Martin Paucar

List of Publications by Year
in descending order

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47 papers	1,243 citations	687363 13 h-index	377865 34 g-index
49 all docs	49 docs citations	49 times ranked	2313 citing authors

#	Article	IF	Citations
1	Are ATXN2 variants modifying our understanding about neural pathogenesis, phenotypes, and diagnostic?. Neural Regeneration Research, 2022, 17, 2445.	3.0	0
2	Hyperkinesias and Echolalia in Primary Familial Brain Calcification. Annals of Neurology, 2021, 89, 418-419.	5.3	1
3	A Novel Duplication in <scp>ATXN2</scp> as Modifier for Spinocerebellar Ataxia 3 (<scp>SCA3</scp>) and <scp>C9ORF72â€ALS</scp>. Movement Disorders, 2021, 36, 508-514.	3.9	8
4	Proenkephalin Decreases in Cerebrospinal Fluid with Symptom Progression of Huntington's Disease. Movement Disorders, 2021, 36, 481-491.	3.9	12
5	Increasing involvement of CAPN1 variants in spastic ataxias and phenotype-genotype correlations. Neurogenetics, 2021, 22, 71-79.	1.4	11
6	V374A KCND3 Pathogenic Variant Associated With Paroxysmal Ataxia Exacerbations. Neurology: Genetics, 2021, 7, e546.	1.9	10
7	Expanding the etiologic spectrum of spastic ataxia syndrome: chronic infection with human T lymphotropic virus type 1. Journal of NeuroVirology, 2021, 27, 345-347.	2.1	0
8	Adult-Onset Ataxia With Neuropathy and White Matter Abnormalities Due to a Novel SAMD9L Variant. Neurology: Genetics, 2021, 7, e628.	1.9	1
9	Predominant Spastic Paraparesis Associated With the D178N Mutation in PRNP. Neurology: Genetics, 2021, 7, e636.	1.9	0
10	Involuntary movements, vocalizations and cognitive decline. Parkinsonism and Related Disorders, 2020, 79, 135-137.	2.2	1
11	SLC1A3 variant associated with hemiplegic migraine and acetazolamide-responsive MRS changes. Neurology: Genetics, 2020, 6, e474.	1.9	9
12	Rare variants in dynein heavy chain genes in two individuals with situs inversus and developmental dyslexia: a case report. BMC Medical Genetics, 2020, 21, 87.	2.1	5
13	Phenotypic variability in chorea-acanthocytosis associated with novel VPS13A mutations. Neurology: Genetics, 2020, 6, e426.	1.9	5
14	Broader phenotypic traits and widespread brain hypometabolism in spinocerebellar ataxia 27. Journal of Internal Medicine, 2020, 288, 103-115.	6.0	16
15	Heterozygous variants in DCC. Neurology: Genetics, 2020, 6, e526.	1.9	4
16	The cerebellar phenotype of Charcot-Marie-Tooth neuropathy type 4C. Cerebellum and Ataxias, 2019, 6, 9.	1.9	9
17	Clinical Reasoning: Leg weakness and stiffness at the emergency room. Neurology, 2019, 92, e622-e625.	1.1	0
18	Altered CSF levels of monoamines in hereditary spastic paraparesis 10. Neurology: Genetics, 2019, 5, e344.	1.9	2

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19	Variant ataxia-telangiectasia with prominent camptocormia. Parkinsonism and Related Disorders, 2019, 62, 253-255.	2.2	7
20	Progressive Ataxia with Elevated Alpha-Fetoprotein: Diagnostic Issues and Review of the Literature. Tremor and Other Hyperkinetic Movements, 2019, 9, .	2.0	6
21	Novel Features and Abnormal Pattern of Cerebral Glucose Metabolism in Spinocerebellar Ataxia 19. Cerebellum, 2018, 17, 465-476.	2.5	13
22	Novel hyperkinetic dystonia-like manifestation and neurological disease course of Swedish Gaucher patients. Blood Cells, Molecules, and Diseases, 2018, 68, 86-92.	1.4	21
23	Novel Imaging Biomarkers for Huntingtonâ€™s Disease and Other Hereditary Choreas. Current Neurology and Neuroscience Reports, 2018, 18, 85.	4.2	13
24	Chorea, psychosis, acanthocytosis, and prolonged survival associated with ELAC2 mutations. Neurology, 2018, 91, 710-712.	1.1	8
25	Pathological Study of a FMR1 Premutation Carrier With Progressive Supranuclear Palsy. Frontiers in Genetics, 2018, 9, 317.	2.3	6
26	GLRA1 mutation and long-term follow-up of the first hyperekplexia family. Neurology: Genetics, 2018, 4, e259.	1.9	2
27	A SLC20A2 gene mutation carrier displaying ataxia and increased levels of cerebrospinal fluid phosphate. Journal of the Neurological Sciences, 2017, 375, 245-247.	0.6	14
28	An unusual cause of fatal rapid-onset ataxia plus syndrome. Cerebellum and Ataxias, 2017, 4, 5.	1.9	0
29	Hereditary spastic paraplegia type 5: natural history, biomarkers and a randomized controlled trial. Brain, 2017, 140, 3112-3127.	7.6	87
30	A KCNC3 mutation causes a neurodevelopmental, non-progressive SCA13 subtype associated with dominant negative effects and aberrant EGFR trafficking. PLoS ONE, 2017, 12, e0173565.	2.5	22
31	Paroxysmal Kinesigenic Dyskinesia. Tremor and Other Hyperkinetic Movements, 2017, 7, 529.	2.0	0
32	J10â€…Chorea, psychotic symptoms and long survival in a subject with ELAC2 mutations. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, A78.3-A79.	1.9	0
33	Hypospadias as a novel feature in spinal bulbar muscle atrophy. Journal of Neurology, 2016, 263, 703-706.	3.6	2
34	Expanding the ataxia with oculomotor apraxia type 4 phenotype. Neurology: Genetics, 2016, 2, e49.	1.9	37
35	Absence of the Autophagy Adaptor SQSTM1/p62 Causes Childhood-Onset Neurodegeneration with Ataxia, Dystonia, and Gaze Palsy. American Journal of Human Genetics, 2016, 99, 735-743.	6.2	99
36	Progressive brain calcifications and signs in a family with the L9R mutation in the PDGFB gene. Neurology: Genetics, 2016, 2, e84.	1.9	4

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37	PSP-CBS with Dopamine Deficiency in a Female with a FMR1 Premutation. Cerebellum, 2016, 15, 636-640.	2.5	7
38	POLG-Associated Ataxia Presenting as a Fragile X Tremor/Ataxia Phenocopy Syndrome. Cerebellum, 2016, 15, 632-635.	2.5	1
39	Teaching Video Neuro Images : Feeding dystonia in chorea-acanthocytosis. Neurology, 2015, 85, e143-4.	1.1	8
40	Mutations in XPR1 cause primary familial brain calcification associated with altered phosphate export. Nature Genetics, 2015, 47, 579-581.	21.4	237
41	7Î±-hydroxy-3-oxo-4-cholestenoic acid in cerebrospinal fluid reflects the integrity of the blood-brain barrier. Journal of Lipid Research, 2014, 55, 313-318.	4.2	37
42	Mutations in the gene encoding PDGF-B cause brain calcifications in humans and mice. Nature Genetics, 2013, 45, 1077-1082.	21.4	273
43	Mutations in SLC20A2 are a major cause of familial idiopathic basal ganglia calcification. Neurogenetics, 2013, 14, 11-22.	1.4	131
44	Genotype-phenotype analysis in inherited prion disease with eight octapeptide repeat insertional mutation. Prion, 2013, 7, 501-510.	1.8	17
45	De Novo Mutations in Ataxin-2 Gene and ALS Risk. PLoS ONE, 2013, 8, e70560.	2.5	28
46	D02â€…A case of Huntington disease-like 1 (HDL1) with long disease duration. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, A19.2-A19.	1.9	1
47	Persistence of viral RNA in the brain of offspring to mice infected with influenza A/WSN/33 virus during pregnancy. Journal of NeuroVirology, 2002, 8, 353-357.	2.1	66