

Jun Mitsui

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	Family with congenital contractural arachnodactyly due to a novel multiexon deletion of the gene.. <i>Clinical Case Reports (discontinued)</i> , 2022 , 10, e05335	0.7	
111	Complex hereditary peripheral neuropathies caused by novel variants in mitochondrial-related nuclear genes.. <i>Journal of Neurology</i> , 2022 , 1	5.5	
110	A case of idiopathic normal pressure hydrocephalus with fragile X-associated tremor/ataxia syndrome.. <i>Clinical Neurology and Neurosurgery</i> , 2022 , 218, 107278	2	
109	Clinical significance of NF2 alteration in grade I meningiomas revisited; prognostic impact integrated with extent of resection, tumour location, and Ki-67 index.. <i>Acta Neuropathologica Communications</i> , 2022 , 10, 76	7.3	0
108	Symptomatic and Stenotic Developmental Venous Anomaly with Pontine Capillary Telangiectasia: A Case Report with Genetic Considerations. <i>NMC Case Report Journal</i> , 2022 , 9, 139-144	0.6	
107	Frequency of FMR1 Premutation Alleles in Patients with Undiagnosed Cerebellar Ataxia and Multiple System Atrophy in the Japanese Population. <i>Cerebellum</i> , 2021 , 1	4.3	1
106	Targeted deep sequencing of DNA from multiple tissue types improves the diagnostic rate and reveals a highly diverse phenotype of mosaic neurofibromatosis type 2. <i>Journal of Medical Genetics</i> , 2021 , 58, 701-711	5.8	4
105	Juvenile amyotrophic lateral sclerosis with complex phenotypes associated with novel mutations. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2021 , 22, 576-578	3.6	4
104	Associations of pathological diagnosis and genetic abnormalities in meningiomas with the embryological origins of the meninges. <i>Scientific Reports</i> , 2021 , 11, 6987	4.9	4
103	A Case of Irreversible Corneal Edema Associated with Dentatorubropallidolusian Atrophy Following Corneal Endothelial Transplantation. <i>SN Comprehensive Clinical Medicine</i> , 2021 , 3, 2029-2032	2.7	
102	Esophageal Dysmotility is Common in Patients With Multiple System Atrophy. <i>Laryngoscope</i> , 2021 , 131, 832-838	3.6	
101	Genetic spectrum of Charcot-Marie-Tooth disease associated with myelin protein zero gene variants in Japan. <i>Clinical Genetics</i> , 2021 , 99, 359-375	4	6
100	Splice-site mutations in KIF5A in the Japanese case series of amyotrophic lateral sclerosis. <i>Neurogenetics</i> , 2021 , 22, 11-17	3	1
99	Loss-of-function variants in NEK1 are associated with an increased risk of sporadic ALS in the Japanese population. <i>Journal of Human Genetics</i> , 2021 , 66, 237-241	4.3	3
98	Multiple system atrophy variant with severe hippocampal pathology. <i>Brain Pathology</i> , 2021 , e13002	6	9
97	Brainstem intraparenchymal schwannoma with genetic analysis: a case report and literature review. <i>BMC Medical Genomics</i> , 2021 , 14, 205	3.7	0
96	Clinical Impact of Copy Number Variation on the Genetic Diagnosis of Syndromic Aortopathies. <i>Circulation Genomic and Precision Medicine</i> , 2021 , 14, e003458	5.2	2

95	COQ2 V393A confers high risk susceptibility for multiple system atrophy in East Asian population. <i>Journal of the Neurological Sciences</i> , 2021 , 429, 117623	3.2	2
94	Reliability and validity of Japanese version of Unified Multiple System Atrophy Rating Scale. <i>Neurology and Clinical Neuroscience</i> , 2021 , 9, 171-180	0.3	0
93	DMD exon 2 duplication due to a complex genomic rearrangement is associated with a somatic mosaicism.. <i>Neuromuscular Disorders</i> , 2021 ,	2.9	0
92	Novel variant of CSF1R in sporadic case with early-onset cognitive impairment. <i>Neurology and Clinical Neuroscience</i> , 2020 , 8, 430-432	0.3	
91	An autopsy case of G gangliosidosis type II in a patient who survived a long duration with artificial respiratory support. <i>Neuropathology</i> , 2020 , 40, 379-388	2	0
90	Clinical and molecular genetic characterization of two female patients harboring the Xq27.3q28 deletion with different ratios of X chromosome inactivation. <i>Human Mutation</i> , 2020 , 41, 1447-1460	4.7	1
89	Germline MICA Polymorphism Is Associated with the Long-Term Outcomes in Patients Undergoing Hepatectomy for Colorectal Liver Metastases. <i>Journal of Gastrointestinal Surgery</i> , 2020 , 24, 2137-2139	3.3	1
88	A novel mutation in ABCD1 gene in a Filipino patient with adult-onset X-linked ALD. <i>Neurology and Clinical Neuroscience</i> , 2020 , 8, 329-331	0.3	
87	Clinical features of inherited neuropathy with BSCL2 mutations in Japan. <i>Journal of the Peripheral Nervous System</i> , 2020 , 25, 125-131	4.7	1
86	DNA sequencing and other methods of exonic and genomic analyses 2020 , 109-120		
85	A Novel de novo KIF1A Mutation in a Patient with Autism, Hyperactivity, Epilepsy, Sensory Disturbance, and Spastic Paraplegia. <i>Internal Medicine</i> , 2020 , 59, 839-842	1.1	8
84	Familial dementia with Lewy bodies with VPS13C mutations. <i>Parkinsonism and Related Disorders</i> , 2020 , 81, 31-33	3.6	3
83	A novel multi-exon deletion in the dysferlin gene of a limb-girdle muscular dystrophy type 2B Filipino patient. <i>Neurology and Clinical Neuroscience</i> , 2020 , 8, 419-421	0.3	
82	Clinical efficacy of haematopoietic stem cell transplantation for adult adrenoleukodystrophy. <i>Brain Communications</i> , 2020 , 2, fcz048	4.5	8
81	Comprehensive investigation of RNF213 nonsynonymous variants associated with intracranial artery stenosis. <i>Scientific Reports</i> , 2020 , 10, 11942	4.9	5
80	CNV analysis using whole exome sequencing identified biallelic CNVs of VPS13B in siblings with intellectual disability. <i>European Journal of Medical Genetics</i> , 2020 , 63, 103610	2.6	9
79	UBAP1 mutations cause juvenile-onset hereditary spastic paraplegias (SPG80) and impair UBAP1 targeting to endosomes. <i>Journal of Human Genetics</i> , 2019 , 64, 1055-1065	4.3	9
78	Oxygen consumption rate for evaluation of COQ2 variants associated with multiple system atrophy. <i>Neurogenetics</i> , 2019 , 20, 51-52	3	0

77	Ataxic phenotype with altered Ca _v 3.1 channel property in a mouse model for spinocerebellar ataxia 42. <i>Neurobiology of Disease</i> , 2019 , 130, 104516	7.5	15
76	Functional evaluation of PDGFB-variants in idiopathic basal ganglia calcification, using patient-derived iPSC cells. <i>Scientific Reports</i> , 2019 , 9, 5698	4.9	5
75	Whole-exome sequencing in a Japanese pedigree implicates a rare non-synonymous single-nucleotide variant in BEST3 as a candidate for mandibular prognathism. <i>Bone</i> , 2019 , 122, 193-198	4.7	5
74	Association of ATXN2 intermediate-length CAG repeats with amyotrophic lateral sclerosis correlates with the distributions of normal CAG repeat alleles among individual ethnic populations. <i>Neurogenetics</i> , 2019 , 20, 65-71	3	1
73	Prominent Spasticity and Hyperreflexia of the Legs in a Nepalese Patient with Friedreich Ataxia. <i>Internal Medicine</i> , 2019 , 58, 2865-2869	1.1	
72	Noncoding CGG repeat expansions in neuronal intranuclear inclusion disease, oculopharyngodistal myopathy and an overlapping disease. <i>Nature Genetics</i> , 2019 , 51, 1222-1232	36.3	132
71	Atypical Familial Amyotrophic Lateral Sclerosis with Slowly Progressing Lower Extremities-predominant Late-onset Muscular Weakness and Atrophy. <i>Internal Medicine</i> , 2019 , 58, 1851-1858	1.1	1
70	Late presented congenital myasthenic syndrome with novel compound heterozygous CHRNE mutations mimicking seronegative myasthenia gravis. <i>Neurology and Clinical Neuroscience</i> , 2019 , 7, 288-290	2.3	
69	Sporadic progressive myoclonic epilepsy with early-onset dementia caused by a de novo mutation in PSEN1. <i>Neurology and Clinical Neuroscience</i> , 2019 , 7, 294-296	0.3	
68	Neuroimaging, genetic, and enzymatic study in a Japanese family with a GBA gross deletion. <i>Parkinsonism and Related Disorders</i> , 2019 , 61, 57-63	3.6	4
67	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	5.5	15
66	PLA2G6-associated neurodegeneration presenting as a complicated form of hereditary spastic paraplegia. <i>Journal of Human Genetics</i> , 2019 , 64, 55-59	4.3	11
65	Generation of D1-1 TALEN isogenic control cell line from Dravet syndrome patient iPSCs using TALEN-mediated editing of the SCN1A gene. <i>Stem Cell Research</i> , 2018 , 28, 100-104	1.6	9
64	Expansions of intronic TTTCA and TTTTA repeats in benign adult familial myoclonic epilepsy. <i>Nature Genetics</i> , 2018 , 50, 581-590	36.3	152
63	Amnionless-mediated glycosylation is crucial for cell surface targeting of cubilin in renal and intestinal cells. <i>Scientific Reports</i> , 2018 , 8, 2351	4.9	14
62	Clinical and genetic features of Charcot-Marie-Tooth disease 2F and hereditary motor neuropathy 2B in Japan. <i>Journal of the Peripheral Nervous System</i> , 2018 , 23, 40-48	4.7	8
61	Frequency and characteristics of the TBK1 gene variants in Japanese patients with sporadic amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2018 , 64, 158.e15-158.e19	5.6	8
60	Mutations in COA7 cause spinocerebellar ataxia with axonal neuropathy. <i>Brain</i> , 2018 , 141, 1622-1636	11.2	27

59	A Homozygous LAMA2 Mutation of c.818G>A Caused Partial Merosin Deficiency in a Japanese Patient. <i>Internal Medicine</i> , 2018 , 57, 877-882	1.1	6
58	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	5.6	22
57	Novel De Novo KCND3 Mutation in a Japanese Patient with Intellectual Disability, Cerebellar Ataxia, Myoclonus, and Dystonia. <i>Cerebellum</i> , 2018 , 17, 237-242	4.3	11
56	A case of late-onset Krabbe disease which showed subacute progression of spastic paresis with bilateral spinal cord lesions. <i>Neurology and Clinical Neuroscience</i> , 2018 , 6, 104-106	0.3	0
55	Refining the clinical phenotype of Okur-Chung neurodevelopmental syndrome. <i>Human Genome Variation</i> , 2018 , 5, 18011	1.8	12
54	Novel mutations in the ALDH18A1 gene in complicated hereditary spastic paraplegia with cerebellar ataxia and cognitive impairment. <i>Journal of Human Genetics</i> , 2018 , 63, 1009-1013	4.3	12
53	A Japanese family with mutation in the proteinase inhibitor 12 L47P gene: A case report. <i>Journal of the Neurological Sciences</i> , 2018 , 384, 126-128	3.2	3
52	The pathogenesis linked to coenzyme Q10 insufficiency in iPSC-derived neurons from patients with multiple-system atrophy. <i>Scientific Reports</i> , 2018 , 8, 14215	4.9	30
51	Simultaneous detection of reduced and oxidized forms of coenzyme Q10 in human cerebral spinal fluid as a potential marker of oxidative stress. <i>Journal of Clinical Biochemistry and Nutrition</i> , 2018 , 63, 205-210	3.1	1
50	Host MICA Polymorphism as a Potential Predictive Marker in Response to Chemotherapy for Colorectal Liver Metastases. <i>Digestive Diseases</i> , 2018 , 36, 437-445	3.2	2
49	Novel mutation in a Japanese family with adult polyglucosan body disease. <i>Neurology: Genetics</i> , 2017 , 3, e138	3.8	6
48	A novel mutation causes X-linked intellectual disability type Nascimento. <i>Human Genome Variation</i> , 2017 , 4, 17019	1.8	20
47	Identification of candidate genes involved in the etiology of sporadic Tourette syndrome by exome sequencing. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2017 , 174, 712-723	3.5	13
46	Ataxic form of autosomal recessive PEX10-related peroxisome biogenesis disorders with a novel compound heterozygous gene mutation and characteristic clinical phenotype. <i>Journal of the Neurological Sciences</i> , 2017 , 375, 424-429	3.2	7
45	Letter re: A genome-wide association study in multiple system atrophy. <i>Neurology</i> , 2017 , 88, 1296	6.5	
44	Slowly progressive d-bifunctional protein deficiency with survival to adulthood diagnosed by whole-exome sequencing. <i>Journal of the Neurological Sciences</i> , 2017 , 372, 6-10	3.2	8
43	Altered expression of Crb2 in podocytes expands a variation of CRB2 mutations in steroid-resistant nephrotic syndrome. <i>Pediatric Nephrology</i> , 2017 , 32, 801-809	3.2	7
42	TBCD may be a causal gene in progressive neurodegenerative encephalopathy with atypical infantile spinal muscular atrophy. <i>Journal of Human Genetics</i> , 2017 , 62, 473-480	4.3	11

41	Novel mutation in the membrane metalloendopeptidase gene in a patient with the autosomal recessive form of Charcot-Marie-Tooth disease. <i>Neurology and Clinical Neuroscience</i> , 2017 , 5, 124-126	0.3	
40	Partial duplication of causes minifascicular neuropathy: A novel mutation detection of. <i>Annals of Clinical and Translational Neurology</i> , 2017 , 4, 415-421	5.3	10
39	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	4.3	24
38	Clinical and genetic diversities of Charcot-Marie-Tooth disease with MFN2 mutations in a large case study. <i>Journal of the Peripheral Nervous System</i> , 2017 , 22, 191-199	4.7	19
37	Novel mutation in the SOD1 gene in a patient with early-onset, rapidly progressive amyotrophic lateral sclerosis. <i>Neurology and Clinical Neuroscience</i> , 2017 , 5, 189-191	0.3	3
36	Human genetic variation database, a reference database of genetic variations in the Japanese population. <i>Journal of Human Genetics</i> , 2016 , 61, 547-53	4.3	212
35	Plasma Coenzyme Q10 Levels and Multiple System Atrophy-Reply. <i>JAMA Neurology</i> , 2016 , 73, 1499-1500	17.2	3
34	Modeling neurological diseases with induced pluripotent cells reprogrammed from immortalized lymphoblastoid cell lines. <i>Molecular Brain</i> , 2016 , 9, 88	4.5	15
33	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. <i>Nature Communications</i> , 2016 , 7, 11253	17.4	126
32	Respiratory and cardiac function in japanese patients with dysferlinopathy. <i>Muscle and Nerve</i> , 2016 , 53, 394-401	3.4	14
31	Plasma Coenzyme Q10 Levels in Patients With Multiple System Atrophy. <i>JAMA Neurology</i> , 2016 , 73, 977-802	17.2	24
30	Tubular aggregate myopathy caused by a novel mutation in the cytoplasmic domain of STIM1. <i>Neurology: Genetics</i> , 2016 , 2, e50	3.8	18
29	Atypical parkinsonism caused by Pro105Leu mutation of prion protein: A broad clinical spectrum. <i>Neurology: Genetics</i> , 2016 , 2, e48	3.8	10
28	AglN: measuring the landscape of CpG methylation of individual repetitive elements. <i>Bioinformatics</i> , 2016 , 32, 2911-9	7.2	22
27	Mutations in MME cause an autosomal-recessive Charcot-Marie-Tooth disease type 2. <i>Annals of Neurology</i> , 2016 , 79, 659-72	9.4	53
26	Novel COL6A2 mutation in a case of limb girdle muscular dystrophy phenotype with autosomal recessive inheritance. <i>Neurology and Clinical Neuroscience</i> , 2016 , 4, 189-191	0.3	
25	A Novel Mutation in ELOVL4 Leading to Spinocerebellar Ataxia (SCA) With the Hot Cross Bun Sign but Lacking Erythrokeratoderma: A Broadened Spectrum of SCA34. <i>JAMA Neurology</i> , 2015 , 72, 797-805	17.2	53
24	Variants associated with Gaucher disease in multiple system atrophy. <i>Annals of Clinical and Translational Neurology</i> , 2015 , 2, 417-26	5.3	76

23	The first Japanese familial case of spinocerebellar ataxia 23 with a novel mutation in the PDYN gene. <i>Parkinsonism and Related Disorders</i> , 2015 , 21, 332-4	3.6	7
22	DNA Sequencing and Other Methods of Exonic and Genomic Analyses 2015 , 77-85		
21	Exome sequencing shows a novel de novo mutation in ATL1. <i>Neurology and Clinical Neuroscience</i> , 2014 , 2, 1-4	0.3	2
20	Genomic aspects of sporadic neurodegenerative diseases. <i>Biochemical and Biophysical Research Communications</i> , 2014 , 452, 221-5	3.4	16
19	Mutant COQ2 in multiple-system atrophy. <i>New England Journal of Medicine</i> , 2014 , 371, 82-3	59.2	62
18	Sensorimotor neuropathy in late-onset Krabbe disease progressing over 40 years after onset. <i>Neurology and Clinical Neuroscience</i> , 2014 , 2, 114-116	0.3	
17	Rapid detection of expanded short tandem repeats in personal genomics using hybrid sequencing. <i>Bioinformatics</i> , 2014 , 30, 815-22	7.2	50
16	Exome analysis reveals a Japanese family with spinocerebellar ataxia, autosomal recessive 1. <i>Journal of the Neurological Sciences</i> , 2013 , 331, 158-60	3.2	2
15	ERBB4 mutations that disrupt the neuregulin-ErbB4 pathway cause amyotrophic lateral sclerosis type 19. <i>American Journal of Human Genetics</i> , 2013 , 93, 900-5	11	95
14	Identification of ATP1A3 mutations by exome sequencing as the cause of alternating hemiplegia of childhood in Japanese patients. <i>PLoS ONE</i> , 2013 , 8, e56120	3.7	61
13	Increased gene dosage of myelin protein zero causes Charcot-Marie-Tooth disease. <i>Annals of Neurology</i> , 2012 , 71, 84-92	9.4	28
12	CSF1R mutations identified in three families with autosomal dominantly inherited leukoencephalopathy. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2012 , 159B, 951-7	3.5	29
11	Clinical features and haplotype analysis of newly identified Japanese patients with gelsolin-related familial amyloidosis of Finnish type. <i>Neurogenetics</i> , 2012 , 13, 237-43	3	13
10	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, 320-9	11	76
9	C9ORF72 repeat expansion in amyotrophic lateral sclerosis in the Kii peninsula of Japan. <i>Archives of Neurology</i> , 2012 , 69, 1154-8		77
8	Posterior column ataxia with retinitis pigmentosa in a Japanese family with a novel mutation in FLVCR1. <i>Neurogenetics</i> , 2011 , 12, 117-21	3	31
7	Adult-onset leukoencephalopathies with vanishing white matter with novel missense mutations in EIF2B2, EIF2B3, and EIF2B5. <i>Neurogenetics</i> , 2011 , 12, 259-61	3	28
6	Multiplexed resequencing analysis to identify rare variants in pooled DNA with barcode indexing using next-generation sequencer. <i>Journal of Human Genetics</i> , 2010 , 55, 448-55	4.3	11

5	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	11	71
4	Mutations for Gaucher disease confer high susceptibility to Parkinson disease. <i>Archives of Neurology</i> , 2009 , 66, 571-6		143
3	SNP haplotype mapping in a small ALS family. <i>PLoS ONE</i> , 2009 , 4, e5687	3-7	3
2	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		37
1	Pathology of the sympathetic nervous system corresponding to the decreased cardiac uptake in 123I-metaiodobenzylguanidine (MIBG) scintigraphy in a patient with Parkinson disease. <i>Journal of the Neurological Sciences</i> , 2006 , 243, 101-4	3-2	57