Tatsushi Toda

List of Publications by Year in Descending Order

208 papers	9,137 citations	43 h-index	93 g-index
230 ext. papers	10,535 ext. citations	6.1 avg, IF	5.39 L-index

#	Paper	IF	Citations
208	Outcomes of gastrointestinal cancer surgeries in Parkinson's disease patients: A nationwide study.. *Parkinsonism and Related Disorders*, **2022**, 96, 45-49	3.6	
207	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*, **2022**, 79, 36-41	3	3
206	Efficacy of canakinumab on AA amyloidosis in late-onset NLRP3-associated autoinflammatory disease with an I574F somatic mosaic mutation.. *Clinical Rheumatology*, **2022**, 1	3.9	
205	CDP-ribitol prodrug treatment ameliorates ISPD-deficient muscular dystrophy mouse model.. *Nature Communications*, **2022**, 13, 1847	17.4	1
204	Clinical and electrophysiological findings of facial palsy in a case of hereditary gelsolin amyloidosis.. *Auris Nasus Larynx*, **2022**,	2.2	
203	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
202	Reply to: On the pathophysiology of takotsubo syndrome triggered by administered adrenergic agonists, noted in the JADER database. *Journal of Cardiology*, **2021**,	3	1
201	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	7.6	1
200	Juvenile amyotrophic lateral sclerosis with complex phenotypes associated with novel mutations. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2021**, 22, 576-578	3.6	4
199	A deep learning method for HLA imputation and trans-ethnic MHC fine-mapping of type 1 diabetes. *Nature Communications*, **2021**, 12, 1639	17.4	15
198	Biallelic variants in LIG3 cause a novel mitochondrial neurogastrointestinal encephalomyopathy. *Brain*, **2021**, 144, 1451-1466	11.2	8
197	Trans-Ethnic Fine-Mapping of the Major Histocompatibility Complex Region Linked to Parkinson's Disease. *Movement Disorders*, **2021**, 36, 1805-1814	7	2
196	Randomized phase 2 study of perampanel for sporadic amyotrophic lateral sclerosis. *Journal of Neurology*, **2021**, 1	5.5	5
195	A Case of Irreversible Corneal Edema Associated with Dentatorubropallidoluysian Atrophy Following Corneal Endothelial Transplantation. *SN Comprehensive Clinical Medicine*, **2021**, 3, 2029-2032	2.7	
194	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	4.7	6
193	Genetic variations and clinical spectrum of dystroglycanopathy in a large cohort of Chinese patients. *Clinical Genetics*, **2021**, 99, 384-395	4	6
192	Splice-site mutations in KIF5A in the Japanese case series of amyotrophic lateral sclerosis. *Neurogenetics*, **2021**, 22, 11-17	3	1

191	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
190	Cranial Nerve Involvement and Dysautonomia in Post-COVID-19 Guillain-Barr Syndrome. *Internal Medicine*, **2021**, 60, 3477-3480	1.1	4
189	Delayed Brachial Plexus Palsy after Clavicular Fracture. *Internal Medicine*, **2021**, 60, 2511-2512	1.1	
188	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 Alzheimerls Disease*, **2021**, 82, 1333-1344	4.3	1
187	Phenotype and Genotype Study of Chinese -Related Dystroglycanopathy. *Frontiers in Genetics*, **2021**, 12, 692479	4.5	1
186	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	6.1	1
185	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	10.5	9
184	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
183	Reliability and validity of Japanese version of Unified Multiple System Atrophy Rating Scale. *Neurology and Clinical Neuroscience*, **2021**, 9, 171-180	0.3	0
182	Predicting amyloid risk by machine learning algorithms based on the A4 screen data: Application to the Japanese Trial-Ready Cohort study. *Alzheimerls and Dementia: Translational Research and Clinical Interventions*, **2021**, 7, e12135	6	4
181	Genetic Variations and Neuropathologic Features of Patients with PRKN Mutations. *Movement Disorders*, **2021**, 36, 1634-1643	7	5
180	DMD exon 2 duplication due to a complex genomic rearrangement is associated with a somatic mosaicism.. *Neuromuscular Disorders*, **2021**,	2.9	0
179	Genome-wide association study identifies zonisamide responsive gene in Parkinson disease patients. *Journal of Human Genetics*, **2020**, 65, 693-704	4.3	5
178	Congenital hearing impairment associated with peripheral cochlear nerve dysmyelination in glycosylation-deficient muscular dystrophy. *PLoS Genetics*, **2020**, 16, e1008826	6	6
177	Arginine is a disease modifier for polyQ disease models that stabilizes polyQ protein conformation. *Brain*, **2020**, 143, 1811-1825	11.2	10
176	Variants in saposin D domain of prosaposin gene linked to Parkinson disease. *Brain*, **2020**, 143, 1190-1205	10.2	37
175	Neuron-specific analysis of histone modifications with post-mortem brains. *Scientific Reports*, **2020**, 10, 3767	4.9	3
174	Isolated seizure as initial presentation of GABA receptor antibody-associated encephalitis. *Journal of the Neurological Sciences*, **2020**, 410, 116666	3.2	3

173	Galectin 3-binding protein suppresses amyloid-β production by modulating β-cleavage of amyloid precursor protein. *Journal of Biological Chemistry*, **2020**, 295, 3678-3691	5.4	18
172	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	9.9	29
171	Autocorrelation-based method to identify disordered rhythm in Parkinson's disease tasks: A novel approach applicable to multimodal devices. *PLoS ONE*, **2020**, 15, e0238486	3.7	
170	Novel mutations in B3GALNT2 gene causing dystroglycanopathy in Chinese patients. *Chinese Medical Journal*, **2020**, 134, 1483-1485	2.9	0
169	Isolated Body Lateropulsion in Supplementary Motor Area Infarction. *Internal Medicine*, **2020**, 59, 3113-3114	1.1	
168	Bardet-Biedl syndrome and related disorders in Japan. *Journal of Human Genetics*, **2020**, 65, 847-853	4.3	3
167	Crystal structures of fukutin-related protein (FKRP), a ribitol-phosphate transferase related to muscular dystrophy. *Nature Communications*, **2020**, 11, 303	17.4	11
166	Rituximab improves not only back stiffness but also "stiff eyes" in stiff person syndrome: Implications for immune-mediated treatment. *Journal of the Neurological Sciences*, **2020**, 408, 116506	3.2	1
165	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
164	Familial dementia with Lewy bodies with VPS13C mutations. *Parkinsonism and Related Disorders*, **2020**, 81, 31-33	3.6	3
163	A Japanese family with primary familial brain calcification presenting with paroxysmal kinesigenic dyskinesia - A comprehensive mutational analysis. *Journal of the Neurological Sciences*, **2020**, 418, 117091	3.2	2
162	Gait improvement after levofloxacin administration in a progressive supranuclear palsy patient. *Clinical Parkinsonism & Related Disorders*, **2020**, 3, 100080	0.9	0
161	Clinical efficacy of haematopoietic stem cell transplantation for adult adrenoleukodystrophy. *Brain Communications*, **2020**, 2, fcz048	4.5	8
160	Attempt to Predict A/T/N-Based Alzheimer's Disease Cerebrospinal Fluid Biomarkers Using a Peripheral Blood DNA Methylation Clock. *Journal of Alzheimer's Disease Reports*, **2020**, 4, 287-296	3.3	0
159	Pembrolizumab on pre-existing inclusion body myositis: a case report. *BMC Rheumatology*, **2020**, 4, 48	2.9	1
158	Clinical Characteristics of Neuronal Intranuclear Inclusion Disease-Related Retinopathy With CGG Repeat Expansions in the NOTCH2NLC Gene **2020**, 61, 27		7
157	Identification of Risk Loci for Parkinson Disease in Asians and Comparison of Risk Between Asians and Europeans: A Genome-Wide Association Study. *JAMA Neurology*, **2020**, 77, 746-754	17.2	84
156	Congenital hearing impairment associated with peripheral cochlear nerve dysmyelination in glycosylation-deficient muscular dystrophy **2020**, 16, e1008826		

155	Congenital hearing impairment associated with peripheral cochlear nerve dysmyelination in glycosylation-deficient muscular dystrophy **2020**, 16, e1008826		
154	Congenital hearing impairment associated with peripheral cochlear nerve dysmyelination in glycosylation-deficient muscular dystrophy **2020**, 16, e1008826		
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152	Congenital hearing impairment associated with peripheral cochlear nerve dysmyelination in glycosylation-deficient muscular dystrophy **2020**, 16, e1008826		
151	Congenital hearing impairment associated with peripheral cochlear nerve dysmyelination in glycosylation-deficient muscular dystrophy **2020**, 16, e1008826		
150	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	4.8	43
149	Genome-wide association studies identify polygenic effects for completed suicide in the Japanese population. *Neuropsychopharmacology*, **2019**, 44, 2119-2124	8.7	19
148	Characterization of dystroglycan binding in adhesion of human induced pluripotent stem cells to laminin-511 E8 fragment. *Scientific Reports*, **2019**, 9, 13037	4.9	6
147	Multinodular and vacuolating neuronal tumor (MVNT): A presumably incidental and asymptomatic case in an intractable epilepsy patient. *Clinical Neurophysiology Practice*, **2019**, 4, 164-167	3.8	3
146	Validation of the Guy□ Neurological Disability Scale as a screening tool for cognitive impairment in multiple sclerosis. *Multiple Sclerosis and Related Disorders*, **2019**, 35, 272-275	4	
145	Estimating acceleration time point of respiratory decline in ALS patients: A novel metric. *Journal of the Neurological Sciences*, **2019**, 403, 7-12	3.2	0
144	Chronic cerebral hypoperfusion shifts the equilibrium of amyloid □oligomers to aggregation-prone species with higher molecular weight. *Scientific Reports*, **2019**, 9, 2827	4.9	21
143	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
142	Fukuyama Congenital Muscular Dystrophy and Related Diseases **2019**, 209-221		
141	Lower Serum Calcium as a Potentially Associated Factor for Conversion of Mild Cognitive Impairment to Early Alzheimer□ Disease in the Japanese Alzheimer□ Disease Neuroimaging Initiative. *Journal of Alzheimerls Disease*, **2019**, 68, 777-788	4.3	11
140	Wide distribution of alpha-synuclein oligomers in multiple system atrophy brain detected by proximity ligation. *Acta Neuropathologica*, **2019**, 137, 455-466	14.3	30
139	Fragility Index in Randomized Controlled Trials of Ischemic Stroke. *Journal of Stroke and Cerebrovascular Diseases*, **2019**, 28, 1290-1294	2.8	2
138	Colocalization of BRCA1 with Tau Aggregates in Human Tauopathies. *Brain Sciences*, **2019**, 10,	3.4	11

137	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	5.3	6
136	Prominent Spasticity and Hyperreflexia of the Legs in a Nepalese Patient with Friedreich Ataxia. *Internal Medicine*, **2019**, 58, 2865-2869	1.1	
135	Noncoding CGG repeat expansions in neuronal intranuclear inclusion disease, oculopharyngodistal myopathy and an overlapping disease. *Nature Genetics*, **2019**, 51, 1222-1232	36.3	132
134	Variable Cardiac Responses to Immunosuppressive Therapy in Anti-Mitochondrial Antibody-Positive Myositis. *Canadian Journal of Cardiology*, **2019**, 35, 1604.e9-1604.e12	3.8	6
133	Quantifying normal and parkinsonian gait features from home movies: Practical application of a deep learning-based 2D pose estimator. *PLoS ONE*, **2019**, 14, e0223549	3.7	23
132	Psychosocial Twin Cohort Studies in Japan: The Keio Twin Research Center (KoTReC). *Twin Research and Human Genetics*, **2019**, 22, 591-596	2.2	4
131	Elimination of fukutin reveals cellular and molecular pathomechanisms in muscular dystrophy-associated heart failure. *Nature Communications*, **2019**, 10, 5754	17.4	14
130	Subtype-Dependent Reporting of Stroke With SGLT2 Inhibitors: Implications From a Japanese Pharmacovigilance Study. *Journal of Clinical Pharmacology*, **2019**, 60, 629	2.9	7
129	Inflammatory myopathy with myasthenia gravis: Thymoma association and polymyositis pathology. *Neurology: Neuroimmunology and NeuroInflammation*, **2019**, 6, e535	9.1	17
128	Effect of istradefylline on mood disorders in Parkinson's disease. *Journal of the Neurological Sciences*, **2019**, 396, 78-83	3.2	21
127	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
126	Altered regulation of serum lysosomal acid hydrolase activities in Parkinson's disease: A potential peripheral biomarker?. *Parkinsonism and Related Disorders*, **2019**, 61, 132-137	3.6	11
125	Aphasic status epilepticus preceding tumefactive left hemisphere lesion in anti-MOG antibody associated disease. *Multiple Sclerosis and Related Disorders*, **2019**, 27, 91-94	4	15
124	Quantifying normal and parkinsonian gait features from home movies: Practical application of a deep learningBased 2D pose estimator **2019**, 14, e0223549		
123	Quantifying normal and parkinsonian gait features from home movies: Practical application of a deep learningBased 2D pose estimator **2019**, 14, e0223549		
122	Quantifying normal and parkinsonian gait features from home movies: Practical application of a deep learningBased 2D pose estimator **2019**, 14, e0223549		
121	Quantifying normal and parkinsonian gait features from home movies: Practical application of a deep learningBased 2D pose estimator **2019**, 14, e0223549		
120	Ribitol-phosphate-a newly identified posttranslational glycosylation unit in mammals: structure, modification enzymes and relationship to human diseases. *Journal of Biochemistry*, **2018**, 163, 359-369	3.1	12

119	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	5.6	13
118	Cell endogenous activities of fukutin and FKRP coexist with the ribitol xylosyltransferase, TMEM5. *Biochemical and Biophysical Research Communications*, **2018**, 497, 1025-1030	3.4	6
117	Paraneoplastic NMOSD associated with EG junction adenocarcinoma expressing unprotected AQP4. *Neurology: Neuroimmunology and NeuroInflammation*, **2018**, 5, e482	9.1	10
116	Modulation of motor learning by a paired associative stimulation protocol inducing LTD-like effects. *Brain Stimulation*, **2018**, 11, 1314-1321	5.1	4
115	National registry of patients with Fukuyama congenital muscular dystrophy in Japan. *Neuromuscular Disorders*, **2018**, 28, 885-893	2.9	7
114	In silico drug screening by using genome-wide association study data repurposed dabrafenib, an anti-melanoma drug, for Parkinson□ disease. *Human Molecular Genetics*, **2018**, 27, 3974-3985	5.6	16
113	Cell surface flip-flop of phosphatidylserine is critical for PIEZO1-mediated myotube formation. *Nature Communications*, **2018**, 9, 2049	17.4	74
112	CDP-glycerol inhibits the synthesis of the functional -mannosyl glycan of □dystroglycan. *Journal of Biological Chemistry*, **2018**, 293, 12186-12198	5.4	11
111	Methylation changes and aberrant expression of FGFR3 in Lewy body disease neurons. *Brain Research*, **2018**, 1697, 59-66	3.7	4
110	Novel FKRP mutations in a Japanese MDC1C sibship clinically diagnosed with Fukuyama congenital muscular dystrophy. *Brain and Development*, **2017**, 39, 869-872	2.2	3
109	Muscular Dystrophy with Ribitol-Phosphate Deficiency: A Novel Post-Translational Mechanism in Dystroglycanopathy. *Journal of Neuromuscular Diseases*, **2017**, 4, 259-267	5	19
108	Cardiac involvement in Fukuyama muscular dystrophy is less severe than in Duchenne muscular dystrophy. *Brain and Development*, **2017**, 39, 861-868	2.2	2
107	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	4.3	4
106	Carbohydrate-binding domain of the POMGnT1 stem region modulates O-mannosylation sites of □dystroglycan. *Proceedings of the National Academy of Sciences of the United States of America*, **2016**, 113, 9280-5	11.5	38
105	Mechanistic aspects of the formation of □dystroglycan and therapeutic research for the treatment of □dystroglycanopathy: A review. *Molecular Aspects of Medicine*, **2016**, 51, 115-24	16.7	45
104	Identification of a Post-translational Modification with Ribitol-Phosphate and Its Defect in Muscular Dystrophy. *Cell Reports*, **2016**, 14, 2209-2223	10.6	139
103	The Muscular Dystrophy Gene TMEM5 Encodes a Ribitol □1,4-Xylosyltransferase Required for the Functional Glycosylation of Dystroglycan. *Journal of Biological Chemistry*, **2016**, 291, 24618-24627	5.4	44
102	□Dystroglycanopathy **2016**, 21-38		1

101	2. Molecular Mechanisms and Therapeutic Strategies for Muscular Dystrophies. *The Journal of the Japanese Society of Internal Medicine*, **2016**, 105, 77a-83a	0	
100	2. Molecular Mechanisms and Therapeutic Strategies for Muscular Dystrophies. *The Journal of the Japanese Society of Internal Medicine*, **2016**, 105, 1578-1587	0	
99	Variants associated with Gaucher disease in multiple system atrophy. *Annals of Clinical and Translational Neurology*, **2015**, 2, 417-26	5.3	76
98	Founder mutation causes classical Fukuyama congenital muscular dystrophy (FCMD) in Chinese patients. *Brain and Development*, **2015**, 37, 880-6	2.2	4
97	ABCG2 variant has opposing effects on onset ages of Parkinson disease and gout. *Annals of Clinical and Translational Neurology*, **2015**, 2, 302-6	5.3	24
96	Fukutin is prerequisite to ameliorate muscular dystrophic phenotype by myofiber-selective LARGE expression. *Scientific Reports*, **2015**, 5, 8316	4.9	20
95	CHCHD2 mutations in autosomal dominant late-onset Parkinson disease: a genome-wide linkage and sequencing study. *Lancet Neurology, The*, **2015**, 14, 274-82	24.1	208
94	TRPV2 is critical for the maintenance of cardiac structure and function in mice. *Nature Communications*, **2014**, 5, 3932	17.4	84
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, 4543-58	5.6	20
92	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	3.7	2
91	Fukutin and Fukutin-Related Protein (FKRP) **2014**, 1181-1190		
90	Novel POMGnT1 mutations cause muscle-eye-brain disease in Chinese patients. *Molecular Genetics and Genomics*, **2013**, 288, 297-308	3.1	9
89	YY1 binds to synuclein 3 flanking region SNP and stimulates antisense noncoding RNA expression. *Journal of Human Genetics*, **2013**, 58, 711-9	4.3	26
88	Modeling Alzheimer disease with iPSCs reveals stress phenotypes associated with intracellular A and differential drug responsiveness. *Cell Stem Cell*, **2013**, 12, 487-96	18	539
87	Inhibition of protein misfolding/aggregation using polyglutamine binding peptide QBP1 as a therapy for the polyglutamine diseases. *Neurotherapeutics*, **2013**, 10, 440-6	6.4	24
86	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, 3003-15	5.6	29
85	AGO61-dependent GlcNAc modification primes the formation of functional glycans on dystroglycan. *Scientific Reports*, **2013**, 3, 3288	4.9	27
84	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, 721-6	5.8	78

83	Human natural killer-1 sulfotransferase (HNK-1ST)-induced sulfate transfer regulates laminin-binding glycans on □dystroglycan. *Journal of Biological Chemistry*, **2012**, 287, 30823-32	5.4	25
82	Large-scale replication and heterogeneity in Parkinson disease genetic loci. *Neurology*, **2012**, 79, 659-67	6.5	106
81	Detection of the dystroglycanopathy protein, fukutin, using a new panel of site-specific monoclonal antibodies. *Biochemical and Biophysical Research Communications*, **2012**, 424, 354-7	3.4	6
80	Hsp40 gene therapy exerts therapeutic effects on polyglutamine disease mice via a non-cell autonomous mechanism. *PLoS ONE*, **2012**, 7, e51069	3.7	29
79	Comprehensive research synopsis and systematic meta-analyses in Parkinson□ disease genetics: The PDGene database. *PLoS Genetics*, **2012**, 8, e1002548	6	420
78	Mislocalization of fukutin protein by disease-causing missense mutations can be rescued with treatments directed at folding amelioration. *Journal of Biological Chemistry*, **2012**, 287, 8398-406	5.4	21
77	Absence of post-phosphoryl modification in dystroglycanopathy mouse models and wild-type tissues expressing non-laminin binding form of □dystroglycan. *Journal of Biological Chemistry*, **2012**, 287, 9560-7	5.4	27
76	Association analysis of LRP8 SNP rs3820198 and rs5174 with Parkinson□ disease in Han Chinese population. *Neurological Research*, **2012**, 34, 725-9	2.7	4
75	The Aggregation Inhibitor Peptide QBP1 as a Therapeutic Molecule for the Polyglutamine Neurodegenerative Diseases. *Journal of Amino Acids*, **2011**, 2011, 265084		16
74	Pathogenic exon-trapping by SVA retrotransposon and rescue in Fukuyama muscular dystrophy. *Nature*, **2011**, 478, 127-31	50.4	127
73	Post-translational maturation of dystroglycan is necessary for pikachurin binding and ribbon synaptic localization. *Journal of Biological Chemistry*, **2010**, 285, 31208-16	5.4	37
72	Residual laminin-binding activity and enhanced dystroglycan glycosylation by LARGE in novel model mice to dystroglycanopathy. *Human Molecular Genetics*, **2009**, 18, 621-31	5.6	64
71	Mutations for Gaucher disease confer high susceptibility to Parkinson disease. *Archives of Neurology*, **2009**, 66, 571-6		143
70	Fukutin gene retrotransposal insertion in a non-Japanese Fukuyama congenital muscular dystrophy (FCMD) patient. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 2403-8	2.5	13
69	Genome-wide association study identifies common variants at four loci as genetic risk factors for Parkinson□ disease. *Nature Genetics*, **2009**, 41, 1303-7	36.3	1045
68	Structure-activity relationship study on polyglutamine binding peptide QBP1. *Bioorganic and Medicinal Chemistry*, **2009**, 17, 1259-63	3.4	18
67	Spinocerebellar ataxia type 31 is associated with "inserted" penta-nucleotide repeats containing (TGGAA)n. *American Journal of Human Genetics*, **2009**, 85, 544-57	11	204
66	Surface plasmon resonance characterization of specific binding of polyglutamine aggregation inhibitors to the expanded polyglutamine stretch. *Biochemical and Biophysical Research Communications*, **2009**, 378, 634-9	3.4	10

65	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	3.3	34
64	Pikachurin, a dystroglycan ligand, is essential for photoreceptor ribbon synapse formation. *Nature Neuroscience*, **2008**, 11, 923-31	25.5	211
63	Seizure-genotype relationship in Fukuyama-type congenital muscular dystrophy. *Brain and Development*, **2008**, 30, 59-67	2.2	15
62	A toxic monomeric conformer of the polyglutamine protein. *Nature Structural and Molecular Biology*, **2007**, 14, 332-40	17.6	258
61	Defective peripheral nerve myelination and neuromuscular junction formation in fukutin-deficient chimeric mice. *Journal of Neurochemistry*, **2007**, 101, 1712-22	6	31
60	Detection of polyglutamine protein oligomers in cells by fluorescence correlation spectroscopy. *Journal of Biological Chemistry*, **2007**, 282, 24039-48	5.4	80
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, 19034-9	11.5	34
58	Multiple candidate gene analysis identifies alpha-synuclein as a susceptibility gene for sporadic Parkinson disease. *Human Molecular Genetics*, **2006**, 15, 1151-8	5.6	185
57	Aberrant neuromuscular junctions and delayed terminal muscle fiber maturation in alpha-dystroglycanopathies. *Human Molecular Genetics*, **2006**, 15, 1279-89	5.6	36
56	Expression profiling of muscles from Fukuyama-type congenital muscular dystrophy and laminin-alpha 2 deficient congenital muscular dystrophy; is congenital muscular dystrophy a primary fibrotic disease?. *Biochemical and Biophysical Research Communications*, **2006**, 342, 489-502	3.4	26
55	Molecular interaction between fukutin and POMGnT1 in the glycosylation pathway of alpha-dystroglycan. *Biochemical and Biophysical Research Communications*, **2006**, 350, 935-41	3.4	62
54	The genetic and molecular basis of muscular dystrophy: roles of cell-matrix linkage in the pathogenesis. *Journal of Human Genetics*, **2006**, 51, 915-926	4.3	100
53	An autosomal dominant cerebellar ataxia linked to chromosome 16q22.1 is associated with a single-nucleotide substitution in the 5untranslated region of the gene encoding a protein with spectrin repeat and Rho guanine-nucleotide exchange-factor domains. *American Journal of Human Genetics*, **2005**, 77, 280-96	11	109
52	Basement membrane fragility underlies embryonic lethality in fukutin-null mice. *Neurobiology of Disease*, **2005**, 19, 208-17	7.5	59
51	beta4GalT-II is a key regulator of glycosylation of the proteins involved in neuronal development. *Biochemical and Biophysical Research Communications*, **2005**, 333, 131-7	3.4	23
50	Effects of fukutin deficiency in the developing mouse brain. *Neuromuscular Disorders*, **2005**, 15, 416-26	2.9	39
49	Unique tauopathy in Fukuyama-type congenital muscular dystrophy. *Journal of Neuropathology and Experimental Neurology*, **2005**, 64, 1118-26	3.1	8
48	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**, 138, 344-8	2.5	31

47	Genomic rearrangement at 10q24 in non-syndromic split-hand/split-foot malformation. *Human Genetics*, **2005**, 118, 477-83	6.3	19
46	Antenatal and postnatal brain magnetic resonance imaging in muscle-eye-brain disease. *Archives of Neurology*, **2004**, 61, 1301-6		22
45	UCHL1 is a Parkinson's disease susceptibility gene. *Annals of Neurology*, **2004**, 55, 512-21	9.4	190
44	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, 1200-6	3.4	26
43	Structure-function analysis of human protein O-linked mannose beta1,2-N-acetylglucosaminyltransferase 1, POMGnT1. *Biochemical and Biophysical Research Communications*, **2004**, 320, 39-44	3.4	33
42	Worldwide distribution and broader clinical spectrum of muscle-eye-brain disease. *Human Molecular Genetics*, **2003**, 12, 527-34	5.6	100
41	Glycosylation in congenital muscular dystrophies. *Biological and Pharmaceutical Bulletin*, **2003**, 26, 1641-7	2.3	58
40	Toward identification of susceptibility genes for sporadic Parkinson's disease. *Journal of Neurology*, **2003**, 250 Suppl 3, III40-3	5.5	26
39	A new mutation of the fukutin gene in a non-Japanese patient. *Annals of Neurology*, **2003**, 53, 392-6	9.4	86
38	Fukuyama-type congenital muscular dystrophy (FCMD) and alpha-dystroglycanopathy. *Congenital Anomalies (discontinued)*, **2003**, 43, 97-104	1.1	42
37	Enzymatic diagnostic test for Muscle-Eye-Brain type congenital muscular dystrophy using commercially available reagents. *Clinical Biochemistry*, **2003**, 36, 339-44	3.5	48
36	Loss-of-function of an N-acetylglucosaminyltransferase, POMGnT1, in muscle-eye-brain disease. *Biochemical and Biophysical Research Communications*, **2003**, 306, 93-7	3.4	90
35	Fukutin is required for maintenance of muscle integrity, cortical histiogenesis and normal eye development. *Human Molecular Genetics*, **2003**, 12, 1449-59	5.6	98
34	Prevention of polyglutamine oligomerization and neurodegeneration by the peptide inhibitor QBP1 in Drosophila. *Human Molecular Genetics*, **2003**, 12, 1253-9	5.6	106
33	Sister and brother with Vici syndrome: agenesis of the corpus callosum, albinism, and recurrent infections. *American Journal of Medical Genetics Part A*, **2002**, 109, 61-6		40
32	Association studies of multiple candidate genes for Parkinson's disease using single nucleotide polymorphisms. *Annals of Neurology*, **2002**, 51, 133-6	9.4	185
31	Isolation and Characterization of the Mouse Ortholog of the Fukuyama-type Congenital Muscular Dystrophy Gene. *Genomics*, **2002**, 80, 482-486	4.3	11
30	Deficiency of alpha-dystroglycan in muscle-eye-brain disease. *Biochemical and Biophysical Research Communications*, **2002**, 291, 1283-6	3.4	105

29	A variant of congenital muscular dystrophy. *Brain and Development*, **2002**, 24, 24-9	2.2	5
28	Isolation and characterization of the mouse ortholog of the Fukuyama-type congenital muscular dystrophy gene. *Genomics*, **2002**, 80, 482-6	4.3	3
27	Muscular dystrophy and neuronal migration disorder caused by mutations in a glycosyltransferase, POMGnT1. *Developmental Cell*, **2001**, 1, 717-24	10.2	590
26	Structural organization, complete genomic sequences and mutational analyses of the Fukuyama-type congenital muscular dystrophy gene, fukutin. *FEBS Letters*, **2001**, 489, 192-6	3.8	34
25	Haplotype-phenotype correlation in Fukuyama congenital muscular dystrophy. *American Journal of Medical Genetics Part A*, **2000**, 92, 184-90		29
24	Age and origin of the FCMD 3Ǫuntranslated-region retrotransposal insertion mutation causing Fukuyama-type congenital muscular dystrophy in the Japanese population. *Human Genetics*, **2000**, 107, 559-67	6.3	26
23	A case of Walker-Warburg syndrome. *Brain and Development*, **2000**, 22, 454-7	2.2	7
22	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, 541-7	2.9	26
21	Genetic heterogeneity in three Chinese children with Fukuyama congenital muscular dystrophy. *Neuromuscular Disorders*, **2000**, 10, 108-12	2.9	9
20	The Fukuyama congenital muscular dystrophy story. *Neuromuscular Disorders*, **2000**, 10, 153-9	2.9	67
19	Walker-Warburg syndrome is genetically distinct from Fukuyama type congenital muscular dystrophy. *Journal of the Neurological Sciences*, **2000**, 177, 150-3	3.2	9
18	Broader clinical spectrum of Fukuyama-type congenital muscular dystrophy manifested by haplotype analysis. *Journal of Child Neurology*, **1999**, 14, 711-5	2.5	10
17	Fukuyama-type congenital muscular dystrophy: the first human disease to be caused by an ancient retrotransposal integration. *Journal of Molecular Medicine*, **1999**, 77, 816-23	5.5	14
16	Genetic variations on the Y chromosome in the Japanese population and implications for modern human Y chromosome lineage. *Journal of Human Genetics*, **1999**, 44, 240-5	4.3	47
15	Spermatogenic ability is different among males in different Y chromosome lineage. *Journal of Human Genetics*, **1999**, 44, 289-92	4.3	83
14	An ancient retrotransposal insertion causes Fukuyama-type congenital muscular dystrophy. *Nature*, **1998**, 394, 388-92	50.4	665
13	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		10
12	Prenatal diagnosis of Fukuyama-type congenital muscular dystrophy by microsatellite analysis. *Human Reproduction*, **1998**, 13, 320-3	5.7	3

11	YAC and cosmid contigs encompassing the Fukuyama-type congenital muscular dystrophy (FCMD) candidate region on 9q31. *Genomics*, **1997**, 40, 284-93	4.3	16
10	Molecular genetic evidence of clinical heterogeneity in Fukuyama-type congenital muscular dystrophy. *Human Genetics*, **1997**, 99, 427-32	6.3	19
9	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 FCMD fetus. *Acta Neuropathologica*, **1996**, 91, 313-21	14.3	71
8	A novel (CA)n polymorphism on 6p21.1-21.2. *Japanese Journal of Human Genetics*, **1996**, 41, 423-5		
7	Fukuyama congenital muscular dystrophy: Cortical dysplasia of the cerebrum in a 20 week fetus. *Neuropathology*, **1996**, 16, 184-189	2	10
6	Prenatal diagnosis of Fukuyama type congenital muscular dystrophy by polymorphism analysis. *American Journal of Medical Genetics Part A*, **1996**, 66, 169-74		12
5	Dinucleotide repeat polymorphism on chromosome 9q32. *Japanese Journal of Human Genetics*, **1995**, 40, 333-4		6
4	Genetic identity of Fukuyama-type congenital muscular dystrophy and Walker-Warburg syndrome. *Annals of Neurology*, **1995**, 37, 99-101	9.4	49
3	Three-dimensional MR imaging of brain surface anomalies in Fukuyama-type congenital muscular dystrophy. *Muscle and Nerve*, **1995**, 18, 508-17	3.4	3
2	A linkage study with DNA markers (D4S95, D4S115, and D4S111) in Japanese Huntington disease families. *Japanese Journal of Human Genetics*, **1993**, 38, 193-201		1
1	Efficacy and Cost-effectiveness of Promotion Methods to Recruit Participants to an Online Screening Registry for Alzheimer Disease Prevention Trials: Observational Study (Preprint)		1