Shoji Tsuji

List of Publications by Year in Descending Order

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	Idiopathic Late Onset Cerebellar Ataxia (ILOCA), and Cerebellar Plus Syndrome **2022**, 2433-2440		
305	Complex hereditary peripheral neuropathies caused by novel variants in mitochondrial-related nuclear genes.. *Journal of Neurology*, **2022**, 1	5.5	
304	The Movement Disorder Society Criteria for the Diagnosis of Multiple System Atrophy.. *Movement Disorders*, **2022**,	7	19
303	An immigrant family with Kii amyotrophic lateral sclerosis/parkinsonism-dementia complex. *Neurological Sciences*, **2021**, 43, 1423	3.5	0
302	Frequency of FMR1 Premutation Alleles in Patients with Undiagnosed Cerebellar Ataxia and Multiple System Atrophy in the Japanese Population. *Cerebellum*, **2021**, 1	4.3	1
301	Candesartan prevents arteriopathy progression in cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy model. *Journal of Clinical Investigation*, **2021**, 131,	15.9	3
300	CAG repeat-binding small molecule improves motor coordination impairment in a mouse model of Dentatorubral-pallidoluysian atrophy.. *Neurobiology of Disease*, **2021**, 163, 105604	7.5	1
299	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	5.8	4
298	Juvenile amyotrophic lateral sclerosis with complex phenotypes associated with novel mutations. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2021**, 22, 576-578	3.6	4
297	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. *Brain*, **2021**, 144, 1422-1434	11.2	7
296	Quantitative Evaluation of Cerebellar Function in Multiple System Atrophy with Transcranial Magnetic Stimulation. *Cerebellum*, **2021**, 1	4.3	2
295	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	7	1
294	Low Prevalence of NOTCH2NLC GGC Repeat Expansion in White Patients with Movement Disorders. *Movement Disorders*, **2021**, 36, 251-255	7	11
293	Genetic spectrum of Charcot-Marie-Tooth disease associated with myelin protein zero gene variants in Japan. *Clinical Genetics*, **2021**, 99, 359-375	4	6
292	Premature saccades: A detailed physiological analysis. *Clinical Neurophysiology*, **2021**, 132, 63-76	4.3	0
291	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	4.3	0
290	Splice-site mutations in KIF5A in the Japanese case series of amyotrophic lateral sclerosis. *Neurogenetics*, **2021**, 22, 11-17	3	1

289	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	4.3	3
288	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	4.9	5
287	SPG9A with the new occurrence of an ALDH18A1 mutation in a CMT1A family with PMP22 duplication: case report. *BMC Neurology*, **2021**, 21, 64	3.1	0
286	Multiple system atrophy variant with severe hippocampal pathology. *Brain Pathology*, **2021**, e13002	6	9
285	Clinicopathologic Features of Oculopharyngodistal Myopathy With LRP12 CGG Repeat Expansions Compared With Other Oculopharyngodistal Myopathy Subtypes. *JAMA Neurology*, **2021**, 78, 853-863	17.2	6
284	Severe dilated cardiomyopathy and ventricular arrhythmia in a patient with Emery-Dreifuss muscular dystrophy harboring a novel frameshift mutation in EMD. *Neurology and Clinical Neuroscience*, **2021**, 9, 490	0.3	
283	COQ2 V393A confers high risk susceptibility for multiple system atrophy in East Asian population. *Journal of the Neurological Sciences*, **2021**, 429, 117623	3.2	2
282	Reliability and validity of Japanese version of Unified Multiple System Atrophy Rating Scale. *Neurology and Clinical Neuroscience*, **2021**, 9, 171-180	0.3	0
281	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	3.3	
280	DMD exon 2 duplication due to a complex genomic rearrangement is associated with a somatic mosaicism.. *Neuromuscular Disorders*, **2021**,	2.9	0
279	An autopsy case of G gangliosidosis type II in a patient who survived a long duration with artificial respiratory support. *Neuropathology*, **2020**, 40, 379-388	2	0
278	A novel mutation in ABCD1 gene in a Filipino patient with adult-onset X-linked ALD. *Neurology and Clinical Neuroscience*, **2020**, 8, 329-331	0.3	
277	Clinical features of inherited neuropathy with BSCL2 mutations in Japan. *Journal of the Peripheral Nervous System*, **2020**, 25, 125-131	4.7	1
276	A novel mutation in the gene in a Japanese patient with SPG46: A case report. *ENeurologicalSci*, **2020**, 19, 100238	2.1	3
275	Neuron-specific analysis of histone modifications with post-mortem brains. *Scientific Reports*, **2020**, 10, 3767	4.9	3
274	Isolated seizure as initial presentation of GABA receptor antibody-associated encephalitis. *Journal of the Neurological Sciences*, **2020**, 410, 116666	3.2	3
273	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	3.6	7
272	DNA sequencing and other methods of exonic and genomic analyses **2020**, 109-120		

271	VPS13D-related disorders presenting as a pure and complicated form of hereditary spastic paraplegia. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1108	2.3	14
270	Rituximab improves not only back stiffness but also "stiff eyes" in stiff person syndrome: Implications for immune-mediated treatment. *Journal of the Neurological Sciences*, **2020**, 408, 116506	3.2	1
269	A Novel de novo KIF1A Mutation in a Patient with Autism, Hyperactivity, Epilepsy, Sensory Disturbance, and Spastic Paraplegia. *Internal Medicine*, **2020**, 59, 839-842	1.1	8
268	Familial dementia with Lewy bodies with VPS13C mutations. *Parkinsonism and Related Disorders*, **2020**, 81, 31-33	3.6	3
267	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	7.4	1
266	Identification of a novel mutation in associated with a complicated form of hereditary spastic paraplegia. *Neurology: Genetics*, **2020**, 6, e514	3.8	7
265	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	3.2	2
264	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
263	Clinical efficacy of haematopoietic stem cell transplantation for adult adrenoleukodystrophy. *Brain Communications*, **2020**, 2, fcz048	4.5	8
262	Do eye movements "age" earlier in progeria?. *Clinical Neurophysiology*, **2020**, 131, 1835-1836	4.3	
261	Comprehensive investigation of RNF213 nonsynonymous variants associated with intracranial artery stenosis. *Scientific Reports*, **2020**, 10, 11942	4.9	5
260	First Report of Multidrug-Resistant Carbapenemase-Producing Bacteria Coharboring Associated with Respiratory Disease Complex in Pets: Potential of Animal-Human Transmission. *Antimicrobial Agents and Chemotherapy*, **2020**, 65,	5.9	8
259	Clinical Characteristics of Neuronal Intranuclear Inclusion Disease-Related Retinopathy With CGG Repeat Expansions in the NOTCH2NLC Gene **2020**, 61, 27		7
258	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	0.8	3
257	CNV analysis using whole exome sequencing identified biallelic CNVs of VPS13B in siblings with intellectual disability. *European Journal of Medical Genetics*, **2020**, 63, 103610	2.6	9
256	Novel variant in a Japanese patient with idiopathic basal ganglia calcification-1 (IBGC1) associated with dopa-responsive parkinsonism. *Human Genome Variation*, **2019**, 6, 44	1.8	1
255	UBAP1 mutations cause juvenile-onset hereditary spastic paraplegias (SPG80) and impair UBAP1 targeting to endosomes. *Journal of Human Genetics*, **2019**, 64, 1055-1065	4.3	9
254	Oxygen consumption rate for evaluation of COQ2 variants associated with multiple system atrophy. *Neurogenetics*, **2019**, 20, 51-52	3	0

253	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
252	A critique of the second consensus criteria for multiple system atrophy. *Movement Disorders*, **2019**, 34, 975-984	7	44
251	Functional evaluation of PDGFB-variants in idiopathic basal ganglia calcification, using patient-derived iPS cells. *Scientific Reports*, **2019**, 9, 5698	4.9	5
250	Chronic cerebral hypoperfusion shifts the equilibrium of amyloid β oligomers to aggregation-prone species with higher molecular weight. *Scientific Reports*, **2019**, 9, 2827	4.9	21
249	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	3	1
248	Prominent Spasticity and Hyperreflexia of the Legs in a Nepalese Patient with Friedreich Ataxia. *Internal Medicine*, **2019**, 58, 2865-2869	1.1	
247	Noncoding CGG repeat expansions in neuronal intranuclear inclusion disease, oculopharyngodistal myopathy and an overlapping disease. *Nature Genetics*, **2019**, 51, 1222-1232	36.3	132
246	Increased facilitation of the primary motor cortex in de novo Parkinson's disease. *Parkinsonism and Related Disorders*, **2019**, 66, 125-129	3.6	12
245	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. *Nature Communications*, **2019**, 10, 4920	17.4	48
244	Neuroimaging, genetic, and enzymatic study in a Japanese family with a GBA gross deletion. *Parkinsonism and Related Disorders*, **2019**, 61, 57-63	3.6	4
243	The novel mutation of gene as the cause for Spastic paraplegia 30 in a Japanese case. *ENeurologicalSci*, **2019**, 14, 34-37	2.1	14
242	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	4.3	3
241	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
240	PLA2G6-associated neurodegeneration presenting as a complicated form of hereditary spastic paraplegia. *Journal of Human Genetics*, **2019**, 64, 55-59	4.3	11
239	Expansions of intronic TTTCA and TTTTA repeats in benign adult familial myoclonic epilepsy. *Nature Genetics*, **2018**, 50, 581-590	36.3	152
238	Safety and efficacy of eculizumab in Guillain-Barré syndrome: a multicentre, double-blind, randomised phase 2 trial. *Lancet Neurology, The*, **2018**, 17, 519-529	24.1	72
237	No novel, high penetrant gene might remain to be found in Japanese patients with unknown MODY. *Journal of Human Genetics*, **2018**, 63, 821-829	4.3	1
236	Amnionless-mediated glycosylation is crucial for cell surface targeting of cubilin in renal and intestinal cells. *Scientific Reports*, **2018**, 8, 2351	4.9	14

235	Clinical and genetic features of Charcot-Marie-Tooth disease 2F and hereditary motor neuropathy 2B in Japan. *Journal of the Peripheral Nervous System*, **2018**, 23, 40-48	4.7	8
234	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
233	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	5.6	8
232	Mutations in COA7 cause spinocerebellar ataxia with axonal neuropathy. *Brain*, **2018**, 141, 1622-1636	11.2	27
231	A Homozygous LAMA2 Mutation of c.818G>A Caused Partial Merosin Deficiency in a Japanese Patient. *Internal Medicine*, **2018**, 57, 877-882	1.1	6
230	Authors' reply to the Drs. Finsterer and Zarrouk-Mahjoub's comments for our case report. *International Journal of Cardiology*, **2018**, 254, 262	3.2	
229	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
228	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
227	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
226	Does the Clock Tick Slower or Faster in Parkinson's Disease? - Insights Gained From the Synchronized Tapping Task. *Frontiers in Psychology*, **2018**, 9, 1178	3.4	7
225	Optic neuropathy and decorticate-like posture as presenting symptoms of Bickerstaff's brainstem encephalitis: A case report and literature review. *Clinical Neurology and Neurosurgery*, **2018**, 173, 159-162	2	1
224	JASPAC: Japan Spastic Paraplegia Research Consortium. *Brain Sciences*, **2018**, 8,	3.4	23
223	An Autopsy Case of Familial Neuronal Intranuclear Inclusion Disease with Dementia and Neuropathy. *Internal Medicine*, **2018**, 57, 3459-3462	1.1	13
222	Degeneration of the Substantia Nigra Following Ipsilateral Striatal Infarction. *Internal Medicine*, **2018**, 57, 767-768	1.1	
221	Methylation changes and aberrant expression of FGFR3 in Lewy body disease neurons. *Brain Research*, **2018**, 1697, 59-66	3.7	4
220	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
219	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
218	Effect of subthalamic nucleus deep brain stimulation on visual scanning. *Clinical Neurophysiology*, **2018**, 129, 2421-2432	4.3	1

217	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	3.1	1
216	HIV Dementia with a Decreased Cardiac I-metaiodobenzylguanidine Uptake Masquerading as Dementia with Lewy Bodies. *Internal Medicine*, **2018**, 57, 3007-3010	1.1	3
215	Host MICA Polymorphism as a Potential Predictive Marker in Response to Chemotherapy for Colorectal Liver Metastases. *Digestive Diseases*, **2018**, 36, 437-445	3.2	2
214	SCA42 mutation analysis in a case series of Japanese patients with spinocerebellar ataxia. *Journal of Human Genetics*, **2017**, 62, 857-859	4.3	19
213	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
212	Tacrolimus-Induced Reversible Cerebral Vasoconstriction Syndrome with Delayed Multi-Segmental Vasoconstriction. *Journal of Stroke and Cerebrovascular Diseases*, **2017**, 26, e75-e77	2.8	10
211	AuthorsPresponse to "Compound heterozygous Fukutin mutation-related non-compaction" by Finsterer and Zarrouk-Mahjoub. *International Journal of Cardiology*, **2017**, 233, 102	3.2	
210	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	3.2	7
209	Slowly progressive d-bifunctional protein deficiency with survival to adulthood diagnosed by whole-exome sequencing. *Journal of the Neurological Sciences*, **2017**, 372, 6-10	3.2	8
208	Altered expression of Crb2 in podocytes expands a variation of CRB2 mutations in steroid-resistant nephrotic syndrome. *Pediatric Nephrology*, **2017**, 32, 801-809	3.2	7
207	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
206	Neuron-specific methylome analysis reveals epigenetic regulation and tau-related dysfunction of BRCA1 in Alzheimerβ disease. *Proceedings of the National Academy of Sciences of the United States of America*, **2017**, 114, E9645-E9654	11.5	52
205	Development of a novel system to quantify the spatialDemporal parameters for crutch-assisted quadrupedal gait. *Advanced Robotics*, **2017**, 31, 80-87	1.7	2
204	Structural Basis and Genotype-Phenotype Correlations of INSR Mutations Causing Severe Insulin Resistance. *Diabetes*, **2017**, 66, 2713-2723	0.9	14
203	Clinicopathologic features of myositis patients with CD8-MHC-1 complex pathology. *Neurology*, **2017**, 89, 1060-1068	6.5	14
202	Partial duplication of causes minifascicular neuropathy: A novel mutation detection of. *Annals of Clinical and Translational Neurology*, **2017**, 4, 415-421	5.3	10
201	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
200	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

199	Successful management of chronic myeloid leukemia with a complication of anti-SRP antibody-associated myopathy. *Leukemia and Lymphoma*, **2017**, 58, 1242-1245	1.9	0
198	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
197	Plasma Coenzyme Q10 Levels and Multiple System Atrophy-Reply. *JAMA Neurology*, **2016**, 73, 1499-1500	17.2	3
196	Modeling neurological diseases with induced pluripotent cells reprogrammed from immortalized lymphoblastoid cell lines. *Molecular Brain*, **2016**, 9, 88	4.5	15
195	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. *Nature Communications*, **2016**, 7, 11253	17.4	126
194	Assessing Cell-to-Cell DNA Methylation Variability on Individual Long Reads. *Scientific Reports*, **2016**, 6, 21317	4.9	9
193	Next-generation sequencing of 28 ALS-related genes in a Japanese ALS cohort. *Neurobiology of Aging*, **2016**, 39, 219.e1-8	5.6	42
192	Genomic Landscape of Esophageal Squamous Cell Carcinoma in´a Japanese Population. *Gastroenterology*, **2016**, 150, 1171-1182	13.3	195
191	Persistent pain as a non-motor symptom in corticobasal syndrome. *Journal of Clinical Neuroscience*, **2016**, 29, 35-7	2.2	1
190	Recovered recall memory after decompression of the fornix by surgical removal of pineal tumor. *Neurology*, **2016**, 86, 790-1	6.5	4
189	Adult onset ictal aphasia with epileptic discharges in Broca□s and Wernicke□s areas. *Clinical Neurophysiology*, **2016**, 127, 1754-1756	4.3	1
188	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
187	Elevated Serum Uric Acid Levels Are Related to Cognitive Deterioration in an Elderly Japanese Population. *Dementia and Geriatric Cognitive Disorders Extra*, **2016**, 6, 580-588	2.5	2
186	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
185	Plasma Coenzyme Q10 Levels in Patients With Multiple System Atrophy. *JAMA Neurology*, **2016**, 73, 977-80	17.2	24
184	Cancer association as a risk factor for anti-HMGCR antibody-positive myopathy. *Neurology: Neuroimmunology and NeuroInflammation*, **2016**, 3, e290	9.1	49
183	Exome sequencing reveals a novel missense mutation in the KIAA0196 gene in a Japanese patient with SPG8. *Clinical Neurology and Neurosurgery*, **2016**, 144, 36-8	2	5
182	Tubular aggregate myopathy caused by a novel mutation in the cytoplasmic domain of STIM1. *Neurology: Genetics*, **2016**, 2, e50	3.8	18

181	Atypical parkinsonism caused by Pro105Leu mutation of prion protein: A broad clinical spectrum. *Neurology: Genetics*, **2016**, 2, e48	3.8	10
180	BIN1 regulates BACE1 intracellular trafficking and amyloid-β production. *Human Molecular Genetics*, **2016**, 25, 2948-2958	5.6	52
179	AgIn: measuring the landscape of CpG methylation of individual repetitive elements. *Bioinformatics*, **2016**, 32, 2911-9	7.2	22
178	Fukutin gene mutations that cause left ventricular noncompaction. *International Journal of Cardiology*, **2016**, 222, 727-729	3.2	13
177	Mutations in MME cause an autosomal-recessive Charcot-Marie-Tooth disease type 2. *Annals of Neurology*, **2016**, 79, 659-72	9.4	53
176	Novel COL6A2 mutation in a case of limb girdle muscular dystrophy phenotype with autosomal recessive inheritance. *Neurology and Clinical Neuroscience*, **2016**, 4, 189-191	0.3	
175	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
174	Variants associated with Gaucher disease in multiple system atrophy. *Annals of Clinical and Translational Neurology*, **2015**, 2, 417-26	5.3	76
173	The first Japanese familial case of spinocerebellar ataxia 23 with a novel mutation in the PDYN gene. *Parkinsonism and Related Disorders*, **2015**, 21, 332-4	3.6	7
172	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
171	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	7.5	7
170	Accumulation of transportin 1 in the fused in sarcoma-positive neuronal inclusions in sporadic amyotrophic lateral sclerosis without FUS mutation. *Neurology and Clinical Neuroscience*, **2015**, 3, 194-196	0.3	
169	The 3-second rule in hereditary pure cerebellar ataxia: a synchronized tapping study. *PLoS ONE*, **2015**, 10, e0118592	3.7	10
168	DNA Sequencing and Other Methods of Exonic and Genomic Analyses **2015**, 77-85		
167	Modulation of error-sensitivity during a prism adaptation task in people with cerebellar degeneration. *Journal of Neurophysiology*, **2015**, 114, 2460-71	3.2	29
166	Novel mutations in the PNPLA6 gene in Boucher-Neuhäuser syndrome. *Journal of Human Genetics*, **2015**, 60, 217-20	4.3	12
165	Adult-onset vanishing white matter disease with novel missense mutations in a subunit of translational regulator, EIF2B4. *Clinical Genetics*, **2015**, 88, 401-3	4	4
164	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	1.4	6

163	Triad-conditioning transcranial magnetic stimulation in Parkinson's disease. *Brain Stimulation*, **2014**, 7, 74-9	5.1	6
162	Exome sequencing shows a novel de novo mutation in ATL1. *Neurology and Clinical Neuroscience*, **2014**, 2, 1-4	0.3	2
161	Modulation of the age at onset in spinocerebellar ataxia by CAG tracts in various genes. *Brain*, **2014** , 137, 2444-55	11.2	108
160	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
159	Genomic aspects of sporadic neurodegenerative diseases. *Biochemical and Biophysical Research Communications*, **2014**, 452, 221-5	3.4	16
158	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
157	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
156	Abadie's sign in adrenomyeloneuropathy. *Journal of the Neurological Sciences*, **2014**, 340, 245-6	3.2	
155	P1-078: BIN1 REGULATES BACE1 TRAFFICKING AND AB PRODUCTION **2014**, 10, P331-P332		1
154	Late-onset spastic ataxia phenotype in a patient with a homozygous DDHD2 mutation. *Scientific Reports*, **2014**, 4, 7132	4.9	16
153	Mutant COQ2 in multiple-system atrophy. *New England Journal of Medicine*, **2014**, 371, 82-3	59.2	62
152	Treatable chorea associated with polycythemia vera. *Neurology and Clinical Neuroscience*, **2014**, 2, 90-91	0.3	
151	Sensorimotor neuropathy in late-onset Krabbe disease progressing over 40 years after onset. *Neurology and Clinical Neuroscience*, **2014**, 2, 114-116	0.3	
150	Rapid detection of expanded short tandem repeats in personal genomics using hybrid sequencing. *Bioinformatics*, **2014**, 30, 815-22	7.2	50
149	Evaluation of SLC20A2 mutations that cause idiopathic basal ganglia calcification in Japan. *Neurology*, **2014**, 82, 705-12	6.5	61
148	Unilateral opercular infarction presenting with Foix-Chavany-Marie Syndrome. *Journal of Stroke and Cerebrovascular Diseases*, **2014**, 23, 179-81	2.8	15
147	Differential effect of HDAC3 on cytoplasmic and nuclear huntingtin aggregates. *PLoS ONE*, **2014**, 9, e111277	3.7	14
146	Top-down but not bottom-up visual scanning is affected in hereditary pure cerebellar ataxia. *PLoS ONE*, **2014**, 9, e116181	3.7	4

145	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
144	An open trial of long-term testosterone suppression in spinal and bulbar muscular atrophy. *Muscle and Nerve*, **2013**, 47, 816-22	3.4	6
143	Effects of acidic phospholipids on antiganglioside antibodies in Guillain□Barr□syndrome: Role of the disialosyl residue. *Clinical and Experimental Neuroimmunology*, **2013**, 4, 70-74	0.4	3
142	O4□6□5: SORL1 is genetically associated with late-onset Alzheimer□s disease in Japanese, Koreans and Caucasians **2013**, 9, P693-P694		1
141	Recurrent K3E mutation in Cu/Zn superoxide dismutase gene associated with amyotrophic lateral sclerosis. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2013**, 14, 608-14	3.6	6
140	The neurogenomics view of neurological diseases. *JAMA Neurology*, **2013**, 70, 689-94	17.2	14
139	Familial amyotrophic lateral sclerosis with novel A4D SOD1 mutation with late age at onset and rapid progressive course. *Neurology and Clinical Neuroscience*, **2013**, 1, 45-47	0.3	8
138	Monitoring daily life activity shows less activity among female dementia patients. *Neurology and Clinical Neuroscience*, **2013**, 1, 91-95	0.3	2
137	Idiopathic Late Onset Cerebellar Ataxia (ILOCA), and Cerebellar plus Syndrome **2013**, 2143-2150		
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