Ludger SchÃ¶ls

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/1328727/publications.pdf

Version: 2024-02-01

435	30,270	4960 84	7517 151
papers	citations	h-index	g-index

460	460	460	22402
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The prodromal phase of hereditary spastic paraplegia type 4: the preSPG4 cohort study. Brain, 2023, 146, 1093-1102.	7.6	6
2	Characterization of Lifestyle in Spinocerebellar Ataxia Type 3 and Association with Disease Severity. Movement Disorders, 2022, 37, 405-410.	3.9	8
3	Allele-specific targeting of mutant ataxin-3 by antisense oligonucleotides in SCA3-iPSC-derived neurons. Molecular Therapy - Nucleic Acids, 2022, 27, 99-108.	5.1	15
4	<scp>BCAS3</scp>â€Related Neurodevelopmental Disorder Shows Magnetic Resonance Imaging Features Resembling Brain Iron Accumulation. Movement Disorders, 2022, 37, 870-872.	3.9	0
5	Realâ€Life Turning Movements Capture Subtle Longitudinal and Preataxic Changes in Cerebellar Ataxia. Movement Disorders, 2022, 37, 1047-1058.	3.9	24
6	Characteristics of serum neurofilament light chain as a biomarker in hereditary spastic paraplegia type 4. Annals of Clinical and Translational Neurology, 2022, 9, 326-338.	3.7	6
7	Modified Delphi procedure-based expert consensus on endpoints for an international disease registry for Metachromatic Leukodystrophy: The European Metachromatic Leukodystrophy initiative (MLDi). Orphanet Journal of Rare Diseases, 2022, 17, 48.	2.7	10
8	Levels of Neurofilament Light at the Preataxic and Ataxic Stages of Spinocerebellar Ataxia Type 1. Neurology, 2022, 98, .	1.1	15
9	Tau and neurofilament lightâ€chain as fluid biomarkers in spinocerebellar ataxia type 3. European Journal of Neurology, 2022, 29, 2439-2452.	3.3	25
10	â€œEars of the lynxâ€•sign and thin corpus callosum on MRI in heterozygous SPG11 mutation carriers. Journal of Neurology, 2022, 269, 6148-6151.	3.6	1
11	Mitochondrial Dysfunction in Spinocerebellar Ataxia Type 3 Is Linked to VDAC1 Deubiquitination. International Journal of Molecular Sciences, 2022, 23, 5933.	4.1	9
12	CNS-associated T-lymphocytes in a mouse model of Hereditary Spastic Paraplegia type 11 (SPG11) are therapeutic targets for established immunomodulators. Experimental Neurology, 2022, 355, 114119.	4.1	3
13	Homeâ€based biofeedback speech treatment improves dysarthria in repeatâ€expansion <scp>SCAs</scp>. Annals of Clinical and Translational Neurology, 2022, 9, 1310-1315.	3.7	12
14	Differential Temporal Dynamics of Axial and Appendicular Ataxia in <scp>SCA3</scp>. Movement Disorders, 2022, 37, 1850-1860.	3.9	11
15	Association of Age at Onset and First Symptoms With Disease Progression in Patients With Metachromatic Leukodystrophy. Neurology, 2021, 96, e255-e266.	1.1	47
16	PolyQ-expanded ataxin-3 protein levels in peripheral blood mononuclear cells correlate with clinical parameters in SCA3: a pilot study. Journal of Neurology, 2021, 268, 1304-1315.	3.6	15
17	Natural History, Phenotypic Spectrum, and Discriminative Features of Multisystemic RFC1 Disease. Neurology, 2021, 96, e1369-e1382.	1.1	93
18	Correspondence on â€œClinical, neuropathological, and genetic characterization of STUB1 variants in cerebellar ataxias: a frequent cause of predominant cognitive impairmentâ€•by Roux et al.. Genetics in Medicine, 2021, 23, 1171-1172.	2.4	2

#	Article	IF	Citations
19	Spinocerebellar ataxia type 14: refining clinicogenetic diagnosis in a rare adultâ€onset disorder. Annals of Clinical and Translational Neurology, 2021, 8, 774-789.	3.7	13
20	Neurofilament light chain is a cerebrospinal fluid biomarker in hereditary spastic paraplegia. Annals of Clinical and Translational Neurology, 2021, 8, 1122-1131.	3.7	11
21	Biallelic loss-of-function variations in PRDX3 cause cerebellar ataxia. Brain, 2021, 144, 1467-1481.	7.6	18
22	Deep mining of oxysterols and cholestenoic acids in human plasma and cerebrospinal fluid: Quantification using isotope dilution mass spectrometry. Analytica Chimica Acta, 2021, 1154, 338259.	5.4	14
23	Solving unsolved rare neurological diseasesâ€"a Solve-RD viewpoint. European Journal of Human Genetics, 2021, 29, 1332-1336.	2.8	4
24	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
25	Progression characteristics of the European Friedreich's Ataxia Consortium for Translational Studies (EFACTS): a 4-year cohort study. Lancet Neurology, The, 2021, 20, 362-372.	10.2	53
26	Developmental Consequences of Defective ATG7-Mediated Autophagy in Humans. New England Journal of Medicine, 2021, 384, 2406-2417.	27.0	84
27	Bi-allelic loss-of-function variants in BCAS3 cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2021, 108, 1069-1082.	6.2	8
28	The ARCA Registry: A Collaborative Global Platform for Advancing Trial Readiness in Autosomal Recessive Cerebellar Ataxias. Frontiers in Neurology, 2021, 12, 677551.	2.4	15
29	Natural History of Polymerase Gammaâ€"Related Ataxia. Movement Disorders, 2021, 36, 2642-2652.	3.9	10
30	Polyglutamineâ€Expanded Ataxinâ€3: A Target Engagement Marker for Spinocerebellar Ataxia Type 3 in Peripheral Blood. Movement Disorders, 2021, 36, 2675-2681.	3.9	22
31	Bi-allelic truncating mutations in VWA1 cause neuromyopathy. Brain, 2021, 144, 574-583.	7.6	16
32	Metabolic profiling in serum, cerebrospinal fluid, and brain of patients with cerebrotendinous xanthomatosis. Journal of Lipid Research, 2021, 62, 100078.	4.2	14
33	Fampridine and Acetazolamide in EA2 and Related Familial EA. Neurology: Clinical Practice, 2021, 11, e438-e446.	1.6	27
34	Pathophysiological interplay between O -GlcNAc transferase and the Machadoâ€"Joseph disease protein ataxin-3. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.1	5
35	Safety and Efficacy of Acetyl-DL-Leucine in Certain Types of Cerebellar Ataxia. JAMA Network Open, 2021, 4, e2135841.	5.9	16
36	Non-motor symptoms are relevant and possibly treatable in hereditary spastic paraplegia type 4 (SPG4). Journal of Neurology, 2020, 267, 369-379.	3.6	11

#	Article	IF	Citations
37	Unraveling the genetic cause of hereditary ophthalmic disorders in Arab societies from Israel and the Palestinian Authority. European Journal of Human Genetics, 2020, 28, 742-753.	2.8	1
38	Single Nucleotide Polymorphisms in Thyroid Hormone Transporter Genes MCT8, MCT10 and Deiodinase DIO2 Contribute to Inter-Individual Variance of Executive Functions and Personality Traits. Experimental and Clinical Endocrinology and Diabetes, 2020, 128, 573-581.	1.2	5
39	Onset features and time to diagnosis in Friedreichâ€™s Ataxia. Orphanet Journal of Rare Diseases, 2020, 15, 198.	2.7	27
40	Clinical, neuropathological, and genetic characterization of STUB1 variants in cerebellar ataxias: a frequent cause of predominant cognitive impairment. Genetics in Medicine, 2020, 22, 1851-1862.	2.4	30
41	Generation of the CRISPR/Cas9-mediated KIF1C knock-out human iPSC line HIHRSi003-A-1. Stem Cell Research, 2020, 49, 102059.	0.7	3
42	Haploinsufficiency due to a novel ACO2 deletion causes mitochondrial dysfunction in fibroblasts from a patient with dominant optic nerve atrophy. Scientific Reports, 2020, 10, 16736.	3.3	12
43	Comparative Transcriptional Profiling of Motor Neuron Disorder-Associated Genes in Various Human Cell Culture Models. Frontiers in Cell and Developmental Biology, 2020, 8, 544043.	3.7	11
44	Defining diagnostic cutoffs in neurological patients for serum very long chain fatty acids (VLCFA) in genetically confirmed X-Adrenoleukodystrophy. Scientific Reports, 2020, 10, 15093.	3.3	12
45	Natural history of Krabbe disease â€“ a nationwide study in Germany using clinical and MRI data. Orphanet Journal of Rare Diseases, 2020, 15, 243.	2.7	17
46	Conversion of individuals at risk for spinocerebellar ataxia types 1, 2, 3, and 6 to manifest ataxia (RISCA): a longitudinal cohort study. Lancet Neurology, The, 2020, 19, 738-747.	10.2	41
47	Multifocal, hypoechogenic nerve thickening in Cerebrotendinous Xanthomatosis. Clinical Neurophysiology, 2020, 131, 1798-1803.	1.5	2
48	Delineating MT-ATP6 -associated disease. Neurology: Genetics, 2020, 6, e393.	1.9	73
49	First-line exome sequencing in Palestinian and Israeli Arabs with neurological disorders is efficient and facilitates disease gene discovery. European Journal of Human Genetics, 2020, 28, 1034-1043.	2.8	20
50	Real-life gait assessment in degenerative cerebellar ataxia. Neurology, 2020, 95, e1199-e1210.	1.1	60
51	Neurofilaments in spinocerebellar ataxia type 3: blood biomarkers at the preataxic and ataxic stage in humans and mice. EMBO Molecular Medicine, 2020, 12, e11803.	6.9	73
52	Clinicoâ€Genetic, Imaging and Molecular Delineation of <scp>COQ8A</scp>â€Ataxia: A Multicenter Study of 59 Patients. Annals of Neurology, 2020, 88, 251-263.	5.3	52
53	ATPase Domain <scp>AFG3L2</scp> Mutations Alter <scp>OPA1</scp> Processing and Cause Optic Neuropathy. Annals of Neurology, 2020, 88, 18-32.	5.3	31
54	FIG4 mutations leading to parkinsonism and a phenotypical continuum between CMT4J and Yunis VarÃ³n syndrome. Parkinsonism and Related Disorders, 2020, 74, 6-11.	2.2	15

#	Article	IF	Citations
55	The European Reference Network for Rare Neurological Diseases. Frontiers in Neurology, 2020, 11, 616569.	2.4	26
56	Clinico-Genetic, Imaging and Molecular Delineation of COQ8A-Ataxia: A Multicenter Study of 59 Patients. , 2020, 88, 251.		1
57	NfL and pNfH are increased in Friedreichâ€™s ataxia. Journal of Neurology, 2020, 267, 1420-1430.	3.6	17
58	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	12.8	35
59	Decreased Na+/K+ ATPase Expression and Depolarized Cell Membrane in Neurons Differentiated from Chorea-Acanthocytosis Patients. Scientific Reports, 2020, 10, 8391.	3.3	9
60	CHIP mutations affect the heat shock response differently in human fibroblasts and iPSC-derived neurons. DMM Disease Models and Mechanisms, 2020, 13, .	2.4	5
61	Confirmation of TACO1 as a Leigh Syndrome Disease Gene in Two Additional Families. Journal of Neuromuscular Diseases, 2020, 7, 301-308.	2.6	8
62	Structural characteristics of the central nervous system in FriedreichÂataxia: an in vivo spinal cord and brain MRI study. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 615-617.	1.9	33
63	Brain MRI features and scoring of leukodystrophy in adult-onset Krabbe disease. Neurology, 2019, 93, e647-e652.	1.1	25
64	Multiparametric rapid screening of neuronal process pathology for drug target identification in HSP patient-specific neurons. Scientific Reports, 2019, 9, 9615.	3.3	30
65	Protocol of a randomized, double-blind, placebo-controlled, parallel-group, multicentre study of the efficacy and safety of nicotinamide in patients with Friedreich ataxia (NICOFA). Neurological Research and Practice, 2019, 1, 33.	2.0	14
66	Application of Quantitative Motor Assessments in Friedreich Ataxia and Evaluation of Their Relation to Clinical Measures. Cerebellum, 2019, 18, 896-909.	2.5	9
67	Clinical, radiological, and genetic characteristics of 16 patients with ACO2 gene defects: Delineation of an emerging neurometabolic syndrome. Journal of Inherited Metabolic Disease, 2019, 42, 264-275.	3.6	18
68	Phenotypic spectrum of autosomal recessive retinitis pigmentosa without posterior column ataxia caused by mutations in the FLVCR1 gene. Graefe's Archive for Clinical and Experimental Ophthalmology, 2019, 257, 629-638.	1.9	13
69	Prediction of Survival With Longâ€Term Disease Progression in Most Common Spinocerebellar Ataxia. Movement Disorders, 2019, 34, 1220-1227.	3.9	14
70	Phenotypic variation between siblings with Metachromatic Leukodystrophy. Orphanet Journal of Rare Diseases, 2019, 14, 136.	2.7	29
71	Loss of paraplegin drives spasticity rather than ataxia in a cohort of 241 patients with SPG7. Neurology, 2019, 92, e2679-e2690.	1.1	49
72	A novel biallelic lossâ€ofâ€function mutation in TMCO1 gene confirming and expanding the phenotype spectrum of cerebroâ€facioâ€thoracic dysplasia. American Journal of Medical Genetics, Part A, 2019, 179, 1338-1345.	1.2	9

#	Article	IF	Citations
73	Biallelic variants in the transcription factor PAX7 are a new genetic cause of myopathy. Genetics in Medicine, 2019, 21, 2521-2531.	2.4	25
74	FAHN/SPG35: a narrow phenotypic spectrum across disease classifications. Brain, 2019, 142, 1561-1572.	7.6	70
75	Patientâ€reported outcomes in Friedreichâ€™s ataxia after withdrawal from idebenone. Acta Neurologica Scandinavica, 2019, 139, 533-539.	2.1	17
76	Speech treatment improves dysarthria in multisystemic ataxia: a rater-blinded, controlled pilot-study in ARSACS. Journal of Neurology, 2019, 266, 1260-1266.	3.6	27
77	Realâ€time use of audioâ€biofeedback can improve postural sway in patients with degenerative ataxia. Annals of Clinical and Translational Neurology, 2019, 6, 285-294.	3.7	7
78	Pattern of Cerebellar Atrophy in Friedreichâ€™s Ataxiaâ€”Using the SUIT Template. Cerebellum, 2019, 18, 435-447.	2.5	23
79	Autosomal Recessive Cerebellar Ataxias: Paving the Way toward Targeted Molecular Therapies. Neuron, 2019, 101, 560-583.	8.1	83
80	mRNA as a Novel Treatment Strategy for Hereditary Spastic Paraplegia Type 5. Molecular Therapy - Methods and Clinical Development, 2019, 15, 359-370.	4.1	23
81	Generation of a homozygous CRISPR/Cas9-mediated knockout human iPSC line for the STUB1 locus. Stem Cell Research, 2019, 34, 101378.	0.7	7
82	A previously identified missense mutation in STYXL1 is likely benign. European Journal of Medical Genetics, 2019, 62, 103582.	1.3	2
83	TSFM mutations cause a complex hyperkinetic movement disorder with strong relief by cannabinoids. Parkinsonism and Related Disorders, 2019, 60, 176-178.	2.2	6
84	The movement disorder spectrum of SCA21 (ATX-TMEM240): 3 novel families and systematic review of the literature. Parkinsonism and Related Disorders, 2019, 62, 215-220.	2.2	18
85	Mechanistic basis of an epistatic interaction reducing age at onset in hereditary spastic paraplegia. Brain, 2018, 141, 1286-1299.	7.6	29
86	Establishment of STUB1/ CHIP mutant induced pluripotent stem cells (iPSCs) from a patient with Gordon Holmes syndrome/SCAR16. Stem Cell Research, 2018, 29, 166-169.	0.7	5
87	Consensus Paper: Neurophysiological Assessments of Ataxias in Daily Practice. Cerebellum, 2018, 17, 628-653.	2.5	30
88	Comprehensive systematic review summary: Treatment of cerebellar motor dysfunction and ataxia. Neurology, 2018, 90, 464-471.	1.1	108
89	To die or not to die SGK1-sensitive ORAI/STIM in cell survival. Cell Calcium, 2018, 74, 29-34.	2.4	21
90	Survival in patients with spinocerebellar ataxia types 1, 2, 3, and 6 (EUROSCA): a longitudinal cohort study. Lancet Neurology, The, 2018, 17, 327-334.	10.2	69

#	Article	IF	Citations
91	Treatable inherited rare movement disorders. Movement Disorders, 2018, 33, 21-35.	3.9	79
92	SERAC1 deficiency causes complicated HSP: evidence from a novel splice mutation in a large family. Journal of Medical Genetics, 2018, 55, 39-47.	3.2	28
93	Case series: Slowing alpha rhythm in late-stage ALS patients. Clinical Neurophysiology, 2018, 129, 406-408.	1.5	14
94	Inhibition of Lithium Sensitive Orai1/ STIM1 Expression and Store Operated Ca2+ Entry in Chorea-Acanthocytosis Neurons by NF-ÎºB Inhibitor Wogonin. Cellular Physiology and Biochemistry, 2018, 51, 278-289.	1.6	9
95	Nerve ultrasound characterizes AMN polyneuropathy as inhomogeneous and focal hypertrophic. Orphanet Journal of Rare Diseases, 2018, 13, 194.	2.7	9
96	Mitochondrial Morphology, Function and Homeostasis Are Impaired by Expression of an N-terminal Calpain Cleavage Fragment of Ataxin-3. Frontiers in Molecular Neuroscience, 2018, 11, 368.	2.9	32
97	NfL is a biomarker for adult-onset leukoencephalopathy with axonal spheroids and pigmented glia. Neurology, 2018, 91, 755-757.	1.1	11
98	Long-term evolution of patient-reported outcome measures in spinocerebellar ataxias. Journal of Neurology, 2018, 265, 2040-2051.	3.6	34
99	Generation of an induced pluripotent stem cell line from a patient with spinocerebellar ataxia type 3 (SCA3): HIHCNi002-A. Stem Cell Research, 2018, 30, 171-174.	0.7	12
100	Coordination and timing deficits in speech and swallowing in autosomal recessive spastic ataxia of Charlevoixâ€“Saguenay (ARSACS). Journal of Neurology, 2018, 265, 2060-2070.	3.6	21
101	Generation of an induced pluripotent stem cell line from a patient with adult-onset leukoencephalopathy with axonal spheroids and pigmented glia (ALSP): HIHCNi003-A. Stem Cell Research, 2018, 30, 206-209.	0.7	3
102	De novo ITPR1 variants are a recurrent cause of early-onset ataxia, acting via loss of channel function. European Journal of Human Genetics, 2018, 26, 1623-1634.	2.8	32
103	Serum neurofilament light is increased in multiple system atrophy of cerebellar type and in repeat-expansion spinocerebellar ataxias: a pilot study. Journal of Neurology, 2018, 265, 1618-1624.	3.6	58
104	Nonataxia symptoms in Friedreich Ataxia. Neurology, 2018, 91, e917-e930.	1.1	46
105	Serum neurofilament light chain is increased in hereditary spastic paraplegias. Annals of Clinical and Translational Neurology, 2018, 5, 876-882.	3.7	26
106	GPT2 mutations cause developmental encephalopathy with microcephaly and features of complicated hereditary spastic paraplegia. Clinical Genetics, 2018, 94, 356-361.	2.0	14
107	Effects of acetyl-DL-leucine on cerebellar ataxia (ALCAT trial): study protocol for a multicenter, multinational, randomized, double-blind, placebo-controlled, crossover phase III trial. BMC Neurology, 2017, 17, 7.	1.8	23
108	STUB1/CHIP mutations cause Gordon Holmes syndrome as part of a widespread multisystemic neurodegeneration: evidence from four novel mutations. Orphanet Journal of Rare Diseases, 2017, 12, 31.	2.7	56

#	Article	IF	Citations
109	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578.	7.6	85
110	Involvement of the cerebellum in Parkinson disease and dementia with Lewy bodies. Annals of Neurology, 2017, 81, 898-903.	5.3	44
111	CNTNAP1 mutations cause CNS hypomyelination and neuropathy with or without arthrogryposis. Neurology: Genetics, 2017, 3, e144.	1.9	24
112	Loss-of-function mutations in theATP13A2/PARK9 gene cause complicated hereditary spastic paraplegia (SPG78). Brain, 2017, 140, 287-305.	7.6	135
113	Uniparental disomy determined by wholeâ€exome sequencing in a spectrum of rare motoneuron diseases and ataxias. Molecular Genetics & Genomic Medicine, 2017, 5, 280-286.	1.2	23
114	A combinatorial approach to identify calpain cleavage sites in the Machado-Joseph disease protein ataxin-3. Brain, 2017, 140, 1280-1299.	7.6	33
115	Individualized exergame training improves postural control in advanced degenerative spinocerebellar ataxia: A rater-blinded, intra-individually controlled trial. Parkinsonism and Related Disorders, 2017, 39, 80-84.	2.2	45
116	TBK1 Mutation Spectrum in an Extended European Patient Cohort with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Human Mutation, 2017, 38, 297-309.	2.5	87
117	Neurochondrin is a neuronal target antigen in autoimmune cerebellar degeneration. Neurology: Neuroimmunology and NeuroInflammation, 2017, 4, e307.	6.0	39
118	Clinical and molecular characterization of hereditary spastic paraplegias: A next-generation sequencing panel approach. Journal of the Neurological Sciences, 2017, 383, 18-25.	0.6	44
119	Lithium Sensitivity of Store Operated Ca2+ Entry and Survival of Fibroblasts Isolated from Chorea-Acanthocytosis Patients. Cellular Physiology and Biochemistry, 2017, 42, 2066-2077.	1.6	24
120	Frequent genes in rare diseases: panelâ€based next generation sequencing to disclose causal mutations in hereditary neuropathies. Journal of Neurochemistry, 2017, 143, 507-522.	3.9	68
121	Body Mass Index Decline Is Related to Spinocerebellar Ataxia Disease Progression. Movement Disorders Clinical Practice, 2017, 4, 689-697.	1.5	25
122	Speech and swallowing abnormalities in adults with POLG associated ataxia (POLG-A). Mitochondrion, 2017, 37, 1-7.	3.4	20
123	Lithium Sensitive ORAI1 Expression, Store Operated Ca2+ Entry and Suicidal Death of Neurons in Chorea-Acanthocytosis. Scientific Reports, 2017, 7, 6457.	3.3	31
124	Enzymatic characterization of novel arylsulfatase A variants using human arylsulfatase Aâ€deficient immortalized mesenchymal stromal cells. Human Mutation, 2017, 38, 1511-1520.	2.5	20
125	Clinical and genetic characteristics of sporadic adult-onset degenerative ataxia. Neurology, 2017, 89, 1043-1049.	1.1	45
126	Neurons, Erythrocytes and Beyond â€“The Diverse Functions of Chorein. NeuroSignals, 2017, 25, 117-126.	0.9	17

#	Article	IF	Citations
127	Hereditary spastic paraplegia type 5: natural history, biomarkers and a randomized controlled trial. Brain, 2017, 140, 3112-3127.	7.6	87
128	Absence of EEG correlates of self-referential processing depth in ALS. PLoS ONE, 2017, 12, e0180136.	2.5	10
129	Hemodialysis in MNGIE transiently reduces serum and urine levels of thymidine and deoxyuridine, but not CSF levels and neurological function. Orphanet Journal of Rare Diseases, 2017, 12, 135.	2.7	23
130	Rare Variants in Neurodegeneration Associated Genes Revealed by Targeted Panel Sequencing in a German ALS Cohort. Frontiers in Molecular Neuroscience, 2016, 9, 92.	2.9	41
131	Hereditary spastic paraplegia: Clinicogenetic lessons from 608 patients. Annals of Neurology, 2016, 79, 646-658.	5.3	218
132	Individual changes in preclinical spinocerebellar ataxia identified via increased motor complexity. Movement Disorders, 2016, 31, 1891-1900.	3.9	54
133	MultisystemicSYNE1ataxia: confirming the high frequency and extending the mutational and phenotypic spectrum. Brain, 2016, 139, e46-e46.	7.6	40
134	SYNE1 ataxia is a common recessive ataxia with major non-cerebellar features: a large multi-centre study. Brain, 2016, 139, 1378-1393.	7.6	87
135	Complicated spastic paraplegia in patients with AP5Z1 mutations (SPG48). Neurology: Genetics, 2016, 2, e98.	1.9	35
136	Induced pluripotent stem cells (iPSCs) derived from cerebrotendinous xanthomatosis (CTX) patient's fibroblasts carrying a R395S mutation. Stem Cell Research, 2016, 17, 433-436.	0.7	1
137	Generation of optic atrophy 1 patient-derived induced pluripotent stem cells (iPS-OPA1-BEHR) for disease modeling of complex optic atrophy syndromes (Behr syndrome). Stem Cell Research, 2016, 17, 426-429.	0.7	5
138	Generation of induced pluripotent stem cells (iPSCs) from a hereditary spastic paraplegia patient carrying a homozygous Y275X mutation in CYP7B1 (SPG5). Stem Cell Research, 2016, 17, 437-440.	0.7	8
139	Generation of induced pluripotent stem cells (iPSCs) from a hereditary spastic paraplegia patient carrying a homozygous R486C mutation in CYP7B1 (SPG5). Stem Cell Research, 2016, 17, 422-425.	0.7	1
140	Establishment of SPAST mutant induced pluripotent stem cells (iPSCs) from a hereditary spastic paraplegia (HSP) patient. Stem Cell Research, 2016, 17, 485-488.	0.7	4
141	CYP2U1 mutations in two Iranian patients with activity induced dystonia, motor regression and spastic paraplegia. European Journal of Paediatric Neurology, 2016, 20, 782-787.	1.6	19
142	Inducing Differentiation of Premalignant Hepatic Cells as a Novel Therapeutic Strategy in Hepatocarcinoma. Cancer Research, 2016, 76, 5550-5561.	0.9	15
143	Thickening of the peripheral nerves in metachromatic leukodystrophy. Journal of the Neurological Sciences, 2016, 368, 399-401.	0.6	12
144	A recurrent mutation in KCNA2 as a novel cause of hereditary spastic paraplegia and ataxia. Annals of Neurology, 2016, 80, .	5.3	49

#	Article	IF	Citations
145	Antisense Oligonucleotide Mediated Splice Correction of a Deep Intronic Mutation in OPA1. Molecular Therapy - Nucleic Acids, 2016, 5, e390.	5.1	43
146	A cognitive brainâ€“computer interface for patients with amyotrophic lateral sclerosis. Progress in Brain Research, 2016, 228, 221-239.	1.4	9
147	Uniparental disomy of chromosome 16 unmasks recessive mutations of FA2H /SPG35 in 4 families. Neurology, 2016, 87, 186-191.	1.1	27
148	Leukoencephalopathy With Axonal Spheroids and Pigmented Glia. JAMA Neurology, 2016, 73, 1400.	9.0	0
149	Progression characteristics of the European Friedreichâ€™s Ataxia Consortium for Translational Studies (EFACTS): a 2 year cohort study. Lancet Neurology, The, 2016, 15, 1346-1354.	10.2	117
150	M4â€…Safety and tolerability of BN82451B in huntingtonâ€™s disease. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, A102.2-A103.	1.9	0
151	Standardized Assessment of Hereditary Ataxia Patients in Clinical Studies. Movement Disorders Clinical Practice, 2016, 3, 230-240.	1.5	13
152	Identifying Niemannâ€“Pick type C in early-onset ataxia: two quick clinical screening tools. Journal of Neurology, 2016, 263, 1911-1918.	3.6	16
153	Parental mosaicism in another case of Dravet syndrome caused by a novel SCN1A deletion: a case report. Journal of Medical Case Reports, 2016, 10, 67.	0.8	5
154	Reply. Annals of Neurology, 2016, 80, 170-171.	5.3	0
155	Expanded phenotypic spectrum of the m.8344A>G â€œMERRFâ€•mutation: data from the German mitoNET registry. Journal of Neurology, 2016, 263, 961-972.	3.6	77
156	Peripheral Neuropathy in Spinocerebellar Ataxia Type 1, 2, 3, and 6. Cerebellum, 2016, 15, 165-173.	2.5	54
157	The spectrum of KIAA0196 variants, and characterization of a murine knockout: implications for the mutational mechanism in hereditary spastic paraplegia type SPG8. Orphanet Journal of Rare Diseases, 2015, 10, 147.	2.7	16
158	Abnormal Paraplegin Expression in Swollen Neurites, Ï„- and Î±-Synuclein Pathology in a Case of Hereditary Spastic Paraplegia SPG7 with an Ala510Val Mutation. International Journal of Molecular Sciences, 2015, 16, 25050-25066.	4.1	18
159	A randomized, placeboâ€controlled trial of AFQ056 for the treatment of chorea in Huntington's disease. Movement Disorders, 2015, 30, 427-431.	3.9	67
160	Ataxia meets chorioretinal dystrophy and hypogonadism: Boucher-NeuhÃuser syndrome due toPNPLA6mutations: FigureÃ1. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 580-581.	1.9	20
161	Brain-computer interfacing in amyotrophic lateral sclerosis: Implications of a resting-state EEG analysis. , 2015, 2015, 6979-82.		9
162	A Cognitive Brain-Computer Interface for Patients with Amyotrophic Lateral Sclerosis. , 2015, , .		1

#	Article	IF	Citations
163	Chorein Sensitive Arrangement of Cytoskeletal Architecture. Cellular Physiology and Biochemistry, 2015, 37, 399-408.	1.6	28
164	Biological and clinical characteristics of the European Friedreich's Ataxia Consortium for Translational Studies (EFACTS) cohort: a cross-sectional analysis of baseline data. Lancet Neurology, The, 2015, 14, 174-182.	10.2	159
165	Comparing speech characteristics in spinocerebellar ataxias type 3 and type 6 with Friedreich ataxia. Journal of Neurology, 2015, 262, 21-26.	3.6	33
166	Absence of BiP Co-chaperone DNAJC3 Causes Diabetes Mellitus and Multisystemic Neurodegeneration. American Journal of Human Genetics, 2015, 96, 514.	6.2	1
167	De novo loss- or gain-of-function mutations in KCNA2 cause epileptic encephalopathy. Nature Genetics, 2015, 47, 393-399.	21.4	224
168	No parkinsonism in SCA2 and SCA3 despite severe neurodegeneration of the dopaminergic substantia nigra. Brain, 2015, 138, 3316-3326.	7.6	54
169	Genetics of Hereditary Spastic Paraplegias (HSP). , 2015, , 353-383.		0
170	Long-term disease progression in spinocerebellar ataxia types 1, 2, 3, and 6: a longitudinal cohort study. Lancet Neurology, The, 2015, 14, 1101-1108.	10.2	213
171	Gray and white matter alterations in hereditary spastic paraplegia type SPG4 and clinical correlations. Journal of Neurology, 2015, 262, 1961-1971.	3.6	30
172	NPC1 is enriched in unexplained early onset ataxia: a targeted high-throughput screening. Journal of Neurology, 2015, 262, 2557-2563.	3.6	25
173	Spinocerebellar ataxia type 36 exists in diverse populations and can be caused by a short hexanucleotide GGCCTG repeat expansion. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 986-995.	1.9	49
174	Ataxia. Neuropsychiatric Symptoms of Neurological Disease, 2015, , 277-292.	0.3	0
175	Motor protein mutations cause a new form of hereditary spastic paraplegia. Neurology, 2014, 82, 2007-2016.	1.1	56
176	Cholestenoic acids regulate motor neuron survival via liver X receptors. Journal of Clinical Investigation, 2014, 124, 4829-4842.	8.2	84
177	Pure and syndromic optic atrophy explained by deep intronic OPA1 mutations and an intralocus modifier. Brain, 2014, 137, 2164-2177.	7.6	62
178	Cerebellar pathology in Friedreich's ataxia: Atrophied dentate nuclei with normal iron content. NeuroImage: Clinical, 2014, 6, 93-99.	2.7	56
179	Absence of BiP Co-chaperone DNAJC3 Causes Diabetes Mellitus and Multisystemic Neurodegeneration. American Journal of Human Genetics, 2014, 95, 689-697.	6.2	100
180	Safety and tolerability of carbamylated erythropoietin in Friedreich's ataxia. Movement Disorders, 2014, 29, 935-939.	3.9	46

#	Article	IF	Citations
181	Dominant spinal muscular atrophy due to BICD2: a novel mutation refines the phenotype. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 590-592.	1.9	37
182	Targeted high-throughput sequencing identifies a TARDBP mutation as a cause of early-onset FTD without motor neuron disease. Neurobiology of Aging, 2014, 35, 1212.e1-1212.e5.	3.1	40
183	X-linked Charcot-Marie-Tooth disease, Arts syndrome, and prelingual non-syndromic deafness form a disease continuum: evidence from a family with a novel PRPS1 mutation. Orphanet Journal of Rare Diseases, 2014, 9, 24.	2.7	42
184	Prediction of the age at onset in spinocerebellar ataxia type 1, 2, 3 and 6. Journal of Medical Genetics, 2014, 51, 479-486.	3.2	85
185	PNPLA6 mutations cause Boucher-NeuhÃuser and Gordon Holmes syndromes as part of a broad neurodegenerative spectrum. Brain, 2014, 137, 69-77.	7.6	189
186	Consensus Paper: Management of Degenerative Cerebellar Disorders. Cerebellum, 2014, 13, 248-268.	2.5	166
187	The tumour suppressor gene WWOX is mutated in autosomal recessive cerebellar ataxia with epilepsy and mental retardation. Brain, 2014, 137, 411-419.	7.6	127
188	AP5Z1/SPG4 8 frequency in autosomal recessive and sporadic spastic paraplegia. Molecular Genetics & Genomic Medicine, 2014, 2, 379-382.	1.2	10
189	Imaging features in conventional MRI, spectroscopy and diffusion weighted images of hereditary diffuse leukoencephalopathy with axonal spheroids (HDLS). Journal of Neurology, 2014, 261, 2351-2359.	3.6	35
190	Leukodystrophies underlying cryptic spastic paraparesis: frequency and phenotype in 76 patients. European Journal of Neurology, 2014, 21, 983-988.	3.3	23
191	Modulation of the age at onset in spinocerebellar ataxia by CAG tracts in various genes. Brain, 2014, 137, 2444-2455.	7.6	144
192	Rare mutations in SQSTM1 modify susceptibility to frontotemporal lobar degeneration. Acta Neuropathologica, 2014, 128, 397-410.	7.7	93
193	Dystonia with aphonia, slow horizontal saccades, epilepsy and photic myoclonus: A novel syndrome?. Parkinsonism and Related Disorders, 2014, 20, 328-331.	2.2	4
194	Phenotype and frequency of STUB1 mutations: next-generation screenings in Caucasian ataxia and spastic paraplegia cohorts. Orphanet Journal of Rare Diseases, 2014, 9, 57.	2.7	54
195	Sanfilippo type A: new clinical manifestations and neuro-imaging findings in patients from the same family in Israel: a case report. Journal of Medical Case Reports, 2014, 8, 78.	0.8	10
196	Autosomal dominant cerebellar ataxia with slow ocular saccades, neuropathy and orthostatism: A novel entity?. Parkinsonism and Related Disorders, 2014, 20, 748-754.	2.2	4
197	A novel missense mutation in CACNA1A evaluated by in silico protein modeling is associated with non-episodic spinocerebellar ataxia with slow progression. European Journal of Medical Genetics, 2014, 57, 207-211.	1.3	22
198	The effectiveness of allied health care in patients with ataxia: a systematic review. Journal of Neurology, 2014, 261, 251-258.	3.6	66

#	Article	IF	Citations
199	High prevalence of <scp>NMDA</scp> receptor IgA/IgM antibodies in different dementia types. Annals of Clinical and Translational Neurology, 2014, 1, 822-832.	3.7	114
200	HereditÃre BewegungsstÃ¶rungen. , 2014, , 155-175.		0
201	Autosomal recessive spastic ataxia of Charlevoix Saguenay (ARSACS): expanding the genetic, clinical and imaging spectrum. Orphanet Journal of Rare Diseases, 2013, 8, 41.	2.7	147
202	Late adult-onset pure spinal muscular atrophy due to a compound HEXB macro-deletion. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 628-629.	1.7	5
203	Experience in a short-term trial with 4-Aminopyridine in cerebellar ataxia. Journal of Neurology, 2013, 260, 2175-2176.	3.6	25
204	PRRT2-related disorders: further PKD and ICCA cases and review of the literature. Journal of Neurology, 2013, 260, 1234-1244.	3.6	63
205	Inventory of Non-Ataxia Signs (INAS): Validation of a New Clinical Assessment Instrument. Cerebellum, 2013, 12, 418-428.	2.5	112
206	Clinical features, neurogenetics and neuropathology of the polyglutamine spinocerebellar ataxias type 1, 2, 3, 6 and 7. Progress in Neurobiology, 2013, 104, 38-66.	5.7	283
207	HereditÃre Ataxien. Medizinische Genetik, 2013, 25, 235-248.	0.2	1
208	Loss of Function of Glucocerebrosidase GBA2 Is Responsible for Motor Neuron Defects in Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2013, 92, 238-244.	6.2	154
209	Chorein sensitivity of cytoskeletal organization and degranulation of platelets. FASEB Journal, 2013, 27, 2799-2806.	0.5	47
210	Biological and clinical characteristics of individuals at risk for spinocerebellar ataxia types 1, 2, 3, and 6 in the longitudinal RISCA study: analysis of baseline data. Lancet Neurology, The, 2013, 12, 650-658.	10.2	167
211	Electrophysiological characterisation of motor and sensory tracts in patients with hereditary spastic paraplegia (HSP). Orphanet Journal of Rare Diseases, 2013, 8, 158.	2.7	31
212	A Panâ€<scp>E</scp>uropean Study of theC9orf72Repeat Associated with<scp>FTLD</scp>: Geographic Prevalence, Genomic Instability, and Intermediate Repeats. Human Mutation, 2013, 34, 363-373.	2.5	247
213	Decreased retinal sensitivity and loss of retinal nerve fibers in multiple system atrophy. Graefe's Archive for Clinical and Experimental Ophthalmology, 2013, 251, 235-241.	1.9	17
214	L-2 hydroxyglutaric aciduria as a rare cause of leukencephalopathy in adults. Clinical Neurology and Neurosurgery, 2013, 115, 765-766.	1.4	6
215	Niemann-Pick type C is frequent in adult ataxia with cognitive decline and vertical gaze palsy. Neurology, 2013, 80, 1169-1170.	1.1	31
216	Friedreich Ataxia: Dysarthria Profile and Clinical Data. Cerebellum, 2013, 12, 475-484.	2.5	39

#	Article	IF	Citations
217	Alteration of Ganglioside Biosynthesis Responsible for Complex Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2013, 93, 118-123.	6.2	151
218	Videogame-based coordinative training can improve advanced, multisystemic early-onset ataxia. Journal of Neurology, 2013, 260, 2656-2658.	3.6	50
219	Genotype-specific patterns of atrophy progression are more sensitive than clinical decline in SCA1, SCA3 and SCA6. Brain, 2013, 136, 905-917.	7.6	128
220	De novo mutations in hereditary diffuse leukoencephalopathy with axonal spheroids (HDLS). Neurology, 2013, 81, 2039-2044.	1.1	62
221	Leukodystrophies in idiopathic adultâ€onset ataxia: Frequency and phenotype in 105 patients. Movement Disorders, 2013, 28, 2033-2035.	3.9	2
222	Mutations in phospholipase DDHD2 cause autosomal recessive hereditary spastic paraplegia (SPG54). European Journal of Human Genetics, 2013, 21, 1214-1218.	2.8	63
223	PLEKHG5 deficiency leads to an intermediate form of autosomal-recessive Charcotâ€“Marieâ€“Tooth disease. Human Molecular Genetics, 2013, 22, 4224-4232.	2.9	31
224	A Hereditary Spastic Paraplegia Mouse Model Supports a Role of ZFYVE26/SPASTIZIN for the Endolysosomal System. PLoS Genetics, 2013, 9, e1003988.	3.5	82
225	Prospective Analysis of Falls in Dominant Ataxias. European Neurology, 2013, 69, 53-57.	1.4	41
226	Teaching Neuro Images : MRI guides genetics. Neurology, 2013, 80, e176-7.	1.1	7
227	No association of GBA mutations and multiple system atrophy. European Journal of Neurology, 2013, 20, e61-2.	3.3	28
228	Sulphatation Does Not Appear to Be a Protective Mechanism to Prevent Oxysterol Accumulation in Humans and Mice. PLoS ONE, 2013, 8, e68031.	2.5	5
229	Spastic Paraplegia Mutation N256S in the Neuronal Microtubule Motor KIF5A Disrupts Axonal Transport in a Drosophila HSP Model. PLoS Genetics, 2012, 8, e1003066.	3.5	50
230	Mutant superoxide dismutase-1 indistinguishable from wild-type causes ALS. Human Molecular Genetics, 2012, 21, 3568-3574.	2.9	47
231	Video gameâ€“based coordinative training improves ataxia in children with degenerative ataxia. Neurology, 2012, 79, 2056-2060.	1.1	155
232	PET and MRI Reveal Early Evidence of Neurodegeneration in Spinocerebellar Ataxia Type 17. Journal of Nuclear Medicine, 2012, 53, 1074-1080.	5.0	54
233	Choreinâ€sensitive polymerization of cortical actin and suicidal cell death in choreaâ€acanthocytosis. FASEB Journal, 2012, 26, 1526-1534.	0.5	75
234	<scp>C</scp>harcotâ€“<scp>M</scp>arieâ€“<scp>T</scp>ooth hereditary neuropathy due to a mitochondrial <scp>ATP</scp>6 mutation. European Journal of Neurology, 2012, 19, e114-6.	3.3	17

#	Article	IF	Citations
235	Pathoanatomy of Cerebellar Degeneration in Spinocerebellar Ataxia Type 2 (SCA2) and Type 3 (SCA3). Cerebellum, 2012, 11, 749-760.	2.5	83
236	POLG and PEO1 (Twinkle) mutations are infrequent in PSP-like atypical parkinsonism: a preliminary screening study. Journal of Neurology, 2012, 259, 2232-2233.	3.6	0
237	D90A-SOD1 ALS mimicking monoclonal gammopathy-associated ALS. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 326-327.	2.1	0
238	A high-throughput resequencing microarray for autosomal dominant spastic paraplegia genes. Neurogenetics, 2012, 13, 215-227.	1.4	5
239	Screening in ALS and FTD patients reveals 3 novel UBQLN2 mutations outside the PXX domain and a pure FTD phenotype. Neurobiology of Aging, 2012, 33, 2949.e13-2949.e17.	3.1	86
240	Spinocerebellar ataxia type 1 (SCA1): new pathoanatomical and clinicoâ€pathological insights. Neuropathology and Applied Neurobiology, 2012, 38, 665-680.	3.2	66
241	Alteration of Fatty-Acid-Metabolizing Enzymes Affects Mitochondrial Form and Function in Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2012, 91, 1051-1064.	6.2	179
242	Characterizing POLG Ataxia: Clinics, Electrophysiology and Imaging. Cerebellum, 2012, 11, 1002-1011.	2.5	63
243	Retinal nerve fibre layer loss in hereditary spastic paraplegias is restricted to complex phenotypes. BMC Neurology, 2012, 12, 143.	1.8	20
244	Polymorphisms in the glial glutamate transporter SLC1A2 are associated with essential tremor. Neurology, 2012, 79, 243-248.	1.1	111
245	Spinocerebellar Ataxia Types 1, 2, 3 and 6: the Clinical Spectrum of Ataxia and Morphometric Brainstem and Cerebellar Findings. Cerebellum, 2012, 11, 155-166.	2.5	74
246	Mutation in the AP4B1 gene cause hereditary spastic paraplegia type 47 (SPG47). Neurogenetics, 2012, 13, 73-76.	1.4	52
247	Axonal transport deficit in a KIF5A â€"/â€" mouse model. Neurogenetics, 2012, 13, 169-179.	1.4	64
248	Disease severity affects quality of life of hereditary spastic paraplegia patients. European Journal of Neurology, 2012, 19, 168-171.	3.3	32
249	Mutations in the ER-shaping protein reticulon 2 cause the axon-degenerative disorder hereditary spastic paraplegia type 12. Journal of Clinical Investigation, 2012, 122, 538-544.	8.2	149
250	Increased [11C]PIB-PET levels in inclusion body myositis are indicative of amyloid Â deposition. Journal of Neurology, Neurosurgery and Psychiatry, 2011, 82, 1060-1062.	1.9	22
251	Acetazolamide-responsive exercise-induced episodic ataxia associated with a novel homozygous DARS2 mutation. Journal of Medical Genetics, 2011, 48, 713-715.	3.2	45
252	A Potential Role for a Genetic Variation of AKAP5 in Human Aggression and Anger Control. Frontiers in Human Neuroscience, 2011, 5, 175.	2.0	23

#	Article	IF	Citations
253	Mutations in the PDYN gene (SCA23) are not a frequent cause of dominant ataxia in Central Europe. Clinical Genetics, 2011, 80, 503-504.	2.0	10
254	Ampliconâ€based highâ€throughput pooled sequencing identifies mutations in CYP7B1 and SPG7 in sporadic spastic paraplegia patients. Clinical Genetics, 2011, 80, 148-160.	2.0	38
255	EFNS guidelines for the molecular diagnosis of neurogenetic disorders: motoneuron, peripheral nerve and muscle disorders. European Journal of Neurology, 2011, 18, 207-217.	3.3	29
256	Restless Legs and Substantia Nigra Hypoechogenicity are Common Features in Friedreichâ€™s Ataxia. Cerebellum, 2011, 10, 9-13.	2.5	15
257	Hepatic mitochondrial dysfunction in Friedreich Ataxia. BMC Neurology, 2011, 11, 145.	1.8	15
258	Atypical juvenile parkinsonism in a consanguineous SPG15 family. Movement Disorders, 2011, 26, 565-566.	3.9	26
259	Retinal nerve fiber layer loss in multiple system atrophy. Movement Disorders, 2011, 26, 914-916.	3.9	23
260	Depression comorbidity in spinocerebellar ataxia. Movement Disorders, 2011, 26, 870-876.	3.9	69
261	The natural history of spinocerebellar ataxia type 1, 2, 3, and 6. Neurology, 2011, 77, 1035-1041.	1.1	259
262	Genetics of Hereditary Spastic Paraplegias. Seminars in Neurology, 2011, 31, 484-493.	1.4	125
263	Paroxysmal choreoathetosis/spasticity (DYT9) is caused by a GLUT1 defect. Neurology, 2011, 77, 959-964.	1.1	114
264	Quantitative Assessment of Brain Stem and Cerebellar Atrophy in Spinocerebellar Ataxia Types 3 and 6: Impact on Clinical Status. American Journal of Neuroradiology, 2011, 32, 890-897.	2.4	56
265	Spinocerebellar ataxia type 15: diagnostic assessment, frequency, and phenotypic features. Journal of Medical Genetics, 2011, 48, 407-412.	3.2	49
266	Transcranial Sonography Reveals Cerebellar, Nigral, and Forebrain Abnormalities in Friedreichâ€™s Ataxia. Neurodegenerative Diseases, 2011, 8, 470-475.	1.4	21
267	Side Chain-oxidized Oxysterols Regulate the Brain Renin-Angiotensin System through a Liver X Receptor-dependent Mechanism. Journal of Biological Chemistry, 2011, 286, 25574-25585.	3.4	47
268	Severe orthostatic dysregulation associated with Wolfram syndrome. Journal of Neurology, 2010, 257, 1751-1753.	3.6	1
269	Comprehensive autonomic assessment does not differentiate between Parkinsonâ€™s disease, multiple system atrophy and progressive supranuclear palsy. Journal of Neural Transmission, 2010, 117, 69-76.	2.8	43
270	The cold hand sign in multiple system atrophy: skin perfusion revisited. Journal of Neural Transmission, 2010, 117, 475-479.	2.8	10

#	Article	IF	Citations
271	Falls in Spinocerebellar Ataxias: Results of the EuroSCA Fall Study. Cerebellum, 2010, 9, 232-239.	2.5	59
272	Leukodystrophies and other genetic metabolic leukoencephalopathies in children and adults. Brain and Development, 2010, 32, 82-89.	1.1	26
273	First appraisal of brain pathology owing to A30P mutant alphaâ€synuclein. Annals of Neurology, 2010, 67, 684-689.	5.3	91
274	Selfâ€rated health status in spinocerebellar ataxiaâ€"Results from a European multicenter study. Movement Disorders, 2010, 25, 587-595.	3.9	74
275	Baroreflex sensitivity and power spectral analysis during autonomic testing in different extrapyramidal syndromes. Movement Disorders, 2010, 25, 315-324.	3.9	36
276	Sustained dopaminergic response of parkinsonism and depression in POLGâ€associated parkinsonism. Movement Disorders, 2010, 25, 243-245.	3.9	26
277	Prevalence of THAP1 sequence variants in German patients with primary dystonia. Movement Disorders, 2010, 25, 1982-1986.	3.9	31
278	Longâ€term effects of coordinative training in degenerative cerebellar disease. Movement Disorders, 2010, 25, 2239-2246.	3.9	148
279	POLG, but not PEO1, is a frequent cause of cerebellar ataxia in Central Europe. Movement Disorders, 2010, 25, 2678-2682.	3.9	31
280	Complex hyperkinetic movement disorders associated with POLG mutations. Movement Disorders, 2010, 25, 2472-2475.	3.9	14
281	EFNS guidelines on the molecular diagnosis of ataxias and spastic paraplegias. European Journal of Neurology, 2010, 17, 179-188.	3.3	49
282	EFNS guidelines on the molecular diagnosis of channelopathies, epilepsies, migraine, stroke, and dementias. European Journal of Neurology, 2010, 17, 641-648.	3.3	10
283	A total of 220 patients with autosomal dominant spastic paraplegia do not display mutations in the SLC33A1 gene (SPG42). European Journal of Human Genetics, 2010, 18, 1065-1067.	2.8	31
284	Dissecting the role of the mitochondrial chaperone mortalin in Parkinson's disease: functional impact of disease-related variants on mitochondrial homeostasis. Human Molecular Genetics, 2010, 19, 4437-4452.	2.9	121
285	Responsiveness of different rating instruments in spinocerebellar ataxia patients. Neurology, 2010, 74, 678-684.	1.1	153
286	The Effects of COMT (Val108/158Met) and DRD4 (SNP -521) Dopamine Genotypes on Brain Activations Related to Valence and Magnitude of Rewards. Cerebral Cortex, 2010, 20, 1985-1996.	2.9	78
287	The human G93A SOD1 phenotype closely resembles sporadic amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, 764-767.	1.9	25
288	Marked accumulation of 27-hydroxycholesterol in SPG5 patients with hereditary spastic paresis. Journal of Lipid Research, 2010, 51, 819-823.	4.2	100

#	Article	IF	Citations
289	Neurophysiological markers of novelty processing are modulated by COMT and DRD4 genotypes. NeuroImage, 2010, 53, 962-969.	4.2	34
290	Visualization, quantification and correlation of brain atrophy with clinical symptoms in spinocerebellar ataxia types 1, 3 and 6. NeuroImage, 2010, 49, 158-168.	4.2	162
291	Spinocerebellar ataxia type 11 (SCA11) is an uncommon cause of dominant ataxia among French and German kindreds. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, 1229-1232.	1.9	47
292	KICK AND RUSH: PARADOXICAL KINESIA IN PARKINSON DISEASE. Neurology, 2009, 73, 328-329.	1.1	26
293	Ataxia with oculomotor apraxia type 2: clinical, biological and genotype/phenotype correlation study of a cohort of 90 patients. Brain, 2009, 132, 2688-2698.	7.6	218
294	ATAXIA WITH OPHTHALMOPLEGIA OR SENSORY NEUROPATHY IS FREQUENTLY CAUSED BY POLG MUTATIONS. Neurology, 2009, 73, 898-900.	1.1	69
295	Autosomal dominant spastic paraplegia with peripheral neuropathy maps to chr12q23-24. Neurology, 2009, 72, 1893-1898.	1.1	25
296	Frequency and phenotype of SPG11 and SPG15 in complicated hereditary spastic paraplegia. Journal of Neurology, Neurosurgery and Psychiatry, 2009, 80, 1402-1404.	1.9	66
297	Effective thalamic deep brain stimulation for neuropathic tremor in a patient with severe demyelinating neuropathy. Journal of Neurology, Neurosurgery and Psychiatry, 2009, 80, 235-236.	1.9	33
298	ADHD candidate gene (DRD4 exon III) affects inhibitory control in a healthy sample. BMC Neuroscience, 2009, 10, 150.	1.9	36
299	Comparison of three clinical rating scales in Friedreich ataxia (FRDA). Movement Disorders, 2009, 24, 1779-1784.	3.9	131
300	Loss of nocturnal blood pressure fall in various extrapyramidal syndromes. Movement Disorders, 2009, 24, 2136-2142.	3.9	95
301	Identification of a heterozygous genomic deletion in the spatacsin gene in SPG11 patients using high-resolution comparative genomic hybridization. Neurogenetics, 2009, 10, 43-48.	1.4	22
302	Analysis of CYP7B1 in non-consanguineous cases of hereditary spastic paraplegia. Neurogenetics, 2009, 10, 97-104.	1.4	54
303	Valsalva manoeuvre in patients with different Parkinsonian disorders. Journal of Neural Transmission, 2009, 116, 875-880.	2.8	40
304	Spinocerebellar ataxia type 6 (SCA6): neurodegeneration goes beyond the known brain predilection sites. Neuropathology and Applied Neurobiology, 2009, 35, 515-527.	3.2	44
305	Variant in the sequence of the LINGO1 gene confers risk of essential tremor. Nature Genetics, 2009, 41, 277-279.	21.4	211
306	EFNS guidelines on the molecular diagnosis of neurogenetic disorders: general issues, Huntingtonâ€™s disease, Parkinsonâ€™s disease and dystonias. European Journal of Neurology, 2009, 16, 777-785.	3.3	51

#	Article	IF	Citations
307	EFNS guidelines on the molecular diagnosis of mitochondrial disorders. European Journal of Neurology, 2009, 16, 1255-1264.	3.3	55
308	Diagnosis and treatment of Friedreich ataxia: a European perspective. Nature Reviews Neurology, 2009, 5, 222-234.	10.1	231
309	Genetic Variability in the Dopamine System (Dopamine Receptor D4, Catechol-O-Methyltransferase) Modulates Neurophysiological Responses to Gains and Losses. Biological Psychiatry, 2009, 66, 154-161.	1.3	82
310	Intensive coordinative training improves motor performance in degenerative cerebellar disease. Neurology, 2009, 73, 1823-1830.	1.1	282
311	SCA3: Neurological features, pathogenesis and animal models. Cerebellum, 2008, 7, 125-137.	2.5	208
312	Electrophysiology in spinocerebellar ataxias: Spread of disease and characteristic findings. Cerebellum, 2008, 7, 198-203.	2.5	42
313	Identification and characterisation of a large Senataxin (SETX) gene duplication in ataxia with ocular apraxia type 2 (AOA2). Neurogenetics, 2008, 9, 295-299.	1.4	21
314	Baroreflex sensitivity and power spectral analysis in different extrapyramidal syndromes. Journal of Neural Transmission, 2008, 115, 1527-1536.	2.8	43
315	Primary central nervous system vasculitis in a child mimicking parasitosis. Journal of Neurology, 2008, 255, 130-132.	3.6	3
316	â€œPseudodominant inheritanceâ€•of ataxia with ocular apraxia type 2 (AOA2). Journal of Neurology, 2008, 255, 495-501.	3.6	33
317	Spinal cord atrophy in spinocerebellar ataxia type 3 and 6. Journal of Neurology, 2008, 255, 1244-1249.	3.6	36
318	Autonomic dysfunction in different subtypes of multiple system atrophy. Movement Disorders, 2008, 23, 1766-1772.	3.9	34
319	Early symptoms in spinocerebellar ataxia type 1, 2, 3, and 6. Movement Disorders, 2008, 23, 2232-2238.	3.9	125
320	Spinocerebellar ataxia type 7 (SCA7): widespread brain damage in an adultâ€•onset patient with progressive visual impairments in comparison with an adultâ€•onset patient without visual impairments. Neuropathology and Applied Neurobiology, 2008, 34, 155-168.	3.2	33
321	Involvement of the auditory brainstem system in spinocerebellar ataxia type 2 (SCA2), type 3 (SCA3) and type 7 (SCA7). Neuropathology and Applied Neurobiology, 2008, 34, 479-491.	3.2	33
322	Screening of hereditary spastic paraplegia patients for alterations at NIPA1 mutational hotspots. Journal of the Neurological Sciences, 2008, 268, 131-135.	0.6	17
323	Spinocerebellar ataxia types 1, 2, 3, and 6. Neurology, 2008, 71, 982-989.	1.1	235
324	Effect of spastic paraplegia mutations in KIF5A kinesin on transport activity. Human Molecular Genetics, 2008, 17, 1245-1252.	2.9	134

#	Article	IF	Citations
325	REEP1 mutation spectrum and genotype/phenotype correlation in hereditary spastic paraplegia type 31. Brain, 2008, 131, 1078-1086.	7.6	163
326	SCA Functional Index. Neurology, 2008, 71, 486-492.	1.1	127
327	SPG10 is a rare cause of spastic paraplegia in European families. Journal of Neurology, Neurosurgery and Psychiatry, 2008, 79, 584-587.	1.9	45
328	Kick and rush. Neurology, 2008, 71, 695-695.	1.1	31
329	Friedreich's ataxia. Drugs of the Future, 2008, 33, 0607.	0.1	0
330	SCA3: Neurological features, pathogenesis and animal models. Cerebellum, 2008, 7, 1-13.	2.5	1
331	Electrophysiology in spinocerebellar ataxias: Spread of disease and characteristic findings. Cerebellum, 2008, 7, 1-6.	2.5	0
332	Chapter 3 Multiple System Atrophy. Blue Books of Neurology, 2007, , 78-112.	0.1	0
333	The Impact of Catechol-O-Methyltransferase and Dopamine D4 Receptor Genotypes on Neurophysiological Markers of Performance Monitoring. Journal of Neuroscience, 2007, 27, 14190-14198.	3.6	113
334	Further evidence for genetic heterogeneity of distal HMN type V, CMT2 with predominant hand involvement and Silver syndrome. Journal of the Neurological Sciences, 2007, 263, 100-106.	0.6	64
335	Longâ€term course and mutational spectrum of spatacsinâ€linked spastic paraplegia. Annals of Neurology, 2007, 62, 656-665.	5.3	113
336	Data protection in biomaterial banks for Parkinson's disease research: The model of GEPARD (Gene bank Parkinson's Disease Germany). Movement Disorders, 2007, 22, 611-618.	3.9	16
337	Cardiomyopathy in Friedreich's ataxia-assessment by cardiac MRI. Movement Disorders, 2007, 22, 1615-1622.	3.9	45
338	Pupil diameter in darkness differentiates progressive supranuclear palsy (PSP) from other extrapyramidal syndromes. Movement Disorders, 2007, 22, 2123-2126.	3.9	23
339	Repeat expansion in spinocerebellar ataxia type 17 alleles of the TATA-box binding protein gene: an evolutionary approach. European Journal of Human Genetics, 2007, 15, 81-87.	2.8	11
340	Consistent affection of the central somatosensory system in spinocerebellar ataxia type 2 and type 3 and its significance for clinical symptoms and rehabilitative therapy. Brain Research Reviews, 2007, 53, 235-249.	9.0	53
341	Late onset vanishing white matter disease. Journal of Neurology, 2007, 254, 544-545.	3.6	11
342	High frequency of partial SPAST deletions in autosomal dominant hereditary spastic paraplegia. Neurology, 2006, 67, 1926-1930.	1.1	133

#	Article	IF	Citations
343	Dissociation of grey and white matter reduction in spinocerebellar ataxia type 3 and 6: A voxel-based morphometry study. Neuroscience Letters, 2006, 408, 230-235.	2.1	81
344	Spinocerebellar Ataxia Type 3 (SCA3): Thalamic Neurodegeneration Occurs Independently from Thalamic Ataxin-3 Immunopositive Neuronal Intranuclear Inclusions. Brain Pathology, 2006, 16, 218-227.	4.1	61
345	Degeneration of ingestion-related brainstem nuclei in spinocerebellar ataxia type 2, 3, 6 and 7. Neuropathology and Applied Neurobiology, 2006, 32, 635-649.	3.2	82
346	Spectrin mutations in spinocerebellar ataxia (SCA). BioEssays, 2006, 28, 785-787.	2.5	15
347	Reliability and validity of the International Cooperative Ataxia Rating Scale: A study in 156 spinocerebellar ataxia patients. Movement Disorders, 2006, 21, 699-704.	3.9	150
348	Mutations inTITF1 are not relevant to sporadic and familial chorea of unknown cause. Movement Disorders, 2006, 21, 1734-1737.	3.9	15
349	CAG repeats in restless legs syndrome. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2006, 141B, 173-176.	1.7	25
350	Regular Insulin Secretory Oscillations Despite Impaired ATP Synthesis in Friedreich Ataxia Patients. Hormone and Metabolic Research, 2006, 38, 683-687.	1.5	4
351	The Spastic Paraplegia Rating Scale (SPRS): A reliable and valid measure of disease severity. Neurology, 2006, 67, 430-434.	1.1	247
352	Cerebrotendinous xanthomatosis. Neurology, 2006, 67, E20-E20.	1.1	1
353	Scale for the assessment and rating of ataxia. Neurology, 2006, 66, 1717-1720.	1.1	1,579
354	Extended pathoanatomical studies point to a consistent affection of the thalamus in spinocerebellar ataxia type 2. Neuropathology and Applied Neurobiology, 2005, 31, 127-140.	3.2	45
355	New mutations in protein kinase CÎ³ associated with spinocerebellar ataxia type 14. Annals of Neurology, 2005, 58, 720-729.	5.3	85
356	Mitochondrial impairment in patients and asymptomatic mutation carriers of Huntington's disease. Movement Disorders, 2005, 20, 674-679.	3.9	162
357	Alphaâ€synuclein and Parkinson's disease: Implications from the screening of more than 1,900 patients. Movement Disorders, 2005, 20, 1191-1194.	3.9	67
358	Involvement of the cranial nerves and their nuclei in spinocerebellar ataxia type 2 (SCA2). Acta Neuropathologica, 2005, 109, 617-631.	7.7	80
359	L-carnitine and creatine in Friedreichâ€™s ataxia. A randomized, placebo-controlled crossover trial. Journal of Neural Transmission, 2005, 112, 789-796.	2.8	48
360	Spinocerebellar ataxias types 2 and 3: degeneration of the precerebellar nuclei isolates the three phylogenetically defined regions of the cerebellum. Journal of Neural Transmission, 2005, 112, 1523-1545.	2.8	51

#	Article	IF	Citations
361	The fragile X tremor ataxia syndrome in the differential diagnosis of multiple system atrophy: data from the EMSA Study Group. Brain, 2005, 128, 1855-1860.	7.6	172
362	Spinocerebellar Ataxia Type 7 (SCA7): First Report of a Systematic Neuropathological Study of the Brain of a Patient with a Very Short Expanded CAGâ€Repeat. Brain Pathology, 2005, 15, 287-295.	4.1	39
363	Damage to the reticulotegmental nucleus of the pons in spinocerebellar ataxia type 1, 2, and 3. Neurology, 2004, 63, 1258-1263.	1.1	59
364	Senataxin, the ortholog of a yeast RNA helicase, is mutant in ataxia-ocular apraxia 2. Nature Genetics, 2004, 36, 225-227.	21.4	454
365	Autosomal dominant cerebellar ataxias: clinical features, genetics, and pathogenesis. Lancet Neurology, The, 2004, 3, 291-304.	10.2	963
366	Supracubital perineurioma misdiagnosed as carpal tunnel syndrome: case report. BMC Neurology, 2004, 4, 19.	1.8	4
367	Mutation at the SCA17 locus is not a common cause of primary dystonia. Journal of Neurology, 2004, 251, 1232-1234.	3.6	6
368	Novel homozygous p.E64D mutation in DJ1 in early onset Parkinson disease (PARK7). Human Mutation, 2004, 24, 321-329.	2.5	117
369	Lack of mutations in the epsilon-sarcoglycan gene in patients with different subtypes of primary dystonias. Movement Disorders, 2004, 19, 1294-1297.	3.9	11
370	Transcranial sonography in spinocerebellar ataxia type 3. Journal of Neural Transmission Supplementum, 2004, , 123-133.	0.5	34
371	Polymorphisms of the Î±-synuclein promoter: expression analyses and association studies in Parkinson's disease. Journal of Neural Transmission, 2003, 110, 67-76.	2.8	53
372	Modulation of disease risk according to a cathepsin D / apolipoprotein E genotype in Parkinson's disease. Journal of Neural Transmission, 2003, 110, 749-755.	2.8	16
373	Do CTG expansions at the SCA8 locus cause ataxia?. Annals of Neurology, 2003, 54, 110-115.	5.3	50
374	Clinical features and neuropathology of autosomal dominant spinocerebellar ataxia (SCA17). Annals of Neurology, 2003, 54, 367-375.	5.3	222
375	Mutation analysis of the neurofilament M gene in Parkinson's disease. Neuroscience Letters, 2003, 351, 125-129.	2.1	28
376	Identification and functional characterization of a novel R621C mutation in the synphilin-1 gene in Parkinson's disease. Human Molecular Genetics, 2003, 12, 1223-1231.	2.9	124
377	Prevalence of antigliadin antibodies in ataxia patients. Neurology, 2003, 60, 1674-1675.	1.1	71
378	Frequency and Phenotypic Variability of the GAG Deletion of the DYT1 Gene in an Unselected Group of Patients With Dystonia. Archives of Neurology, 2003, 60, 1266-70.	4.5	79

#	Article	IF	Citations
379	Neurophysiological findings in SPG4 patients differ from other types of spastic paraplegia. Neurology, 2003, 60, 1529-1532.	1.1	35
380	The aetiology of sporadic adultâ€onset ataxia. Brain, 2002, 125, 961-968.	7.6	228
381	Vertebral body infarction indicating midthoracic spinal stroke. Spinal Cord, 2002, 40, 244-247.	1.9	25
382	Motor cortex activation by transcranial magnetic stimulation in ataxia patients depends on the genetic defect. Brain, 2002, 125, 301-309.	7.6	87
383	Polymorphisms in the interleukin-1 alpha and beta genes and the risk for Parkinson's disease. Neuroscience Letters, 2002, 326, 70-72.	2.1	96
384	Neurofilament L gene is not a genetic factor of sporadic and familial Parkinsonâ€™s disease. Brain Research, 2002, 951, 82-86.	2.2	16
385	Protein surveillance machinery in brains with spinocerebellar ataxia type 3: Redistribution and differential recruitment of 26S proteasome subunits and chaperones to neuronal intranuclear inclusions. Annals of Neurology, 2002, 51, 302-310.	5.3	133
386	Mutation analysis of thespastingene (SPG4) in patients in Germany with autosomal dominant hereditary spastic paraplegia. Human Mutation, 2002, 20, 127-132.	2.5	92
387	Mutation analysis and association studies of nuclear factor-Î²B1 in sporadic Parkinson's disease patients. Journal of Neural Transmission, 2002, 109, 1181-1188.	2.8	12
388	Idebenone in patients with Friedreich ataxia. Neuroscience Letters, 2001, 306, 169-172.	2.1	90
389	Evaluation of the Î³-synuclein gene in German Parkinson's disease patients. Neuroscience Letters, 2001, 310, 191-193.	2.1	16
390	Association of Moderate Polyglutamine Tract Expansions in the Slow Calcium-Activated Potassium Channel Type 3 With Ataxia. Archives of Neurology, 2001, 58, 1649.	4.5	24
391	The GAA repeat expansion in intron 1 of the frataxin gene is related to the severity of cardiac manifestation in patients with Friedreich's ataxia. Journal of Molecular Medicine, 2001, 78, 626-632.	3.9	46
392	Double-blind Crossover Trial of Trimethoprim-Sulfamethoxazole in Spinocerebellar Ataxia Type 3/Machado-Joseph Disease. Archives of Neurology, 2001, 58, 1451.	4.5	46
393	Familial parkinsonism with synuclein pathology. Neurology, 2001, 56, 1355-1362.	1.1	146
394	Extrapyramidal Motor Signs in Degenerative Ataxias. Archives of Neurology, 2000, 57, 1495-500.	4.5	76
395	Genetic analysis of the Î±2-macroglobulin gene in early-and late-onset ParkinsonÊ¼s disease. NeuroReport, 2000, 11, 2439-2442.	1.2	16
396	Mutation analysis and association studies of the UCHL1 gene in German ParkinsonÊ¼s disease patients. NeuroReport, 2000, 11, 2079-2082.	1.2	143

#	Article	IF	Citations
397	Is the sural nerve a pure sensory nerve?. Muscle and Nerve, 2000, 23, 1895-1895.	2.2	2
398	The dopamine D2 receptor (DRD2) gene-a genetic risk factor in heavy smoking?. Addiction Biology, 2000, 5, 429-436.	2.6	23
399	Genetic analysis of immunomodulating factors in sporadic Parkinson's disease. Journal of Neural Transmission, 2000, 107, 553-562.	2.8	86
400	Genetic background of apparently idiopathic sporadic cerebellar ataxia. Human Genetics, 2000, 107, 132-137.	3.8	128
401	Oxidative stress in patients with Friedreich ataxia. Neurology, 2000, 55, 1719-1721.	1.1	312
402	Heterozygous expansion of the GAA tract of the X25/frataxin gene is associated with insulin resistance in humans. Diabetes, 2000, 49, 1604-1607.	0.6	20
403	Mitochondrial impairment of human muscle in Friedreich ataxia in vivo. Neuromuscular Disorders, 2000, 10, 430-435.	0.6	87
404	Age related axonal neuropathy in spinocerebellar ataxia type 3/Machado-Joseph disease (SCA3/MJD). Journal of Neurology, Neurosurgery and Psychiatry, 1999, 66, 222-224.	1.9	64
405	Analysis of the parkin deletion in sporadic and familial Parkinson's disease. Journal of Neural Transmission, 1999, 106, 159-163.	2.8	16
406	Increased susceptibility to sporadic Parkinson's disease by a certain combined ?-synuclein/apolipoprotein E genotype. Annals of Neurology, 1999, 45, 611-617.	5.3	273
407	An Isoform of Ataxinâ€3 Accumulates in the Nucleus of Neuronal Cells in Affected Brain Regions of SCA3 Patients. Brain Pathology, 1998, 8, 669-679.	4.1	189
408	AlaSOPro mutation in the gene encoding Î±-synuclein in Parkinson's disease. Nature Genetics, 1998, 18, 106-108.	21.4	3,711
409	Collision technique in Martin-Gruber anastomosis. , 1998, 21, 1354-1355.		3
410	Sleep disturbance in spinocerebellar ataxias. Neurology, 1998, 51, 1603-1607.	1.1	172
411	Spinocerebellar ataxia type 6: genotype and phenotype in German kindreds. Journal of Neurology, Neurosurgery and Psychiatry, 1998, 64, 67-73.	1.9	167
412	The natural history of degenerative ataxia: a retrospective study in 466 patients. Brain, 1998, 121, 589-600.	7.6	316
413	Spinocerebellar Ataxia Type 2. Archives of Neurology, 1997, 54, 1073.	4.5	161
414	Motor fibers in the sural nerve of humans. Neurology, 1997, 49, 1725-1728.	1.1	43

#	Article	IF	Citations
415	SCA6 is caused by moderate CAG expansion in the alpha1A-voltage- dependent calcium channel gene. Human Molecular Genetics, 1997, 6, 1289-1293.	2.9	104
416	Friedreich's ataxia. Revision of the phenotype according to molecular genetics. Brain, 1997, 120, 2131-2140.	7.6	147
417	Transmission distortion of the mutant alleles in spinocerebellar ataxia. Human Genetics, 1997, 99, 282-284.	3.8	33
418	Polymorphisms in the PMP-22 gene region (17p11.2-12) are crucial for simplified diagnosis of duplications/deletions. Human Genetics, 1997, 99, 688-691.	3.8	20
419	Differential stability of the (GAA) n tract in the Friedreich ataxia (STM7) gene. Human Genetics, 1997, 99, 834-836.	3.8	125
420	SCA2 trinucleotide expansion in German SCA patients. Neurogenetics, 1997, 1, 59-64.	1.4	98
421	Autosomal dominant cerebellar ataxia: Phenotypic differences in genetically defined subtypes?. Annals of Neurology, 1997, 42, 924-932.	5.3	298
422	Motor evoked potentials in the spinocerebellar ataxias type 1 and type 3. , 1997, 20, 226-228.		17
423	EXTENSOR DIGITORUM BREVIS INNERVATED BY THE TIBIAL NERVE ("ALL TIBIAL FOOT"): ANOMALOUS INNERVATION OR TECHNICAL PITFALL? (A REPLY). Muscle and Nerve, 1997, 20, 907-908.	2.2	1
424	Repeat length and disease progression in spinocerebellar ataxia type 3. Lancet, The, 1996, 348, 830.	13.7	21
425	Mitochondrial disorders in degenerative ataxias. European Journal of Neurology, 1996, 3, 55-60.	3.3	9
426	Letters to the editor. Muscle and Nerve, 1996, 19, 1227-1231.	2.2	6
427	Relations between genotype and phenotype in German patients with the Machado-Joseph disease mutation.. Journal of Neurology, Neurosurgery and Psychiatry, 1996, 61, 466-470.	1.9	73
428	Reversible urinary retention as the main symptom in the first manifestation of a syringomyelia.. Journal of Neurology, Neurosurgery and Psychiatry, 1996, 61, 407-408.	1.9	8
429	Head tremor due to bilateral thalamic and midbrain infarction. Journal of Neurology, 1995, 242, 608-611.	3.6	19
430	Machado-Joseph disease mutations as the genetic basis of most spinocerebellar ataxias in Germany.. Journal of Neurology, Neurosurgery and Psychiatry, 1995, 59, 449-450.	1.9	31
431	Trinucleotide expansion within the MJD1 gene presents clinically as spinocerebellar ataxia and occurs most frequently in German SCA patients. Human Molecular Genetics, 1995, 4, 1001-1005.	2.9	124
432	Spinocerebellar ataxia type 1: clinical and neurophysiological characteristics in German kindreds. Acta Neurologica Scandinavica, 1995, 92, 478-485.	2.1	28

#	Article	IF	Citations
433	Unaltered Respiratory Chain Enzyme Activity and Mitochondrial DNA in Skeletal Muscle from Patients with Idiopathic Parkinsonâ€™s Syndrome. European Neurology, 1994, 34, 263-267.	1.4	26
434	Continuous subcutaneous lisuride infusion in OPCA. Journal of Neural Transmission, 1992, 90, 145-150.	2.8	2
435	Reestablishment of the heterogeneous distribution of hepatic glutamine synthetase during regeneration after CCl4-intoxication. Histochemistry, 1990, 94, 49-54.	1.9	40