Shoji Tsuji

List of Publications by Citations

306 papers	12,978 citations	55 h-index	107 g-index
326 ext. papers	14,769 ext. citations	6.8 avg, IF	5.59 L-index

#	Paper	IF	Citations
306	Interference by huntingtin and atrophin-1 with cbp-mediated transcription leading to cellular toxicity. *Science*, **2001**, 291, 2423-8	33.3	944
305	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, 323-30	24.1	830
304	Hereditary progressive dystonia with marked diurnal fluctuation caused by mutations in the GTP cyclohydrolase I gene. *Nature Genetics*, **1994**, 8, 236-42	36.3	707
303	Alpha-synuclein immunoreactivity in glial cytoplasmic inclusions in multiple system atrophy. *Neuroscience Letters*, **1998**, 249, 180-2	3.3	515
302	SCA17, a novel autosomal dominant cerebellar ataxia caused by an expanded polyglutamine in TATA-binding protein. *Human Molecular Genetics*, **2001**, 10, 1441-8	5.6	465
301	Expanded polyglutamine stretches interact with TAFII130, interfering with CREB-dependent transcription. *Nature Genetics*, **2000**, 26, 29-36	36.3	359
300	Suppression of aggregate formation and apoptosis by transglutaminase inhibitors in cells expressing truncated DRPLA protein with an expanded polyglutamine stretch. *Nature Genetics*, **1998**, 18, 111-7	36.3	350
299	Early-onset ataxia with ocular motor apraxia and hypoalbuminemia is caused by mutations in a new HIT superfamily gene. *Nature Genetics*, **2001**, 29, 184-8	36.3	342
298	Association of HTRA1 mutations and familial ischemic cerebral small-vessel disease. *New England Journal of Medicine*, **2009**, 360, 1729-39	59.2	324
297	Mis-sense mutation Val----Ile in exon 17 of amyloid precursor protein gene in Japanese familial Alzheimerƀ disease. *Lancet, The*, **1991**, 337, 978-9	40	280
296	Synphilin-1 is present in Lewy bodies in Parkinsonƀ disease. *Annals of Neurology*, **2000**, 47, 521-523	9.4	225
295	Human genetic variation database, a reference database of genetic variations in the Japanese population. *Journal of Human Genetics*, **2016**, 61, 547-53	4.3	212
294	Spinocerebellar ataxia type 31 is associated with "inserted" penta-nucleotide repeats containing (TGGAA)n. *American Journal of Human Genetics*, **2009**, 85, 544-57	11	204
293	Genomic Landscape of Esophageal Squamous Cell Carcinoma in´a Japanese Population. *Gastroenterology*, **2016**, 150, 1171-1182	13.3	195
292	An autopsy case of autosomal-recessive juvenile parkinsonism with a homozygous exon 4 deletion in the parkin gene. *Movement Disorders*, **2000**, 15, 884-8	7	195
291	Genetic association of the very low density lipoprotein (VLDL) receptor gene with sporadic Alzheimerƀ disease. *Nature Genetics*, **1995**, 11, 207-9	36.3	153
290	Expansions´of´intronic TTTCA and TTTTA repeats in benign adult familial myoclonic epilepsy. *Nature Genetics*, **2018**, 50, 581-590	36.3	152

289	Mutations for Gaucher disease confer high susceptibility to Parkinson disease. *Archives of Neurology*, **2009**, 66, 571-6		143
288	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, 875-84	24.1	135
287	ApoE-epsilon 4 and early-onset Alzheimer's. *Nature Genetics*, **1994**, 7, 10-1	36.3	133
286	Noncoding CGG repeat expansions in neuronal intranuclear inclusion disease, oculopharyngodistal myopathy and an overlapping disease. *Nature Genetics*, **2019**, 51, 1222-1232	36.3	132
285	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. *Nature Communications*, **2016**, 7, 11253	17.4	126
284	Dentatorubral-pallidoluysian atrophy: clinical features are closely related to unstable expansions of trinucleotide (CAG) repeat. *Annals of Neurology*, **1995**, 37, 769-75	9.4	125
283	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, 156-63	9.4	121
282	Sporadic ataxias in Japan--a population-based epidemiological study. *Cerebellum*, **2008**, 7, 189-97	4.3	109
281	Modulation of the age at onset in spinocerebellar ataxia by CAG tracts in various genes. *Brain*, **2014** , 137, 2444-55	11.2	108
280	Multiplex families with multiple system atrophy. *Archives of Neurology*, **2007**, 64, 545-51		101
279	ERBB4 mutations that disrupt the neuregulin-ErbB4 pathway cause amyotrophic lateral sclerosis type 19. *American Journal of Human Genetics*, **2013**, 93, 900-5	11	95
278	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	9.4	90
277	Novel mutations, pseudo-dominant inheritance, and possible familial affects in patients with autosomal recessive juvenile parkinsonism. *Annals of Neurology*, **2000**, 48, 245-250	9.4	90
276	Trinucleotide repeat length and rate of progression of Huntington's disease. *Annals of Neurology*, **1994**, 36, 630-5	9.4	89
275	Gene locus for autosomal recessive distal myopathy with rimmed vacuoles maps to chromosome 9. *Annals of Neurology*, **1997**, 41, 432-7	9.4	88
274	Age at onset influences on wide-ranged clinical features of sporadic amyotrophic lateral sclerosis. *Journal of the Neurological Sciences*, **2009**, 276, 163-9	3.2	81
273	Hereditary dentatorubral-pallidoluysian atrophy: detection of widespread ubiquitinated neuronal and glial intranuclear inclusions in the brain. *Acta Neuropathologica*, **1998**, 96, 547-52	14.3	80
272	Analysis of the expression level of alpha-synuclein mRNA using postmortem brain samples from pathologically confirmed cases of multiple system atrophy. *Acta Neuropathologica*, **2001**, 102, 188-90	14.3	79

271	C9ORF72 repeat expansion in amyotrophic lateral sclerosis in the Kii peninsula of Japan. *Archives of Neurology*, **2012**, 69, 1154-8		77
270	Variants associated with Gaucher disease in multiple system atrophy. *Annals of Clinical and Translational Neurology*, **2015**, 2, 417-26	5.3	76
269	The TRK-fused gene is mutated in hereditary motor and sensory neuropathy with proximal dominant involvement. *American Journal of Human Genetics*, **2012**, 91, 320-9	11	76
268	Safety and efficacy of eculizumab in Guillain-Barr□syndrome: a multicentre, double-blind, randomised phase 2 trial. *Lancet Neurology, The*, **2018**, 17, 519-529	24.1	72
267	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	11	71
266	Aprataxin, the causative protein for EAOH is a nuclear protein with a potential role as a DNA repair protein. *Annals of Neurology*, **2004**, 55, 241-9	9.4	69
265	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	9.4	69
264	A homozygous mutation of C12orf65 causes spastic paraplegia with optic atrophy and neuropathy (SPG55). *Journal of Medical Genetics*, **2012**, 49, 777-84	5.8	66
263	Pathology of CAG repeat diseases. *Neuropathology*, **2000**, 20, 319-325	2	65
262	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		65
261	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. *Annals of Neurology*, **1998**, 43, 288-96	9.4	64
260	Mutant COQ2 in multiple-system atrophy. *New England Journal of Medicine*, **2014**, 371, 82-3	59.2	62
259	Evaluation of SLC20A2 mutations that cause idiopathic basal ganglia calcification in Japan. *Neurology*, **2014**, 82, 705-12	6.5	61
258	Exome sequencing reveals a homozygous SYT14 mutation in adult-onset, autosomal-recessive spinocerebellar ataxia with psychomotor retardation. *American Journal of Human Genetics*, **2011**, 89, 320-7	11	60
257	A novel ferritin light chain gene mutation in a Japanese family with neuroferritinopathy: description of clinical features and implications for genotype-phenotype correlations. *Movement Disorders*, **2009**, 24, 441-5	7	59
256	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		58
255	Genetics of neurodegenerative diseases: insights from high-throughput resequencing. *Human Molecular Genetics*, **2010**, 19, R65-70	5.6	57
254	Pathology of CAG repeat diseases. *Neuropathology*, **2000**, 20, 319-25	2	57

253	Adult onset globoid cell leukodystrophy (Krabbe disease): analysis of galactosylceramidase cDNA from four Japanese patients. *Human Genetics*, **1997**, 100, 450-6	6.3	56
252	No mutation in the entire coding region of the alpha-synuclein gene in pathologically confirmed cases of multiple system atrophy. *Neuroscience Letters*, **1999**, 270, 110-2	3.3	56
251	A Novel Mutation in ELOVL4 Leading to Spinocerebellar Ataxia (SCA) With the Hot Cross Bun Sign but Lacking Erythrokeratodermia: A Broadened Spectrum of SCA34. *JAMA Neurology*, **2015**, 72, 797-805	17.2	53
250	A novel monoclonal antibody reveals a conformational alteration shared by amyotrophic lateral sclerosis-linked SOD1 mutants. *Annals of Neurology*, **2012**, 72, 739-49	9.4	53
249	Mutations in MME cause an autosomal-recessive Charcot-Marie-Tooth disease type 2. *Annals of Neurology*, **2016**, 79, 659-72	9.4	53
248	Neuron-specific methylome analysis reveals epigenetic regulation and tau-related dysfunction of BRCA1 in Alzheimer□s disease. *Proceedings of the National Academy of Sciences of the United States of America*, **2017**, 114, E9645-E9654	11.5	52
247	Autosomal dominant cerebellar ataxia (SCA6): clinical, genetic and neuropathological study in a family. *Acta Neuropathologica*, **1998**, 95, 333-7	14.3	52
246	BIN1 regulates BACE1 intracellular trafficking and amyloid-□production. *Human Molecular Genetics* , **2016**, 25, 2948-2958	5.6	52
245	Aprataxin, causative gene product for EAOH/AOA1, repairs DNA single-strand breaks with damaged 3Pphosphate and 3Pphosphoglycolate ends. *Nucleic Acids Research*, **2007**, 35, 3797-809	20.1	51
244	The FHA domain of aprataxin interacts with the C-terminal region of XRCC1. *Biochemical and Biophysical Research Communications*, **2004**, 325, 1279-85	3.4	51
243	Mutational analysis and genotype-phenotype correlation of 29 unrelated Japanese patients with X-linked adrenoleukodystrophy. *Archives of Neurology*, **1999**, 56, 295-300		51
242	Fatty acid elongation activity in fibroblasts from patients with adrenoleukodystrophy (ALD). *Journal of Biochemistry*, **1984**, 96, 1241-7	3.1	51
241	Rapid detection of expanded short tandem repeats in personal genomics using hybrid sequencing. *Bioinformatics*, **2014**, 30, 815-22	7.2	50
240	Dentatorubral-pallidoluysian atrophy. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2012**, 103, 587-94	3	50
239	Cancer association as a risk factor for anti-HMGCR antibody-positive myopathy. *Neurology: Neuroimmunology and NeuroInflammation*, **2016**, 3, e290	9.1	49
238	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. *Nature Communications*, **2019**, 10, 4920	17.4	48
237	Intronic pentanucleotide TTTCA repeat insertion in the SAMD12 gene causes familial cortical myoclonic tremor with epilepsy type 1. *Brain*, **2018**, 141, 2280-2288	11.2	45
236	A critique of the second consensus criteria for multiple system atrophy. *Movement Disorders*, **2019**, 34, 975-984	7	44

235	Next-generation sequencing of 28 ALS-related genes in a Japanese ALS cohort. *Neurobiology of Aging*, **2016**, 39, 219.e1-8	5.6	42
234	A functional variant in ZNF512B is associated with susceptibility to amyotrophic lateral sclerosis in Japanese. *Human Molecular Genetics*, **2011**, 20, 3684-92	5.6	42
233	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		41
232	Epidemiology of X-linked adrenoleukodystrophy in Japan. *Journal of Human Genetics*, **2002**, 47, 590-3	4.3	40
231	Toxicity of expanded polyglutamine-domain proteins in Escherichia coli. *FEBS Letters*, **1996**, 399, 135-9	3.8	40
230	Molecular epidemiology and clinical spectrum of hereditary spastic paraplegia in the Japanese population based on comprehensive mutational analyses. *Journal of Human Genetics*, **2014**, 59, 163-72	4.3	38
229	Hereditary dentatorubral-pallidoluysian atrophy: ubiquitinated filamentous inclusions in the cerebellar dentate nucleus neurons. *Acta Neuropathologica*, **1998**, 95, 479-82	14.3	38
228	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, 723-36	5.6	37
227	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		37
226	Interaction of expanded polyglutamine stretches with nuclear transcription factors leads to aberrant transcriptional regulation in polyglutamine diseases. *Neuropathology*, **2000**, 20, 326-333	2	36
225	Autosomal dominant diffuse Lewy body disease. *Acta Neuropathologica*, **1998**, 96, 207-10	14.3	35
224	Mitochondrial DNA mutation in family with Leberß hereditary optic neuropathy. *Lancet, The*, **1989**, 1, 1076-7	40	35
223	TDP-43 M337V mutation in familial amyotrophic lateral sclerosis in Japan. *Internal Medicine*, **2010**, 49, 331-4	1.1	34
222	Appropriate data cleaning methods for genome-wide association study. *Journal of Human Genetics*, **2008**, 53, 886-893	4.3	34
221	Lack of association of very low density lipoprotein receptor gene polymorphism with Caucasian Alzheimerß disease. *Annals of Neurology*, **1996**, 40, 251-4	9.4	33
220	Apolipoprotein E epsilon4 allele and progression of cortical Lewy body pathology in Parkinsonß disease. *Acta Neuropathologica*, **1998**, 95, 450-4	14.3	32
219	Posterior column ataxia with retinitis pigmentosa in a Japanese family with a novel mutation in FLVCR1. *Neurogenetics*, **2011**, 12, 117-21	3	31
218	X-linked nonprogressive congenital cerebellar hypoplasia: clinical description and mapping to chromosome Xq. *Annals of Neurology*, **1996**, 40, 75-83	9.4	30

217	The pathogenesis linked to coenzyme Q10 insufficiency in iPSC-derived neurons from patients with multiple-system atrophy. *Scientific Reports*, **2018**, 8, 14215	4.9	30
216	Modulation of error-sensitivity during a prism adaptation task in people with cerebellar degeneration. *Journal of Neurophysiology*, **2015**, 114, 2460-71	3.2	29
215	CSF1R mutations identified in three families with autosomal dominantly inherited leukoencephalopathy. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2012**, 159B, 951-7	3.5	29
214	Effect of intravenous immunoglobulin in Lambert-Eaton myasthenic syndrome with small-cell lung cancer: correlation with the titer of anti-voltage-gated calcium channel antibody. *Muscle and Nerve*, **1994**, 17, 1073-5	3.4	29
213	Increased gene dosage of myelin protein zero causes Charcot-Marie-Tooth disease. *Annals of Neurology*, **2012**, 71, 84-92	9.4	28
212	Adult-onset leukoencephalopathies with vanishing white matter with novel missense mutations in EIF2B2, EIF2B3, and EIF2B5. *Neurogenetics*, **2011**, 12, 259-61	3	28
211	Mitochondrial ND3 as the novel causative gene for Leber hereditary optic neuropathy and dystonia. *Neurogenetics*, **2009**, 10, 337-45	3	28
210	Mutations in COA7 cause spinocerebellar ataxia with axonal neuropathy. *Brain*, **2018**, 141, 1622-1636	11.2	27
209	Diffusion tensor tract-specific analysis of the uncinate fasciculus in patients with amyotrophic lateral sclerosis. *Neuroradiology*, **2010**, 52, 729-33	3.2	27
208	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	3	25
207	Genotype-phenotype correlations in early onset ataxia with ocular motor apraxia and hypoalbuminaemia. *Brain*, **2011**, 134, 1387-99	11.2	25
206	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	3.4	25
205	Assignment of the human ST2 gene to chromosome 2 at q11.2. *Human Genetics*, **1996**, 97, 561-3	6.3	25
204	A 3-year cohort study of the natural history of spinocerebellar ataxia type 6 in Japan. *Orphanet Journal of Rare Diseases*, **2014**, 9, 118	4.2	24
203	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	4.3	24
202	Thalamic form of Creutzfeldt-Jakob disease or fatal insomnia? Report of a sporadic case with normal prion protein genotype. *Acta Neuropathologica*, **1997**, 93, 317-22	14.3	24
201	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		24
200	Lack of association between dopamine D2 receptor gene Cys311 variant and schizophrenia. *American Journal of Medical Genetics Part A*, **1996**, 67, 208-11		24

199	Plasma Coenzyme Q10 Levels in Patients With Multiple System Atrophy. *JAMA Neurology*, **2016**, 73, 977-80	17.2	24
198	JASPAC: Japan Spastic Paraplegia Research Consortium. *Brain Sciences*, **2018**, 8,	3.4	23
197	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	5.6	22
196	Neuronal atrophy and synaptic alteration in a mouse model of dentatorubral-pallidoluysian atrophy. *Brain*, **2006**, 129, 2353-62	11.2	22
195	AgIn: measuring the landscape of CpG methylation of individual repetitive elements. *Bioinformatics*, **2016**, 32, 2911-9	7.2	22
194	Chronic cerebral hypoperfusion shifts the equilibrium of amyloid □oligomers to aggregation-prone species with higher molecular weight. *Scientific Reports*, **2019**, 9, 2827	4.9	21
193	SNP HiTLink: a high-throughput linkage analysis system employing dense SNP data. *BMC Bioinformatics*, **2009**, 10, 121	3.6	21
192	Transcortical sensory aphasia following left frontal infarction. *Journal of Neurology*, **1998**, 245, 69-76	5.5	21
191	A rapid functional decline type of amyotrophic lateral sclerosis is linked to low expression of TTN. *Journal of Neurology, Neurosurgery and Psychiatry*, **2016**, 87, 851-8	5.5	20
190	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, 336-50	7.5	20
189	SCA42 mutation analysis in a case series of Japanese patients with spinocerebellar ataxia. *Journal of Human Genetics*, **2017**, 62, 857-859	4.3	19
188	Clinical and genetic diversities of Charcot-Marie-Tooth disease with MFN2 mutations in a large case study. *Journal of the Peripheral Nervous System*, **2017**, 22, 191-199	4.7	19
187	Molecular genetic evidence of clinical heterogeneity in Fukuyama-type congenital muscular dystrophy. *Human Genetics*, **1997**, 99, 427-32	6.3	19
186	Quantitation of heteroplasmy of mitochondrial tRNA(Leu(UUR)) gene using PCR-SSCP. *Muscle and Nerve*, **1995**, 18, 1390-7	3.4	19
185	The Movement Disorder Society Criteria for the Diagnosis of Multiple System Atrophy.. *Movement Disorders*, **2022**,	7	19
184	TRPM7 is not associated with amyotrophic lateral sclerosis-parkinsonism dementia complex in the Kii peninsula of Japan. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2010**, 153B, 310-3	3.5	18
183	Ataxia with isolated vitamin E deficiency and retinitis pigmentosa. *Annals of Neurology*, **1998**, 43, 273	9.4	18
182	Lack of association between dopamine D4 receptor gene and schizophrenia. *American Journal of Medical Genetics Part A*, **1995**, 60, 580-2		18

181	Tubular aggregate myopathy caused by a novel mutation in the cytoplasmic domain of STIM1. *Neurology: Genetics*, **2016**, 2, e50	3.8	18
180	Autosomal-recessive complicated spastic paraplegia with a novel lysosomal trafficking regulator gene mutation. *Journal of Neurology, Neurosurgery and Psychiatry*, **2014**, 85, 1024-8	5.5	17
179	Unstable expansion of triplet repeats as a new disease mechanism for neurodegenerative diseases. *Japanese Journal of Human Genetics*, **1996**, 41, 279-90		17
178	Association study between schizophrenia and dopamine D3 receptor gene polymorphism. *American Journal of Medical Genetics Part A*, **1996**, 67, 366-8		17
177	A Prospective, Multicenter, Randomized Phase II Study to Evaluate the Efficacy and Safety of Eculizumab in Patients with Guillain-Barr Syndrome (GBS): Protocol of Japanese Eculizumab Trial for GBS (JET-GBS). *JMIR Research Protocols*, **2016**, 5, e210	2	17
176	Genomic aspects of sporadic neurodegenerative diseases. *Biochemical and Biophysical Research Communications*, **2014**, 452, 221-5	3.4	16
175	Late-onset spastic ataxia phenotype in a patient with a homozygous DDHD2 mutation. *Scientific Reports*, **2014**, 4, 7132	4.9	16
174	Mutational analysis of familial and sporadic amyotrophic lateral sclerosis with OPTN mutations in Japanese population. *Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders*, **2012**, 13, 562-6		16
173	Ataxic phenotype with altered Ca3.1 channel property in a mouse model for spinocerebellar ataxia 42. *Neurobiology of Disease*, **2019**, 130, 104516	7.5	15
172	Modeling neurological diseases with induced pluripotent cells reprogrammed from immortalized lymphoblastoid cell lines. *Molecular Brain*, **2016**, 9, 88	4.5	15
171	Unilateral opercular infarction presenting with Foix-Chavany-Marie Syndrome. *Journal of Stroke and Cerebrovascular Diseases*, **2014**, 23, 179-81	2.8	15
170	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	5.5	15
169	Epidemiology and molecular mechanism of frontotemporal lobar degeneration/amyotrophic lateral sclerosis with repeat expansion mutation in C9orf72. *Journal of Neurogenetics*, **2015**, 29, 85-94	1.6	14
168	Amnionless-mediated glycosylation is crucial for cell surface targeting of cubilin in renal and intestinal cells. *Scientific Reports*, **2018**, 8, 2351	4.9	14
167	Structural Basis and Genotype-Phenotype Correlations of INSR Mutations Causing Severe Insulin Resistance. *Diabetes*, **2017**, 66, 2713-2723	0.9	14
166	Clinicopathologic features of myositis patients with CD8-MHC-1 complex pathology. *Neurology*, **2017**, 89, 1060-1068	6.5	14
165	The neurogenomics view of neurological diseases. *JAMA Neurology*, **2013**, 70, 689-94	17.2	14
164	Differential effect of HDAC3 on cytoplasmic and nuclear huntingtin aggregates. *PLoS ONE*, **2014**, 9, e111277	3.7	14

163	VPS13D-related disorders presenting as a pure and complicated form of hereditary spastic paraplegia. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1108	2.3	14
162	The novel mutation of gene as the cause for Spastic paraplegia 30 in a Japanese case. *ENeurologicalSci*, **2019**, 14, 34-37	2.1	14
161	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	3.5	13
160	An Autopsy Case of Familial Neuronal Intranuclear Inclusion Disease with Dementia and Neuropathy. *Internal Medicine*, **2018**, 57, 3459-3462	1.1	13
159	Clinical features and haplotype analysis of newly identified Japanese patients with gelsolin-related familial amyloidosis of Finnish type. *Neurogenetics*, **2012**, 13, 237-43	3	13
158	Human leukocyte antigens in Fisher syndrome. *Annals of Neurology*, **1993**, 33, 655-7	9.4	13
157	Fukutin gene mutations that cause left ventricular noncompaction. *International Journal of Cardiology*, **2016**, 222, 727-729	3.2	13
156	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	4.3	12
155	Increased facilitation of the primary motor cortex in de novo Parkinson disease. *Parkinsonism and Related Disorders*, **2019**, 66, 125-129	3.6	12
154	Novel mutations in the PNPLA6 gene in Boucher-Neuhäuser syndrome. *Journal of Human Genetics*, **2015**, 60, 217-20	4.3	12
153	Mutational analysis of the anion exchanger 3 gene in familial paroxysmal dystonic choreoathetosis linked to chromosome 2q. *American Journal of Medical Genetics Part A*, **1999**, 88, 733-7		12
152	TBCD may be a causal gene in progressive neurodegenerative encephalopathy with atypical infantile spinal muscular atrophy. *Journal of Human Genetics*, **2017**, 62, 473-480	4.3	11
151	Novel De Novo KCND3 Mutation in a Japanese Patient with Intellectual Disability, Cerebellar Ataxia, Myoclonus, and Dystonia. *Cerebellum*, **2018**, 17, 237-242	4.3	11
150	Molecular genetics of triplet repeats: unstable expansion of triplet repeats as a new mechanism for neurodegenerative diseases. *Internal Medicine*, **1997**, 36, 3-8	1.1	11
149	Advances in repeat expansion diseases and a new concept of repeat motif-phenotype correlation. *Current Opinion in Genetics and Development*, **2020**, 65, 176-185	4.9	11
148	PLA2G6-associated neurodegeneration presenting as a complicated form of hereditary spastic paraplegia. *Journal of Human Genetics*, **2019**, 64, 55-59	4.3	11
147	Low Prevalence of NOTCH2NLC GGC Repeat Expansion in White Patients with Movement Disorders. *Movement Disorders*, **2021**, 36, 251-255	7	11
146	Tacrolimus-Induced Reversible Cerebral Vasoconstriction Syndrome with Delayed Multi-Segmental Vasoconstriction. *Journal of Stroke and Cerebrovascular Diseases*, **2017**, 26, e75-e77	2.8	10

145	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	3.3	10
144	Partial duplication of causes minifascicular neuropathy: A novel mutation detection of. *Annals of Clinical and Translational Neurology*, **2017**, 4, 415-421	5.3	10
143	The 3-second rule in hereditary pure cerebellar ataxia: a synchronized tapping study. *PLoS ONE*, **2015**, 10, e0118592	3.7	10
142	Two Japanese cases with aspartylglycosaminuria: clinical and morphological features. *Clinical Genetics*, **1991**, 40, 318-25	4	10
141	Shy-Drager syndrome with abnormal circadian rhythm of plasma antidiuretic hormone secretion and urinary excretion. *Internal Medicine*, **1993**, 32, 225-7	1.1	10
140	Stereotyped hand clasping: an unusual tardive movement disorder. *Movement Disorders*, **1993**, 8, 230-1	7	10
139	Atypical parkinsonism caused by Pro105Leu mutation of prion protein: A broad clinical spectrum. *Neurology: Genetics*, **2016**, 2, e48	3.8	10
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