Jun Mitsui

List of Publications by Citations

112 papers	2,245 citations	24 h-index	45 g-index
129 ext. papers	2,856 ext. citations	5 avg, IF	4.24 L-index

#	Paper	IF	Citations
112	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
111	Expansions of intronic TTTCA and TTTTA repeats in benign adult familial myoclonic epilepsy. *Nature Genetics*, **2018**, 50, 581-590	36.3	152
110	Mutations for Gaucher disease confer high susceptibility to Parkinson disease. *Archives of Neurology*, **2009**, 66, 571-6		143
109	Noncoding CGG repeat expansions in neuronal intranuclear inclusion disease, oculopharyngodistal myopathy and an overlapping disease. *Nature Genetics*, **2019**, 51, 1222-1232	36.3	132
108	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. *Nature Communications*, **2016**, 7, 11253	17.4	126
107	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
106	C9ORF72 repeat expansion in amyotrophic lateral sclerosis in the Kii peninsula of Japan. *Archives of Neurology*, **2012**, 69, 1154-8		77
105	Variants associated with Gaucher disease in multiple system atrophy. *Annals of Clinical and Translational Neurology*, **2015**, 2, 417-26	5.3	76
104	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
103	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
102	Mutant COQ2 in multiple-system atrophy. *New England Journal of Medicine*, **2014**, 371, 82-3	59.2	62
101	Identification of ATP1A3 mutations by exome sequencing as the cause of alternating hemiplegia of childhood in Japanese patients. *PLoS ONE*, **2013**, 8, e56120	3.7	61
100	Pathology of the sympathetic nervous system corresponding to the decreased cardiac uptake in 123I-metaiodobenzylguanidine (MIBG) scintigraphy in a patient with Parkinson disease. *Journal of the Neurological Sciences*, **2006**, 243, 101-4	3.2	57
99	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
98	Mutations in MME cause an autosomal-recessive Charcot-Marie-Tooth disease type 2. *Annals of Neurology*, **2016**, 79, 659-72	9.4	53
97	Rapid detection of expanded short tandem repeats in personal genomics using hybrid sequencing. *Bioinformatics*, **2014**, 30, 815-22	7.2	50
96	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

95	Posterior column ataxia with retinitis pigmentosa in a Japanese family with a novel mutation in FLVCR1. *Neurogenetics*, **2011**, 12, 117-21	3	31
94	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
93	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
92	Increased gene dosage of myelin protein zero causes Charcot-Marie-Tooth disease. *Annals of Neurology*, **2012**, 71, 84-92	9.4	28
91	Adult-onset leukoencephalopathies with vanishing white matter with novel missense mutations in EIF2B2, EIF2B3, and EIF2B5. *Neurogenetics*, **2011**, 12, 259-61	3	28
90	Mutations in COA7 cause spinocerebellar ataxia with axonal neuropathy. *Brain*, **2018**, 141, 1622-1636	11.2	27
89	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
88	Plasma Coenzyme Q10 Levels in Patients With Multiple System Atrophy. *JAMA Neurology*, **2016**, 73, 977-80	8.2	24
87	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
86	AgIn: measuring the landscape of CpG methylation of individual repetitive elements. *Bioinformatics*, **2016**, 32, 2911-9	7.2	22
85	A novel mutation causes X-linked intellectual disability type Nascimento. *Human Genome Variation*, **2017**, 4, 17019	1.8	20
84	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
83	Tubular aggregate myopathy caused by a novel mutation in the cytoplasmic domain of STIM1. *Neurology: Genetics*, **2016**, 2, e50	3.8	18
82	Genomic aspects of sporadic neurodegenerative diseases. *Biochemical and Biophysical Research Communications*, **2014**, 452, 221-5	3.4	16
81	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
80	Modeling neurological diseases with induced pluripotent cells reprogrammed from immortalized lymphoblastoid cell lines. *Molecular Brain*, **2016**, 9, 88	4.5	15
79	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
78	Amnionless-mediated glycosylation is crucial for cell surface targeting of cubilin in renal and intestinal cells. *Scientific Reports*, **2018**, 8, 2351	4.9	14

77	Respiratory and cardiac function in japanese patients with dysferlinopathy. *Muscle and Nerve*, **2016**, 53, 394-401	3.4	14
76	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
75	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
74	Refining the clinical phenotype of Okur-Chung neurodevelopmental syndrome. *Human Genome Variation*, **2018**, 5, 18011	1.8	12
73	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
72	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
71	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
70	Multiplexed resequencing analysis to identify rare variants in pooled DNA with barcode indexing using next-generation sequencer. *Journal of Human Genetics*, **2010**, 55, 448-55	4.3	11
69	PLA2G6-associated neurodegeneration presenting as a complicated form of hereditary spastic paraplegia. *Journal of Human Genetics*, **2019**, 64, 55-59	4.3	11
68	Partial duplication of causes minifascicular neuropathy: A novel mutation detection of. *Annals of Clinical and Translational Neurology*, **2017**, 4, 415-421	5.3	10
67	Atypical parkinsonism caused by Pro105Leu mutation of prion protein: A broad clinical spectrum. *Neurology: Genetics*, **2016**, 2, e48	3.8	10
66	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
65	Generation of D1-1 TALEN isogenic control cell line from Dravet syndrome patient iPSCs using TALEN-mediated editing of the SCN1A gene. *Stem Cell Research*, **2018**, 28, 100-104	1.6	9
64	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
63	Multiple system atrophy variant with severe hippocampal pathology. *Brain Pathology*, **2021**, e13002	6	9
62	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
61	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
60	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

59	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
58	Clinical efficacy of haematopoietic stem cell transplantation for adult adrenoleukodystrophy. *Brain Communications*, **2020**, 2, fcz048	4.5	8
57	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
56	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
55	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
54	Novel mutation in a Japanese family with adult polyglucosan body disease. *Neurology: Genetics*, **2017**, 3, e138	3.8	6
53	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
52	Genetic spectrum of Charcot-Marie-Tooth disease associated with myelin protein zero gene variants in Japan. *Clinical Genetics*, **2021**, 99, 359-375	4	6
51	Functional evaluation of PDGFB-variants in idiopathic basal ganglia calcification, using patient-derived iPS cells. *Scientific Reports*, **2019**, 9, 5698	4.9	5
50	Whole-exome sequencing in a Japanese pedigree implicates a rare non-synonymous single-nucleotide variant in BEST3 as a candidate for mandibular prognathism. *Bone*, **2019**, 122, 193-198	4.7	5
49	Comprehensive investigation of RNF213 nonsynonymous variants associated with intracranial artery stenosis. *Scientific Reports*, **2020**, 10, 11942	4.9	5
48	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
47	Juvenile amyotrophic lateral sclerosis with complex phenotypes associated with novel mutations. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2021**, 22, 576-578	3.6	4
46	Associations of pathological diagnosis and genetic abnormalities in meningiomas with the embryological origins of the meninges. *Scientific Reports*, **2021**, 11, 6987	4.9	4
45	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
44	Plasma Coenzyme Q10 Levels and Multiple System Atrophy-Reply. *JAMA Neurology*, **2016**, 73, 1499-1500	17.2	3
43	Novel mutation in the SOD1 gene in a patient with early-onset, rapidly progressive amyotrophic lateral sclerosis. *Neurology and Clinical Neuroscience*, **2017**, 5, 189-191	0.3	3
42	SNP haplotype mapping in a small ALS family. *PLoS ONE*, **2009**, 4, e5687	3.7	3

41	Familial dementia with Lewy bodies with VPS13C mutations. *Parkinsonism and Related Disorders*, **2020**, 81, 31-33	3.6	3
40	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
39	A Japanese family with mutation in the proteinase inhibitor 12 L47P gene: A case report. *Journal of the Neurological Sciences*, **2018**, 384, 126-128	3.2	3
38	Exome sequencing shows a novel de novo mutation in ATL1. *Neurology and Clinical Neuroscience*, **2014**, 2, 1-4	0.3	2
37	Exome analysis reveals a Japanese family with spinocerebellar ataxia, autosomal recessive 1. *Journal of the Neurological Sciences*, **2013**, 331, 158-60	3.2	2
36	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
35	Clinical Impact of Copy Number Variation on the Genetic Diagnosis of Syndromic Aortopathies. *Circulation Genomic and Precision Medicine*, **2021**, 14, e003458	5.2	2
34	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
33	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
32	Clinical and molecular genetic characterization of two female patients harboring the Xq27.3q28 deletion with different ratios of X chromosome inactivation. *Human Mutation*, **2020**, 41, 1447-1460	4.7	1
31	Germline MICA Polymorphism Is Associated with the Long-Term Outcomes in Patients Undergoing Hepatectomy for Colorectal Liver Metastases. *Journal of Gastrointestinal Surgery*, **2020**, 24, 2137-2139	3.3	1
30	Clinical features of inherited neuropathy with BSCL2 mutations in Japan. *Journal of the Peripheral Nervous System*, **2020**, 25, 125-131	4.7	1
29	Atypical Familial Amyotrophic Lateral Sclerosis with Slowly Progressing Lower Extremities-predominant Late-onset Muscular Weakness and Atrophy. *Internal Medicine*, **2019**, 58, 1851-1858	1.1	1
28	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
27	Splice-site mutations in KIF5A in the Japanese case series of amyotrophic lateral sclerosis. *Neurogenetics*, **2021**, 22, 11-17	3	1
26	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
25	Oxygen consumption rate for evaluation of COQ2 variants associated with multiple system atrophy. *Neurogenetics*, **2019**, 20, 51-52	3	0
24	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

23	A case of late-onset Krabbe disease which showed subacute progression of spastic paresis with bilateral spinal cord lesions. *Neurology and Clinical Neuroscience*, **2018**, 6, 104-106	0.3	0
22	Brainstem intraparenchymal schwannoma with genetic analysis: a case report and literature review. *BMC Medical Genomics*, **2021**, 14, 205	3.7	0
21	Reliability and validity of Japanese version of Unified Multiple System Atrophy Rating Scale. *Neurology and Clinical Neuroscience*, **2021**, 9, 171-180	0.3	0
20	DMD exon 2 duplication due to a complex genomic rearrangement is associated with a somatic mosaicism.. *Neuromuscular Disorders*, **2021**,	2.9	0
19	Clinical significance of NF2 alteration in grade I meningiomas revisited; prognostic impact integrated with extent of resection, tumour location, and Ki-67 index.. *Acta Neuropathologica Communications*, **2022**, 10, 76	7.3	0
18	Letter re: A genome-wide association study in multiple system atrophy. *Neurology*, **2017**, 88, 1296	6.5	
17	Novel variant of CSF1R in sporadic case with early-onset cognitive impairment. *Neurology and Clinical Neuroscience*, **2020**, 8, 430-432	0.3	
16	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	
15	Prominent Spasticity and Hyperreflexia of the Legs in a Nepalese Patient with Friedreich Ataxia. *Internal Medicine*, **2019**, 58, 2865-2869	1.1	
14	Late presented congenital myasthenic syndrome with novel compound heterozygous CHRNE mutations mimicking seronegative myasthenia gravis. *Neurology and Clinical Neuroscience*, **2019**, 7, 288-290	0.3	
13	Sporadic progressive myoclonic epilepsy with early-onset dementia caused by a de novo mutation in PSEN1. *Neurology and Clinical Neuroscience*, **2019**, 7, 294-296	0.3	
12	Novel mutation in the membrane metalloendopeptidase gene in a patient with the autosomal recessive form of Charcot-Marie-Tooth disease. *Neurology and Clinical Neuroscience*, **2017**, 5, 124-126	0.3	
11	DNA Sequencing and Other Methods of Exonic and Genomic Analyses **2015**, 77-85		
10	Sensorimotor neuropathy in late-onset Krabbe disease progressing over 40 years after onset. *Neurology and Clinical Neuroscience*, **2014**, 2, 114-116	0.3	
9	Family with congenital contractural arachnodactyly due to a novel multiexon deletion of the gene.. *Clinical Case Reports (discontinued)*, **2022**, 10, e05335	0.7	
8	DNA sequencing and other methods of exonic and genomic analyses **2020**, 109-120		
7	A novel multi-exon deletion in the dysferlin gene of a limb-girdle muscular dystrophy type 2B Filipino patient. *Neurology and Clinical Neuroscience*, **2020**, 8, 419-421	0.3	
6	A Case of Irreversible Corneal Edema Associated with Dentatorubropallidoluysian Atrophy Following Corneal Endothelial Transplantation. *SN Comprehensive Clinical Medicine*, **2021**, 3, 2029-2032	2.7	

5	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
4	Esophageal Dysmotility is Common in Patients With Multiple System Atrophy. *Laryngoscope*, **2021**, 131, 832-838	3.6
3	Complex hereditary peripheral neuropathies caused by novel variants in mitochondrial-related nuclear genes.. *Journal of Neurology*, **2022**, 1	5.5
2	A case of idiopathic normal pressure hydrocephalus with fragile X-associated tremor/ataxia syndrome.. *Clinical Neurology and Neurosurgery*, **2022**, 218, 107278	2
1	Symptomatic and Stenotic Developmental Venous Anomaly with Pontine Capillary Telangiectasia: A Case Report with Genetic Considerations. *NMC Case Report Journal*, **2022**, 9, 139-144	0.6