Jun Mitsui

List of Publications by Year
in descending order

120 papers	3,313 citations	201575 27 h-index	168321 53 g-index
129 all docs	129 docs citations	129 times ranked	8368 citing authors

#	Article	IF	Citations
1	Human genetic variation database, a reference database of genetic variations in the Japanese population. Journal of Human Genetics, 2016, 61, 547-553.	1.1	270
2	Noncoding CGG repeat expansions in neuronal intranuclear inclusion disease, oculopharyngodistal myopathy and an overlapping disease. Nature Genetics, 2019, 51, 1222-1232.	9.4	265
3	ExpansionsÂofÂintronic TTTCA and TTTTA repeats in benign adult familial myoclonic epilepsy. Nature Genetics, 2018, 50, 581-590.	9.4	238
4	Mutations for Gaucher Disease Confer High Susceptibility to Parkinson Disease. Archives of Neurology, 2009, 66, 571-6.	4.9	183
5	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. Nature Communications, 2016, 7, 11253.	5.8	174
6	ERBB4 Mutations that Disrupt the Neuregulin-ErbB4 Pathway Cause Amyotrophic Lateral Sclerosis Type 19. American Journal of Human Genetics, 2013, 93, 900-905.	2.6	123
7	The TRK-Fused Gene Is Mutated in Hereditary Motor and Sensory Neuropathy with Proximal Dominant Involvement. American Journal of Human Genetics, 2012, 91, 320-329.	2.6	98
8	Variants associated with Gaucher disease in multiple system atrophy. Annals of Clinical and Translational Neurology, 2015, 2, 417-426.	1.7	90
9	C9ORF72 Repeat Expansion in Amyotrophic Lateral Sclerosis in the Kii Peninsula of Japan. Archives of Neurology, 2012, 69, 1154-8.	4.9	88
10	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.	2.6	85
11	Mutations in MME cause an autosomalâ€recessive Charcotâ€“Marieâ€“Tooth disease type 2. Annals of Neurology, 2016, 79, 659-672.	2.8	82
12	Mutant COQ2 in Multiple-System Atrophy. New England Journal of Medicine, 2014, 371, 80-83.	13.9	81
13	A Novel Mutation inELOVL4Leading to Spinocerebellar Ataxia (SCA) With the Hot Cross Bun Sign but Lacking Erythrokeratodermia. JAMA Neurology, 2015, 72, 797.	4.5	79
14	Identification of ATP1A3 Mutations by Exome Sequencing as the Cause of Alternating Hemiplegia of Childhood in Japanese Patients. PLoS ONE, 2013, 8, e56120.	1.1	79
15	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, 101-104.	0.3	69
16	Rapid detection of expanded short tandem repeats in personal genomics using hybrid sequencing. Bioinformatics, 2014, 30, 815-822.	1.8	61
17	The pathogenesis linked to coenzyme Q10 insufficiency in iPSC-derived neurons from patients with multiple-system atrophy. Scientific Reports, 2018, 8, 14215.	1.6	50
18	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.	4.9	44

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19	Plasma Coenzyme Q10 Levels in Patients With Multiple System Atrophy. JAMA Neurology, 2016, 73, 977.	4.5	42
20	Posterior column ataxia with retinitis pigmentosa in a Japanese family with a novel mutation in FLVCR1. Neurogenetics, 2011, 12, 117-121.	0.7	38
21	Mutations in COA7 cause spinocerebellar ataxia with axonal neuropathy. Brain, 2018, 141, 1622-1636.	3.7	38
22	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.	1.5	37
23	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.	1.4	35
24	CSF1R mutations identified in three families with autosomal dominantly inherited leukoencephalopathy. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2012, 159B, 951-957.	1.1	34
25	Adult-onset leukoencephalopathies with vanishing white matter with novel missense mutations in EIF2B2, EIF2B3, and EIF2B5. Neurogenetics, 2011, 12, 259-261.	0.7	32
26	Clinical and genetic diversities of Charcotâ€Marieâ€Tooth disease with MFN2 mutations in a large case study. Journal of the Peripheral Nervous System, 2017, 22, 191-199.	1.4	31
27	Increased gene dosage of myelin protein zero causes Charcotâ€Marieâ€Tooth disease. Annals of Neurology, 2012, 71, 84-92.	2.8	30
28	AgIn: measuring the landscape of CpG methylation of individual repetitive elements. Bioinformatics, 2016, 32, 2911-2919.	1.8	29
29	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.	0.9	28
30	Tubular aggregate myopathy caused by a novel mutation in the cytoplasmic domain of STIM1. Neurology: Genetics, 2016, 2, e50.	0.9	27
31	Amnionless-mediated glycosylation is crucial for cell surface targeting of cubilin in renal and intestinal cells. Scientific Reports, 2018, 8, 2351.	1.6	27
32	A novel UBE2A mutation causes X-linked intellectual disability type Nascimento. Human Genome Variation, 2017, 4, 17019.	0.4	26
33	Associations of pathological diagnosis and genetic abnormalities in meningiomas with the embryological origins of the meninges. Scientific Reports, 2021, 11, 6987.	1.6	23
34	Respiratory and cardiac function in japanese patients with dysferlinopathy. Muscle and Nerve, 2016, 53, 394-401.	1.0	22
35	Modeling neurological diseases with induced pluripotent cells reprogrammed from immortalized lymphoblastoid cell lines. Molecular Brain, 2016, 9, 88.	1.3	21
36	Novel De Novo KCND3 Mutation in a Japanese Patient with Intellectual Disability, Cerebellar Ataxia, Myoclonus, and Dystonia. Cerebellum, 2018, 17, 237-242.	1.4	21

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37	Refining the clinical phenotype of Okurâ€“Chung neurodevelopmental syndrome. Human Genome Variation, 2018, 5, 18011.	0.4	21
38	Genomic aspects of sporadic neurodegenerative diseases. Biochemical and Biophysical Research Communications, 2014, 452, 221-225.	1.0	20
39	Ataxic phenotype with altered CaV3.1 channel property in a mouse model for spinocerebellar ataxia 42. Neurobiology of Disease, 2019, 130, 104516.	2.1	20
40	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.	1.1	18
41	Genetic spectrum of <scp>Charcotâ€“Marieâ€“Tooth</scp> disease associated with myelin protein zero gene variants in Japan. Clinical Genetics, 2021, 99, 359-375.	1.0	18
42	Multiple system atrophy variant with severe hippocampal pathology. Brain Pathology, 2022, 32, e13002.	2.1	18
43	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.	1.1	17
44	Clinical and genetic features of Charcotâ€Marieâ€Tooth disease 2F and hereditary motor neuropathy 2B in Japan. Journal of the Peripheral Nervous System, 2018, 23, 40-48.	1.4	17
45	PLA2G6-associated neurodegeneration presenting as a complicated form of hereditary spastic paraplegia. Journal of Human Genetics, 2019, 64, 55-59.	1.1	17
46	COQ2 V393A confers high risk susceptibility for multiple system atrophy in East Asian population. Journal of the Neurological Sciences, 2021, 429, 117623.	0.3	17
47	Clinical features and haplotype analysis of newly identified Japanese patients with gelsolin-related familial amyloidosis of Finnish type. Neurogenetics, 2012, 13, 237-243.	0.7	15
48	TBCD may be a causal gene in progressive neurodegenerative encephalopathy with atypical infantile spinal muscular atrophy. Journal of Human Genetics, 2017, 62, 473-480.	1.1	15
49	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.	0.3	15
50	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.	1.5	15
51	UBAP1 mutations cause juvenile-onset hereditary spastic paraplegias (SPG80) and impair UBAP1 targeting to endosomes. Journal of Human Genetics, 2019, 64, 1055-1065.	1.1	15
52	Partial duplication of <scp>DHH</scp> causes minifascicular neuropathy. Annals of Clinical and Translational Neurology, 2017, 4, 415-421.	1.7	14
53	CNV analysis using whole exome sequencing identified biallelic CNVs of VPS13B in siblings with intellectual disability. European Journal of Medical Genetics, 2020, 63, 103610.	0.7	14
54	Clinical efficacy of haematopoietic stem cell transplantation for adult adrenoleukodystrophy. Brain Communications, 2020, 2, fcz048.	1.5	14

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55	Multiplexed resequencing analysis to identify rare variants in pooled DNA with barcode indexing using next-generation sequencer. Journal of Human Genetics, 2010, 55, 448-455.	1.1	13
56	A Novel de novo KIF1A Mutation in a Patient with Autism, Hyperactivity, Epilepsy, Sensory Disturbance, and Spastic Paraplegia. Internal Medicine, 2020, 59, 839-842.	0.3	13
57	Atypical parkinsonism caused by Pro105Leu mutation of prion protein. Neurology: Genetics, 2016, 2, e48.	0.9	12
58	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.	0.3	12
59	Altered expression of Crb2 in podocytes expands a variation of CRB2 mutations in steroid-resistant nephrotic syndrome. Pediatric Nephrology, 2017, 32, 801-809.	0.9	12
60	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.	1.5	12
61	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.	1.1	12
62	Comprehensive investigation of RNF213 nonsynonymous variants associated with intracranial artery stenosis. Scientific Reports, 2020, 10, 11942.	1.6	11
63	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.	2.4	10
64	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.	1.4	9
65	Juvenile amyotrophic lateral sclerosis with complex phenotypes associated with novel SYNE1 mutations. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2021, 22, 576-578.	1.1	9
66	Muscle Transcriptomics Shows Overexpression of Cadherin 1 in Inclusion Body Myositis. Annals of Neurology, 2022, 91, 317-328.	2.8	9
67	The first Japanese familial case of spinocerebellar ataxia 23 with a novel mutation in the PDYN gene. Parkinsonism and Related Disorders, 2015, 21, 332-334.	1.1	8
68	Novel GBE1 mutation in a Japanese family with adult polyglucosan body disease. Neurology: Genetics, 2017, 3, e138.	0.9	8
69	Slowly progressive d-bifunctional protein deficiency with survival to adulthood diagnosed by whole-exome sequencing. Journal of the Neurological Sciences, 2017, 372, 6-10.	0.3	8
70	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.	0.6	8
71	Functional evaluation of PDGFB-variants in idiopathic basal ganglia calcification, using patient-derived iPS cells. Scientific Reports, 2019, 9, 5698.	1.6	8
72	Clinical features of inherited neuropathy with BSCL2 mutations in Japan. Journal of the Peripheral Nervous System, 2020, 25, 125-131.	1.4	7

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73	Clinical Impact of Copy Number Variation on the Genetic Diagnosis of Syndromic Aortopathies. Circulation Genomic and Precision Medicine, 2021, 14, e003458.	1.6	7
74	Novel de novo <scp>POLR3B</scp> mutations responsible for demyelinating Charcotâ€“Marieâ€“Tooth disease in Japan. Annals of Clinical and Translational Neurology, 2022, 9, 747-755.	1.7	7
75	A Homozygous LAMA2 Mutation of c.818G>A Caused Partial Merosin Deficiency in a Japanese Patient. Internal Medicine, 2018, 57, 877-882.	0.3	6
76	A Japanese family with mutation in the proteinase inhibitor 12 L47P gene: A case report. Journal of the Neurological Sciences, 2018, 384, 126-128.	0.3	6
77	Neuroimaging, genetic, and enzymatic study in a Japanese family with a GBA gross deletion. Parkinsonism and Related Disorders, 2019, 61, 57-63.	1.1	6
78	Splice-site mutations in KIF5A in the Japanese case series of amyotrophic lateral sclerosis. Neurogenetics, 2021, 22, 11-17.	0.7	6
79	Exome analysis reveals a Japanese family with spinocerebellar ataxia, autosomal recessive 1. Journal of the Neurological Sciences, 2013, 331, 158-160.	0.3	5
80	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.	0.7	5
81	Reliability and validity of Japanese version of Unified Multiple System Atrophy Rating Scale. Neurology and Clinical Neuroscience, 2021, 9, 171-180.	0.2	5
82	SNP Haplotype Mapping in a Small ALS Family. PLoS ONE, 2009, 4, e5687.	1.1	5
83	Randomized, doubleâ€blind, placeboâ€controlled phase 1 study to evaluate the safety and pharmacokinetics of high doses of ubiquinol in healthy adults. Neurology and Clinical Neuroscience, 2022, 10, 14-24.	0.2	5
84	Divergent variant patterns among 19 patients with<scp>Rubinsteinâ€Taybi</scp>syndrome uncovered by comprehensive genetic analysis including whole genome sequencing. Clinical Genetics, 2022, 101, 335-345.	1.0	5
85	An NEFH founder mutation causes broad phenotypic spectrum in multiple Japanese families. Journal of Human Genetics, 2022, 67, 399-403.	1.1	5
86	DMD exon 2 duplication due to a complex genomic rearrangement is associated with a somatic mosaicism. Neuromuscular Disorders, 2022, 32, 263-269.	0.3	5
87	Plasma Coenzyme Q10 Levels and Multiple System Atrophyâ€”Reply. JAMA Neurology, 2016, 73, 1499.	4.5	4
88	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.	1.1	4
89	Elderly patients with suspected Charcot-Marie-Tooth disease should be tested for the TTR gene for effective treatments. Journal of Human Genetics, 2022, , .	1.1	4
90	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.	0.2	3

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91	Oxygen consumption rate for evaluation of COQ2 variants associated with multiple system atrophy. Neurogenetics, 2019, 20, 51-52.	0.7	3
92	Familial dementia with Lewy bodies with VPS13C mutations. Parkinsonism and Related Disorders, 2020, 81, 31-33.	1.1	3
93	Frequency of FMR1 Premutation Alleles in Patients with Undiagnosed Cerebellar Ataxia and Multiple System Atrophy in the Japanese Population. Cerebellum, 2021, , 1.	1.4	3
94	Exome sequencing shows a novel de novo mutation in <scp>ATL</scp>1. Neurology and Clinical Neuroscience, 2014, 2, 1-4.	0.2	2
95	Host MICA Polymorphism as a Potential Predictive Marker in Response to Chemotherapy for Colorectal Liver Metastases. Digestive Diseases, 2018, 36, 437-445.	0.8	2
96	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.	0.2	2
97	Esophageal Dysmotility is Common in Patients With Multiple System Atrophy. Laryngoscope, 2021, 131, 832-838.	1.1	2
98	Complex hereditary peripheral neuropathies caused by novel variants in mitochondrial-related nuclear genes. Journal of Neurology, 2022, 269, 4129-4140.	1.8	2
99	Symptomatic and Stenotic Developmental Venous Anomaly with Pontine Capillary Telangiectasia: A Case Report with Genetic Considerations. NMC Case Report Journal, 2022, 9, 139-144.	0.2	2
100	Numerical Analysis on Hydraulic Characteristics of Tsunami Overtopping Caisson Breakwaters. , 2018, , .		1
101	Atypical Familial Amyotrophic Lateral Sclerosis with Slowly Progressing Lower Extremities-predominant Late-onset Muscular Weakness and Atrophy. Internal Medicine, 2019, 58, 1851-1858.	0.3	1
102	An autopsy case of G M1 gangliosidosis type II in a patient who survived a long duration with artificial respiratory support. Neuropathology, 2020, 40, 379-388.	0.7	1
103	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.	0.9	1
104	Brainstem intraparenchymal schwannoma with genetic analysis: a case report and literature review. BMC Medical Genomics, 2021, 14, 205.	0.7	1
105	Sensorimotor neuropathy in lateâ€onset <scp>K</scp>rabbe disease progressing over 40Âyears after onset. Neurology and Clinical Neuroscience, 2014, 2, 114-116.	0.2	0
106	DNA Sequencing and Other Methods of Exonic and Genomic Analyses. , 2015, , 77-85.		0
107	Novel COL6A2 mutation in a case of limb girdle muscular dystrophy phenotype with autosomal recessive inheritance. Neurology and Clinical Neuroscience, 2016, 4, 189-191.	0.2	0
108	Letter re: A genome-wide association study in multiple system atrophy. Neurology, 2017, 88, 1296-1296.	1.5	0

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109	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.	0.2	0
110	Prominent Spasticity and Hyperreflexia of the Legs in a Nepalese Patient with Friedreich Ataxia. Internal Medicine, 2019, 58, 2865-2869.	0.3	0
111	Late presented congenital myasthenic syndrome with novel compound heterozygous CHRNE mutations mimicking seronegative myasthenia gravis. Neurology and Clinical Neuroscience, 2019, 7, 288-290.	0.2	0
112	Sporadic progressive myoclonic epilepsy with earlyâ€onset dementia caused by a de novo mutation in PSEN1. Neurology and Clinical Neuroscience, 2019, 7, 294-296.	0.2	0
113	A novel multiâ€exon deletion in the dysferlin gene of a limbâ€girdle muscular dystrophy type 2B Filipino patient. Neurology and Clinical Neuroscience, 2020, 8, 419-421.	0.2	0
114	Novel variant of CSF1R in sporadic case with earlyâ€onset cognitive impairment. Neurology and Clinical Neuroscience, 2020, 8, 430-432.	0.2	0
115	A novel mutation in ABCD1 gene in a Filipino patient with adultâ€onset Xâ€linked ALD. Neurology and Clinical Neuroscience, 2020, 8, 329-331.	0.2	0
116	A Case of Irreversible Corneal Edema Associated with Dentatorubropallidoluysian Atrophy Following Corneal Endothelial Transplantation. SN Comprehensive Clinical Medicine, 2021, 3, 2029-2032.	0.3	0
117	DNA sequencing and other methods of exonic and genomic analyses. , 2020, , 109-120.		0
118	Family with congenital contractural arachnodactyly due to a novel multiexon deletion of the FBN2 gene. Clinical Case Reports (discontinued), 2022, 10, e05335.	0.2	0
119	Expression profile analysis in cells overexpressing <scp>DRPLA cDNA</scp> to explore the roles of <scp>DRPLAp</scp> as a transcriptional coregulator. Neurology and Clinical Neuroscience, 0, , .	0.2	0
120	A case of idiopathic normal pressure hydrocephalus with fragile X-associated tremor/ataxia syndrome. Clinical Neurology and Neurosurgery, 2022, 218, 107278.	0.6	0