Tatsushi Toda

List of Publications by Year
in descending order

219	11,617	46918 47	31759 101
papers	citations	h-index	g-index

230	230	230	14435
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Genome-wide association study identifies common variants at four loci as genetic risk factors for Parkinson's disease. Nature Genetics, 2009, 41, 1303-1307.	9.4	1,217
2	An ancient retrotransposal insertion causes Fukuyama-type congenital muscular dystrophy. Nature, 1998, 394, 388-392.	13.7	758
3	Muscular Dystrophy and Neuronal Migration Disorder Caused by Mutations in a Glycosyltransferase, POMGnT1. Developmental Cell, 2001, 1, 717-724.	3.1	675
4	Modeling Alzheimerâ€™s Disease with iPSCs Reveals Stress Phenotypes Associated with Intracellular AÎ² and Differential Drug Responsiveness. Cell Stem Cell, 2013, 12, 487-496.	5.2	652
5	Comprehensive Research Synopsis and Systematic Meta-Analyses in Parkinson's Disease Genetics: The PDGene Database. PLoS Genetics, 2012, 8, e1002548.	1.5	495
6	A toxic monomeric conformer of the polyglutamine protein. Nature Structural and Molecular Biology, 2007, 14, 332-340.	3.6	296
7	CHCHD2 mutations in autosomal dominant late-onset Parkinson's disease: a genome-wide linkage and sequencing study. Lancet Neurology, The, 2015, 14, 274-282.	4.9	285
8	Noncoding CGG repeat expansions in neuronal intranuclear inclusion disease, oculopharyngodistal myopathy and an overlapping disease. Nature Genetics, 2019, 51, 1222-1232.	9.4	265
9	Spinocerebellar Ataxia Type 31 Is Associated with â€œInsertedâ€•Penta-Nucleotide Repeats Containing (TGGAA)n. American Journal of Human Genetics, 2009, 85, 544-557.	2.6	260
10	Pikachurin, a dystroglycan ligand, is essential for photoreceptor ribbon synapse formation. Nature Neuroscience, 2008, 11, 923-931.	7.1	255
11	UCHL1 is a Parkinson's disease susceptibility gene. Annals of Neurology, 2004, 55, 512-521.	2.8	227
12	Multiple candidate gene analysis identifies Î±-synuclein as a susceptibility gene for sporadic Parkinson's disease. Human Molecular Genetics, 2006, 15, 1151-1158.	1.4	210
13	Association studies of multiple candidate genes for Parkinson's disease using single nucleotide polymorphisms. Annals of Neurology, 2002, 51, 133-136.	2.8	200
14	Mutations for Gaucher Disease Confer High Susceptibility to Parkinson Disease. Archives of Neurology, 2009, 66, 571-6.	4.9	183
15	Identification of a Post-translational Modification with Ribitol-Phosphate and Its Defect in Muscular Dystrophy. Cell Reports, 2016, 14, 2209-2223.	2.9	180
16	Identification of Risk Loci for Parkinson Disease in Asians and Comparison of Risk Between Asians and Europeans. JAMA Neurology, 2020, 77, 746.	4.5	170
17	Pathogenic exon-trapping by SVA retrotransposon and rescue in Fukuyama muscular dystrophy. Nature, 2011, 478, 127-131.	13.7	162
18	Worldwide distribution and broader clinical spectrum of muscle-eye-brain disease. Human Molecular Genetics, 2003, 12, 527-534.	1.4	133

#	Article	IF	Citations
19	Cell surface flip-flop of phosphatidylserine is critical for PIEZO1-mediated myotube formation. Nature Communications, 2018, 9, 2049.	5.8	127
20	An Autosomal Dominant Cerebellar Ataxia Linked to Chromosome 16q22.1 Is Associated with a Single-Nucleotide Substitution in the 5â€² Untranslated Region of the Gene Encoding a Protein with Spectrin Repeat and Rho Guanine-Nucleotide Exchange-Factor Domains. American Journal of Human Genetics, 2005, 77, 280-296.	2.6	124
21	Prevention of polyglutamine oligomerization and neurodegeneration by the peptide inhibitor QBP1 in Drosophila. Human Molecular Genetics, 2003, 12, 1253-1259.	1.4	122
22	Large-scale replication and heterogeneity in Parkinson disease genetic loci. Neurology, 2012, 79, 659-667.	1.5	119
23	Fukutin is required for maintenance of muscle integrity, cortical histiogenesis and normal eye development. Human Molecular Genetics, 2003, 12, 1449-1459.	1.4	116
24	Deficiency of Î±-Dystroglycan in Muscleâ€“Eyeâ€“Brain Disease. Biochemical and Biophysical Research Communications, 2002, 291, 1283-1286.	1.0	115
25	The genetic and molecular basis of muscular dystrophy: roles of cellâ€“matrix linkage in the pathogenesis. Journal of Human Genetics, 2006, 51, 915-926.	1.1	113
26	TRPV2 is critical for the maintenance of cardiac structure and function in mice. Nature Communications, 2014, 5, 3932.	5.8	109
27	Loss-of-function of an N-acetylglucosaminyltransferase, POMGnT1, in muscleâ€“eyeâ€“brain disease. Biochemical and Biophysical Research Communications, 2003, 306, 93-97.	1.0	99
28	Spermatogenic ability is different among males in different Y chromosome lineage. Journal of Human Genetics, 1999, 44, 289-292.	1.1	98
29	A new mutation of thefukutin gene in a non-Japanese patient. Annals of Neurology, 2003, 53, 392-396.	2.8	98
30	A multi-centre clinico-genetic analysis of the VPS35 gene in Parkinson disease indicates reduced penetrance for disease-associated variants. Journal of Medical Genetics, 2012, 49, 721-726.	1.5	94
31	Variants associated with Gaucher disease in multiple system atrophy. Annals of Clinical and Translational Neurology, 2015, 2, 417-426.	1.7	90
32	Detection of Polyglutamine Protein Oligomers in Cells by Fluorescence Correlation Spectroscopy. Journal of Biological Chemistry, 2007, 282, 24039-24048.	1.6	89
33	Are breaches in the glia limitans the primary cause of the micropolygyria in Fukuyama-type congenital muscular dystrophy (FCMD)? - Pathological study of the cerebral cortex of an FCDM fetus. Acta Neuropathologica, 1996, 91, 313-321.	3.9	82
34	The Fukuyama congenital muscular dystrophy story. Neuromuscular Disorders, 2000, 10, 153-159.	0.3	81
35	Residual laminin-binding activity and enhanced dystroglycan glycosylation by LARGE in novel model mice to dystroglycanopathy. Human Molecular Genetics, 2009, 18, 621-631.	1.4	76
36	Variants in saposin D domain of prosaposin gene linked to Parkinsonâ€™s disease. Brain, 2020, 143, 1190-1205.	3.7	72

#	Article	IF	Citations
37	Molecular interaction between fukutin and POMGnT1 in the glycosylation pathway of Î±-dystroglycan. Biochemical and Biophysical Research Communications, 2006, 350, 935-941.	1.0	69
38	Mechanistic aspects of the formation of Î±-dystroglycan and therapeutic research for the treatment of Î±-dystroglycanopathy: A review. Molecular Aspects of Medicine, 2016, 51, 115-124.	2.7	69
39	Neurological and related adverse events in immune checkpoint inhibitors: a pharmacovigilance study from the Japanese Adverse Drug Event Report database. Journal of Neuro-Oncology, 2019, 145, 1-9.	1.4	69
40	Basement membrane fragility underlies embryonic lethality in fukutin-null mice. Neurobiology of Disease, 2005, 19, 208-217.	2.1	68
41	Glycosylation in Congenital Muscular Dystrophies. Biological and Pharmaceutical Bulletin, 2003, 26, 1641-1647.	0.6	67
42	The Muscular Dystrophy Gene TMEM5 Encodes a Ribitol Î²1,4-Xylosyltransferase Required for the Functional Glycosylation of Dystroglycan. Journal of Biological Chemistry, 2016, 291, 24618-24627.	1.6	62
43	Carbohydrate-binding domain of the POMGnT1 stem region modulates O -mannosylation sites of Î±-dystroglycan. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 9280-9285.	3.3	59
44	Enzymatic diagnostic test for Muscle-Eye-Brain type congenital muscular dystrophy using commercially available reagents. Clinical Biochemistry, 2003, 36, 339-344.	0.8	54
45	Quantifying normal and parkinsonian gait features from home movies: Practical application of a deep learningâ€"based 2D pose estimator. PLoS ONE, 2019, 14, e0223549.	1.1	52
46	Genetic variations on the Y chromosome in the Japanese population and implications for modern human Y chromosome lineage. Journal of Human Genetics, 1999, 44, 240-245.	1.1	51
47	Genetic identity of Fukuyama-type congenital muscular dystrophy and Walker-Warburg syndrome. Annals of Neurology, 1995, 37, 99-101.	2.8	50
48	Fukuyamaâ€type congenital muscular dystrophy (FCMD) and Î±â€dystroglycanopathy. Congenital Anomalies (discontinued), 2003, 43, 97-104.	0.3	50
49	Facial nerve palsy following the administration of COVID-19 mRNA vaccines: analysis of a self-reporting database. International Journal of Infectious Diseases, 2021, 111, 310-312.	1.5	48
50	Post-translational Maturation of Dystroglycan Is Necessary for Pikachurin Binding and Ribbon Synaptic Localization*. Journal of Biological Chemistry, 2010, 285, 31208-31216.	1.6	46
51	Wide distribution of alpha-synuclein oligomers in multiple system atrophy brain detected by proximity ligation. Acta Neuropathologica, 2019, 137, 455-466.	3.9	45
52	Sister and brother with Vici syndrome: Agenesis of the corpus callosum, albinism, and recurrent infections. American Journal of Medical Genetics Part A, 2002, 109, 61-66.	2.4	44
53	Delivery of the aggregate inhibitor peptide QBP1 into the mouse brain using PTDs and its therapeutic effect on polyglutamine disease mice. Neuroscience Letters, 2009, 449, 87-92.	1.0	44
54	A deep learning method for HLA imputation and trans-ethnic MHC fine-mapping of type 1 diabetes. Nature Communications, 2021, 12, 1639.	5.8	44

#	Article	IF	Citations
55	Effects of fukutin deficiency in the developing mouse brain. Neuromuscular Disorders, 2005, 15, 416-426.	0.3	43
56	Aberrant neuromuscular junctions and delayed terminal muscle fiber maturation in Î±-dystroglycanopathies. Human Molecular Genetics, 2006, 15, 1279-1289.	1.4	41
57	Impaired viability of muscle precursor cells in muscular dystrophy with glycosylation defects and amelioration of its severe phenotype by limited gene expression. Human Molecular Genetics, 2013, 22, 3003-3015.	1.4	40
58	Structural organization, complete genomic sequences and mutational analyses of the Fukuyama-type congenital muscular dystrophy gene, fukutin. FEBS Letters, 2001, 489, 192-196.	1.3	39
59	Genetically regulated epigenetic transcriptional activation of retrotransposon insertion confers mouse dactylaplasia phenotype. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 19034-19039.	3.3	38
60	Hsp40 Gene Therapy Exerts Therapeutic Effects on Polyglutamine Disease Mice via a Non-Cell Autonomous Mechanism. PLoS ONE, 2012, 7, e51069.	1.1	38
61	Structureâ€“function analysis of human protein O-linked mannose Î²1,2-N-acetylglucosaminyltransferase 1, POMGnT1. Biochemical and Biophysical Research Communications, 2004, 320, 39-44.	1.0	37
62	Haplotype-Phenotype correlation in Fukuyama congenital muscular dystrophy. , 2000, 92, 184-190.		36
63	Congenital muscular dystrophy associated with calf hypertrophy, microcephaly and severe mental retardation in three Italian families: evidence for a novel CMD syndrome. Neuromuscular Disorders, 2000, 10, 541-547.	0.3	36
64	Founder SVA retrotransposal insertion in Fukuyama-type congenital muscular dystrophy and its origin in Japanese and Northeast Asian populations. American Journal of Medical Genetics, Part A, 2005, 138A, 344-348.	0.7	35
65	Effect of istradefylline on mood disorders in Parkinson's disease. Journal of the Neurological Sciences, 2019, 396, 78-83.	0.3	35
66	Neuropsychiatric adverse events of chloroquine: a real-world pharmacovigilance study using the FDA Adverse Event Reporting System (FAERS) database. BioScience Trends, 2020, 14, 139-143.	1.1	35
67	Inflammatory myopathy with myasthenia gravis. Neurology: Neuroimmunology and NeuroInflammation, 2019, 6, e535.	3.1	33
68	Defective peripheral nerve myelination and neuromuscular junction formation in fukutin-deficient chimeric mice. Journal of Neurochemistry, 2007, 101, 1712-1722.	2.1	32
69	AGO61-dependent GlcNAc modification primes the formation of functional glycans on Î±-dystroglycan. Scientific Reports, 2013, 3, 3288.	1.6	32
70	Genome-wide association studies identify polygenic effects for completed suicide in the Japanese population. Neuropsychopharmacology, 2019, 44, 2119-2124.	2.8	32
71	Expression profiling of muscles from Fukuyama-type congenital muscular dystrophy and laminin-Î±2 deficient congenital muscular dystrophy; is congenital muscular dystrophy a primary fibrotic disease?. Biochemical and Biophysical Research Communications, 2006, 342, 489-502.	1.0	31
72	Age and origin of the FCMD 3â€²-untranslated-region retrotransposal insertion mutation causing Fukuyama-type congenital muscular dystrophy in the Japanese population. Human Genetics, 2000, 107, 559-567.	1.8	30

#	Article	IF	Citations
73	YY1 binds to Î±-synuclein 3â€²-flanking region SNP and stimulates antisense noncoding RNA expression. Journal of Human Genetics, 2013, 58, 711-719.	1.1	30
74	Inhibition of Protein Misfolding/Aggregation Using Polyglutamine Binding Peptide QBP1 as a Therapy for the Polyglutamine Diseases. Neurotherapeutics, 2013, 10, 440-446.	2.1	30
75	Human Natural Killer-1 Sulfotransferase (HNK-1ST)-induced Sulfate Transfer Regulates Laminin-binding Glycans on Î±-Dystroglycan. Journal of Biological Chemistry, 2012, 287, 30823-30832.	1.6	29
76	Galectin 3â€“binding protein suppresses amyloid-Î² production by modulating Î²-cleavage of amyloid precursor protein. Journal of Biological Chemistry, 2020, 295, 3678-3691.	1.6	29
77	Toward identification of susceptibility genes for sporadic Parkinson?s disease. Journal of Neurology, 2003, 250, 1-1.	1.8	28
78	Absence of Post-phosphoryl Modification in Dystroglycanopathy Mouse Models and Wild-type Tissues Expressing Non-laminin Binding Form of Î±-Dystroglycan. Journal of Biological Chemistry, 2012, 287, 9560-9567.	1.6	28
79	ABCG2 variant has opposing effects on onset ages of Parkinson's disease and gout. Annals of Clinical and Translational Neurology, 2015, 2, 302-306.	1.7	28
80	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
81	Biallelic variants in LIG3 cause a novel mitochondrial neurogastrointestinal encephalomyopathy. Brain, 2021, 144, 1451-1466.	3.7	28
82	Chronic cerebral hypoperfusion shifts the equilibrium of amyloid Î² oligomers to aggregation-prone species with higher molecular weight. Scientific Reports, 2019, 9, 2827.	1.6	27
83	Disruption of the toxic conformation of the expanded polyglutamine stretch leads to suppression of aggregate formation and cytotoxicity. Biochemical and Biophysical Research Communications, 2004, 317, 1200-1206.	1.0	26
84	Muscular Dystrophy with Ribitol-Phosphate Deficiency: A Novel Post-Translational Mechanism in Dystroglycanopathy. Journal of Neuromuscular Diseases, 2017, 4, 259-267.	1.1	26
85	Elimination of fukutin reveals cellular and molecular pathomechanisms in muscular dystrophy-associated heart failure. Nature Communications, 2019, 10, 5754.	5.8	26
86	Antenatal and Postnatal Brain Magnetic Resonance Imaging in Muscle-Eye-Brain Disease. Archives of Neurology, 2004, 61, 1301-6.	4.9	25
87	Mislocalization of Fukutin Protein by Disease-causing Missense Mutations Can Be Rescued with Treatments Directed at Folding Amelioration. Journal of Biological Chemistry, 2012, 287, 8398-8406.	1.6	25
88	In silico drug screening by using genome-wide association study data repurposed dabrafenib, an anti-melanoma drug, for Parkinsonâ€™s disease. Human Molecular Genetics, 2018, 27, 3974-3985.	1.4	25
89	Î²4GalT-II is a key regulator of glycosylation of the proteins involved in neuronal development. Biochemical and Biophysical Research Communications, 2005, 333, 131-137.	1.0	24
90	Fukutin is prerequisite to ameliorate muscular dystrophic phenotype by myofiber-selective LARGE expression. Scientific Reports, 2015, 5, 8316.	1.6	24

#	Article	IF	Citations
91	Efficacy and Safety of Ultrahigh-Dose Methylcobalamin in Early-Stage Amyotrophic Lateral Sclerosis. JAMA Neurology, 2022, 79, 575.	4.5	24
92	Seizureâ€“genotype relationship in Fukuyama-type congenital muscular dystrophy. Brain and Development, 2008, 30, 59-67.	0.6	23
93	Overexpression of LARGE suppresses muscle regeneration via down-regulation of insulin-like growth factor 1 and aggravates muscular dystrophy in mice. Human Molecular Genetics, 2014, 23, 4543-4558.	1.4	23
94	Need of care in interpreting Google Trends-based COVID-19 infodemiological study results: potential risk of false-positivity. BMC Medical Research Methodology, 2021, 21, 147.	1.4	23
95	Fukuyama-type congenital muscular dystrophy: the first human disease to be caused by an ancient retrotransposal integration. Journal of Molecular Medicine, 1999, 77, 816-823.	1.7	22
96	Molecular genetic evidence of clinical heterogeneity in Fukuyama-type congenital muscular dystrophy. Human Genetics, 1997, 99, 427-432.	1.8	21
97	Genomic rearrangement at 10q24 in non-syndromic split-hand/split-foot malformation. Human Genetics, 2005, 118, 477-483.	1.8	21
98	Structureâ€“activity relationship study on polyglutamine binding peptide QBP1. Bioorganic and Medicinal Chemistry, 2009, 17, 1259-1263.	1.4	21
99	Aphasic status epilepticus preceding tumefactive left hemisphere lesion in anti-MOG antibody associated disease. Multiple Sclerosis and Related Disorders, 2019, 27, 91-94.	0.9	21
100	Crystal structures of fukutin-related protein (FKRP), a ribitol-phosphate transferase related to muscular dystrophy. Nature Communications, 2020, 11, 303.	5.8	21
101	The Aggregation Inhibitor Peptide QBP1 as a Therapeutic Molecule for the Polyglutamine Neurodegenerative Diseases. Journal of Amino Acids, 2011, 2011, 1-10.	5.8	20
102	Temporal requirement of dystroglycan glycosylation during brain development and rescue of severe cortical dysplasia via gene delivery in the fetal stage. Human Molecular Genetics, 2018, 27, 1174-1185.	1.4	20
103	CDP-glycerol inhibits the synthesis of the functional O-mannosyl glycan of Î±-dystroglycan. Journal of Biological Chemistry, 2018, 293, 12186-12198.	1.6	20
104	Arginine is a disease modifier for polyQ disease models that stabilizes polyQ protein conformation. Brain, 2020, 143, 1811-1825.	3.7	20
105	YAC and Cosmid Contigs Encompassing the Fukuyama-Type Congenital Muscular Dystrophy (FCMD) Candidate Region on 9q31. Genomics, 1997, 40, 284-293.	1.3	19
106	Ribitol-phosphateâ€”a newly identified posttranslational glycosylation unit in mammals: structure, modification enzymes and relationship to human diseases. Journal of Biochemistry, 2018, 163, 359-369.	0.9	19
107	Clinical Characteristics of Neuronal Intranuclear Inclusion Disease-Related Retinopathy With CGG Repeat Expansions in the NOTCH2NLC Gene. , 2020, 61, 27.		19
108	Colocalization of BRCA1 with Tau Aggregates in Human Tauopathies. Brain Sciences, 2020, 10, 7.	1.1	19

#	Article	IF	Citations
109	Genetic variations and clinical spectrum of dystroglycanopathy in a large cohort of Chinese patients. Clinical Genetics, 2021, 99, 384-395.	1.0	19
110	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
111	Prenatal diagnosis of Fukuyama type congenital muscular dystrophy by polymorphism analysis. , 1996, 66, 169-174.		16
112	National registry of patients with Fukuyama congenital muscular dystrophy in Japan. Neuromuscular Disorders, 2018, 28, 885-893.	0.3	16
113	Altered regulation of serum lysosomal acid hydrolase activities in Parkinson's disease: A potential peripheral biomarker?. Parkinsonism and Related Disorders, 2019, 61, 132-137.	1.1	16
114	Lower Serum Calcium as a Potentially Associated Factor for Conversion of Mild Cognitive Impairment to Early Alzheimerâ€™s Disease in the Japanese Alzheimerâ€™s Disease Neuroimaging Initiative. Journal of Alzheimer's Disease, 2019, 68, 777-788.	1.2	15
115	Subtypeâ€Dependent Reporting of Stroke With SGLT2 Inhibitors: Implications From a Japanese Pharmacovigilance Study. Journal of Clinical Pharmacology, 2020, 60, 629-635.	1.0	15
116	Randomized phase 2 study of perampanel for sporadic amyotrophic lateral sclerosis. Journal of Neurology, 2022, 269, 885-896.	1.8	15
117	Fukutin gene retrotransposal insertion in a nonâ€Japanese Fukuyama congenital muscular dystrophy (FCMD) patient. American Journal of Medical Genetics, Part A, 2009, 149A, 2403-2408.	0.7	14
118	Surface plasmon resonance characterization of specific binding of polyglutamine aggregation inhibitors to the expanded polyglutamine stretch. Biochemical and Biophysical Research Communications, 2009, 378, 634-639.	1.0	14
119	Paraneoplastic NMOSD associated with EG junction adenocarcinoma expressing unprotected AQP4. Neurology: Neuroimmunology and NeuroInflammation, 2018, 5, e482.	3.1	14
120	Clinical efficacy of haematopoietic stem cell transplantation for adult adrenoleukodystrophy. Brain Communications, 2020, 2, fcz048.	1.5	14
121	Transâ€Ethnic Fineâ€Mapping of the Major Histocompatibility Complex Region Linked to Parkinson's Disease. Movement Disorders, 2021, 36, 1805-1814.	2.2	14
122	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
123	Broader Clinical Spectrum of Fukuyama-Type Congenital Muscular Dystrophy Manifested by Haplotype Analysis. Journal of Child Neurology, 1999, 14, 711-715.	0.7	12
124	Modulation of motor learning by a paired associative stimulation protocol inducing LTD-like effects. Brain Stimulation, 2018, 11, 1314-1321.	0.7	12
125	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
126	Prenatal diagnosis of Fukuyama type congenital muscular dystrophy in eight Japanese families by haplotype analysis using new markers closest to the gene. American Journal of Medical Genetics Part A, 1998, 77, 310-316.	2.4	11

#	Article	IF	Citations
127	Isolation and Characterization of the Mouse Ortholog of the Fukuyama-type Congenital Muscular Dystrophy Gene. Genomics, 2002, 80, 482-486.	1.3	11
128	Novel POMGnT1 mutations cause muscle-eye-brain disease in Chinese patients. Molecular Genetics and Genomics, 2013, 288, 297-308.	1.0	11
129	Visualizing modules of coordinated structural brain atrophy during the course of conversion to Alzheimer's disease by applying methodology from gene co-expression analysis. NeuroImage: Clinical, 2019, 24, 101957.	1.4	11
130	Congenital hearing impairment associated with peripheral cochlear nerve dysmyelination in glycosylation-deficient muscular dystrophy. PLoS Genetics, 2020, 16, e1008826.	1.5	11
131	Predicting amyloid risk by machine learning algorithms based on the A4 screen data: Application to the Japanese Trialâ€Ready Cohort study. Alzheimer's and Dementia: Translational Research and Clinical Interventions, 2021, 7, e12135.	1.8	11
132	Fukuyama congenital muscular dystrophy: Cortical dysplasia of the cerebrum in a 20 week fetus. Neuropathology, 1996, 16, 184-189.	0.7	10
133	A case of Walkerâ€“Warburg syndrome. Brain and Development, 2000, 22, 454-457.	0.6	10
134	Genetic heterogeneity in three Chinese children with Fukuyama congenital muscular dystrophy. Neuromuscular Disorders, 2000, 10, 108-112.	0.3	10
135	Deep-intronic variant of fukutin is the most prevalent point mutation of Fukuyama congenital muscular dystrophy in Japan. Journal of Human Genetics, 2017, 62, 945-948.	1.1	10
136	Variable Cardiac Responses to Immunosuppressive Therapy in Anti-Mitochondrial Antibody-Positive Myositis. Canadian Journal of Cardiology, 2019, 35, 1604.e9-1604.e12.	0.8	10
137	Neuron-specific analysis of histone modifications with post-mortem brains. Scientific Reports, 2020, 10, 3767.	1.6	10
138	Walkerâ€“Warburg syndrome is genetically distinct from Fukuyama type congenital muscular dystrophy. Journal of the Neurological Sciences, 2000, 177, 150-153.	0.3	9
139	Unique Tauopathy in Fukuyama-Type Congenital Muscular Dystrophy. Journal of Neuropathology and Experimental Neurology, 2005, 64, 1118-1126.	0.9	9
140	Characterization of dystroglycan binding in adhesion of human induced pluripotent stem cells to laminin-511 E8 fragment. Scientific Reports, 2019, 9, 13037.	1.6	9
141	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
142	Genome-wide association study identifies zonisamide responsive gene in Parkinsonâ€™s disease patients. Journal of Human Genetics, 2020, 65, 693-704.	1.1	9
143	Bardetâ€“Biedl syndrome and related disorders in Japan. Journal of Human Genetics, 2020, 65, 847-853.	1.1	9
144	Muscle Transcriptomics Shows Overexpression of Cadherin 1 in Inclusion Body Myositis. Annals of Neurology, 2022, 91, 317-328.	2.8	9

#	Article	IF	Citations
145	CDP-ribitol prodrug treatment ameliorates ISPD-deficient muscular dystrophy mouse model. Nature Communications, 2022, 13, 1847.	5.8	9
146	Cell endogenous activities of fukutin and FKRP coexist with the ribitol xylosyltransferase, TMEM5. Biochemical and Biophysical Research Communications, 2018, 497, 1025-1030.	1.0	8
147	Cranial Nerve Involvement and Dysautonomia in Post-COVID-19 Guillain-BarrÃ© Syndrome. Internal Medicine, 2021, 60, 3477-3480.	0.3	8
148	Transcription Factor c-Maf Promotes Immunoregulation of Programmed Cell Death 1â€“Expressed CD8⁺ T Cells in Multiple Sclerosis. Neurology: Neuroimmunology and NeuroInflammation, 2022, 9, e1166.	3.1	8
149	Dinucleotide repeat polymorphism on chromosome 9q32. Japanese Journal of Human Genetics, 1995, 40, 333-334.	0.8	7
150	A variant of congenital muscular dystrophy. Brain and Development, 2002, 24, 24-29.	0.6	7
151	Detection of the dystroglycanopathy protein, fukutin, using a new panel of site-specific monoclonal antibodies. Biochemical and Biophysical Research Communications, 2012, 424, 354-357.	1.0	7
152	Founder mutation causes classical Fukuyama congenital muscular dystrophy (FCMD) in Chinese patients. Brain and Development, 2015, 37, 880-886.	0.6	7
153	Methylation changes and aberrant expression of FGFR3 in Lewy body disease neurons. Brain Research, 2018, 1697, 59-66.	1.1	7
154	Genetic Variations and Neuropathologic Features of Patients with PRKN Mutations. Movement Disorders, 2021, 36, 1634-1643.	2.2	7
155	Isolated Paravermal Hyperintensities in Neuronal Intranuclear Inclusion Disease. Neurology, 2022, 98, 938-939.	1.5	7
156	Prenatal diagnosis of Fukuyama-type congenital muscular dystrophy by microsatellite analysis. Human Reproduction, 1998, 13, 320-323.	0.4	6
157	Novel FKRP mutations in a Japanese MDC1C sibship clinically diagnosed with Fukuyama congenital muscular dystrophy. Brain and Development, 2017, 39, 869-872.	0.6	6
158	Cardiac involvement in Fukuyama muscular dystrophy is less severe than in Duchenne muscular dystrophy. Brain and Development, 2017, 39, 861-868.	0.6	6
159	Fragility Index in Randomized Controlled Trials of Ischemic Stroke. Journal of Stroke and Cerebrovascular Diseases, 2019, 28, 1290-1294.	0.7	6
160	Attempt to Predict A/T/N-Based Alzheimerâ€™s Disease Cerebrospinal Fluid Biomarkers Using a Peripheral Blood DNA Methylation Clock. Journal of Alzheimer's Disease Reports, 2020, 4, 287-296.	1.2	6
161	Splice-site mutations in KIF5A in the Japanese case series of amyotrophic lateral sclerosis. Neurogenetics, 2021, 22, 11-17.	0.7	6
162	Association analysis of LRP8 SNP rs3820198 and rs5174 with Parkinsonâ€™s disease in Han Chinese population. Neurological Research, 2012, 34, 725-729.	0.6	5

#	Article	IF	Citations
163	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
164	Isolated seizure as initial presentation of GABAA receptor antibody-associated encephalitis. Journal of the Neurological Sciences, 2020, 410, 116666.	0.3	5
165	Safety of Memantine in Combination with Potentially Interactive Drugs in the Real World: A Pharmacovigilance Study Using the Japanese Adverse Drug Event Report (JADER) Database. Journal of Alzheimer's Disease, 2021, 82, 1333-1344.	1.2	5
166	Stress cardiomyopathy (Takotsubo syndrome) in patients who received adrenergic agonist drugs: A pharmacovigilance study using the Japanese Adverse Drug Event Report (JADER) database. Journal of Cardiology, 2021, 79, 36-41.	0.8	5
167	Restoration of the defect in radial glial fiber migration and cortical plate organization in a brain organoid model of Fukuyama muscular dystrophy. IScience, 2021, 24, 103140.	1.9	5
168	Reliability and validity of Japanese version of Unified Multiple System Atrophy Rating Scale. Neurology and Clinical Neuroscience, 2021, 9, 171-180.	0.2	5
169	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
170	Three-dimensional MR imaging of brain surface anomalies in fukuyama-type congenital muscular dystrophy. Muscle and Nerve, 1995, 18, 508-517.	1.0	4
171	Psychosocial Twin Cohort Studies in Japan: The Keio Twin Research Center (KoTReC). Twin Research and Human Genetics, 2019, 22, 591-596.	0.3	4
172	Pembrolizumab on pre-existing inclusion body myositis: a case report. BMC Rheumatology, 2020, 4, 48.	0.6	4
173	Efficacy and Cost-effectiveness of Promotion Methods to Recruit Participants to an Online Screening Registry for Alzheimer Disease Prevention Trials: Observational Study. Journal of Medical Internet Research, 2021, 23, e26284.	2.1	4
174	Testing of the therapeutic efficacy and safety of AMPA receptor RNA aptamers in an ALS mouse model. Life Science Alliance, 2022, 5, e202101193.	1.3	4
175	Multinodular and vacuolating neuronal tumor (MVNT): A presumably incidental and asymptomatic case in an intractable epilepsy patient. Clinical Neurophysiology Practice, 2019, 4, 164-167.	0.6	3
176	Familial dementia with Lewy bodies with VPS13C mutations. Parkinsonism and Related Disorders, 2020, 81, 31-33.	1.1	3
177	Phenotype and Genotype Study of Chinese POMT2-Related Î±-Dystroglycanopathy. Frontiers in Genetics, 2021, 12, 692479.	1.1	3
178	Anti-Ku antibody-positive myositis presenting as a wide range of axial myopathies and myocarditis: A case report and review of the literature. Modern Rheumatology Case Reports, 2022, 6, 64-68.	0.3	3
179	Contribution of Dysferlin Deficiency to Skeletal Muscle Pathology in Asymptomatic and Severe Dystroglycanopathy Models: Generation of a New Model for Fukuyama Congenital Muscular Dystrophy. PLoS ONE, 2014, 9, e106721.	1.1	3
180	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

#	Article	IF	Citations
181	Isolation and characterization of the mouse ortholog of the Fukuyama-type congenital muscular dystrophy gene. Genomics, 2002, 80, 482-6.	1.3	3
182	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.	0.3	2
183	Diagnostic Values of Venous Peak Lactate, Lactate-to-pyruvate Ratio, and Fold Increase in Lactate from Baseline in Aerobic Exercise Tests in Patients with Mitochondrial Diseases. Internal Medicine, 2022, 61, 1939-1946.	0.3	2
184	Isolated Abducens Nerve Palsy Caused by Anterior Inferior Cerebellar Artery Compression. Internal Medicine, 2022, 61, 2991-2992.	0.3	2
185	Efficacy of canakinumab on AA amyloidosis in late-onset NLRP3-associated autoinflammatory disease with an I574F somatic mosaic mutation. Clinical Rheumatology, 2022, 41, 2233-2237.	1.0	2
186	Assessment of the upper limb muscles in patients with Fukuyama muscular dystrophy: Noninvasive assessment using visual ultrasound muscle analysis and shear wave elastography. Neuromuscular Disorders, 2022, 32, 754-762.	0.3	2
187	A linkage study with DNA markers (D4S95, D4S115, and D4S111) in Japanese Huntington disease families. Japanese Journal of Human Genetics, 1993, 38, 193-201.	0.8	1
188	Validation of the Guy's Neurological Disability Scale as a screening tool for cognitive impairment in multiple sclerosis. Multiple Sclerosis and Related Disorders, 2019, 35, 272-275.	0.9	1
189	Estimating acceleration time point of respiratory decline in ALS patients: A novel metric. Journal of the Neurological Sciences, 2019, 403, 7-12.	0.3	1
190	Rituximab improves not only back stiffness but also â€œstiff eyesâ€•in stiff person syndrome: Implications for immune-mediated treatment. Journal of the Neurological Sciences, 2020, 408, 116506.	0.3	1
191	Gait improvement after levofloxacin administration in a progressive supranuclear palsy patient. Clinical Parkinsonism & Related Disorders, 2020, 3, 100080.	0.5	1
192	Delayed Brachial Plexus Palsy after Clavicular Fracture. Internal Medicine, 2021, 60, 2511-2512.	0.3	1
193	Î±-Dystroglycanopathy. , 2016, , 21-38.		1
194	Alanine transaminase is predominantly increased in the active phase of anti-HMGCR myopathy. Neuromuscular Disorders, 2021, , .	0.3	1
195	Novel mutations in B3GALNT2 gene causing Î±-dystroglycanopathy in Chinese patients. Chinese Medical Journal, 2021, 134, 1483-1485.	0.9	1
196	Rippling Muscle Disease with Irregular Toe Jerks and Anti-acetylcholine Receptor Antibodies: Remission after Extended Thymectomy. Internal Medicine, 2022, , .	0.3	1
197	Reply to: On the pathophysiology of takotsubo syndrome triggered by administered adrenergic agonists, noted in the JADER database. Journal of Cardiology, 2021, , .	0.8	1
198	Outcomes of gastrointestinal cancer surgeries in Parkinson's disease patients: A nationwide study. Parkinsonism and Related Disorders, 2022, 96, 45-49.	1.1	1

#	Article	IF	Citations
199	Comparative whole transcriptome analysis of Parkinsonâ€™s disease focusing on the efficacy of zonisamide. Journal of Neurology, Neurosurgery and Psychiatry, 2022, , jnnp-2021-328742.	0.9	1
200	A novel (CA)n polymorphism on 6p21.1-21.2. Japanese Journal of Human Genetics, 1996, 41, 423-425.	0.8	0
201	2. Molecular Mechanisms and Therapeutic Strategies for Muscular Dystrophies. The Journal of the Japanese Society of Internal Medicine, 2016, 105, 1578-1587.	0.0	0
202	Prominent Spasticity and Hyperreflexia of the Legs in a Nepalese Patient with Friedreich Ataxia. Internal Medicine, 2019, 58, 2865-2869.	0.3	0
203	Fukuyama Congenital Muscular Dystrophy and Related Diseases. , 2019, , 209-221.		0
204	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
205	Fukutin and Fukutin-Related Protein (FKRP). , 2014, , 1181-1190.		0
206	2. Molecular Mechanisms and Therapeutic Strategies for Muscular Dystrophies. The Journal of the Japanese Society of Internal Medicine, 2016, 105, 77a-83a.	0.0	0
207	Isolated Body Lateropulsion in Supplementary Motor Area Infarction. Internal Medicine, 2020, 59, 3113-3114.	0.3	0
208	Autocorrelation-based method to identify disordered rhythm in Parkinsonâ€™s disease tasks: A novel approach applicable to multimodal devices. PLoS ONE, 2020, 15, e0238486.	1.1	0
209	Title is missing!. , 2020, 16, e1008826.		0
210	Title is missing!. , 2020, 16, e1008826.		0
211	Title is missing!. , 2020, 16, e1008826.		0
212	Title is missing!. , 2020, 16, e1008826.		0
213	Title is missing!. , 2020, 16, e1008826.		0
214	Title is missing!. , 2020, 16, e1008826.		0
215	Title is missing!. , 2019, 14, e0223549.		0
216	Title is missing!. , 2019, 14, e0223549.		0

#	Article	IF	Citations
217	Title is missing!. , 2019, 14, e0223549.		0
218	Title is missing!. , 2019, 14, e0223549.		0
219	Clinical and electrophysiological findings of facial palsy in a case of hereditary gelsolin amyloidosis. Auris Nasus Larynx, 2022, , .	0.5	0