Ludger SchÃ¶ls

List of Publications by Year
in descending order

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435	30,270	5782	8627
papers	citations	84 h-index	151 g-index

460	460	460	24282
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.	3.7	6
2	Characterization of Lifestyle in Spinocerebellar Ataxia Type 3 and Association with Disease Severity. Movement Disorders, 2022, 37, 405-410.	2.2	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.	2.3	15
4	<scp>BCAS3</scp>â€Related Neurodevelopmental Disorder Shows Magnetic Resonance Imaging Features Resembling Brain Iron Accumulation. Movement Disorders, 2022, 37, 870-872.	2.2	0
5	Realâ€Life Turning Movements Capture Subtle Longitudinal and Preataxic Changes in Cerebellar Ataxia. Movement Disorders, 2022, 37, 1047-1058.	2.2	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.	1.7	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.	1.2	10
8	Levels of Neurofilament Light at the Preataxic and Ataxic Stages of Spinocerebellar Ataxia Type 1. Neurology, 2022, 98, .	1.5	15
9	Tau and neurofilament lightâ€chain as fluid biomarkers in spinocerebellar ataxia type 3. European Journal of Neurology, 2022, 29, 2439-2452.	1.7	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.	1.8	1
11	Mitochondrial Dysfunction in Spinocerebellar Ataxia Type 3 Is Linked to VDAC1 Deubiquitination. International Journal of Molecular Sciences, 2022, 23, 5933.	1.8	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.	2.0	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.	1.7	12
14	Differential Temporal Dynamics of Axial and Appendicular Ataxia in <scp>SCA3</scp>. Movement Disorders, 2022, 37, 1850-1860.	2.2	11
15	Association of Age at Onset and First Symptoms With Disease Progression in Patients With Metachromatic Leukodystrophy. Neurology, 2021, 96, e255-e266.	1.5	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.	1.8	15
17	Natural History, Phenotypic Spectrum, and Discriminative Features of Multisystemic RFC1 Disease. Neurology, 2021, 96, e1369-e1382.	1.5	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.	1.1	2

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19	Spinocerebellar ataxia type 14: refining clinicogenetic diagnosis in a rare adultâ€onset disorder. Annals of Clinical and Translational Neurology, 2021, 8, 774-789.	1.7	13
20	Neurofilament light chain is a cerebrospinal fluid biomarker in hereditary spastic paraplegia. Annals of Clinical and Translational Neurology, 2021, 8, 1122-1131.	1.7	11
21	Biallelic loss-of-function variations in PRDX3 cause cerebellar ataxia. Brain, 2021, 144, 1467-1481.	3.7	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.	2.6	14
23	Solving unsolved rare neurological diseasesâ€"a Solve-RD viewpoint. European Journal of Human Genetics, 2021, 29, 1332-1336.	1.4	4
24	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	3.7	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.	4.9	53
26	Developmental Consequences of Defective ATG7-Mediated Autophagy in Humans. New England Journal of Medicine, 2021, 384, 2406-2417.	13.9	84
27	Bi-allelic loss-of-function variants in BCAS3 cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2021, 108, 1069-1082.	2.6	8
28	The ARCA Registry: A Collaborative Global Platform for Advancing Trial Readiness in Autosomal Recessive Cerebellar Ataxias. Frontiers in Neurology, 2021, 12, 677551.	1.1	15
29	Natural History of Polymerase Gammaâ€"Related Ataxia. Movement Disorders, 2021, 36, 2642-2652.	2.2	10
30	Polyglutamineâ€Expanded Ataxinâ€3: A Target Engagement Marker for Spinocerebellar Ataxia Type 3 in Peripheral Blood. Movement Disorders, 2021, 36, 2675-2681.	2.2	22
31	Bi-allelic truncating mutations in VWA1 cause neuromyopathy. Brain, 2021, 144, 574-583.	3.7	16
32	Metabolic profiling in serum, cerebrospinal fluid, and brain of patients with cerebrotendinous xanthomatosis. Journal of Lipid Research, 2021, 62, 100078.	2.0	14
33	Fampridine and Acetazolamide in EA2 and Related Familial EA. Neurology: Clinical Practice, 2021, 11, e438-e446.	0.8	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, .	3.3	5
35	Safety and Efficacy of Acetyl-DL-Leucine in Certain Types of Cerebellar Ataxia. JAMA Network Open, 2021, 4, e2135841.	2.8	16
36	Non-motor symptoms are relevant and possibly treatable in hereditary spastic paraplegia type 4 (SPG4). Journal of Neurology, 2020, 267, 369-379.	1.8	11

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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.	1.4	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.	0.6	5
39	Onset features and time to diagnosis in Friedreichâ€™s Ataxia. Orphanet Journal of Rare Diseases, 2020, 15, 198.	1.2	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.	1.1	30
41	Generation of the CRISPR/Cas9-mediated KIF1C knock-out human iPSC line HIHRSi003-A-1. Stem Cell Research, 2020, 49, 102059.	0.3	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.	1.6	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.	1.8	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.	1.6	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.	1.2	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.	4.9	41
47	Multifocal, hypoechogenic nerve thickening in Cerebrotendinous Xanthomatosis. Clinical Neurophysiology, 2020, 131, 1798-1803.	0.7	2
48	Delineating MT-ATP6 -associated disease. Neurology: Genetics, 2020, 6, e393.	0.9	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.	1.4	20
50	Real-life gait assessment in degenerative cerebellar ataxia. Neurology, 2020, 95, e1199-e1210.	1.5	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.	3.3	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.	2.8	52
53	ATPase Domain <scp>AFG3L2</scp> Mutations Alter <scp>OPA1</scp> Processing and Cause Optic Neuropathy. Annals of Neurology, 2020, 88, 18-32.	2.8	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.	1.1	15

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55	The European Reference Network for Rare Neurological Diseases. Frontiers in Neurology, 2020, 11, 616569.	1.1	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.	1.8	17
58	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	5.8	35
59	Decreased Na+/K+ ATPase Expression and Depolarized Cell Membrane in Neurons Differentiated from Chorea-Acanthocytosis Patients. Scientific Reports, 2020, 10, 8391.	1.6	9
60	CHIP mutations affect the heat shock response differently in human fibroblasts and iPSC-derived neurons. DMM Disease Models and Mechanisms, 2020, 13, .	1.2	5
61	Confirmation of TACO1 as a Leigh Syndrome Disease Gene in Two Additional Families. Journal of Neuromuscular Diseases, 2020, 7, 301-308.	1.1	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.	0.9	33
63	Brain MRI features and scoring of leukodystrophy in adult-onset Krabbe disease. Neurology, 2019, 93, e647-e652.	1.5	25
64	Multiparametric rapid screening of neuronal process pathology for drug target identification in HSP patient-specific neurons. Scientific Reports, 2019, 9, 9615.	1.6	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.	1.0	14
66	Application of Quantitative Motor Assessments in Friedreich Ataxia and Evaluation of Their Relation to Clinical Measures. Cerebellum, 2019, 18, 896-909.	1.4	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.	1.7	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.	1.0	13
69	Prediction of Survival With Longâ€Term Disease Progression in Most Common Spinocerebellar Ataxia. Movement Disorders, 2019, 34, 1220-1227.	2.2	14
70	Phenotypic variation between siblings with Metachromatic Leukodystrophy. Orphanet Journal of Rare Diseases, 2019, 14, 136.	1.2	29
71	Loss of paraplegin drives spasticity rather than ataxia in a cohort of 241 patients with SPG7. Neurology, 2019, 92, e2679-e2690.	1.5	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.	0.7	9

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73	Biallelic variants in the transcription factor PAX7 are a new genetic cause of myopathy. Genetics in Medicine, 2019, 21, 2521-2531.	1.1	25
74	FAHN/SPG35: a narrow phenotypic spectrum across disease classifications. Brain, 2019, 142, 1561-1572.	3.7	70
75	Patientâ€reported outcomes in Friedreichâ€™s ataxia after withdrawal from idebenone. Acta Neurologica Scandinavica, 2019, 139, 533-539.	1.0	17
76	Speech treatment improves dysarthria in multisystemic ataxia: a rater-blinded, controlled pilot-study in ARSACS. Journal of Neurology, 2019, 266, 1260-1266.	1.8	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.	1.7	7
78	Pattern of Cerebellar Atrophy in Friedreichâ€™s Ataxiaâ€”Using the SUIT Template. Cerebellum, 2019, 18, 435-447.	1.4	23
79	Autosomal Recessive Cerebellar Ataxias: Paving the Way toward Targeted Molecular Therapies. Neuron, 2019, 101, 560-583.	3.8	83
80	mRNA as a Novel Treatment Strategy for Hereditary Spastic Paraplegia Type 5. Molecular Therapy - Methods and Clinical Development, 2019, 15, 359-370.	1.8	23
81	Generation of a homozygous CRISPR/Cas9-mediated knockout human iPSC line for the STUB1 locus. Stem Cell Research, 2019, 34, 101378.	0.3	7
82	A previously identified missense mutation in STYXL1 is likely benign. European Journal of Medical Genetics, 2019, 62, 103582.	0.7	2
83	TSFM mutations cause a complex hyperkinetic movement disorder with strong relief by cannabinoids. Parkinsonism and Related Disorders, 2019, 60, 176-178.	1.1	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.	1.1	18
85	Mechanistic basis of an epistatic interaction reducing age at onset in hereditary spastic paraplegia. Brain, 2018, 141, 1286-1299.	3.7	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.	0.3	5
87	Consensus Paper: Neurophysiological Assessments of Ataxias in Daily Practice. Cerebellum, 2018, 17, 628-653.	1.4	30
88	Comprehensive systematic review summary: Treatment of cerebellar motor dysfunction and ataxia. Neurology, 2018, 90, 464-471.	1.5	108
89	To die or not to die SGK1-sensitive ORAI/STIM in cell survival. Cell Calcium, 2018, 74, 29-34.	1.1	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.	4.9	69

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91	Treatable inherited rare movement disorders. Movement Disorders, 2018, 33, 21-35.	2.2	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.	1.5	28
93	Case series: Slowing alpha rhythm in late-stage ALS patients. Clinical Neurophysiology, 2018, 129, 406-408.	0.7	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.	1.1	9
95	Nerve ultrasound characterizes AMN polyneuropathy as inhomogeneous and focal hypertrophic. Orphanet Journal of Rare Diseases, 2018, 13, 194.	1.2	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.	1.4	32
97	NfL is a biomarker for adult-onset leukoencephalopathy with axonal spheroids and pigmented glia. Neurology, 2018, 91, 755-757.	1.5	11
98	Long-term evolution of patient-reported outcome measures in spinocerebellar ataxias. Journal of Neurology, 2018, 265, 2040-2051.	1.8	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.	0.3	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.	1.8	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.	0.3	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.	1.4	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.	1.8	58
104	Nonataxia symptoms in Friedreich Ataxia. Neurology, 2018, 91, e917-e930.	1.5	46
105	Serum neurofilament light chain is increased in hereditary spastic paraplegias. Annals of Clinical and Translational Neurology, 2018, 5, 876-882.	1.7	26
106	GPT2 mutations cause developmental encephalopathy with microcephaly and features of complicated hereditary spastic paraplegia. Clinical Genetics, 2018, 94, 356-361.	1.0	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.	0.8	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.	1.2	56

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109	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578.	3.7	85
110	Involvement of the cerebellum in Parkinson disease and dementia with Lewy bodies. Annals of Neurology, 2017, 81, 898-903.	2.8	44
111	CNTNAP1 mutations cause CNS hypomyelination and neuropathy with or without arthrogryposis. Neurology: Genetics, 2017, 3, e144.	0.9	24
112	Loss-of-function mutations in theATP13A2/PARK9 gene cause complicated hereditary spastic paraplegia (SPG78). Brain, 2017, 140, 287-305.	3.7	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.	0.6	23
114	A combinatorial approach to identify calpain cleavage sites in the Machado-Joseph disease protein ataxin-3. Brain, 2017, 140, 1280-1299.	3.7	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.	1.1	45
116	TBK1 Mutation Spectrum in an Extended European Patient Cohort with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Human Mutation, 2017, 38, 297-309.	1.1	87
117	Neurochondrin is a neuronal target antigen in autoimmune cerebellar degeneration. Neurology: Neuroimmunology and NeuroInflammation, 2017, 4, e307.	3.1	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.	0.3	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.	1.1	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.	2.1	68
121	Body Mass Index Decline Is Related to Spinocerebellar Ataxia Disease Progression. Movement Disorders Clinical Practice, 2017, 4, 689-697.	0.8	25
122	Speech and swallowing abnormalities in adults with POLG associated ataxia (POLG-A). Mitochondrion, 2017, 37, 1-7.	1.6	20
123	Lithium Sensitive ORAI1 Expression, Store Operated Ca2+ Entry and Suicidal Death of Neurons in Chorea-Acanthocytosis. Scientific Reports, 2017, 7, 6457.	1.6	31
124	Enzymatic characterization of novel arylsulfatase A variants using human arylsulfatase Aâ€deficient immortalized mesenchymal stromal cells. Human Mutation, 2017, 38, 1511-1520.	1.1	20
125	Clinical and genetic characteristics of sporadic adult-onset degenerative ataxia. Neurology, 2017, 89, 1043-1049.	1.5	45
126	Neurons, Erythrocytes and Beyond â€“The Diverse Functions of Chorein. NeuroSignals, 2017, 25, 117-126.	0.5	17

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127	Hereditary spastic paraplegia type 5: natural history, biomarkers and a randomized controlled trial. Brain, 2017, 140, 3112-3127.	3.7	87
128	Absence of EEG correlates of self-referential processing depth in ALS. PLoS ONE, 2017, 12, e0180136.	1.1	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.	1.2	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.	1.4	41
131	Hereditary spastic paraplegia: Clinicogenetic lessons from 608 patients. Annals of Neurology, 2016, 79, 646-658.	2.8	218
132	Individual changes in preclinical spinocerebellar ataxia identified via increased motor complexity. Movement Disorders, 2016, 31, 1891-1900.	2.2	54
133	MultisystemicSYNE1ataxia: confirming the high frequency and extending the mutational and phenotypic spectrum. Brain, 2016, 139, e46-e46.	3.7	40
134	SYNE1 ataxia is a common recessive ataxia with major non-cerebellar features: a large multi-centre study. Brain, 2016, 139, 1378-1393.	3.7	87
135	Complicated spastic paraplegia in patients with AP5Z1 mutations (SPG48). Neurology: Genetics, 2016, 2, e98.	0.9	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.	0.3	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.	0.3	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.	0.3	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.	0.3	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.	0.3	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.	0.7	19
142	Inducing Differentiation of Premalignant Hepatic Cells as a Novel Therapeutic Strategy in Hepatocarcinoma. Cancer Research, 2016, 76, 5550-5561.	0.4	15
143	Thickening of the peripheral nerves in metachromatic leukodystrophy. Journal of the Neurological Sciences, 2016, 368, 399-401.	0.3	12
144	A recurrent mutation in KCNA2 as a novel cause of hereditary spastic paraplegia and ataxia. Annals of Neurology, 2016, 80, .	2.8	49

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145	Antisense Oligonucleotide Mediated Splice Correction of a Deep Intronic Mutation in OPA1. Molecular Therapy - Nucleic Acids, 2016, 5, e390.	2.3	43
146	A cognitive brainâ€“computer interface for patients with amyotrophic lateral sclerosis. Progress in Brain Research, 2016, 228, 221-239.	0.9	9
147	Uniparental disomy of chromosome 16 unmasks recessive mutations of FA2H /SPG35 in 4 families. Neurology, 2016, 87, 186-191.	1.5	27
148	Leukoencephalopathy With Axonal Spheroids and Pigmented Glia. JAMA Neurology, 2016, 73, 1400.	4.5	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.	4.9	117
150	M4â€…Safety and tolerability of BN82451B in huntingtonâ€™s disease. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, A102.2-A103.	0.9	0
151	Standardized Assessment of Hereditary Ataxia Patients in Clinical Studies. Movement Disorders Clinical Practice, 2016, 3, 230-240.	0.8	13
152	Identifying Niemannâ€“Pick type C in early-onset ataxia: two quick clinical screening tools. Journal of Neurology, 2016, 263, 1911-1918.	1.8	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.	0.4	5
154	Reply. Annals of Neurology, 2016, 80, 170-171.	2.8	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.	1.8	77
156	Peripheral Neuropathy in Spinocerebellar Ataxia Type 1, 2, 3, and 6. Cerebellum, 2016, 15, 165-173.	1.4	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.	1.2	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.	1.8	18
159	A randomized, placeboâ€controlled trial of AFQ056 for the treatment of chorea in Huntington's disease. Movement Disorders, 2015, 30, 427-431.	2.2	67
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