

Paulo V Souza

List of Publications by Year in descending order

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Version: 2024-02-01

103
papers

771
citations

687363

13
h-index

610901

24
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103
all docs

103
docs citations

103
times ranked

1489
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical and Genetic Aspects of Childhood-Onset Demyelinating Charcot-Marie-Tooth's Disease in Brazil. <i>Journal of Pediatric Genetics</i> , 2023, 12, 301-307.	0.7	0
2	Clinical and radiological profile of patients with spinal muscular atrophy type 4. <i>European Journal of Neurology</i> , 2021, 28, 609-619.	3.3	23
3	<scp><i>GBE1</i></scp>-related disorders: Adult polyglucosan body disease and its neuromuscular phenotypes. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 534-543.	3.6	15
4	Cervical Spondylotic Myelopathy Secondary to Ochronotic Vertebral Arthropathy. <i>Neurology</i> , 2021, 96, 627-628.	1.1	1
5	MR imaging of inherited myopathies: a review and proposal of imaging algorithms. <i>European Radiology</i> , 2021, 31, 8498-8512.	4.5	10
6	Neuromuscular choristoma: a rare cause of congenital non-progressive lower limb amyotrophy. <i>Arquivos De Neuro-Psiquiatria</i> , 2021, 79, 465-466.	0.8	0
7	Progressive spastic tetraplegia and axial hypotonia (STAHP) due to SOD1 deficiency: is it really a new entity?. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 360.	2.7	5
8	Acute hepatic porphyrias for the neurologist: current concepts and perspectives. <i>Arquivos De Neuro-Psiquiatria</i> , 2021, 79, 68-80.	0.8	8
9	Adult-onset non-5q proximal spinal muscular atrophy: a comprehensive review. <i>Arquivos De Neuro-Psiquiatria</i> , 2021, 79, 912-923.	0.8	3
10	DRPLA: An unusual disease or an underestimated cause of ataxia in Brazil?. <i>Parkinsonism and Related Disorders</i> , 2021, 92, 67-71.	2.2	2
11	Immunosuppressors and immunomodulators in Neurology - Part I: a guide for management of patients under immunotherapy. <i>Arquivos De Neuro-Psiquiatria</i> , 2021, 79, 1012-1025.	0.8	1
12	Motor neuron disease with leukodystrophy due to CSF1R mutation. <i>Revue Neurologique</i> , 2020, 176, 219-221.	1.5	1
13	Rapidly progressive bulbar-onset ALS due to SS18L1 mutation. <i>Revue Neurologique</i> , 2020, 176, 217-219.	1.5	1
14	Myasthenia Gravis and COVID-19: Clinical Characteristics and Outcomes. <i>Frontiers in Neurology</i> , 2020, 11, 1053.	2.4	78
15	Intragenic variants in the <i>SMN1</i> gene determine the clinical phenotype in 5q spinal muscular atrophy. <i>Neurology: Genetics</i> , 2020, 6, e505.	1.9	24
16	Teaching NeuroImages: Slowly progressive hypertrophic brachial plexopathy due to SEPT9 mutation. <i>Neurology</i> , 2020, 95, e109-e110.	1.1	0
17	Huntington's disease as an unexpected cause of deafness with dystonia and chorea. <i>Parkinsonism and Related Disorders</i> , 2020, 76, 10-12.	2.2	0
18	Teaching NeuroImages: Hopkins syndrome. <i>Neurology</i> , 2020, 94, e996-e997.	1.1	0

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19	Immune-mediated inflammatory polyneuropathy overlapping Charcot-Marie-Tooth 1B. <i>Journal of Clinical Neuroscience</i> , 2020, 75, 228-231.	1.5	5
20	Adult-onset cerebral X-linked adrenoleukodystrophy presenting as obsessive-compulsive disorder. <i>Arquivos De Neuro-Psiquiatria</i> , 2020, 78, 126-127.	0.8	0
21	Motor unit number index (MUNIX) in myopathic disorders: Clinical correlations and potential pitfalls. <i>Neurophysiologie Clinique</i> , 2019, 49, 329-334.	2.2	7
22	Leigh syndrome caused by mitochondrial DNA-maintenance defects revealed by whole exome sequencing. <i>Mitochondrion</i> , 2019, 49, 25-34.	3.4	3
23	Clinical and molecular findings in a cohort of <i>ANO5</i> -related myopathy. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1225-1238.	3.7	32
24	SPG76: An extremely rare hereditary spastic paraplegia with a new expanding complicated phenotype. <i>Revue Neurologique</i> , 2019, 175, 572-574.	1.5	5
25	Atypical Motor Neuron Disease variants: Still a diagnostic challenge in Neurology. <i>Revue Neurologique</i> , 2019, 175, 221-232.	1.5	21
26	O'Sullivan's McLeod syndrome: Unmasking a rare atypical motor neuron disease. <i>Revue Neurologique</i> , 2019, 175, 81-86.	1.5	4
27	Paraneoplastic motor neuronopathy and malignant acanthosis nigricans. <i>Arquivos De Neuro-Psiquiatria</i> , 2019, 77, 527-527.	0.8	3
28	A complex association of cardiomyopathy, mild dysmorphisms and leukoencephalopathy. <i>Arquivos De Neuro-Psiquiatria</i> , 2019, 77, 292-293.	0.8	0
29	Pseudoxanthoma elasticum presenting as akinetic-rigid parkinsonism and dementia. <i>Arquivos De Neuro-Psiquiatria</i> , 2019, 77, 525-526.	0.8	0
30	Leukodystrophy with disorders of sex development due to WT1 mutations. <i>Journal of the Neurological Sciences</i> , 2018, 390, 94-98.	0.6	3
31	NFU1-Related Disorders as Key Differential Diagnosis of Cavitating Leukoencephalopathy. <i>Journal of Pediatric Genetics</i> , 2018, 07, 040-042.	0.7	9
32	Distal myopathy due to BICD2 mutations. <i>Clinical Neurology and Neurosurgery</i> , 2018, 165, 47-49.	1.4	5
33	Early-onset axonal Charcot-Marie-Tooth disease due to SACS mutation. <i>Neuromuscular Disorders</i> , 2018, 28, 169-172.	0.6	11
34	Proximal limb weakness and amyotrophy in a man with silicosis. <i>Arquivos De Neuro-Psiquiatria</i> , 2018, 76, 59-59.	0.8	0
35	Rapidly progressive subacute motor neuronopathy disclosing type B2 thymoma. <i>Arquivos De Neuro-Psiquiatria</i> , 2018, 76, 62-62.	0.8	0
36	Perforating palmar disease in TTR-related familial amyloid polyneuropathy. <i>Arquivos De Neuro-Psiquiatria</i> , 2018, 76, 569-569.	0.8	0

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37	Non-motor and Extracerebellar Features in Spinocerebellar Ataxia Type 2. <i>Cerebellum</i> , 2017, 16, 34-39.	2.5	27
38	Hereditary Spastic Paraplegia: Clinical and Genetic Hallmarks. <i>Cerebellum</i> , 2017, 16, 525-551.	2.5	169
39	Letter re: Acute intermittent porphyria-related leukoencephalopathy. <i>Neurology</i> , 2017, 88, 718-718.	1.1	2
40	Postictal thoracocervicofacial purpura. <i>Practical Neurology</i> , 2017, 17, 306-306.	1.1	0
41	New genetic causes for complex hereditary spastic paraplegia. <i>Journal of the Neurological Sciences</i> , 2017, 379, 283-292.	0.6	24
42	Collagen type VI-related myopathy. <i>Practical Neurology</i> , 2017, 17, 406-407.	1.1	3
43	Familial progressive bilateral facial paralysis in Finnish type hereditary amyloidosis. <i>Practical Neurology</i> , 2017, 17, 408-409.	1.1	3
44	Prognostication in <sc>MELAS</sc> syndrome and other m.3243Aâ€G mutationâ€associated disorders. <i>European Journal of Neurology</i> , 2017, 24, 231-232.	3.3	3
45	Teaching NeuroImages: MR neurography for the diagnosis of hypertrophic neuropathies. <i>Neurology</i> , 2017, 89, e201-e201.	1.1	0
46	Teaching Neuro <i>Images</i> : Early-onset dementia and demyelinating neuropathy disclosing cerebrotendinous xanthomatosis. <i>Neurology</i> , 2017, 89, e134.	1.1	2
47	Progressive hearing loss and cerebellar ataxia in anti-Ma2-associated autoimmune encephalitis. <i>Arquivos De Neuro-Psiquiatria</i> , 2017, 75, 74-75.	0.8	6
48	Duchenne muscular dystrophy: classical and new therapeutic purposes and future perspectives. <i>Arquivos De Neuro-Psiquiatria</i> , 2017, 75, 495-496.	0.8	1
49	Abnormal tongue features as a clinical clue for late-onset Pompe's disease. <i>Arquivos De Neuro-Psiquiatria</i> , 2017, 75, 835-836.	0.8	2
50	Epilepsy and early-onset overgrowth syndrome revealing Sotos syndrome. <i>Arquivos De Neuro-Psiquiatria</i> , 2017, 75, 134-134.	0.8	0
51	Burning pain attacks and red skin in a young woman. <i>Arquivos De Neuro-Psiquiatria</i> , 2017, 75, 491-491.	0.8	0
52	Far beyond the motor neuron: the role of glial cells in amyotrophic lateral sclerosis. <i>Arquivos De Neuro-Psiquiatria</i> , 2016, 74, 849-854.	0.8	1
53	Clinical and epidemiological profiles of non-traumatic myelopathies. <i>Arquivos De Neuro-Psiquiatria</i> , 2016, 74, 161-165.	0.8	8
54	Clinical and genetic basis of congenital myasthenic syndromes. <i>Arquivos De Neuro-Psiquiatria</i> , 2016, 74, 750-760.	0.8	27

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55	Retinitis pigmentosa in Lafora disease: Expanding findings of progressive myoclonic epilepsy. <i>Neurology</i> , 2016, 86, 1563-1563.	1.1	1
56	Teaching Neuro <i>Images</i> : Macrocerebellum and optic atrophy in a young boy. <i>Neurology</i> , 2016, 86, e201.	1.1	0
57	Anterior horn degeneration in Machado-Joseph disease. <i>Journal of the Neurological Sciences</i> , 2016, 368, 290-291.	0.6	4
58	Teaching Neuro <i>Images</i> : An extremely rare cause of treatable acute encephalopathy. <i>Neurology</i> , 2016, 87, e116.	1.1	3
59	Teaching Neuro <i>Images</i> : Coats disease revealing facioscapulohumeral muscular dystrophy. <i>Neurology</i> , 2016, 87, e39.	1.1	1
60	Infantile-onset ascending spastic paraplegia phenotype associated with SPAST mutation. <i>Journal of the Neurological Sciences</i> , 2016, 371, 34-35.	0.6	8
61	Teaching Neuro <i>Images</i> : Facial grimacing and sensorineural hearing loss in a woman with cirrhosis of the liver. <i>Neurology</i> , 2016, 87, e239.	1.1	0
62	Teaching Neuro <i>Images</i> : Longitudinally extensive transverse myelitis in MELAS. <i>Neurology</i> , 2016, 86, e37.	1.1	2
63	Teaching Neuro <i>Images</i> : Leukodystrophy and progressive myoclonic epilepsy disclosing DRPLA. <i>Neurology</i> , 2016, 86, e58-9.	1.1	3
64	Lumbago and alopecia in a patient with leukodystrophy: think on CARASIL. <i>Arquivos De Neuro-Psiquiatria</i> , 2016, 74, 599-600.	0.8	1
65	Coiling-associated delayed cerebral hypersensitivity: Is nickel the link?. <i>Neurology</i> , 2015, 85, 204-204.	1.1	1
66	A reversible cause of longitudinally extensive transverse myelopathy: question. <i>Journal of Clinical Neuroscience</i> , 2015, 22, 1809.	1.5	1
67	Retinitis pigmentosa in Lafora disease. <i>Neurology</i> , 2015, 85, 1087-1087.	1.1	2
68	Non-progressive cerebellar ataxia and previous undetermined acute cerebellar injury: a mysterious clinical condition. <i>Arquivos De Neuro-Psiquiatria</i> , 2015, 73, 823-827.	0.8	2
69	C9orf72-related disorders: expanding the clinical and genetic spectrum of neurodegenerative diseases. <i>Arquivos De Neuro-Psiquiatria</i> , 2015, 73, 246-256.	0.8	21
70	Clinical and genetic basis of familial amyotrophic lateral sclerosis. <i>Arquivos De Neuro-Psiquiatria</i> , 2015, 73, 1026-1037.	0.8	23
71	Ossifying fibroma of the maxilla and tuberous sclerosis complex. <i>Neurology</i> , 2015, 84, 1611-1612.	1.1	0
72	Metastatic breast cancer in a man with nonprogressive ataxia and epilepsy. <i>Neurology</i> , 2015, 85, 1183-1184.	1.1	1

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73	Whole exome sequencing identifies three recessive FIG4-mutations in an apparently dominant pedigree with Charcot-Marie-Tooth disease. <i>Neuromuscular Disorders</i> , 2015, 25, 359-360.	0.6	5
74	SCA1 patients may present as hereditary spastic paraplegia and must be included in spastic-ataxias group. <i>Parkinsonism and Related Disorders</i> , 2015, 21, 1243-1246.	2.2	14
75	Normal muscle structure, growth, development, and regeneration. <i>Current Reviews in Musculoskeletal Medicine</i> , 2015, 8, 176-181.	3.5	16
76	When should we test patients with familial ataxias for SCA31? A misdiagnosed condition outside Japan?. <i>Journal of the Neurological Sciences</i> , 2015, 355, 206-208.	0.6	11
77	Hypomelanosis of Ito presenting with adult-onset dementia and marked enlarged Virchow-Robin spaces. <i>Arquivos De Neuro-Psiquiatria</i> , 2015, 73, 366-368.	0.8	3
78	Neuroimaging features of progressive ataxia and palatal tremor. <i>Arquivos De Neuro-Psiquiatria</i> , 2015, 73, 633-633.	0.8	3
79	Longitudinally extensive transverse myelopathy in a patient with CADASIL. <i>Arquivos De Neuro-Psiquiatria</i> , 2015, 73, 812-812.	0.8	1
80	The ear as a diagnostic key in a headache syndrome. <i>Arquivos De Neuro-Psiquiatria</i> , 2015, 73, 728-728.	0.8	0
81	Non-aneurysmal non-traumatic subarachnoid hemorrhage: think also in acute promyelocytic leukemia. <i>Arquivos De Neuro-Psiquiatria</i> , 2015, 73, 1044-1044.	0.8	0
82	Brain MRI features in late-onset nonketotic hyperglycinemia. <i>Arquivos De Neuro-Psiquiatria</i> , 2015, 73, 891-891.	0.8	1
83	Clinical applications of immunoglobulin in neuromuscular diseases: focus on inflammatory myopathies. <i>Arquivos De Neuro-Psiquiatria</i> , 2014, 72, 966-971.	0.8	1
84	DARS2 gene clinical spectrum: new ideas regarding an underdiagnosed leukoencephalopathy. <i>Brain</i> , 2014, 137, e289-e289.	7.6	5
85	Urbach-Wiethe disease presenting with partial seizures, skin lesions and typical neuroimaging features. <i>Clinical Neurology and Neurosurgery</i> , 2014, 126, 169-170.	1.4	5
86	Vitamin B12 deficiency mimicking neuroimaging features of motor neuron disease. <i>Arquivos De Neuro-Psiquiatria</i> , 2014, 72, 85-85.	0.8	3
87	Broad thumbs and broad hallux: the hallmarks for the Rubinstein-Taybi syndrome. <i>Arquivos De Neuro-Psiquiatria</i> , 2014, 72, 81-82.	0.8	2
88	Neurogenic T waves as clues for diagnosing hemorrhagic stroke. <i>Arquivos De Neuro-Psiquiatria</i> , 2014, 72, 472-472.	0.8	1
89	Bright tongue sign: a diagnostic marker for amyotrophic lateral sclerosis. <i>Arquivos De Neuro-Psiquiatria</i> , 2014, 72, 572-572.	0.8	6
90	Brain MRI features in Lhermitte-Duclos disease. <i>Arquivos De Neuro-Psiquiatria</i> , 2014, 72, 645-645.	0.8	6

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91	Paracoccidioidomycosis: a rare cause of infectious encephalomyelopathy. <i>Arquivos De Neuro-Psiquiatria</i> , 2014, 72, 904-905.	0.8	6
92	Basilar invagination in headache associated with physical exertion and recurrent torticollis. <i>Arquivos De Neuro-Psiquiatria</i> , 2014, 72, 902-903.	0.8	5
93	Tethered cord syndrome resembling Charcot-Marie-Tooth disease in closed spinal dysraphism. <i>Arquivos De Neuro-Psiquiatria</i> , 2014, 72, 170-171.	0.8	1
94	Eyelid retraction is not a pathognomonic sign of Machado-Joseph disease in the context of spinocerebellar ataxias. <i>Arquivos De Neuro-Psiquiatria</i> , 2014, 72, 326-327.	0.8	0
95	Subacute compressive myelopathy secondary to extramedullary hematopoiesis. <i>Arquivos De Neuro-Psiquiatria</i> , 2014, 72, 740-740.	0.8	0
96	Not all sellar masses are macroadenomas: think also in metastasis. <i>Arquivos De Neuro-Psiquiatria</i> , 2014, 72, 906-906.	0.8	1
97	Early-onset familial Alzheimer's disease related to presenilin 1 mutation resembling autosomal dominant spinocerebellar ataxia. <i>Journal of Neurology</i> , 2013, 260, 1177-1179.	3.6	9
98	Variable phenotype and severity of sialidosis expressed in two siblings presenting with ataxia and macular cherry-red spots. <i>Journal of Clinical Neuroscience</i> , 2013, 20, 1327-1328.	1.5	6
99	Phelan-McDermid syndrome presenting with autistic spectrum: are we underdiagnosing chromosomal diseases in patients with autism?. <i>Journal of Neurology</i> , 2013, 260, 2900-2902.	3.6	1
100	The cerebellum in Parkinson's disease and parkinsonism in cerebellar disorders. <i>Brain</i> , 2013, 136, e248-e248.	7.6	12
101	DykeDavidoffMasson syndrome: a combination of clinical and radiological signs not to be missed. <i>Arquivos De Neuro-Psiquiatria</i> , 2013, 71, 911-911.	0.8	3
102	Congenital ataxia due to cerebellar malformation presenting with unilateral hypoplasia. <i>Arquivos De Neuro-Psiquiatria</i> , 2013, 71, 198-198.	0.8	0
103	Early-onset epilepsy as the main neurological manifestation of cerebrotendinous xanthomatosis. <i>Epilepsy and Behavior</i> , 2012, 24, 380-381.	1.7	12