Mathieu Anheim

List of Publications by Year
in descending order

60	3,119	196777	198040
papers	citations	29 h-index	52 g-index

60	60	60	4172
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A Homozygous Missense Variant in <scp>PPP1R1B/DARPPâ€32</scp> Is Associated With Generalized Complex Dystonia. Movement Disorders, 2022, 37, 365-374.	2.2	7
2	Safety and efficacy of riluzole in spinocerebellar ataxia type 2 in France (ATRIL): a multicentre, randomised, double-blind, placebo-controlled trial. Lancet Neurology, The, 2022, 21, 225-233.	4.9	24
3	â€œPhalanx signâ€•helps to discriminate MSA-C from idiopathic late onset cerebellar ataxia. Journal of Neurology, 2022, 269, 3900-3903.	1.8	1
4	Progression of Nigrostriatal Denervation in Cerebellar Multiple System Atrophy. Neurology, 2022, 98, 232-236.	1.5	6
5	Author Response: Progression of Nigrostriatal Denervation in Cerebellar Multiple System Atrophy: A Prospective Study. Neurology, 2022, 98, 1034-1034.	1.5	0
6	Reply to â€œ<scp>PPP2R5D</scp> Genetic Mutations and Early Onset Parkinsonismâ€• Annals of Neurology, 2021, 89, 195-196.	2.8	1
7	Natural History, Phenotypic Spectrum, and Discriminative Features of Multisystemic RFC1 Disease. Neurology, 2021, 96, e1369-e1382.	1.5	93
8	Biallelic RFC1-expansion in a French multicentric sporadic ataxia cohort. Journal of Neurology, 2021, 268, 3337-3343.	1.8	24
9	Reply to: â€œAutosomalâ€Recessive Cerebellar Ataxias With Elevated Alphaâ€Fetoprotein: Uncommon Diseases, Common Biomarkerâ€• Movement Disorders, 2021, 36, 789-790.	2.2	3
10	Implementation of a Magnetic Resonance Imaging scanner dedicated to emergencies in cases of binocular diplopia: Impact on patient management. Journal of Neuroradiology, 2021, , .	0.6	0
11	Expanding the clinical spectrum of STIP1 homology and U-box containing protein 1-associated ataxia. Journal of Neurology, 2021, 268, 1927-1937.	1.8	15
12	Autosomal Recessive Cerebellar Ataxias With Elevated Alphaâ€Fetoprotein: Uncommon Diseases, Common Biomarker. Movement Disorders, 2020, 35, 2139-2149.	2.2	17
13	The <scp>GRIA3</scp> c.<scp>2477G</scp> >â€‰A Variant Causes an Exaggerated Startle Reflex, Chorea, and Multifocal Myoclonus. Movement Disorders, 2020, 35, 1224-1232.	2.2	13
14	Characterization of Recessive Parkinson Disease in a Large Multicenter Study. Annals of Neurology, 2020, 88, 843-850.	2.8	40
15	Clonidine GH stimulation test to differentiate MSA from idiopathic late onset cerebellar ataxia: a prospective, controlled study. Journal of Neurology, 2020, 267, 855-859.	1.8	3
16	Recessive Ataxia Differential Diagnosis Algorithm (RADIAL) Versus Specific Niemann-Pick Type C Suspicion Indices: A Retrospective Algorithm Comparison. Cerebellum, 2020, 19, 243-251.	1.4	1
17	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
18	Is Motor Side Onset of Parkinson's Disease a Risk Factor for Developing <scp>Impulsiveâ€Compulsive</scp> Behavior? A <scp>Crossâ€Sectional</scp> Study. Movement Disorders, 2020, 35, 1080-1081.	2.2	6

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19	Loss of paraplegin drives spasticity rather than ataxia in a cohort of 241 patients with SPG7. Neurology, 2019, 92, e2679-e2690.	1.5	49
20	Sleep in ADCY5 -Related Dyskinesia: Prolonged Awakenings Caused by Abnormal Movements. Journal of Clinical Sleep Medicine, 2019, 15, 1021-1029.	1.4	12
21	Efficacy of Exome-Targeted Capture Sequencing to Detect Mutations in Known Cerebellar Ataxia Genes. JAMA Neurology, 2018, 75, 591.	4.5	93
22	Clinical, Biomarker, and Molecular Delineations and Genotype-Phenotype Correlations of Ataxia With Oculomotor Apraxia Type 1. JAMA Neurology, 2018, 75, 495.	4.5	28
23	Adult Niemann-Pick disease type C in France: clinical phenotypes and long-term miglustat treatment effect. Orphanet Journal of Rare Diseases, 2018, 13, 175.	1.2	38
24	The genetic nomenclature of recessive cerebellar ataxias. Movement Disorders, 2018, 33, 1056-1076.	2.2	61
25	Assessment of a Targeted Gene Panel for Identification of Genes Associated With Movement Disorders. JAMA Neurology, 2018, 75, 1234.	4.5	64
26	Low cancer prevalence in polyglutamine expansion diseases. Neurology, 2017, 88, 1114-1119.	1.5	21
27	SCA13 causes dominantly inherited non-progressive myoclonus ataxia. Parkinsonism and Related Disorders, 2017, 38, 80-84.	1.1	8
28	The hidden Niemann-Pick type C patient: clinical niches for a rare inherited metabolic disease. Current Medical Research and Opinion, 2017, 33, 877-890.	0.9	25
29	Parkinsonian-Pyramidal syndromes: A systematic review. Parkinsonism and Related Disorders, 2017, 39, 4-16.	1.1	20
30	A recessive ataxia diagnosis algorithm for the next generation sequencing era. Annals of Neurology, 2017, 82, 892-899.	2.8	27
31	Recommendations for the detection and diagnosis of Niemann-Pick disease type C. Neurology: Clinical Practice, 2017, 7, 499-511.	0.8	119
32	Deciphering the causes of sporadic late-onset cerebellar ataxias: a prospective study with implications for diagnostic work. Journal of Neurology, 2017, 264, 1118-1126.	1.8	34
33	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
34	Validation of a clinical practice-based algorithm for the diagnosis of autosomal recessive cerebellar ataxias based on NGS identified cases. Journal of Neurology, 2016, 263, 1314-1322.	1.8	15
35	Delayedâ€onset Friedreich's ataxia revisited. Movement Disorders, 2016, 31, 62-69.	2.2	54
36	Mini-Exome Coupled to Read-Depth Based Copy Number Variation Analysis in Patients with Inherited Ataxias. Human Mutation, 2016, 37, 1340-1353.	1.1	33

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37	New practical definitions for the diagnosis of autosomal recessive spastic ataxia of <scp>C</scp>harlevoixâ€“<scp>S</scp>aguenay. Annals of Neurology, 2015, 78, 871-886.	2.8	62
38	Loss of AP-5 results in accumulation of aberrant endolysosomes: defining a new type of lysosomal storage disease. Human Molecular Genetics, 2015, 24, 4984-4996.	1.4	80
39	ADCY5 -related dyskinesia. Neurology, 2015, 85, 2026-2035.	1.5	163
40	Subthalamic stimulation or subthalamic lesion for Parkinson's disease ? A case report. Parkinsonism and Related Disorders, 2015, 21, 1485-1487.	1.1	2
41	Dalfampridine in hereditary spastic paraplegia: a prospective, open study. Journal of Neurology, 2015, 262, 1285-1288.	1.8	21
42	Relevance of corpus callosum splenium versus middle cerebellar peduncle hyperintensity for FXTAS diagnosis in clinical practice. Journal of Neurology, 2015, 262, 435-442.	1.8	80
43	Genetics of Recessive Ataxias. , 2015, , 235-261.		0
44	The pleiotropic movement disorders phenotype of adult ataxia-telangiectasia. Neurology, 2014, 83, 1087-1095.	1.5	130
45	Autosomal Recessive Cerebellar Ataxia Type 3 Due toANO10Mutations. JAMA Neurology, 2014, 71, 1305.	4.5	57
46	Heterogeneity and frequency of movement disorders in juvenile and adult-onset Niemann-Pick C disease. Journal of Neurology, 2014, 261, 174-179.	1.8	43
47	SPG15: a cause of juvenile atypical levodopa responsive parkinsonism. Journal of Neurology, 2014, 261, 435-437.	1.8	23
48	Overall mutational spectrum of SLC20A2, PDGFB and PDGFRB in idiopathic basal ganglia calcification. Neurogenetics, 2014, 15, 215-216.	0.7	22
49	The tumour suppressor gene WWOX is mutated in autosomal recessive cerebellar ataxia with epilepsy and mental retardation. Brain, 2014, 137, 411-419.	3.7	127
50	Emerging topics in FXTAS. Journal of Neurodevelopmental Disorders, 2014, 6, 31.	1.5	76
51	From anti-GAD to ataxia with ocular motor apraxia type 2: through the looking glass. Journal of Neurology, 2013, 260, 1158-1159.	1.8	1
52	Phenotypic variability in ARCA2 and identification of a core ataxic phenotype with slow progression. Orphanet Journal of Rare Diseases, 2013, 8, 173.	1.2	63
53	Factors Influencing Disease Progression in Autosomal Dominant Cerebellar Ataxia and Spastic Paraplegia. Archives of Neurology, 2012, 69, 500.	4.9	56
54	Exonic Deletions of FXN and Early-Onset Friedreich Ataxia. Archives of Neurology, 2012, 69, 912-6.	4.9	37

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55	FXTAS. Neurology, 2012, 79, 1898-1907.	1.5	221
56	The Autosomal Recessive Cerebellar Ataxias. New England Journal of Medicine, 2012, 366, 636-646.	13.9	307
57	Annual change in Friedreich's ataxia evaluated by the Scale for the Assessment and Rating of Ataxia (SARA) is independent of disease severity. Movement Disorders, 2012, 27, 135-139.	2.2	30
58	SPG11 spastic paraplegia. Journal of Neurology, 2009, 256, 104-108.	1.8	96
59	ADCK3, an Ancestral Kinase, Is Mutated in a Form of Recessive Ataxia Associated with Coenzyme Q10 Deficiency. American Journal of Human Genetics, 2008, 82, 661-672.	2.6	290
60	Clinical and Molecular Findings of Ataxia With Oculomotor Apraxia Type 2 in 4 Families. Archives of Neurology, 2008, 65, 958-62.	4.9	38