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	Beyond the comparator model: A multifactorial two-step account of agency. Consciousness and Cognition, 2008, 17, 219-239.	1.5	695
2	Targeted next generation sequencing as a diagnostic tool in epileptic disorders. Epilepsia, 2012, 53, 1387-1398.	5.1	299
3	Misattributions of agency in schizophrenia are based on imprecise predictions about the sensory consequences of one's actions. Brain, 2010, 133, 262-271.	7.6	295
4	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
5	De novo loss- or gain-of-function mutations in KCNA2 cause epileptic encephalopathy. Nature Genetics, 2015, 47, 393-399.	21.4	224
6	I move, therefore I am: A new theoretical framework to investigate agency and ownership. Consciousness and Cognition, 2008, 17, 411-424.	1.5	220
7	The experience of agency: an interplay between prediction and postdiction. Frontiers in Psychology, 2013, 4, 127.	2.1	208
8	The Cerebellum Updates Predictions about the Visual Consequences of One's Behavior. Current Biology, 2008, 18, 814-818.	3.9	190
9	PNPLA6 mutations cause Boucher-NeuhÃuser and Gordon Holmes syndromes as part of a broad neurodegenerative spectrum. Brain, 2014, 137, 69-77.	7.6	189
10	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
11	S100B is increased in Parkinsonâ€™s disease and ablation protects against MPTP-induced toxicity through the RAGE and TNF-Î± pathway. Brain, 2012, 135, 3336-3347.	7.6	159
12	Stimulating personality: Ethical criteria for deep brain stimulation in psychiatric patients and for enhancement purposes. Biotechnology Journal, 2008, 3, 1511-1520.	3.5	155
13	Video gameâ€“based coordinative training improves ataxia in children with degenerative ataxia. Neurology, 2012, 79, 2056-2060.	1.1	155
14	Longâ€term effects of coordinative training in degenerative cerebellar disease. Movement Disorders, 2010, 25, 2239-2246.	3.9	148
15	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
16	Overcoming the divide between ataxias and spastic paraplegias: Shared phenotypes, genes, and pathways. Movement Disorders, 2017, 32, 332-345.	3.9	144
17	Loss-of-function mutations in theATP13A2/PARK9 gene cause complicated hereditary spastic paraplegia (SPG78). Brain, 2017, 140, 287-305.	7.6	135
18	Serum neurofilament light chain in genetic frontotemporal dementia: a longitudinal, multicentre cohort study. Lancet Neurology, The, 2019, 18, 1103-1111.	10.2	128

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19	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
20	Evidence for altered transport of insulin across the bloodâ€“brain barrier in insulin-resistant humans. Acta Diabetologica, 2014, 51, 679-681.	2.5	123
21	Clinical spectrum and genotypeâ€“phenotype associations of KCNA2-related encephalopathies. Brain, 2017, 140, 2337-2354.	7.6	117
22	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
23	How Physicians Allocate Scarce Resources at the Bedside: A Systematic Review of Qualitative Studies. Journal of Medicine and Philosophy, 2008, 33, 80-99.	0.8	111
24	Mutations inPOLR3AandPOLR3Bare a major cause of hypomyelinating leukodystrophies with or without dental abnormalities and/or hypogonadotropic hypogonadism. Journal of Medical Genetics, 2013, 50, 194-197.	3.2	109
25	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
26	Absence of BiP Co-chaperone DNAJC3 Causes Diabetes Mellitus and Multisystemic Neurodegeneration. American Journal of Human Genetics, 2014, 95, 689-697.	6.2	100
27	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
28	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
29	The Cerebellum Optimizes Perceptual Predictions about External Sensory Events. Current Biology, 2013, 23, 930-935.	3.9	95
30	Internalizing Agency of Self-Action: Perception of One's Own Hand Movements Depends on an Adaptable Prediction About the Sensory Action Outcome. Journal of Neurophysiology, 2006, 96, 1592-1601.	1.8	94
31	Rare mutations in SQSTM1 modify susceptibility to frontotemporal lobar degeneration. Acta Neuropathologica, 2014, 128, 397-410.	7.7	93
32	Natural History, Phenotypic Spectrum, and Discriminative Features of Multisystemic RFC1 Disease. Neurology, 2021, 96, e1369-e1382.	1.1	93
33	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
34	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
35	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
36	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

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37	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578.	7.6	85
38	Developmental Consequences of Defective ATG7-Mediated Autophagy in Humans. New England Journal of Medicine, 2021, 384, 2406-2417.	27.0	84
39	Autosomal Recessive Cerebellar Ataxias: Paving the Way toward Targeted Molecular Therapies. Neuron, 2019, 101, 560-583.	8.1	83
40	Validation of a Step Detection Algorithm during Straight Walking and Turning in Patients with Parkinsonâ€™s Disease and Older Adults Using an Inertial Measurement Unit at the Lower Back. Frontiers in Neurology, 2017, 8, 457.	2.4	79
41	Me or not me â€“ An optimal integration of agency cues?. Consciousness and Cognition, 2009, 18, 1065-1068.	1.5	77
42	Recessive ataxias. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 155, 73-89.	1.8	73
43	Delineating MT-ATP6 -associated disease. Neurology: Genetics, 2020, 6, e393.	1.9	73
44	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
45	Towards a common framework of grounded action cognition: Relating motor control, perception and cognition. Cognition, 2016, 146, 81-89.	2.2	72
46	ATAXIA WITH OPHTHALMOPLEGIA OR SENSORY NEUROPATHY IS FREQUENTLY CAUSED BY POLG MUTATIONS. Neurology, 2009, 73, 898-900.	1.1	69
47	Motor Training in Degenerative Spinocerebellar Disease: Ataxia-Specific Improvements by Intensive Physiotherapy and Exergames. BioMed Research International, 2014, 2014, 1-11.	1.9	69
48	SIL1 mutations and clinical spectrum in patients with Marinesco-SjÃ¶gren syndrome. Brain, 2013, 136, 3634-3644.	7.6	65
49	Neurofilament light chain in FTD is elevated not only in cerebrospinal fluid, but also in serum. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 1270-1272.	1.9	65
50	Characterizing POLG Ataxia: Clinics, Electrophysiology and Imaging. Cerebellum, 2012, 11, 1002-1011.	2.5	63
51	Progressive deafnessâ€“dystonia due to SERAC1 mutations: A study of 67 cases. Annals of Neurology, 2017, 82, 1004-1015.	5.3	63
52	Pure and syndromic optic atrophy explained by deep intronic OPA1 mutations and an intralocus modifier. Brain, 2014, 137, 2164-2177.	7.6	62
53	Fluid biomarkers in frontotemporal dementia: past, present and future. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 204-215.	1.9	62
54	The genetic nomenclature of recessive cerebellar ataxias. Movement Disorders, 2018, 33, 1056-1076.	3.9	61

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55	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
56	Real-life gait assessment in degenerative cerebellar ataxia. Neurology, 2020, 95, e1199-e1210.	1.1	60
57	Are intrinsic neural timescales related to sensory processing? Evidence from abnormal behavioral states. NeuroImage, 2021, 226, 117579.	4.2	60
58	Mutation screen reveals novel variants and expands the phenotypes associated with DYNC1H1. Journal of Neurology, 2015, 262, 2124-2134.	3.6	59
59	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
60	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
61	Autoantibodies Against Amyloid and Glial-Derived Antigens are Increased in Serum and Cerebrospinal Fluid of Lewy Body-Associated Dementias. Journal of Alzheimer's Disease, 2011, 26, 171-179.	2.6	55
62	Electrodes in the brainâ€"Ethical criteria for research and treatment with deep brain stimulation for neuropsychiatric disorders. Brain Stimulation, 2011, 4, 7-16.	1.6	55
63	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
64	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
65	Individual changes in preclinical spinocerebellar ataxia identified via increased motor complexity. Movement Disorders, 2016, 31, 1891-1900.	3.9	54
66	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
67	Plasma Neurofilament Light for Prediction of Disease Progression in Familial Frontotemporal Lobar Degeneration. Neurology, 2021, 96, e2296-e2312.	1.1	52
68	Videogame-based coordinative training can improve advanced, multisystemic early-onset ataxia. Journal of Neurology, 2013, 260, 2656-2658.	3.6	50
69	Spinocerebellar ataxia type 15: diagnostic assessment, frequency, and phenotypic features. Journal of Medical Genetics, 2011, 48, 407-412.	3.2	49
70	A recurrent mutation in KCNA2 as a novel cause of hereditary spastic paraplegia and ataxia. Annals of Neurology, 2016, 80, .	5.3	49
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	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

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73	Recommendations for whole genome sequencing in diagnostics for rare diseases. European Journal of Human Genetics, 2022, 30, 1017-1021.	2.8	48
74	Mutant superoxide dismutase-1 indistinguishable from wild-type causes ALS. Human Molecular Genetics, 2012, 21, 3568-3574.	2.9	47
75	First de novo KCND3 mutation causes severe Kv4.3 channel dysfunction leading to early onset cerebellar ataxia, intellectual disability, oral apraxia and epilepsy. BMC Medical Genetics, 2015, 16, 51.	2.1	46
76	Acetazolamide-responsive exercise-induced episodic ataxia associated with a novel homozygous DARS2 mutation. Journal of Medical Genetics, 2011, 48, 713-715.	3.2	45
77	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
78	Clinical and genetic characteristics of sporadic adult-onset degenerative ataxia. Neurology, 2017, 89, 1043-1049.	1.1	45
79	Affective coding: the emotional dimension of agency. Frontiers in Human Neuroscience, 2014, 8, 608.	2.0	43
80	Antisense Oligonucleotide Mediated Splice Correction of a Deep Intronic Mutation in OPA1. Molecular Therapy - Nucleic Acids, 2016, 5, e390.	5.1	43
81	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
82	Progression of Behavioral Disturbances and Neuropsychiatric Symptoms in Patients With Genetic Frontotemporal Dementia. JAMA Network Open, 2021, 4, e2030194.	5.9	42
83	Ethically Justified, Clinically Applicable Criteria for Physician Decision-Making in Psychopharmacological Enhancement. Neuroethics, 2009, 2, 89-102.	2.8	41
84	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
85	How Happy Is Too Happy? Euphoria, Neuroethics, and Deep Brain Stimulation of the Nucleus Accumbens. AJOB Neuroscience, 2012, 3, 30-36.	1.1	40
86	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
87	MultisystemicSYNE1ataxia: confirming the high frequency and extending the mutational and phenotypic spectrum. Brain, 2016, 139, e46-e46.	7.6	40
88	Atypical parkinsonism in C9orf72 expansions: a case report and systematic review of 45 cases from the literature. Journal of Neurology, 2016, 263, 558-574.	3.6	40
89	4-Aminopyridine is a promising treatment option for patients with gain-of-function KCNA2 -encephalopathy. Science Translational Medicine, 2021, 13, eaaz4957.	12.4	40
90	Friedreich Ataxia: Dysarthria Profile and Clinical Data. Cerebellum, 2013, 12, 475-484.	2.5	39

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91	Neurochondrin is a neuronal target antigen in autoimmune cerebellar degeneration. Neurology: Neuroimmunology and NeuroInflammation, 2017, 4, e307.	6.0	39
92	Truncating Mutations in UBAP1 Cause Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2019, 104, 767-773.	6.2	39
93	Pilot whole-exome sequencing of a German early-onset Alzheimer's disease cohort reveals a substantial frequency of PSEN2 variants. Neurobiology of Aging, 2016, 37, 208.e11-208.e17.	3.1	38
94	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
95	Counterregulation of cAMP-directed kinase activities controls ciliogenesis. Nature Communications, 2018, 9, 1224.	12.8	37
96	Regional Brain and Spinal Cord Volume Loss in Spinocerebellar Ataxia Type 3. Movement Disorders, 2021, 36, 2273-2281.	3.9	37
97	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
98	Lack of enzyme activity in GBA2 mutants associated with hereditary spastic paraplegia/cerebellar ataxia (SPG46). Biochemical and Biophysical Research Communications, 2015, 465, 35-40.	2.1	35
99	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
100	Total tau is increased, but phosphorylated tau not decreased, in cerebrospinal fluid in amyotrophic lateral sclerosis. Neurobiology of Aging, 2015, 36, 1072-1074.	3.1	34
101	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. European Journal of Human Genetics, 2021, 29, 1337-1347.	2.8	34
102	Comparing speech characteristics in spinocerebellar ataxias type 3 and type 6 with Friedreich ataxia. Journal of Neurology, 2015, 262, 21-26.	3.6	33
103	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
104	The inner fluctuations of the brain in presymptomatic Frontotemporal Dementia: The chronnectome fingerprint. NeuroImage, 2019, 189, 645-654.	4.2	33
105	The CST3 BB Genotype and Low Cystatin C Cerebrospinal Fluid Levels are Associated with Dementia in Lewy Body Disease. Journal of Alzheimer's Disease, 2010, 19, 937-942.	2.6	32
106	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
107	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
108	POLG, but not PEO1, is a frequent cause of cerebellar ataxia in Central Europe. Movement Disorders, 2010, 25, 2678-2682.	3.9	31

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109	Niemann-Pick type C is frequent in adult ataxia with cognitive decline and vertical gaze palsy. Neurology, 2013, 80, 1169-1170.	1.1	31
110	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
111	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
112	Serum Levels of Progranulin Do Not Reflect Cerebrospinal Fluid Levels in Neurodegenerative Disease. Current Alzheimer Research, 2016, 13, 654-662.	1.4	31
113	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
114	Network structure and transcriptomic vulnerability shape atrophy in frontotemporal dementia. Brain, 2023, 146, 321-336.	7.6	30
115	Voice in Friedreich Ataxia. Journal of Voice, 2017, 31, 243.e9-243.e19.	1.5	29
116	Feedback inhibition of cAMP effector signaling by a chaperone-assisted ubiquitin system. Nature Communications, 2019, 10, 2572.	12.8	29
117	Characterizing the Clinical Features and Atrophy Patterns of MAPT -Related Frontotemporal Dementia With Disease Progression Modeling. Neurology, 2021, 97, e941-e952.	1.1	29
118	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
119	Differential early subcortical involvement in genetic FTD within the GENFI cohort. NeuroImage: Clinical, 2021, 30, 102646.	2.7	28
120	Sensorimotor Recalibration Depends on Attribution of Sensory Prediction Errors to Internal Causes. PLoS ONE, 2013, 8, e54925.	2.5	28
121	A recessive ataxia diagnosis algorithm for the next generation sequencing era. Annals of Neurology, 2017, 82, 892-899.	5.3	27
122	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
123	White matter hyperintensities in progranulin-associated frontotemporal dementia: A longitudinal GENFI study. NeuroImage: Clinical, 2019, 24, 102077.	2.7	27
124	Fampridine and Acetazolamide in EA2 and Related Familial EA. Neurology: Clinical Practice, 2021, 11, e438-e446.	1.6	27
125	A data-driven disease progression model of fluid biomarkers in genetic frontotemporal dementia. Brain, 2022, 145, 1805-1817.	7.6	27
126	Soluble TAM receptors sAXL and sTyro3 predict structural and functional protection in Alzheimerâ€™s disease. Neuron, 2022, 110, 1009-1022.e4.	8.1	27

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127	Sustained dopaminergic response of parkinsonism and depression in POLGâ€associated parkinsonism. Movement Disorders, 2010, 25, 243-245.	3.9	26
128	Atypical juvenile parkinsonism in a consanguineous SPG15 family. Movement Disorders, 2011, 26, 565-566.	3.9	26
129	Serum neurofilament light chain is increased in hereditary spastic paraplegias. Annals of Clinical and Translational Neurology, 2018, 5, 876-882.	3.7	26
130	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
131	Social cognition impairment in genetic frontotemporal dementia within the GENFI cohort. Cortex, 2020, 133, 384-398.	2.4	26
132	The human G93A SOD1 phenotype closely resembles sporadic amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, 764-767.	1.9	25
133	Doing good or bad: How interactions between action and emotion expectations shape the sense of agency. Social Neuroscience, 2015, 10, 1-13.	1.3	25
134	NPC1 is enriched in unexplained early onset ataxia: a targeted high-throughput screening. Journal of Neurology, 2015, 262, 2557-2563.	3.6	25
135	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
136	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
137	Early symptoms in symptomatic and preclinical genetic frontotemporal lobar degeneration. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 975-984.	1.9	25
138	Tau and neurofilament lightâ€chain as fluid biomarkers in spinocerebellar ataxia type 3. European Journal of Neurology, 2022, 29, 2439-2452.	3.3	25
139	Guidelines and quality measures for the diagnosis of optic ataxia. Frontiers in Human Neuroscience, 2013, 7, 324.	2.0	24
140	Realâ€Life Turning Movements Capture Subtle Longitudinal and Preataxic Changes in Cerebellar Ataxia. Movement Disorders, 2022, 37, 1047-1058.	3.9	24
141	Conceptual framework for the definition of preclinical and prodromal frontotemporal dementia. Alzheimer's and Dementia, 2022, 18, 1408-1423.	0.8	24
142	Retinal nerve fiber layer loss in multiple system atrophy. Movement Disorders, 2011, 26, 914-916.	3.9	23
143	Effects of Cellular Pathway Disturbances on Misfolded Superoxide Dismutase-1 in Fibroblasts Derived from ALS Patients. PLoS ONE, 2016, 11, e0150133.	2.5	23
144	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

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145	Cerebrospinal Fluid Progranulin, but Not Serum Progranulin, Is Reduced in <i>GRN</i>-Negative Frontotemporal Dementia. Neurodegenerative Diseases, 2017, 17, 83-88.	1.4	23
146	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
147	Validation and Psychometric Properties of the German Version of the SWAL-QOL. Dysphagia, 2018, 33, 431-440.	1.8	23
148	Education modulates brain maintenance in presymptomatic frontotemporal dementia. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 1124-1130.	1.9	23
149	Pattern of Cerebellar Atrophy in Friedreichâ€™s Ataxiaâ€”Using the SUIT Template. Cerebellum, 2019, 18, 435-447.	2.5	23
150	Weighting models and weighting factors. Consciousness and Cognition, 2012, 21, 55-58.	1.5	22
151	Features of speech and swallowing dysfunction in pre-ataxic spinocerebellar ataxia type 2. Neurology, 2020, 95, e194-e205.	1.1	22
152	Intraindividual Neurofilament Dynamics in Serum Mark the Conversion to Sporadic Parkinson's Disease. Movement Disorders, 2020, 35, 1233-1238.	3.9	22
153	Biallelic variants in <i>HPDL</i> cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
154	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
155	Transcranial Sonography Reveals Cerebellar, Nigral, and Forebrain Abnormalities in Friedreichâ€™s Ataxia. Neurodegenerative Diseases, 2011, 8, 470-475.	1.4	21
156	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
157	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
158	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
159	Paving the Way Toward Meaningful Trials in Ataxias: An Ataxia Global Initiative Perspective. Movement Disorders, 2022, 37, 1125-1130.	3.9	21
160	Suicide Attempt as the Presenting Symptom of<i>C9orf72</i>Dementia. American Journal of Psychiatry, 2012, 169, 1211-1213.	7.2	20
161	The valence of action outcomes modulates the perception of oneâ€™s actions. Consciousness and Cognition, 2012, 21, 18-29.	1.5	20
162	Ataxia meets chorioretinal dystrophy and hypogonadism: Boucher-NeuhÃuser syndrome due to<i>PNPLA6</i>mutations: FigureÃ1. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 580-581.	1.9	20

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163	Speech and swallowing abnormalities in adults with POLG associated ataxia (POLG-A). Mitochondrion, 2017, 37, 1-7.	3.4	20
164	Analysis of brain atrophy and local gene expression in genetic frontotemporal dementia. Brain Communications, 2020, 2, .	3.3	20
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