Matthis Synofzik

List of Publications by Year
in descending order

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290 papers	11,985 citations	30070 54 h-index	45317 90 g-index
317 all docs	317 docs citations	317 times ranked	13945 citing authors

#	Article	IF	Citations
1	Network structure and transcriptomic vulnerability shape atrophy in frontotemporal dementia. Brain, 2023, 146, 321-336.	7.6	30
2	The prodromal phase of hereditary spastic paraplegia type 4: the preSPG4 cohort study. Brain, 2023, 146, 1093-1102.	7.6	6
3	A modified Camel and Cactus Test detects presymptomatic semantic impairment in genetic frontotemporal dementia within the GENFI cohort. Applied Neuropsychology Adult, 2022, 29, 112-119.	1.2	18
4	Comparison of clinical rating scales in genetic frontotemporal dementia within the GENFI cohort. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, 158-168.	1.9	7
5	Preparing n-of-1 Antisense Oligonucleotide Treatments for Rare Neurological Diseases in Europe: Genetic, Regulatory, and Ethical Perspectives. Nucleic Acid Therapeutics, 2022, 32, 83-94.	3.6	35
6	A data-driven disease progression model of fluid biomarkers in genetic frontotemporal dementia. Brain, 2022, 145, 1805-1817.	7.6	27
7	Characterization of Lifestyle in Spinocerebellar Ataxia Type 3 and Association with Disease Severity. Movement Disorders, 2022, 37, 405-410.	3.9	8
8	Effects of exergaming on hippocampal volume and brainâ€derived neurotrophic factor levels in Parkinsonâ€™s disease. European Journal of Neurology, 2022, 29, 441-449.	3.3	17
9	Stratifying the Presymptomatic Phase of Genetic Frontotemporal Dementia by Serum <scp>NfL</scp> and <scp>pNfH</scp>: A Longitudinal Multicentre Study. Annals of Neurology, 2022, 91, 33-47.	5.3	21
10	Sensory axonal neuropathy in RFC1-disease: tip of the iceberg of broad subclinical multisystemic neurodegeneration. Brain, 2022, 145, e6-e9.	7.6	6
11	Cognitive composites for genetic frontotemporal dementia: GENFI-Cog. Alzheimer's Research and Therapy, 2022, 14, 10.	6.2	4
12	An Automated Toolbox to Predict Single Subject Atrophy in Presymptomatic Granulin Mutation Carriers. Journal of Alzheimer's Disease, 2022, , 1-14.	2.6	3
13	Soluble TAM receptors sAXL and sTyro3 predict structural and functional protection in Alzheimerâ€™s disease. Neuron, 2022, 110, 1009-1022.e4.	8.1	27
14	Cerebellar Bottom of Fissure Hyperintensities in <scp>MTâ€ATP6</scp>â€Associated Ataxia. Annals of Neurology, 2022, 91, 438-440.	5.3	1
15	Realâ€Life Turning Movements Capture Subtle Longitudinal and Preataxic Changes in Cerebellar Ataxia. Movement Disorders, 2022, 37, 1047-1058.	3.9	24
16	Examining empathy deficits across familial forms of frontotemporal dementia within the GENFI cohort. Cortex, 2022, 150, 12-28.	2.4	2
17	Dataâ€driven staging of genetic frontotemporal dementia using multiâ€modal <scp>MRI</scp>. Human Brain Mapping, 2022, 43, 1821-1835.	3.6	7
18	De Novo and Dominantly Inherited <scp>SPTAN1</scp> Mutations Cause Spastic Paraplegia and Cerebellar Ataxia. Movement Disorders, 2022, 37, 1175-1186.	3.9	9

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19	Conceptual framework for the definition of preclinical and prodromal frontotemporal dementia. Alzheimer's and Dementia, 2022, 18, 1408-1423.	0.8	24
20	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
21	Structural brain splitting is a hallmark of Granulin-related frontotemporal dementia. Neurobiology of Aging, 2022, , .	3.1	1
22	Anomia is present pre-symptomatically in frontotemporal dementia due to MAPT mutations. Journal of Neurology, 2022, 269, 4322-4332.	3.6	1
23	The <scp>CBIâ€R</scp> detects early behavioural impairment in genetic frontotemporal dementia. Annals of Clinical and Translational Neurology, 2022, 9, 644-658.	3.7	1
24	RFC1 repeat expansions: A recurrent cause of sensory and autonomic neuropathy with cough and ataxia. European Journal of Neurology, 2022, 29, 2156-2161.	3.3	14
25	Development of a sensitive trial-ready poly(GP) CSF biomarker assay forC9orf72-associated frontotemporal dementia and amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, 761-771.	1.9	12
26	The CCAS-scale in hereditary ataxias: helpful on the group level, particularly in SCA3, but limited in individual patients. Journal of Neurology, 2022, 269, 4363-4374.	3.6	13
27	Consensus Paper: Ataxic Gait. Cerebellum, 2022, , 1.	2.5	9
28	Levels of Neurofilament Light at the Preataxic and Ataxic Stages of Spinocerebellar Ataxia Type 1. Neurology, 2022, 98, .	1.1	15
29	Paving the Way Toward Meaningful Trials in Ataxias: An Ataxia Global Initiative Perspective. Movement Disorders, 2022, 37, 1125-1130.	3.9	21
30	Tau and neurofilament lightâ€chain as fluid biomarkers in spinocerebellar ataxia type 3. European Journal of Neurology, 2022, 29, 2439-2452.	3.3	25
31	Autosomal Recessive Cerebellar Ataxias in South America: A Multicenter Study of 1338 Patients. Movement Disorders, 2022, 37, 1773-1774.	3.9	12
32	Comparative analysis of machine learning algorithms for multi-syndrome classification of neurodegenerative syndromes. Alzheimer's Research and Therapy, 2022, 14, 62.	6.2	9
33	Longitudinal Cognitive Changes in Genetic Frontotemporal Dementia Within the GENFI Cohort. Neurology, 2022, 99, .	1.1	5
34	Recommendations for whole genome sequencing in diagnostics for rare diseases. European Journal of Human Genetics, 2022, 30, 1017-1021.	2.8	48
35	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
36	Adultâ€Onset Neurodegeneration in Nucleotide Excision Repair Disorders (<scp>NERD_{ND}</scp>): Time to Move Beyond the Skin. Movement Disorders, 2022, 37, 1707-1718.	3.9	7

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37	Differential Temporal Dynamics of Axial and Appendicular Ataxia in <scp>SCA3</scp>. Movement Disorders, 2022, 37, 1850-1860.	3.9	11
38	Expanding PRDX3 disease: broad range of onset age and infratentorial MRI signal changes. Brain, 2022, 145, e95-e98.	7.6	3
39	Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in POLR3A, POLR3B, and POLR1C. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e660-e674.	3.6	26
40	Fluid biomarkers in frontotemporal dementia: past, present and future. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 204-215.	1.9	62
41	Are intrinsic neural timescales related to sensory processing? Evidence from abnormal behavioral states. NeuroImage, 2021, 226, 117579.	4.2	60
42	Brain functional network integrity sustains cognitive function despite atrophy in presymptomatic genetic frontotemporal dementia. Alzheimer's and Dementia, 2021, 17, 500-514.	0.8	36
43	PCYT2 mutations disrupting etherlipid biosynthesis: phenotypes converging on the CDP-ethanolamine pathway. Brain, 2021, 144, e17-e17.	7.6	6
44	Apathy in presymptomatic genetic frontotemporal dementia predicts cognitive decline and is driven by structural brain changes. Alzheimer's and Dementia, 2021, 17, 969-983.	0.8	31
45	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
46	Invasive brain stimulation in the treatment of psychiatric illnessâ€"proposed indications and approaches. Deutsches Ärzteblatt International, 2021, 118, 31-36.	0.9	3
47	Natural History, Phenotypic Spectrum, and Discriminative Features of Multisystemic RFC1 Disease. Neurology, 2021, 96, e1369-e1382.	1.1	93
48	Progression of Behavioral Disturbances and Neuropsychiatric Symptoms in Patients With Genetic Frontotemporal Dementia. JAMA Network Open, 2021, 4, e2030194.	5.9	42
49	Early Recognition of Ball Catching Success in Clinical Trials with RNN-Based Predictive Classification. Lecture Notes in Computer Science, 2021, , 444-456.	1.3	1
50	Effects of Levodopa on quality of sleep and nocturnal movements in Parkinsonâ€™s Disease. Journal of Neurology, 2021, 268, 2506-2514.	3.6	11
51	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
52	Outcomes of <scp>SARSâ€CoVâ€2</scp> Infections in Patients with Neurodegenerative Diseases in the <scp>LEOSS</scp> Cohort. Movement Disorders, 2021, 36, 791-793.	3.9	13
53	MRI data-driven algorithm for the diagnosis of behavioural variant frontotemporal dementia. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 608-616.	1.9	10
54	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

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55	Pitfalls in Genetic Diagnostics: Why Phenotyping is Essential. Neuropediatrics, 2021, 52, 274-283.	0.6	1
56	Biallelic loss-of-function variations in PRDX3 cause cerebellar ataxia. Brain, 2021, 144, 1467-1481.	7.6	18
57	Approaches to Understanding <scp>COVID</scp>â€19 and its Neurological Associations. Annals of Neurology, 2021, 89, 1059-1067.	5.3	16
58	Plasma Neurofilament Light for Prediction of Disease Progression in Familial Frontotemporal Lobar Degeneration. Neurology, 2021, 96, e2296-e2312.	1.1	52
59	A de novo STUB1 variant associated with an early adult-onset multisystemic ataxia phenotype. Journal of Neurology, 2021, 268, 3845-3851.	3.6	7
60	Solving unsolved rare neurological diseasesâ€"a Solve-RD viewpoint. European Journal of Human Genetics, 2021, 29, 1332-1336.	2.8	4
61	Regional Brain and Spinal Cord Volume Loss in Spinocerebellar Ataxia Type 3. Movement Disorders, 2021, 36, 2273-2281.	3.9	37
62	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
63	Characterizing the Clinical Features and Atrophy Patterns of MAPT -Related Frontotemporal Dementia With Disease Progression Modeling. Neurology, 2021, 97, e941-e952.	1.1	29
64	Developmental Consequences of Defective ATG7-Mediated Autophagy in Humans. New England Journal of Medicine, 2021, 384, 2406-2417.	27.0	84
65	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
66	Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases. European Journal of Human Genetics, 2021, 29, 1325-1331.	2.8	49
67	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. European Journal of Human Genetics, 2021, 29, 1337-1347.	2.8	34
68	Natural History of Polymerase Gammaâ€"Related Ataxia. Movement Disorders, 2021, 36, 2642-2652.	3.9	10
69	The Revised Self-Monitoring Scale detects early impairment of social cognition in genetic frontotemporal dementia within the GENFI cohort. Alzheimer's Research and Therapy, 2021, 13, 127.	6.2	12
70	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
71	4-Aminopyridine is a promising treatment option for patients with gain-of-function KCNA2 -encephalopathy. Science Translational Medicine, 2021, 13, eaaz4957.	12.4	40
72	Dissemination in time and space in presymptomatic granulin mutation carriers: a GENFI spatial chronnectome study. Neurobiology of Aging, 2021, 108, 155-167.	3.1	3

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73	Differential early subcortical involvement in genetic FTD within the GENFI cohort. NeuroImage: Clinical, 2021, 30, 102646.	2.7	28
74	Disease-related cortical thinning in presymptomatic granulin mutation carriers. NeuroImage: Clinical, 2021, 29, 102540.	2.7	8
75	Reference values for the Cerebellar Cognitive Affective Syndrome Scale: age and education matter. Brain, 2021, 144, e20-e20.	7.6	14
76	Fampridine and Acetazolamide in EA2 and Related Familial EA. Neurology: Clinical Practice, 2021, 11, e438-e446.	1.6	27
77	Intracellular Lipid Accumulation and Mitochondrial Dysfunction Accompanies Endoplasmic Reticulum Stress Caused by Loss of the Co-chaperone DNAJC3. Frontiers in Cell and Developmental Biology, 2021, 9, 710247.	3.7	13
78	A panel of CSF proteins separates genetic frontotemporal dementia from presymptomatic mutation carriers: a GENFI study. Molecular Neurodegeneration, 2021, 16, 79.	10.8	9
79	Predicting disease progression in behavioral variant frontotemporal dementia. Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring, 2021, 13, e12262.	2.4	4
80	A dataâ€driven disease progression model of fluid biomarkers in genetic FTD. Alzheimer's and Dementia, 2021, 17, .	0.8	0
81	Differential synaptic marker involvement in the different genetic forms of frontotemporal dementia. Alzheimer's and Dementia, 2021, 17, .	0.8	1
82	Age at symptom onset and death and disease duration in genetic frontotemporal dementia: an international retrospective cohort study. Lancet Neurology, The, 2020, 19, 145-156.	10.2	175
83	Disentangling brain functional network remodeling in corticobasal syndrome â€“ A multimodal MRI study. NeuroImage: Clinical, 2020, 25, 102112.	2.7	10
84	Documenting the psychometric properties of the scale for the assessment and rating of ataxia to advance trial readiness of Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay. Journal of the Neurological Sciences, 2020, 417, 117050.	0.6	9
85	Network Localization of Alien Limb in Patients with Corticobasal Syndrome. Annals of Neurology, 2020, 88, 1118-1131.	5.3	11
86	Case Report: Expanding the Genetic and Phenotypic Spectrum of Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay. Frontiers in Genetics, 2020, 11, 585136.	2.3	7
87	Early symptoms in symptomatic and preclinical genetic frontotemporal lobar degeneration. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 975-984.	1.9	25
88	Abnormal pain perception is associated with thalamo-cortico-striatal atrophy in C9orf72 expansion carriers in the GENFI cohort. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 1325-1328.	1.9	12
89	A guide to writing systematic reviews of rare disease treatments to generate FAIR-compliant datasets: building a Treatabolome. Orphanet Journal of Rare Diseases, 2020, 15, 206.	2.7	21
90	Analysis of brain atrophy and local gene expression in genetic frontotemporal dementia. Brain Communications, 2020, 2, .	3.3	20

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91	Validation of a German version of the Cerebellar Cognitive Affective/ Schmahmann Syndrome Scale: preliminary version and study protocol. Neurological Research and Practice, 2020, 2, 39.	2.0	13
92	The genetic landscape of axonal neuropathies in the middle-aged and elderly. Neurology, 2020, 95, e3163-e3179.	1.1	19
93	Biallelic mutations in SORD cause a common and potentially treatable hereditary neuropathy with implications for diabetes. Nature Genetics, 2020, 52, 473-481.	21.4	97
94	Features of speech and swallowing dysfunction in pre-ataxic spinocerebellar ataxia type 2. Neurology, 2020, 95, e194-e205.	1.1	22
95	Multifocal, hypoechogenic nerve thickening in Cerebrotendinous Xanthomatosis. Clinical Neurophysiology, 2020, 131, 1798-1803.	1.5	2
96	Delineating MT-ATP6 -associated disease. Neurology: Genetics, 2020, 6, e393.	1.9	73
97	Real-life gait assessment in degenerative cerebellar ataxia. Neurology, 2020, 95, e1199-e1210.	1.1	60
98	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
99	Plasma glial fibrillary acidic protein is raised in progranulin-associated frontotemporal dementia. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 263-270.	1.9	106
100	Neuronal pentraxin 2: a synapse-derived CSF biomarker in genetic frontotemporal dementia. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 612-621.	1.9	55
101	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
102	Intraindividual Neurofilament Dynamics in Serum Mark the Conversion to Sporadic Parkinson's Disease. Movement Disorders, 2020, 35, 1233-1238.	3.9	22
103	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
104	Faster Cortical Thinning and Surface Area Loss in Presymptomatic and Symptomatic C9orf72 Repeat Expansion Adult Carriers. Annals of Neurology, 2020, 88, 113-122.	5.3	19
105	Clinico-Genetic, Imaging and Molecular Delineation of COQ8A-Ataxia: A Multicenter Study of 59 Patients. , 2020, 88, 251.		1
106	Social cognition impairment in genetic frontotemporal dementia within the GENFI cohort. Cortex, 2020, 133, 384-398.	2.4	26
107	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
108	Effects of Exergaming on Attentional Deficits and Dual-Tasking in Parkinson's Disease. Frontiers in Neurology, 2019, 10, 646.	2.4	15

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109	Correlations between serum and CSF pNfH levels in ALS, FTD and controls: a comparison of three analytical approaches. Clinical Chemistry and Laboratory Medicine, 2019, 57, 1556-1564.	2.3	32
110	Serum neurofilament light chain in genetic frontotemporal dementia: a longitudinal, multicentre cohort study. Lancet Neurology, The, 2019, 18, 1103-1111.	10.2	128
111	Application of Quantitative Motor Assessments in Friedreich Ataxia and Evaluation of Their Relation to Clinical Measures. Cerebellum, 2019, 18, 896-909.	2.5	9
112	The motor band sign in ALS: presentations and frequencies in a consecutive series of ALS patients. Journal of the Neurological Sciences, 2019, 406, 116440.	0.6	25
113	The inner fluctuations of the brain in presymptomatic Frontotemporal Dementia: The chronnectome fingerprint. NeuroImage, 2019, 189, 645-654.	4.2	33
114	Education modulates brain maintenance in presymptomatic frontotemporal dementia. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 1124-1130.	1.9	23
115	Feedback inhibition of cAMP effector signaling by a chaperone-assisted ubiquitin system. Nature Communications, 2019, 10, 2572.	12.8	29
116	Loss of paraplegin drives spasticity rather than ataxia in a cohort of 241 patients with SPG7. Neurology, 2019, 92, e2679-e2690.	1.1	49
117	Novel likely pathogenic variants in TMEM126A identified in non-syndromic autosomal recessive optic atrophy: two case reports. BMC Medical Genetics, 2019, 20, 62.	2.1	11
118	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
119	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
120	The molecular pathogenesis of superoxide dismutase 1-linked ALS is promoted by low oxygen tension. Acta Neuropathologica, 2019, 138, 85-101.	7.7	18
121	Unraveling corticobasal syndrome and alien limb syndrome with structural brain imaging. Cortex, 2019, 117, 33-40.	2.4	17
122	Recommendations for patient screening in ultra-rare inherited metabolic diseases: what have we learned from Niemann-Pick disease type C?. Orphanet Journal of Rare Diseases, 2019, 14, 20.	2.7	15
123	ARSACS as a Worldwide Disease: Novel SACS Mutations Identified in a Consanguineous Family from the Remote Tribal Jammu and Kashmir Region in India. Cerebellum, 2019, 18, 807-812.	2.5	18
124	Truncating Mutations in UBAP1 Cause Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2019, 104, 767-773.	6.2	39
125	Genome-wide analyses as part of the international FTLD-TDP whole-genome sequencing consortium reveals novel disease risk factors and increases support for immune dysfunction in FTLD. Acta Neuropathologica, 2019, 137, 879-899.	7.7	90
126	Pattern of Cerebellar Atrophy in Friedreichâ€™s Ataxiaâ€”Using the SUIT Template. Cerebellum, 2019, 18, 435-447.	2.5	23

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127	Autosomal Recessive Cerebellar Ataxias: Paving the Way toward Targeted Molecular Therapies. Neuron, 2019, 101, 560-583.	8.1	83
128	Parkinsonism in neurodegenerative diseases predominantly presenting with ataxia. International Review of Neurobiology, 2019, 149, 277-298.	2.0	7
129	White matter hyperintensities in progranulin-associated frontotemporal dementia: A longitudinal GENFI study. NeuroImage: Clinical, 2019, 24, 102077.	2.7	27
130	Atypical parkinsonism with severely reduced striatal dopamine uptake associated with a 16p11.2 duplication syndrome. Journal of Neurology, 2019, 266, 775-776.	3.6	1
131	Fall Risk in Relation to Individual Physical Activity Exposure in Patients with Different Neurodegenerative Diseases: a Pilot Study. Cerebellum, 2019, 18, 340-348.	2.5	16
132	TSFM mutations cause a complex hyperkinetic movement disorder with strong relief by cannabinoids. Parkinsonism and Related Disorders, 2019, 60, 176-178.	2.2	6
133	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
134	Validity and Reliability of Outcome Measures Assessing Dexterity, Coordination, and Upper Limb Strength in Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay. Archives of Physical Medicine and Rehabilitation, 2018, 99, 1747-1754.	0.9	15
135	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
136	Validation and Psychometric Properties of the German Version of the SWAL-QOL. Dysphagia, 2018, 33, 431-440.	1.8	23
137	Reply: POLR3A variants in hereditary spastic paraplegia and ataxia. Brain, 2018, 141, e2-e2.	7.6	10
138	Assessing mobility and balance in Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay population: Validity and reliability of four outcome measures. Journal of the Neurological Sciences, 2018, 390, 4-9.	0.6	15
139	Potential genetic modifiers of disease risk and age at onset in patients with frontotemporal lobar degeneration and GRN mutations: a genome-wide association study. Lancet Neurology, The, 2018, 17, 548-558.	10.2	97
140	Counterregulation of cAMP-directed kinase activities controls ciliogenesis. Nature Communications, 2018, 9, 1224.	12.8	37
141	The wide genetic landscape of clinical frontotemporal dementia: systematic combined sequencing of 121 consecutive subjects. Genetics in Medicine, 2018, 20, 240-249.	2.4	60
142	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
143	Case series: Slowing alpha rhythm in late-stage ALS patients. Clinical Neurophysiology, 2018, 129, 406-408.	1.5	14
144	Beyond ALS and FTD: the phenotypic spectrum of TBK1 mutations includes PSP-like and cerebellar phenotypes. Neurobiology of Aging, 2018, 62, 244.e9-244.e13.	3.1	30

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145	Nerve ultrasound characterizes AMN polyneuropathy as inhomogeneous and focal hypertrophic. Orphanet Journal of Rare Diseases, 2018, 13, 194.	2.7	9
146	The genetic nomenclature of recessive cerebellar ataxias. Movement Disorders, 2018, 33, 1056-1076.	3.9	61
147	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
148	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
149	Biallelic Parkin (PARK2) mutations can cause a bvFTD phenotype without clinically relevant parkinsonism. Parkinsonism and Related Disorders, 2018, 55, 145-147.	2.2	6
150	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
151	Recessive ataxias. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 155, 73-89.	1.8	73
152	Serum neurofilament light chain is increased in hereditary spastic paraplegias. Annals of Clinical and Translational Neurology, 2018, 5, 876-882.	3.7	26
153	No supportive evidence for TIA1 gene mutations in a European cohort of ALS-FTD spectrum patients. Neurobiology of Aging, 2018, 69, 293.e9-293.e11.	3.1	15
154	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
155	The hidden Niemann-Pick type C patient: clinical niches for a rare inherited metabolic disease. Current Medical Research and Opinion, 2017, 33, 877-890.	1.9	25
156	Overcoming the divide between ataxias and spastic paraplegias: Shared phenotypes, genes, and pathways. Movement Disorders, 2017, 32, 332-345.	3.9	144
157	Clinical assessment of dysphagia in neurodegeneration (CADN): development, validity and reliability of a bedside tool for dysphagia assessment. Journal of Neurology, 2017, 264, 1107-1117.	3.6	17
158	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578.	7.6	85
159	Loss-of-function mutations in theATP13A2/PARK9 gene cause complicated hereditary spastic paraplegia (SPG78). Brain, 2017, 140, 287-305.	7.6	135
160	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
161	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
162	Moving Beyond Syndromic Classifications in Neurodegenerative Disease: The Example of <scp>PLA</scp>2G6. Movement Disorders Clinical Practice, 2017, 4, 8-11.	1.5	6

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