJD1 gene in Machado-Joseph disease. <i>Annals of Neurology</i> , 1998, 43, 288-296.	5.3	70
58	Linkage of autosomal recessive hereditary spastic paraplegia with mental impairment and thin corpus callosum to chromosome 15q13-15. <i>Annals of Neurology</i> , 2000, 48, 108-112.	5.3	70
59	BIN1 regulates BACE1 intracellular trafficking and amyloid- $\beta$ production. <i>Human Molecular Genetics</i> , 2016, 25, ddw146.	2.9	67
60	Pathology of CAG repeat diseases. <i>Neuropathology</i> , 2000, 20, 319-325.	1.2	67
61	No mutation in the entire coding region of the $\alpha$ -synuclein gene in pathologically confirmed cases of multiple system atrophy. <i>Neuroscience Letters</i> , 1999, 270, 110-112.	2.1	66
62	A novel monoclonal antibody reveals a conformational alteration shared by amyotrophic lateral sclerosis-linked SOD1 mutants. <i>Annals of Neurology</i> , 2012, 72, 739-749.	5.3	65
63	A novel ferritin light chain gene mutation in a Japanese family with neuroferritinopathy: Description of clinical features and implications for genotype-phenotype correlations. <i>Movement Disorders</i> , 2009, 24, 441-445.	3.9	64
64	Dentatorubral-pallidoluysian atrophy. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2012, 103, 587-594.	1.8	64
65	Rapid detection of expanded short tandem repeats in personal genomics using hybrid sequencing. <i>Bioinformatics</i> , 2014, 30, 815-822.	4.1	61
66	Adult onset globoid cell leukodystrophy (Krabbe disease): analysis of galactosylceramidase cDNA from four Japanese patients. <i>Human Genetics</i> , 1997, 100, 450-456.	3.8	60
67	14-3-3 protein $\gamma$ chain gene (YWHAH) polymorphism and its genetic association with schizophrenia. <i>American Journal of Medical Genetics Part A</i> , 1999, 88, 164-167.	2.4	60
68	Aprataxin, causative gene product for EAOH/AOA1, repairs DNA single-strand breaks with damaged 3'-phosphate and 3'-phosphoglycolate ends. <i>Nucleic Acids Research</i> , 2007, 35, 3797-3809.	14.5	60
69	Fatty Acid Elongation Activity in Fibroblasts from Patients with Adrenoleukodystrophy (ALD)1. <i>Journal of Biochemistry</i> , 1984, 96, 1241-1247.	1.7	58
70	Autosomal dominant cerebellar ataxia (SCA6): clinical, genetic and neuropathological study in a family. <i>Acta Neuropathologica</i> , 1998, 95, 333-337.	7.7	58
71	Mutational Analysis and Genotype-Phenotype Correlation of 29 Unrelated Japanese Patients With X-linked Adrenoleukodystrophy. <i>Archives of Neurology</i> , 1999, 56, 295.	4.5	58
72	The FHA domain of aprataxin interacts with the C-terminal region of XRCC1. <i>Biochemical and Biophysical Research Communications</i> , 2004, 325, 1279-1285.	2.1	56

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73	A functional variant in ZNF512B is associated with susceptibility to amyotrophic lateral sclerosis in Japanese. <i>Human Molecular Genetics</i> , 2011, 20, 3684-3692.	2.9	53
74	Molecular epidemiology and clinical spectrum of hereditary spastic paraplegia in the Japanese population based on comprehensive mutational analyses. <i>Journal of Human Genetics</i> , 2014, 59, 163-172.	2.3	53
75	The pathogenesis linked to coenzyme Q10 insufficiency in iPSC-derived neurons from patients with multiple-system atrophy. <i>Scientific Reports</i> , 2018, 8, 14215.	3.3	50
76	Dentatorubral-pallidoluysian atrophy (DRPLA): Close correlation of CAG repeat expansions with the wide spectrum of clinical presentations and prominent anticipation. <i>Seminars in Cell Biology</i> , 1995, 6, 37-44.	3.4	49
77	Next-generation sequencing of 28 ALS-related genes in a Japanese ALS cohort. <i>Neurobiology of Aging</i> , 2016, 39, 219.e1-219.e8.	3.1	49
78	Epidemiology of X-linked adrenoleukodystrophy in Japan. <i>Journal of Human Genetics</i> , 2002, 47, 0590-0593.	2.3	45
79	Toxicity of expanded polyglutamine-domain proteins in <i>Escherichia coli</i> . <i>FEBS Letters</i> , 1996, 399, 135-139.	2.8	44
80	Development of a High-Throughput Microarray-Based Resequencing System for Neurological Disorders and Its Application to Molecular Genetics of Amyotrophic Lateral Sclerosis. <i>Archives of Neurology</i> , 2008, 65, 1326-32.	4.5	44
81	Modulation of error-sensitivity during a prism adaptation task in people with cerebellar degeneration. <i>Journal of Neurophysiology</i> , 2015, 114, 2460-2471.	1.8	43
82	TDP-43 M337V Mutation in Familial Amyotrophic Lateral Sclerosis in Japan. <i>Internal Medicine</i> , 2010, 49, 331-334.	0.7	42
83	Plasma Coenzyme Q10 Levels in Patients With Multiple System Atrophy. <i>JAMA Neurology</i> , 2016, 73, 977.	9.0	42
84	Hereditary dentatorubral-pallidoluysian atrophy: ubiquitinated filamentous inclusions in the cerebellar dentate nucleus neurons. <i>Acta Neuropathologica</i> , 1998, 95, 479-482.	7.7	41
85	JASPAC: Japan Spastic Paraplegia Research Consortium. <i>Brain Sciences</i> , 2018, 8, 153.	2.3	41
86	MITOCHONDRIAL DNA MUTATION IN FAMILY WITH LEBER'S HEREDITARY OPTIC NEUROPATHY. <i>Lancet</i> , The, 1989, 333, 1076-1077.	13.7	40
87	Appropriate data cleaning methods for genome-wide association study. <i>Journal of Human Genetics</i> , 2008, 53, 886-893.	2.3	40
88	Apolipoprotein E $\epsilon$ 4 allele and progression of cortical Lewy body pathology in Parkinson's disease. <i>Acta Neuropathologica</i> , 1998, 95, 450-454.	7.7	38
89	Autosomal dominant diffuse Lewy body disease. <i>Acta Neuropathologica</i> , 1998, 96, 207-210.	7.7	38
90	Severe neurological phenotypes of Q129 DRPLA transgenic mice serendipitously created by en masse expansion of CAG repeats in Q76 DRPLA mice. <i>Human Molecular Genetics</i> , 2009, 18, 723-736.	2.9	38

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91	Posterior column ataxia with retinitis pigmentosa in a Japanese family with a novel mutation in FLVCR1. <i>Neurogenetics</i> , 2011, 12, 117-121.	1.4	38
92	Mutations in COA7 cause spinocerebellar ataxia with axonal neuropathy. <i>Brain</i> , 2018, 141, 1622-1636.	7.6	38
93	Lack of association of very low density lipoprotein receptor gene polymorphism with caucasian Alzheimer's disease. <i>Annals of Neurology</i> , 1996, 40, 251-254.	5.3	37
94	Molecular epidemiological study of familial amyotrophic lateral sclerosis in Japanese population by whole-exome sequencing and identification of novel HNRNPA1 mutation. <i>Neurobiology of Aging</i> , 2018, 61, 255.e9-255.e16.	3.1	37
95	Interaction of expanded polyglutamine stretches with nuclear transcription factors leads to aberrant transcriptional regulation in polyglutamine diseases. <i>Neuropathology</i> , 2000, 20, 326-333.	1.2	36
96	Mitochondrial ND3 as the novel causative gene for Leber hereditary optic neuropathy and dystonia. <i>Neurogenetics</i> , 2009, 10, 337-345.	1.4	35
97	A 3-year cohort study of the natural history of spinocerebellar ataxia type 6 in Japan. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 118.	2.7	35
98	Three-Year Follow-Up of High-Dose Ubiquinol Supplementation in a Case of Familial Multiple System Atrophy with Compound Heterozygous COQ2 Mutations. <i>Cerebellum</i> , 2017, 16, 664-672.	2.5	35
99	<i>CSF1R</i> mutations identified in three families with autosomal dominantly inherited leukoencephalopathy. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2012, 159B, 951-957.	1.7	34
100	X-linked nonprogressive congenital cerebellar hypoplasia: Clinical description and mapping to chromosome Xq. <i>Annals of Neurology</i> , 1996, 40, 75-83.	5.3	33
101	A rapid functional decline type of amyotrophic lateral sclerosis is linked to low expression of <i>TTN</i> . <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, 851-858.	1.9	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. <i>Muscle and Nerve</i> , 1994, 17, 1073-1075.	2.2	32
103	Adult-onset leukoencephalopathies with vanishing white matter with novel missense mutations in EIF2B2, EIF2B3, and EIF2B5. <i>Neurogenetics</i> , 2011, 12, 259-261.	1.4	32
104	<i>TRPM7</i> is not associated with amyotrophic lateral sclerosisâ€‘parkinsonism dementia complex in the Kii peninsula of Japan. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2010, 153B, 310-313.	1.7	31
105	Clinical and genetic diversities of Charcotâ€‘Marieâ€‘Tooth disease with <i>MFN2</i> mutations in a large case study. <i>Journal of the Peripheral Nervous System</i> , 2017, 22, 191-199.	3.1	31
106	Thalamic form of Creutzfeldt-Jakob disease or fatal insomnia? Report of a sporadic case with normal prion protein genotype. <i>Acta Neuropathologica</i> , 1997, 93, 317-322.	7.7	30
107	Phosphorylation in the C-terminal domain of Aquaporin-4 is required for Golgi transition in primary cultured astrocytes. <i>Biochemical and Biophysical Research Communications</i> , 2008, 377, 463-468.	2.1	30
108	Diffusion tensor tract-specific analysis of the uncinata fasciculus in patients with amyotrophic lateral sclerosis. <i>Neuroradiology</i> , 2010, 52, 729-733.	2.2	30



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109	Genotype-phenotype correlations in early onset ataxia with ocular motor apraxia and hypoalbuminaemia. <i>Brain</i> , 2011, 134, 1387-1399.	7.6	30
110	Increased gene dosage of myelin protein zero causes Charcot-Marie-Tooth disease. <i>Annals of Neurology</i> , 2012, 71, 84-92.	5.3	30
111	Advances in repeat expansion diseases and a new concept of repeat motif-phenotype correlation. <i>Current Opinion in Genetics and Development</i> , 2020, 65, 176-185.	3.3	30
112	Clinicopathologic Features of Oculopharyngodistal Myopathy With LRP12 CCG Repeat Expansions Compared With Other Oculopharyngodistal Myopathy Subtypes. <i>JAMA Neurology</i> , 2021, 78, 853.	9.0	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. <i>Neurogenetics</i> , 2011, 12, 41-50.	1.4	29
114	Late-onset spastic ataxia phenotype in a patient with a homozygous DDHD2 mutation. <i>Scientific Reports</i> , 2015, 4, 7132.	3.3	29
115	AgIn: measuring the landscape of CpG methylation of individual repetitive elements. <i>Bioinformatics</i> , 2016, 32, 2911-2919.	4.1	29
116	Clinicopathologic features of myositis patients with CD8-MHC-1 complex pathology. <i>Neurology</i> , 2017, 89, 1060-1068.	1.1	29
117	VPS13D-related disorders presenting as a pure and complicated form of hereditary spastic paraplegia. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e1108.	1.2	29
118	SNP HiTLink: a high-throughput linkage analysis system employing dense SNP data. <i>BMC Bioinformatics</i> , 2009, 10, 121.	2.6	28
119	Structural Basis and Genotype-Phenotype Correlations of INSR Mutations Causing Severe Insulin Resistance. <i>Diabetes</i> , 2017, 66, 2713-2723.	0.6	28
120	Burden of rare variants in causative genes for amyotrophic lateral sclerosis (ALS) accelerates age at onset of ALS. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2019, 90, 537-542.	1.9	28
121	Assignment of the human ST2 gene to chromosome 2 at q11.2. <i>Human Genetics</i> , 1996, 97, 561-563.	3.8	27
122	Decrease in benzodiazepine receptor binding in a patient with Angelman syndrome detected by iodine-123 iomazenil and single-photon emission tomography. <i>European Journal of Nuclear Medicine and Molecular Imaging</i> , 1996, 23, 598-604.	2.1	27
123	Autosomal-recessive complicated spastic paraplegia with a novel lysosomal trafficking regulator gene mutation. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014, 85, 1024-1028.	1.9	27
124	Tubular aggregate myopathy caused by a novel mutation in the cytoplasmic domain of STIM1. <i>Neurology: Genetics</i> , 2016, 2, e50.	1.9	27
125	Amnionless-mediated glycosylation is crucial for cell surface targeting of cubilin in renal and intestinal cells. <i>Scientific Reports</i> , 2018, 8, 2351.	3.3	27
126	Chronic cerebral hypoperfusion shifts the equilibrium of amyloid $\beta^2$ oligomers to aggregation-prone species with higher molecular weight. <i>Scientific Reports</i> , 2019, 9, 2827.	3.3	27



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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.	3.6	26
129	SCA42 mutation analysis in a case series of Japanese patients with spinocerebellar ataxia. Journal of Human Genetics, 2017, 62, 857-859.	2.3	25
130	Neuronal atrophy and synaptic alteration in a mouse model of dentatorubral-pallidoluysian atrophy. Brain, 2006, 129, 2353-2362.	7.6	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.	4.4	23
132	Low Prevalence of NOTCH2NLC GGC Repeat Expansion in White Patients with Movement Disorders. Movement Disorders, 2021, 36, 251-255.	3.9	23
133	Quantitation of heteroplasmy of mitochondrial trnaLeu(UUR) gene using PCR-SSCP. Muscle and Nerve, 1995, 18, 1390-1397.	2.2	22
134	Ataxia with isolated vitamin E deficiency and retinitis pigmentosa. Annals of Neurology, 1998, 43, 273-273.	5.3	22
135	Biallelic variants in <i>HPDL</i> cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
136	Molecular genetic evidence of clinical heterogeneity in Fukuyama-type congenital muscular dystrophy. Human Genetics, 1997, 99, 427-432.	3.8	21
137	Modeling neurological diseases with induced pluripotent cells reprogrammed from immortalized lymphoblastoid cell lines. Molecular Brain, 2016, 9, 88.	2.6	21
138	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
139	An Autopsy Case of Familial Neuronal Intranuclear Inclusion Disease with Dementia and Neuropathy. Internal Medicine, 2018, 57, 3459-3462.	0.7	21
140	Genomic aspects of sporadic neurodegenerative diseases. Biochemical and Biophysical Research Communications, 2014, 452, 221-225.	2.1	20
141	Increased facilitation of the primary motor cortex in de novo Parkinson's disease. Parkinsonism and Related Disorders, 2019, 66, 125-129.	2.2	20
142	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
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

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145	Mutational analysis of familial and sporadic amyotrophic lateral sclerosis with <i>OPTN</i> mutations in Japanese population. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2012, 13, 562-566.	2.1	19
146	The Neurogenomics View of Neurological Diseases. <i>JAMA Neurology</i> , 2013, 70, 689.	9.0	19
147	Clinical Characteristics of Neuronal Intranuclear Inclusion Disease-Related Retinopathy With CCG Repeat Expansions in the <i>NOTCH2NLC</i> Gene. , 2020, 61, 27.		19
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