## Hideshi Kawakami

List of Publications by Year in descending order

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162 papers 10,164 citations

43 h-index 97 g-index

166 all docs

166
docs citations

166 times ranked 11606 citing authors

#	Article	IF	Citations
1	Knockdown of optineurin controls C2C12 myoblast differentiation via regulating myogenin and MyoD expressions. Differentiation, 2022, 123, 1-8.	1.9	2
2	Channelopathies and Cerebellar Disease. , 2022, , 1399-1413.		0
3	Kv11 ( <i>etherâ€Ãâ€goâ€go</i> â€related gene) voltageâ€dependent K <sup>+</sup> channels promote resonal and oscillation of subthreshold membrane potentials. Journal of Physiology, 2021, 599, 547-569.	nce 2.9	7
4	Optineurin defects cause TDP43-pathology with autophagic vacuolar formation. Neurobiology of Disease, 2021, 148, 105215.	4.4	15
5	An autopsy report of a familial amyotrophic lateral sclerosis case carrying VCP Arg487His mutation with a unique TDPâ€43 proteinopathy. Neuropathology, 2021, 41, 118-126.	1.2	11
6	Long-term MRI findings of adult-onset neuronal intranuclear inclusion disease. Clinical Neurology and Neurosurgery, 2021, 201, 106456.	1.4	4
7	Prediction Model of Amyotrophic Lateral Sclerosis by Deep Learning with Patient Induced Pluripotent Stem Cells. Annals of Neurology, 2021, 89, 1226-1233.	5 <b>.</b> 3	22
8	Analysis of genetic risk factors in Japanese patients with Parkinson's disease. Journal of Human Genetics, 2021, 66, 957-964.	2.3	4
9	Prospects and status of the dosimetry system for atomic bomb survivor cohort study conducted at Research Institute for Radiation Biology and Medicine of Hiroshima University. Journal of Radiation Research, 2021, 62, i107-i113.	1.6	2
10	Novel monoallelic variant in ERLIN2 causes spastic paraplegia converted to amyotrophic lateral sclerosis. Journal of the Neurological Sciences, 2021, 430, 119984.	0.6	2
11	FXTAS is difficult to differentiate from neuronal intranuclear inclusion disease through skin biopsy: a case report. BMC Neurology, 2021, 21, 396.	1.8	21
12	Retinitis pigmentosa prior to familial ALS caused by a homozygous cilia and flagella-associated protein 410 mutation. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 220-222.	1.9	6
13	Zonisamide can ameliorate the voltage-dependence alteration of the T-type calcium channel CaV3.1 caused by a mutation responsible for spinocerebellar ataxia. Molecular Brain, 2020, 13, 163.	2.6	3
14	Aggressive periodontitis and NOD2 variants. Journal of Human Genetics, 2020, 65, 841-846.	2.3	7
15	Genetic screening for potassium channel mutations in Japanese autosomal dominant spinocerebellar ataxia. Journal of Human Genetics, 2020, 65, 363-369.	2.3	8
16	Biallelic mutation of <i>HSD17B4</i> induces middle age–onset spinocerebellar ataxia. Neurology: Genetics, 2020, 6, e396.	1.9	6
17	Optineurin regulates osteoblastogenesis through STAT1. Biochemical and Biophysical Research Communications, 2020, 525, 889-894.	2.1	8
18	Middle-age-onset cerebellar ataxia caused by a homozygous TWNK variant: a case report. BMC Medical Genetics, 2020, 21, 68.	2.1	10

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19	The first Japanese case of primary familial brain calcification caused by an MYORG variant. Journal of Human Genetics, 2020, 65, 917-920.	2.3	5
20	Co-morbidity of progressive supranuclear palsy and amyotrophic lateral sclerosis: a clinical-pathological case report. BMC Neurology, 2019, 19, 168.	1.8	10
21	C-terminal mutations in SYNE1 are associated with motor neuron disease in patients with SCAR8. Journal of the Neurological Sciences, 2019, 402, 118-120.	0.6	4
22	Treatment of intractable resting tremor of spinocerebellar ataxia 42 with zonisamide. Journal of the Neurological Sciences, 2019, 396, 119-120.	0.6	3
23	Amyotrophic lateral sclerosis of long clinical course clinically presenting with progressive muscular atrophy. Neuropathology, 2019, 39, 47-53.	1.2	3
24	Novel compound heterozygous mutations in the <i><scp>PARK</scp>2</i> gene identified in a Chinese pedigree with earlyâ€onset Parkinson's disease. Brain and Behavior, 2018, 8, e00901.	2.2	7
25	Multiple Proteinopathies in Familial ALS Cases With Optineurin Mutations. Journal of Neuropathology and Experimental Neurology, 2018, 77, 128-138.	1.7	25
26	Compound heterozygote mutations in the <i>SIGMAR1</i> gene in an oldestâ€old patient with amyotrophic lateral sclerosis. Geriatrics and Gerontology International, 2018, 18, 1519-1520.	1.5	14
27	RELATIVE BIOLOGICAL EFFECTIVENESS OF NEUTRONS DERIVED FROM THE EXCESS RELATIVE RISK MODEL WITH THE ATOMIC BOMB SURVIVORS DATA MANAGED BY HIROSHIMA UNIVERSITY. Radiation Protection Dosimetry, 2018, 180, 346-350.	0.8	8
28	PLK1-mediated phosphorylation of WDR62/MCPH2 ensures proper mitotic spindle orientation. Human Molecular Genetics, 2017, 26, 4429-4440.	2.9	32
29	Second derivative of the finger photoplethysmogram and cardiovascular mortality in middle-aged and elderly Japanese women. Hypertension Research, 2017, 40, 207-211.	2.7	22
30	First report of a Japanese family with spinocerebellar ataxia type 10: The second report from Asia after a report from China. PLoS ONE, 2017, 12, e0177955.	2.5	17
31	Linear ubiquitination is involved in the pathogenesis of optineurin-associated amyotrophic lateral sclerosis. Nature Communications, 2016, 7, 12547.	12.8	109
32	Vulnerability of Purkinje Cells Generated from Spinocerebellar Ataxia Type 6 Patient-Derived iPSCs. Cell Reports, 2016, 17, 1482-1490.	6.4	91
33	A mutation in the low voltage-gated calcium channel CACNA1G alters the physiological properties of the channel, causing spinocerebellar ataxia. Molecular Brain, 2015, 8, 89.	2.6	80
34	Self-Organization of Polarized Cerebellar Tissue in 3D Culture of Human Pluripotent Stem Cells. Cell Reports, 2015, 10, 537-550.	6.4	531
35	Neurogenetics. Neurology, 2015, 84, 1070-1071.	1.1	1
36	An autopsy case of familial amyotrophic lateral sclerosis with <b><i>FUS</i></b> R521G mutation. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2014, 15, 305-308.	1.7	3

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37	Clinicopathologic features of autosomal recessive amyotrophic lateral sclerosis associated with optineurin mutation. Neuropathology, 2014, 34, 64-70.	1.2	37
38	Exome sequencing reveals a novel <i><scp>ANO10</scp></i> mutation in a Japanese patient with autosomal recessive spinocerebellar ataxia. Clinical Genetics, 2014, 85, 296-297.	2.0	22
39	Genomeâ€wide association study in musician's dystonia: A risk variant at the arylsulfatase G locus?. Movement Disorders, 2014, 29, 921-927.	3.9	53
40	Neuropathological features of <scp>J</scp> apanese familial amyotrophic lateral sclerosis with p. <scp>N</scp> 352 <scp>S</scp> mutation in <scp><i>TARDBP</i></scp> . Neuropathology and Applied Neurobiology, 2014, 40, 231-236.	3.2	9
41	DYT6 in Japanâ€"genetic screening and clinical characteristics of the patients. Movement Disorders, 2014, 29, 278-280.	3.9	2
42	Authors' reply to <scp>D</scp> rs <scp>M</scp> van <scp>B</scp> litterswijk, <scp>R R</scp> ademakers and <scp>LH</scp> van den <scp>B</scp> erg. Neuropathology and Applied Neurobiology, 2014, 40, 359-360.	3.2	0
43	Immunoreactivity of valosin-containing protein in sporadic amyotrophic lateral sclerosis and in a case of its novel mutant. Acta Neuropathologica Communications, 2014, 2, 172.	5.2	39
44	Exome sequencing reveals a novel TTC19 mutation in an autosomal recessive spinocerebellar ataxia patient. BMC Neurology, 2014, 14, 5.	1.8	30
45	Exome sequencing reveals a novel MRE11 mutation in a patient with progressive myoclonic ataxia. Journal of the Neurological Sciences, 2014, 337, 219-223.	0.6	16
46	Mutations in Twinkle primase-helicase cause Perrault syndrome with neurologic features. Neurology, 2014, 83, 2054-2061.	1.1	86
47	An autopsy case of sporadic amyotrophic lateral sclerosis associated with the <scp>Il13T</scp> â€ <i><scp>SOD1</scp></i> mutation. Neuropathology, 2014, 34, 58-63.	1.2	19
48	A <scp>J</scp> apanese patient with familial <scp>ALS</scp> and a p. <scp>K510M</scp> mutation in the gene for <scp>FUS</scp> ( <i><scp>FUS</scp></i> ) resulting in the totally lockedâ€in state. Neuropathology, 2014, 34, 504-509.	1.2	7
49	Detecting gene mutations in Japanese Alzheimer's patients by semiconductor sequencing. Neurobiology of Aging, 2014, 35, 1780.e1-1780.e5.	3.1	19
50	Autosomal recessive Andersen-Tawil syndrome with a novel mutation L94P in Kir2.1. Neurology and Clinical Neuroscience, 2013, 1, 131-137.	0.4	8
51	Cerebellar ataxia with <i>SYNE1</i> mutation accompanying motor neuron disease. Neurology, 2013, 80, 600-601.	1.1	55
52	Optineurin and amyotrophic lateral sclerosis. Geriatrics and Gerontology International, 2013, 13, 528-532.	1.5	43
53	Optineurin suppression causes neuronal cell death via ⟨scp⟩NF⟨/scp⟩â€PB pathway. Journal of Neurochemistry, 2013, 126, 699-704.	3.9	80
54	Oromandibular dystonia associated with SCA36. Movement Disorders, 2013, 28, 558-559.	3.9	15

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55	Convenient diagnosis of spinal and bulbar muscular atrophy using a microchip electrophoresis system. American Journal of Neurodegenerative Disease, 2013, 2, 35-9.	0.1	O
56	The clinical characteristics of spinocerebellar ataxia 36: A study of 2121 Japanese ataxia patients. Movement Disorders, 2012, 27, 1158-1163.	3.9	22
57	Investigation on circular asymmetry of geographical distribution in cancer mortality of Hiroshima atomic bomb survivors based on risk maps: analysis of spatial survival data. Radiation and Environmental Biophysics, 2012, 51, 133-141.	1.4	12
58	Optineurin with amyotrophic lateral sclerosis-related mutations abrogates inhibition of interferon regulatory factor-3 activation. Neuroscience Letters, 2011, 505, 279-281.	2.1	29
59	The UCHL1 S18Y polymorphism and Parkinson's disease in a Japanese population. Parkinsonism and Related Disorders, 2011, 17, 473-475.	2.2	5
60	Screening for OPTN mutations in amyotrophic lateral sclerosis in a mainly Caucasian population. Neurobiology of Aging, 2011, 32, 1923.e9-1923.e10.	3.1	20
61	Optineurin is co-localized with FUS in basophilic inclusions of ALS with FUS mutation and in basophilic inclusion body disease. Acta Neuropathologica, 2011, 121, 555-557.	7.7	53
62	Clinicopathologic study on an ALS family with a heterozygous E478G optineurin mutation. Acta Neuropathologica, 2011, 122, 223-229.	7.7	60
63	Severe brain atrophy after long-term survival seen in siblings with familial amyotrophic lateral sclerosis and a mutation in the optineurin gene: a case series. Journal of Medical Case Reports, 2011, 5, 573.	0.8	6
64	Homozygosity Mapping on Homozygosity Haplotype Analysis to Detect Recessive Disease-Causing Genes from a Small Number of Unrelated, Outbred Patients. PLoS ONE, 2011, 6, e25059.	2.5	9
65	A quantitatively-modeled homozygosity mapping algorithm, qHomozygosityMapping, utilizing whole genome single nucleotide polymorphism genotyping data. BMC Bioinformatics, 2010, 11, S5.	2.6	9
66	Mutations of optineurin in amyotrophic lateral sclerosis. Nature, 2010, 465, 223-226.	27.8	1,097
67	Autopsy Case of Later-Onset Pontocerebellar Hypoplasia Type 1: Pontine Atrophy and Pyramidal Tract Involvement. Journal of Child Neurology, 2010, 25, 1429-1434.	1.4	3
68	Analysis on the Susceptibility Genes in Two Chinese Pedigrees with Familial Parkinson's Disease. Neurology Research International, 2010, 2010, 1-4.	1.3	2
69	<i>LRRK2</i> mutations and risk variants in Japanese patients with Parkinson's disease. Movement Disorders, 2009, 24, 1034-1041.	3.9	60
70	Genome-wide association study identifies common variants at four loci as genetic risk factors for Parkinson's disease. Nature Genetics, 2009, 41, 1303-1307.	21.4	1,217
71	Screening for TARDBP mutations in Japanese familial amyotrophic lateral sclerosis. Journal of the Neurological Sciences, 2009, 284, 69-71.	0.6	45
72	Aortic Pulse Wave Velocity Predicts Cardiovascular Mortality in Middle-Aged and Elderly Japanese Men. Circulation Journal, 2009, 73, 549-553.	1.6	78

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73	Sporadic ataxias in Japan – a population-based epidemiological study. Cerebellum, 2008, 7, 189-197.	2.5	131
74	Pathogenic expansions of the SCA6 locus are associated with a common CACNA1A haplotype across the globe: founder effect or predisposing chromosome?. European Journal of Human Genetics, 2008, 16, 841-847.	2.8	8
75	The CNTN4 c.4256C>T mutation is rare in Japanese with inherited spinocerebellar ataxia. Journal of the Neurological Sciences, 2008, 266, 180-181.	0.6	8
76	A polymorphism of LOC387715 gene is associated with age-related macular degeneration in the Japanese population. Neuroscience Letters, 2007, 414, 71-74.	2.1	43
77	Voltage-gated potassium channel antibodies associated limbic encephalitis in a patient with invasive thymoma. Journal of the Neurological Sciences, 2006, 250, 167-169.	0.6	31
78	High frequency of open-angle glaucoma in Japanese patients with Alzheimer's disease. Journal of the Neurological Sciences, 2006, 246, 79-83.	0.6	216
79	Clinicopathologic investigation of a family with expanded SCA8 CTA/CTG repeats. Neurology, 2006, 67, 1479-1481.	1.1	34
80	NO ASSOCIATION OF COMPLEMENT FACTOR H GENE POLYMORPHISM AND AGE-RELATED MACULAR DEGENERATION IN THE JAPANESE POPULATION. Retina, 2006, 26, 985-987.	1.7	62
81	Collaborative Analysis of α-Synuclein Gene Promoter Variability and Parkinson Disease. JAMA - Journal of the American Medical Association, 2006, 296, 661.	7.4	467
82	Lack of replication of thirteen single-nucleotide polymorphisms implicated in Parkinson's disease: a large-scale international study. Lancet Neurology, The, 2006, 5, 917-923.	10.2	83
83	Identification of a new family of spinocerebellar ataxia type 14 in the japanese spinocerebellar ataxia population by the screening of PRKCG exon 4. Movement Disorders, 2006, 21, 1355-1360.	3.9	29
84	Identification and haplotype analysis of <i>LRRK2</i> G2019S in Japanese patients with Parkinson disease. Neurology, 2006, 67, 697-699.	1.1	54
85	Activation of human SII cortex during exploratory finger movement and hand clenching tasks. NeuroReport, 2005, 16, 145-148.	1.2	4
86	Glutathioneâ€∢i>Sâ€transferaseâ€1 and interleukinâ€1β gene polymorphisms in Japanese patients with Parkinson's disease. Movement Disorders, 2005, 20, 901-902.	3.9	39
87	Brainâ€derived neurotrophic factor gene polymorphisms in Japanese patients with sporadic Alzheimer's disease, Parkinson's disease, and multiple system atrophy. Movement Disorders, 2005, 20, 1031-1033.	3.9	39
88	Mutant Protein Kinase $\hat{Cl}^3$ Found in Spinocerebellar Ataxia Type 14 Is Susceptible to Aggregation and Causes Cell Death. Journal of Biological Chemistry, 2005, 280, 29096-29106.	3.4	64
89	Influence of a tumor necrosis factor gene polymorphism in Japanese patients with multiple system atrophy. Neuroscience Letters, 2005, 374, 218-221.	2.1	40
90	Possible Reduced Penetrance of Expansion of 44 to 47 CAG/CAA Repeats in the TATA-Binding Protein Gene in Spinocerebellar Ataxia Type 17. Archives of Neurology, 2004, 61, 209.	<b>4.</b> 5	81

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91	Dinucleotide repeat polymorphism in interferon-? gene is not associated with sporadic Alzheimer's disease. American Journal of Medical Genetics Part A, 2004, 124B, 48-49.	2.4	3
92	A novel haplotype of spinocerebellar ataxia type 6 contributes to the highest prevalence in Western Japan. Neuroscience Letters, 2004, 358, 107-110.	2.1	4
93	Influence of polymorphisms in the genes for cytokines and glutathione S-transferase omega on sporadic Alzheimer's disease. Neuroscience Letters, 2004, 368, 140-143.	2.1	51
94	Disinhibition of the somatosensory cortex in cervical dystonia—decreased amplitudes of high-frequency oscillations. Clinical Neurophysiology, 2004, 115, 1624-1630.	1.5	39
95	Quantitative Assessment of Cerebral Blood Flow in Genetically Confirmed Spinocerebellar Ataxia Type 6. Archives of Neurology, 2004, 61, 933.	4.5	29
96	Influence of monocyte chemoattractant protein 1 gene polymorphism on age at onset of sporadic Parkinson's disease. Movement Disorders, 2003, 18, 953-955.	3.9	37
97	Influence of cytokine and mannose binding protein gene polymorphisms on human t-cell leukemia virus type i (hTLV-i) provirus load in HTLV-I asymptomatic carriers. Human Immunology, 2003, 64, 453-457.	2.4	17
98	SCA8 Repeat Expansion: Large CTA/CTG Repeat Alleles Are More Common in Ataxic Patients, Including Those with SCA6. American Journal of Human Genetics, 2003, 72, 704-709.	6.2	65
99	Novel mutation in X-linked Charcot-Marie-Tooth disease associated with CNS impairment. Neurology, 2002, 59, 923-926.	1.1	34
100	Association between interleukin-6 gene polymorphism and human T-Cell leukemia virus type I associated myelopathy. Human Immunology, 2002, 63, 696-700.	2.4	25
101	Dinucleotide repeat polymorphisms in the Neprilysin gene are not associated with sporadic Alzheimer's disease. Neuroscience Letters, 2002, 320, 105-107.	2.1	32
102	Difference in disease-free survival curve and regional distribution according to subtype of spinocerebellar ataxia: A study of 1,286 Japanese patients. American Journal of Medical Genetics Part A, 2002, 114, 578-583.	2.4	97
103	Contribution of the interleukinâ€1β gene polymorphism in multiple system atrophy. Movement Disorders, 2002, 17, 808-811.	3.9	68
104	Dopamine Transporter and Parkinson's Disease. Advances in Behavioral Biology, 2002, , 467-470.	0.2	0
105	Genetic studies in Parkinson's disease with an α-synuclein/NACP gene polymorphism in Japan. Neuroscience Letters, 2001, 300, 125-127.	2.1	65
106	Influence of interleukin- $1\hat{l}^2$ gene polymorphism on age-at-onset of spinocerebellar ataxia 6 (SCA6) in Japanese patients. Neuroscience Letters, 2001, 307, 128-130.	2.1	8
107	Nicotinic acetylcholine receptors and neurodegenerative disease. Alcohol, 2001, 24, 79-81.	1.7	13
108	Lack of an association between cystatin C gene polymorphisms in Japanese patients with Alzheimer's disease. Neurology, 2001, 57, 337-339.	1.1	45

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109	ã,¢ãf«ãf"ãfã,ãfžãf¼ç—…ã«é−¢ã™ã,‹ã,°ãf«ã,¿ãfŸãf³é…¸è⅓,é€è›‹ç™½ã®å^†åç"Ÿç‰ ©å¦çš"ç"ç©¶. Japanese	Joournal of	<b>Ce</b> riatrics, 2
110	Risk factors for dementia., 2001,, 279-286.		0
111	Electrophysiological studies in spinocerebellar ataxia type 6. NeuroReport, 2000, 11, 969-972.	1.2	15
112	A single nucleotide polymorphism of dopamine transporter gene is associated with Parkinson's disease. Annals of Neurology, 2000, 47, 528-531.	5.3	39
113	Cerebral white matter lesions are not associated with apoE genotype but with age and female sex in Alzheimer's disease. Journal of Neurology, Neurosurgery and Psychiatry, 2000, 68, 653-656.	1.9	51
114	Lack of an Association of Estrogen Receptor $\hat{l}_{\pm}$ Gene Polymorphisms and Transcriptional Activity With Alzheimer Disease. Archives of Neurology, 2000, 57, 236.	4.5	144
115	Tumor necrosis factor, tumor necrosis factor receptors type 1 and 2, lymphotoxin- $\hat{l}_{\pm}$ , and HLA-DRB1 gene polymorphisms in human T-Cell lymphotropic virus type I associated myelopathy. Human Immunology, 2000, 61, 1262-1269.	2.4	26
116	A single nucleotide polymorphism of dopamine transporter gene is associated with Parkinson's disease. Annals of Neurology, 2000, 47, 528-531.	<b>5.</b> 3	3
117	Direct Alteration of the P/Q-Type Ca <sup>2+</sup> Channel Property by Polyglutamine Expansion in Spinocerebellar Ataxia 6. Journal of Neuroscience, 1999, 19, RC14-RC14.	3.6	107
118	Spinocerebellar ataxia type 6 in relation to CAG repeat length. Acta Neurologica Scandinavica, 1999, 99, 209-212.	2.1	10
119	Identification and analysis of the promoter region of the human NeuroD-related factor (NDRF). Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 1999, 1445, 142-147.	2.4	5
120	Apolipoprotein E promoter polymorphism and sporadic Alzheimer's disease in a Japanese population. Neuroscience Letters, 1999, 259, 56-58.	2.1	44
121	Organization of the human orphan nuclear receptor Nurr1 gene. Gene, 1999, 230, 225-232.	2.2	40
122	Effects of nerve growth factor and nicotine on the expression of nicotinic acetylcholine receptor subunits in PC12 cells. Neuroscience Research, 1999, 35, 175-181.	1.9	27
123	Structure and regulation of the human NeuroD (BETA2/BHF1) gene. Molecular Brain Research, 1999, 69, 223-231.	2.3	21
124	Variation in the number of CAG repeats in the Machado–Joseph disease gene (MJD1) in the Japanese population. Journal of the Neurological Sciences, 1999, 166, 71-73.	0.6	2
125	HLA-DRB1 and tumor necrosis factor gene polymorphisms in Japanese patients with multiple sclerosis. Journal of Neuroimmunology, 1998, 92, 109-112.	2.3	40
126	Genetic contribution of the tumor necrosis factor region in guillainâ€barré syndrome. Annals of Neurology, 1998, 44, 815-818.	<b>5.</b> 3	32

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127	Spinocerebellar ataxia type 6: MRI of three Japanese patients. Neuroradiology, 1998, 40, 222-227.	2.2	27
128	Effect of amino acid ergot alkaloids on glutamate transport via human glutamate transporter hGluT-1. Journal of the Neurological Sciences, 1998, 155, 31-36.	0.6	14
129	No association between apolipoprotein E alleles and olivopontocerebellar atrophy. Journal of the Neurological Sciences, 1998, 158, 110-112.	0.6	6
130	Characteristic Magnetic Resonance Imaging Findings in Machado-Joseph Disease. Archives of Neurology, 1998, 55, 33.	4.5	142
131	<i>HLA</i> and T-cell receptor gene polymorphisms in Guillain-Barrelesyndrome. Neurology, 1998, 51, 379-384.	1.1	52
132	A necropsied case of Machado-Joseph disease with a hyperintense signal of transverse pontine fibres on long TR sequences of magnetic resonance images. Journal of Neurology, Neurosurgery and Psychiatry, 1998, 64, 140-141.	1.9	9
133	Characteristic Magnetic Resonance Imaging Findings in Spinocerebellar Ataxia 6. Archives of Neurology, 1998, 55, 1348.	4.5	77
134	Molecular features of the CAG repeats of spinocerebellar ataxia 6 (SCA6). Human Molecular Genetics, 1997, 6, 1283-1287.	2.9	129
135	A new mitochondrial DNA mutation associated with mitochondrial myopathy: tRNA <sup>Leu(UUR) </sup> 3254C-to-G. Neurology, 1997, 49, 598-600.	1.1	37
136	CAG repeat length and disease duration in Machado-Joseph disease: a new clinical classification. Journal of the Neurological Sciences, 1997, 152, 166-171.	0.6	27
137	Structure and organization of the gene encoding human dopamine transporter. Gene, 1997, 195, 11-18.	2.2	98
138	Coagulation and vascular abnormalities in Crow-Fukase syndrome., 1997, 20, 486-492.		50
139	Expression of three glutamate transporter subtype mRNAs in human brain regions and peripheral tissues. Molecular Brain Research, 1996, 36, 189-192.	2.3	42
140	Serum deprivation alters the expression and the splicing at exons 7, 8 and 15 of the $\hat{l}^2$ -amyloid precursor protein in the C6 glioma cell line. Molecular Brain Research, 1996, 39, 12-22.	2.3	3
141	Molecular Cloning and Characterization of a cDNA Encoding a Novel Basic Helix-Loop-Helix Protein Structurally Related to NeuroD/BHF1. Biochemical and Biophysical Research Communications, 1996, 220, 754-758.	2.1	43
142	Cloning and Expression of a Rat Brain Basic Helix–Loop–Helix Factor. Biochemical and Biophysical Research Communications, 1996, 221, 199-204.	2.1	13
143	Reevaluation of the exact CAG repeat length in hereditary cerebellar ataxias using highly denaturing conditions and long PCR. Human Genetics, 1996, 97, 591-595.	3.8	21
144	Cytokines and myelin antibodies in Crow-Fukase syndrome. , 1996, 19, 1620-1622.		7

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145	A family with Machado-Joseph disease, previously diagnosed as dentatorubral-pallidoluysian atrophy. Neurology, 1996, 46, 1154-1156.	1.1	13
146	Homozygosity for Machado-Joseph disease gene enhances phenotypic severity Journal of Neurology, Neurosurgery and Psychiatry, 1996, 60, 354-356.	1.9	55
147	Cytokines and myelin antibodies in Crowâ€Fukase syndrome. Muscle and Nerve, 1996, 19, 1620-1622.	2.2	1
148	Clinical Manifestations of Autosomal Recessive Early-Onset Parkinsonism with Diurnal Fluctuation. Advances in Behavioral Biology, 1996, , 485-489.	0.2	5
149	Reevaluation of the exact CAG repeat length in hereditary cerebellar ataxias using highly denaturing conditions and long PCR. Human Genetics, 1996, 97, 591-595.	3.8	1
150	Localization of the gene encoding the human L-glutamate transporter (GLT-1) to 11p11.2-p13 by fluorescence in situ hybridization. Human Genetics, 1996, 97, 387-389.	3.8	1
151	Localization of the gene (SLC1A3) encoding human glutamate transporter (GluT-1) to 5p13 by fluorescence in situ hybridization. Cytogenetic and Genome Research, 1995, 69, 209-210.	1.1	25
152	Unique features of the CAG repeats in Machado–Joseph disease. Nature Genetics, 1995, 9, 344-345.	21.4	41
153	Inhibition by folded isomers of L-2-(carboxycyclopropyl) glycine of glutamate uptake via the human glutamate transporter hGluT-1. European Journal of Pharmacology, 1995, 289, 387-390.	2.6	16
154	Molecular features of the CAG repeats and clinical manifestation of Machado-Joseph disease. Human Molecular Genetics, 1995, 4, 807-812.	2.9	191
155	Neuroprotective mechanism of bromocriptine. Lancet, The, 1995, 346, 1305.	13.7	14
156	CAG expansions in a novel gene for Machado-Joseph disease at chromosome 14q32.1. Nature Genetics, 1994, 8, 221-228.	21.4	1,717
157	Cloning and Expression of a Human Glutamate Transporter. Biochemical and Biophysical Research Communications, 1994, 199, 171-176.	2.1	56
158	Ciliary Neurotrophic Factor Induced Increase in $\hat{l}^2$ -Amyloid Precursor Protein mRNA in Rat C6 Glioma Cells. Biochemical and Biophysical Research Communications, 1994, 204, 391-398.	2.1	4
159	Structural Organization and Expression of the Gene for Bovine Myosin I Heavy Chain1. Journal of Biochemistry, 1992, 111, 302-309.	1.7	8
160	HLAs and genes in japanese patients with multiple sclerosis: evidence for increased frequencies of HLA-Cw3, HLA-DR2, and HLA-DQB1â—0602. Human Immunology, 1992, 35, 116-124.	2.4	27
161	Generation of transgenic mice with elevated blood pressure by introduction of the rat renin and angiotensinogen genes Proceedings of the National Academy of Sciences of the United States of America, 1990, 87, 5153-5157.	7.1	142
162	Comparison of two families with and without ataxia harboring novel variants in PRKCG. Journal of Human Genetics, $0$ , , .	2.3	1