Paulo V Souza

List of Publications by Year
in descending order

103	771	687363 13	610901 24
papers	citations	h-index	g-index

103	103	103	1489
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Hereditary Spastic Paraplegia: Clinical and Genetic Hallmarks. Cerebellum, 2017, 16, 525-551.	2.5	169
2	Myasthenia Gravis and COVID-19: Clinical Characteristics and Outcomes. Frontiers in Neurology, 2020, 11, 1053.	2.4	78
3	Clinical and molecular findings in a cohort of ANO5â€related myopathy. Annals of Clinical and Translational Neurology, 2019, 6, 1225-1238.	3.7	32
4	Clinical and genetic basis of congenital myasthenic syndromes. Arquivos De Neuro-Psiquiatria, 2016, 74, 750-760.	0.8	27
5	Non-motor and Extracerebellar Features in Spinocerebellar Ataxia Type 2. Cerebellum, 2017, 16, 34-39.	2.5	27
6	New genetic causes for complex hereditary spastic paraplegia. Journal of the Neurological Sciences, 2017, 379, 283-292.	0.6	24
7	Intragenic variants in the SMN1 gene determine the clinical phenotype in 5q spinal muscular atrophy. Neurology: Genetics, 2020, 6, e505.	1.9	24
8	Clinical and genetic basis of familial amyotrophic lateral sclerosis. Arquivos De Neuro-Psiquiatria, 2015, 73, 1026-1037.	0.8	23
9	Clinical and radiological profile of patients with spinal muscular atrophy type 4. European Journal of Neurology, 2021, 28, 609-619.	3.3	23
10	C9orf72-related disorders: expanding the clinical and genetic spectrum of neurodegenerative diseases. Arquivos De Neuro-Psiquiatria, 2015, 73, 246-256.	0.8	21
11	Atypical Motor Neuron Disease variants: Still a diagnostic challenge in Neurology. Revue Neurologique, 2019, 175, 221-232.	1.5	21
12	Normal muscle structure, growth, development, and regeneration. Current Reviews in Musculoskeletal Medicine, 2015, 8, 176-181.	3.5	16
13	<scp>GBE1</scp>â€related disorders: Adult polyglucosan body disease and its neuromuscular phenotypes. Journal of Inherited Metabolic Disease, 2021, 44, 534-543.	3.6	15
14	SCA1 patients may present as hereditary spastic paraplegia and must be included in spastic-ataxias group. Parkinsonism and Related Disorders, 2015, 21, 1243-1246.	2.2	14
15	Earlyâ€onset epilepsy as the main neurological manifestation of cerebrotendinous xanthomatosis. Epilepsy and Behavior, 2012, 24, 380-381.	1.7	12
16	The cerebellum in Parkinsonâ€™s disease and parkinsonism in cerebellar disorders. Brain, 2013, 136, e248-e248.	7.6	12
17	When should we test patients with familial ataxias for SCA31? A misdiagnosed condition outside Japan?. Journal of the Neurological Sciences, 2015, 355, 206-208.	0.6	11
18	Early-onset axonal Charcot-Marie-Tooth disease due to SACS mutation. Neuromuscular Disorders, 2018, 28, 169-172.	0.6	11

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19	MR imaging of inherited myopathies: a review and proposal of imaging algorithms. European Radiology, 2021, 31, 8498-8512.	4.5	10
20	Early-onset familial Alzheimerâ€™s disease related to presenilin 1 mutation resembling autosomal dominant spinocerebellar ataxia. Journal of Neurology, 2013, 260, 1177-1179.	3.6	9
21	NFU1-Related Disorders as Key Differential Diagnosis of Cavitating Leukoencephalopathy. Journal of Pediatric Genetics, 2018, 07, 040-042.	0.7	9
22	Clinical and epidemiological profiles of non-traumatic myelopathies. Arquivos De Neuro-Psiquiatria, 2016, 74, 161-165.	0.8	8
23	Infantile-onset ascending spastic paraplegia phenotype associated with SPAST mutation. Journal of the Neurological Sciences, 2016, 371, 34-35.	0.6	8
24	Acute hepatic porphyrias for the neurologist: current concepts and perspectives. Arquivos De Neuro-Psiquiatria, 2021, 79, 68-80.	0.8	8
25	Motor unit number index (MUNIX) in myopathic disorders: Clinical correlations and potential pitfalls. Neurophysiologie Clinique, 2019, 49, 329-334.	2.2	7
26	Variable phenotype and severity of sialidosis expressed in two siblings presenting with ataxia and macular cherry-red spots. Journal of Clinical Neuroscience, 2013, 20, 1327-1328.	1.5	6
27	Bright tongue sign: a diagnostic marker for amyotrophic lateral sclerosis. Arquivos De Neuro-Psiquiatria, 2014, 72, 572-572.	0.8	6
28	Brain MRI features in Lhermitte-Duclos disease. Arquivos De Neuro-Psiquiatria, 2014, 72, 645-645.	0.8	6
29	Paracoccidioidomycosis: a rare cause of infectious encephalomyelopathy. Arquivos De Neuro-Psiquiatria, 2014, 72, 904-905.	0.8	6
30	Progressive hearing loss and cerebellar ataxia in anti-Ma2-associated autoimmune encephalitis. Arquivos De Neuro-Psiquiatria, 2017, 75, 74-75.	0.8	6
31	DARS2 gene clinical spectrum: new ideas regarding an underdiagnosed leukoencephalopathy. Brain, 2014, 137, e289-e289.	7.6	5
32	Urbach-Wiethe disease presenting with partial seizures, skin lesions and typical neuroimaging features. Clinical Neurology and Neurosurgery, 2014, 126, 169-170.	1.4	5
33	Whole exome sequencing identifies three recessive FIG4-mutations in an apparently dominant pedigree with Charcot-Marie-Tooth disease. Neuromuscular Disorders, 2015, 25, 359-360.	0.6	5
34	Distal myopathy due to BICD2 mutations. Clinical Neurology and Neurosurgery, 2018, 165, 47-49.	1.4	5
35	SPG76: An extremely rare hereditary spastic paraplegia with a new expanding complicated phenotype. Revue Neurologique, 2019, 175, 572-574.	1.5	5
36	Immune-mediated inflammatory polyneuropathy overlapping Charcot-Marie-Tooth 1B. Journal of Clinical Neuroscience, 2020, 75, 228-231.	1.5	5

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37	Progressive spastic tetraplegia and axial hypotonia (STAHP) due to SOD1 deficiency: is it really a new entity?. Orphanet Journal of Rare Diseases, 2021, 16, 360.	2.7	5
38	Basilar invagination in headache associated with physical exertion and recurrent torticollis. Arquivos De Neuro-Psiquiatria, 2014, 72, 902-903.	0.8	5
39	Anterior horn degeneration in Machado-Joseph disease. Journal of the Neurological Sciences, 2016, 368, 290-291.	0.6	4
40	O'Sullivanâ€"McLeod syndrome: Unmasking a rare atypical motor neuron disease. Revue Neurologique, 2019, 175, 81-86.	1.5	4
41	Teaching Neuro Images : An extremely rare cause of treatable acute encephalopathy. Neurology, 2016, 87, e116.	1.1	3
42	Teaching Neuro Images : Leukodystrophy and progressive myoclonic epilepsy disclosing DRPLA. Neurology, 2016, 86, e58-9.	1.1	3
43	Collagen type VI-related myopathy. Practical Neurology, 2017, 17, 406-407.	1.1	3
44	Familial progressive bilateral facial paralysis in Finnish type hereditary amyloidosis. Practical Neurology, 2017, 17, 408-409.	1.1	3
45	Prognostication in <scp>MELAS</scp> syndrome and other m.3243Aâ€€G mutationâ€€associated disorders. European Journal of Neurology, 2017, 24, 231-232.	3.3	3
46	Leukodystrophy with disorders of sex development due to WT1 mutations. Journal of the Neurological Sciences, 2018, 390, 94-98.	0.6	3
47	Leigh syndrome caused by mitochondrial DNA-maintenance defects revealed by whole exome sequencing. Mitochondrion, 2019, 49, 25-34.	3.4	3
48	DykeDavidoffMasson syndrome: a combination of clinical and radiological signs not to be missed. Arquivos De Neuro-Psiquiatria, 2013, 71, 911-911.	0.8	3
49	Vitamin B12 deficiency mimicking neuroimaging features of motor neuron disease. Arquivos De Neuro-Psiquiatria, 2014, 72, 85-85.	0.8	3
50	Hypomelanosis of Ito presenting with adult-onset dementia and marked enlarged Virchow-Robin spaces. Arquivos De Neuro-Psiquiatria, 2015, 73, 366-368.	0.8	3
51	Neuroimaging features of progressive ataxia and palatal tremor. Arquivos De Neuro-Psiquiatria, 2015, 73, 633-633.	0.8	3
52	Paraneoplastic motor neuronopathy and malignant acanthosis nigricans. Arquivos De Neuro-Psiquiatria, 2019, 77, 527-527.	0.8	3
53	Adult-onset non-5q proximal spinal muscular atrophy: a comprehensive review. Arquivos De Neuro-Psiquiatria, 2021, 79, 912-923.	0.8	3
54	Retinitis pigmentosa in Lafora disease. Neurology, 2015, 85, 1087-1087.	1.1	2

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55	Non-progressive cerebellar ataxia and previous undetermined acute cerebellar injury: a mysterious clinical condition. Arquivos De Neuro-Psiquiatria, 2015, 73, 823-827.	0.8	2
56	Teaching Neuro Images : Longitudinally extensive transverse myelitis in MELAS. Neurology, 2016, 86, e37.	1.1	2
57	Letter re: Acute intermittent porphyria-related leukoencephalopathy. Neurology, 2017, 88, 718-718.	1.1	2
58	Teaching Neuro Images : Early-onset dementia and demyelinating neuropathy disclosing cerebrotendinous xanthomatosis. Neurology, 2017, 89, e134.	1.1	2
59	Broad thumbs and broad hallux: the hallmarks for the Rubinstein-Taybi syndrome. Arquivos De Neuro-Psiquiatria, 2014, 72, 81-82.	0.8	2
60	Abnormal tongue features as a clinical clue for late-onset Pompe's disease. Arquivos De Neuro-Psiquiatria, 2017, 75, 835-836.	0.8	2
61	DRPLA: An unusual disease or an underestimated cause of ataxia in Brazil?. Parkinsonism and Related Disorders, 2021, 92, 67-71.	2.2	2
62	Phelan-McDermid syndrome presenting with autistic spectrum: are we underdiagnosing chromosomal diseases in patients with autism?. Journal of Neurology, 2013, 260, 2900-2902.	3.6	1
63	Clinical applications of immunoglobulin in neuromuscular diseases: focus on inflammatory myopathies. Arquivos De Neuro-Psiquiatria, 2014, 72, 966-971.	0.8	1
64	Coiling-associated delayed cerebral hypersensitivity: Is nickel the link?. Neurology, 2015, 85, 204-204.	1.1	1
65	A reversible cause of longitudinally extensive transverse myelopathy: question. Journal of Clinical Neuroscience, 2015, 22, 1809.	1.5	1
66	Metastatic breast cancer in a man with nonprogressive ataxia and epilepsy. Neurology, 2015, 85, 1183-1184.	1.1	1
67	Far beyond the motor neuron: the role of glial cells in amyotrophic lateral sclerosis. Arquivos De Neuro-Psiquiatria, 2016, 74, 849-854.	0.8	1
68	Retinitis pigmentosa in Lafora disease: Expanding findings of progressive myoclonic epilepsy. Neurology, 2016, 86, 1563-1563.	1.1	1
69	Teaching Neuro Images : Coats disease revealing facioscapulohumeral muscular dystrophy. Neurology, 2016, 87, e39.	1.1	1
70	Motor neuron disease with leukodystrophy due to CSF1R mutation. Revue Neurologique, 2020, 176, 219-221.	1.5	1
71	Rapidly progressive bulbar-onset ALS due to SS18L1 mutation. Revue Neurologique, 2020, 176, 217-219.	1.5	1
72	Cervical Spondylotic Myelopathy Secondary to Ochronotic Vertebral Arthropathy. Neurology, 2021, 96, 627-628.	1.1	1

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73	Neurogenic T waves as clues for diagnosing hemorrhagic stroke. Arquivos De Neuro-Psiquiatria, 2014, 72, 472-472.	0.8	1
74	Longitudinally extensive transverse myelopathy in a patient with CADASIL. Arquivos De Neuro-Psiquiatria, 2015, 73, 812-812.	0.8	1
75	Duchenne muscular dystrophy: classical and new therapeutic purposes and future perspectives. Arquivos De Neuro-Psiquiatria, 2017, 75, 495-496.	0.8	1
76	Tethered cord syndrome resembling Charcot-Marie-Tooth disease in closed spinal dysraphism. Arquivos De Neuro-Psiquiatria, 2014, 72, 170-171.	0.8	1
77	Not all sellar masses are macroadenomas: think also in metastasis. Arquivos De Neuro-Psiquiatria, 2014, 72, 906-906.	0.8	1
78	Brain MRI features in late-onset nonketotic hyperglycinemia. Arquivos De Neuro-Psiquiatria, 2015, 73, 891-891.	0.8	1
79	Lumbago and alopecia in a patient with leukodystrophy: think on CARASIL. Arquivos De Neuro-Psiquiatria, 2016, 74, 599-600.	0.8	1
80	Immunosuppressors and immunomodulators in Neurology - Part I: a guide for management of patients underimmunotherapy. Arquivos De Neuro-Psiquiatria, 2021, 79, 1012-1025.	0.8	1
81	Ossifying fibroma of the maxilla and tuberous sclerosis complex. Neurology, 2015, 84, 1611-1612.	1.1	0
82	Teaching Neuro Images : Macrocerebellum and optic atrophy in a young boy. Neurology, 2016, 86, e201.	1.1	0
83	Teaching Neuro Images : Facial grimacing and sensorineural hearing loss in a woman with cirrhosis of the liver. Neurology, 2016, 87, e239.	1.1	0
84	Postictal thoracocervicofacial purpura. Practical Neurology, 2017, 17, 306-306.	1.1	0
85	Teaching NeuroImages: MR neurography for the diagnosis of hypertrophic neuropathies. Neurology, 2017, 89, e201-e201.	1.1	0
86	Teaching NeuroImages: Slowly progressive hypertrophic brachial plexopathy due to SEPT9 mutation. Neurology, 2020, 95, e109-e110.	1.1	0
87	Huntington's disease as an unexpected cause of deafness with dystonia and chorea. Parkinsonism and Related Disorders, 2020, 76, 10-12.	2.2	0
88	Teaching NeuroImages: Hopkins syndrome. Neurology, 2020, 94, e996-e997.	1.1	0
89	Neuromuscular choristoma: a rare cause of congenital non-progressive lower limb amyotrophy. Arquivos De Neuro-Psiquiatria, 2021, 79, 465-466.	0.8	0
90	Congenital ataxia due to cerebellar malformation presenting with unilateral hypoplasia. Arquivos De Neuro-Psiquiatria, 2013, 71, 198-198.	0.8	0

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91	Eyelid retraction is not a pathognomonic sign of Machado-Joseph disease in the context of spinocerebellar ataxias. Arquivos De Neuro-Psiquiatria, 2014, 72, 326-327.	0.8	0
92	Subacute compressive myelopathy secondary to extramedullary hematopoiesis. Arquivos De Neuro-Psiquiatria, 2014, 72, 740-740.	0.8	0
93	The ear as a diagnostic key in a headache syndrome. Arquivos De Neuro-Psiquiatria, 2015, 73, 728-728.	0.8	0
94	Non-aneurysmal non-traumatic subarachnoid hemorrhage: think also in acute promyelocytic leukemia. Arquivos De Neuro-Psiquiatria, 2015, 73, 1044-1044.	0.8	0
95	Epilepsy and early-onset overgrowth syndrome revealing Sotos syndrome. Arquivos De Neuro-Psiquiatria, 2017, 75, 134-134.	0.8	0
96	Burning pain attacks and red skin in a young woman. Arquivos De Neuro-Psiquiatria, 2017, 75, 491-491.	0.8	0
97	Proximal limb weakness and amyotrophy in a man with silicosis. Arquivos De Neuro-Psiquiatria, 2018, 76, 59-59.	0.8	0
98	Rapidly progressive subacute motor neuronopathy disclosing type B2 thymoma. Arquivos De Neuro-Psiquiatria, 2018, 76, 62-62.	0.8	0
99	Perforating palmar disease in TTR-related familial amyloid polyneuropathy. Arquivos De Neuro-Psiquiatria, 2018, 76, 569-569.	0.8	0
100	A complex association of cardiomyopathy, mild dysmorphisms and leukoencephalopathy. Arquivos De Neuro-Psiquiatria, 2019, 77, 292-293.	0.8	0
101	Pseudoxanthoma elasticum presenting as akinetic-rigid parkinsonism and dementia. Arquivos De Neuro-Psiquiatria, 2019, 77, 525-526.	0.8	0
102	Adult-onset cerebral X-linked adrenoleukodystrophy presenting as obsessive-compulsive disorder. Arquivos De Neuro-Psiquiatria, 2020, 78, 126-127.	0.8	0
103	Clinical and Genetic Aspects of Childhood-Onset Demyelinating Charcotâ€“Marieâ€“Tooth's Disease in Brazil. Journal of Pediatric Genetics, 2023, 12, 301-307.	0.7	0