Antonio Federico

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/4907229/publications.pdf

Version: 2024-02-01

101	3,048	279798	175258
papers	citations	23 h-index	52 g-index

105	105	105	5755
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mitochondria, oxidative stress and neurodegeneration. Journal of the Neurological Sciences, 2012, 322, 254-262.	0.6	621
2	Apoptosis and Oxidative Stress in Neurodegenerative Diseases. Journal of Alzheimer's Disease, 2014, 42, S125-S152.	2.6	467
3	Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (CADASIL) as a model of small vessel disease: update on clinical, diagnostic, and management aspects. BMC Medicine, 2017, 15, 41.	5.5	212
4	A suspicion index for early diagnosis and treatment of cerebrotendinous xanthomatosis. Journal of Inherited Metabolic Disease, 2014, 37, 421-429.	3.6	109
5	The effect of NOTCH3 pathogenic variant position on CADASIL disease severity: NOTCH3 EGFr 1â€“6 pathogenic variant are associated with a more severe phenotype and lower survival compared with EGFr 7â€“34 pathogenic variant. Genetics in Medicine, 2019, 21, 676-682.	2.4	102
6	Structural and Functional Brain Changes beyond Visual System in Patients with Advanced Glaucoma. PLoS ONE, 2014, 9, e105931.	2.5	91
7	WRNMutation Update: Mutation Spectrum, Patient Registries, and Translational Prospects. Human Mutation, 2017, 38, 7-15.	2.5	79
8	Cerebrotendinous Xanthomatosis. Journal of Child Neurology, 2003, 18, 633-638.	1.4	77
9	Hereditary cerebral small vessel diseases: A review. Journal of the Neurological Sciences, 2012, 322, 25-30.	0.6	76
10	Next Generation Molecular Diagnosis of Hereditary Spastic Paraplegias: An Italian Cross-Sectional Study. Frontiers in Neurology, 2018, 9, 981.	2.4	64
11	Long term follow-up to evaluate the efficacy of miglustat treatment in Italian patients with Niemann-Pick disease type C. Orphanet Journal of Rare Diseases, 2015, 10, 22.	2.7	54
12	Two novel HTRA1 mutations in a European CARASIL patient. Neurology, 2014, 82, 898-900.	1.1	53
13	Evaluation of cholesterol metabolism in cerebrotendinous xanthomatosis. Journal of Inherited Metabolic Disease, 2016, 39, 75-83.	3.6	52
14	No lockdown for neurological diseases during COVID19 pandemic infection. Neurological Sciences, 2020, 41, 999-1001.	1.9	47
15	Heterozygous mutations of <scp>HTRA</scp>1 gene in patients with familial cerebral small vessel disease. CNS Neuroscience and Therapeutics, 2017, 23, 759-765.	3.9	46
16	The spectrum of magnetic resonance findings in cerebrotendinous xanthomatosis: redefinition and evidence of new markers of disease progression. Journal of Neurology, 2017, 264, 862-874.	3.6	43
17	Can untreated PKU patients escape from intellectual disability? A systematic review. Orphanet Journal of Rare Diseases, 2018, 13, 149.	2.7	36
18	Operationalizing mild cognitive impairment criteria in small vessel disease: the VMCI-Tuscany Study. , 2016, 12, 407-418.		34

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19	Location, number and factors associated with cerebral microbleeds in an Italian-British cohort of CADASIL patients. PLoS ONE, 2018, 13, e0190878.	2.5	33
20	Next-generation sequencing approach to hyperCKemia. Neurology: Genetics, 2019, 5, e352.	1.9	31
21	Is the time ripe for new diagnostic criteria of cognitive impairment due to cerebrovascular disease? Consensus report of the International Congress on Vascular Dementia working group. BMC Medicine, 2016, 14, 162.	5.5	30
22	Cerebrotendinous xanthomatosis with predominant parkinsonian syndrome: Further confirmation of the clinical heterogeneity. Movement Disorders, 2000, 15, 1017-1019.	3.9	26
23	The safety and effectiveness of chenodeoxycholic acid treatment in patients with cerebrotendinous xanthomatosis: two retrospective cohort studies. Neurological Sciences, 2020, 41, 943-949.	1.9	26
24	The European Reference Network for Rare Neurological Diseases. Frontiers in Neurology, 2020, 11, 616569.	2.4	26
25	Hereditary diffuse leukoencephalopathy with axonal spheroids (HDLS): update on molecular genetics. Neurological Sciences, 2016, 37, 1565-1569.	1.9	25
26	Mitochondrial dysfunction in hereditary spastic paraparesis with mutations in DDHD1/SPG28. Journal of the Neurological Sciences, 2016, 362, 287-291.	0.6	24
27	Schnyder corneal crystalline dystrophy: Description of a new family with evidence of abnormal lipid storage in skin fibroblasts. , 1998, 75, 35-39.		23
28	Perilesional edema in brain metastasis from non-small cell lung cancer (NSCLC) as predictor of response to radiosurgery (SRS). Neurological Sciences, 2017, 38, 975-982.	1.9	23
29	Primary cilium alterations and expression changes of Patched1 proteins in niemannâ€pick type C disease. Journal of Cellular Physiology, 2018, 233, 663-672.	4.1	22
30	Alzheimerâ€™s disease: the controversial approval of Aducanumab. Neurological Sciences, 2021, 42, 3069-3070.	1.9	20
31	SPG2 mimicking multiple sclerosis in a family identified using next generation sequencing. Journal of the Neurological Sciences, 2017, 375, 198-202.	0.6	18
32	Double trouble? Progranulin mutation and C9ORF72 repeat expansion in a case of primary non-fluent aphasia. Journal of the Neurological Sciences, 2014, 341, 176-178.	0.6	17
33	Treatment of SPG5 with cholesterol-lowering drugs. Journal of Neurology, 2015, 262, 2783-2785.	3.6	17
34	Novel POLG mutations and variable clinical phenotypes in 13 Italian patients. Neurological Sciences, 2017, 38, 563-570.	1.9	17
35	Effects of Sapropterin on Endothelium-Dependent Vasodilation in Patients With CADASIL. Stroke, 2014, 45, 2959-2966.	2.0	16
36	The role of dentate nuclei in human oculomotor control: insights from cerebrotendinous xanthomatosis. Journal of Physiology, 2017, 595, 3607-3620.	2.9	16

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37	Changes in grey matter volume and functional connectivity in cluster headache versus migraine. Brain Imaging and Behavior, 2020, 14, 496-504.	2.1	16
38	Exploring the role of music therapy in multiple sclerosis: brief updates from research to clinical practice. Neurological Sciences, 2019, 40, 2277-2285.	1.9	15
39	A Novel CSF1R Mutation in a Patient with Clinical and Neuroradiological Features of Hereditary Diffuse Leukoencephalopathy with Axonal Spheroids. Journal of Alzheimer's Disease, 2015, 47, 319-322.	2.6	14
40	Characteristic Eye Movements in Ataxia-Telangiectasia-Like Disorder: An Explanatory Hypothesis. Frontiers in Neurology, 2017, 8, 596.	2.4	14
41	Application of the DSM-5 Criteria for Major Neurocognitive Disorder to Vascular MCI Patients. Dementia and Geriatric Cognitive Disorders Extra, 2018, 8, 104-116.	1.3	13
42	Primary familial brain calcification with a novel SLC20A2 mutation: Analysis of PiTâ€2 expression and localization. Journal of Cellular Physiology, 2018, 233, 2324-2331.	4.1	13
43	Anti-Saccades in Cerebellar Ataxias Reveal a Contribution of the Cerebellum in Executive Functions. Frontiers in Neurology, 2018, 9, 274.	2.4	13
44	Expert opinion on diagnosing, treating and managing patients with cerebrotendinous xanthomatosis (CTX): a modified Delphi study. Orphanet Journal of Rare Diseases, 2021, 16, 353.	2.7	13
45	Circulating Biomarkers in Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy Patients. Journal of Stroke and Cerebrovascular Diseases, 2017, 26, 823-833.	1.6	12
46	Relevance of brain lesion location for cognition in vascular mild cognitive impairment. NeuroImage: Clinical, 2019, 22, 101789.	2.7	12
47	HTRA1 expression profile and activity on TGFâ€Î²2 signaling in HTRA1 mutation carriers. Journal of Cellular Physiology, 2020, 235, 7120-7127.	4.1	12
48	Update on Several/Certain Adult-Onset Genetic Leukoencephalopathies: Clinical Signs and Molecular Confirmation. Journal of Alzheimer's Disease, 2014, 42, S27-S35.	2.6	11
49	Oculodentodigital Dysplasia with Massive Brain Calcification and a New Mutation of GJA1 Gene. Journal of Alzheimer's Disease, 2015, 49, 27-30.	2.6	11
50	Eye movement changes in autosomal dominant spinocerebellar ataxias. Neurological Sciences, 2020, 41, 1719-1734.	1.9	11
51	Brain Awareness Week, CoVID-19 infection and Neurological Sciences. Neurological Sciences, 2020, 41, 747-748.	1.9	9
52	Temporal lobe abnormalities in neurosyphilis. Practical Neurology, 2014, 14, 449-450.	1.1	8
53	Analysis of opa1 isoforms expression and apoptosis regulation in autosomal dominant optic atrophy (ADOA) patients with mutations in the opa1 gene. Journal of the Neurological Sciences, 2015, 351, 99-108.	0.6	8
54	Imaging of the thymus in myotonic dystrophy type 1. Neurological Sciences, 2018, 39, 347-351.	1.9	8

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55	New disease modifying therapies for two genetic childhood-onset neurometabolic disorders (metachromatic leucodystrophy and adrenoleucodystrophy). Neurological Sciences, 2021, 42, 2603-2606.	1.9	8
56	POLR3A variants in hereditary spastic paraparesis and ataxia: clinical, genetic, and neuroradiological findings in a cohort of Italian patients. Neurological Sciences, 2022, 43, 1071-1077.	1.9	8
57	Expanding the clinical and genetic spectrum of pathogenic variants in <scp>STIM1</scp>. Muscle and Nerve, 2021, 64, 567-575.	2.2	7
58	Mitochondrial recessive ataxia syndrome: A neurological rarity not to be missed. Journal of the Neurological Sciences, 2015, 349, 254-255.	0.6	6
59	Oxidative stress-induced apoptosis in peripheral blood lymphocytes from patients with POLG-related disorders. Journal of the Neurological Sciences, 2016, 368, 359-368.	0.6	6
60	Primary familial brain calcification caused by MYORG mutations in an Italian family. Parkinsonism and Related Disorders, 2019, 67, 24-26.	2.2	6
61	Neurological music therapy during the COVID-19 outbreak: updates and future challenges. Neurological Sciences, 2022, 43, 3473-3478.	1.9	6
62	A case of 3243A>G mutation in mtDNA presenting as apparently idiopathic hyperCKemia. Journal of the Neurological Sciences, 2014, 338, 232-234.	0.6	5
63	Eye movements in genetic parkinsonisms affecting the Î±-synuclein, PARK9, and manganese network. Clinical Neurophysiology, 2017, 128, 2450-2453.	1.5	5
64	Myoclonus epilepsy, retinitis pigmentosa, leukoencephalopathy and cerebral calcifications associated with a novel m.5513G>A mutation in the MT-TW gene. Biochemical and Biophysical Research Communications, 2018, 500, 158-162.	2.1	5
65	Functional magnetic resonance imaging with encoding task in patients with mild cognitive impairment and different severity of leukoaraiosis. Psychiatry Research - Neuroimaging, 2018, 282, 126-131.	1.8	5
66	Tarlovâ€™s cyst as an underestimated cause of persistent genital arousal disorder: a case report and review. Neurological Sciences, 2020, 41, 3337-3339.	1.9	5
67	Eye movement changes in mitochondrial neurogastrointestinal encephalomyopathy (MNGIE). Journal of the Neurological Sciences, 2015, 350, 107-109.	0.6	4
68	A case of pneumocephalus as complication of ozone therapy: diagnosis and treatment. Neurological Sciences, 2020, 41, 481-483.	1.9	4
69	Italian neurology: past, present and future. Functional Neurology, 2011, 26, 73-6.	1.3	4
70	Sporadic hereditary motor and sensory neuropathies: Advances in the diagnosis using next generation sequencing technology. Journal of the Neurological Sciences, 2015, 359, 409-417.	0.6	3
71	Vitamin D levels in cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL). Neurological Sciences, 2017, 38, 1333-1336.	1.9	3
72	Progression of oculomotor deficit in a patient with posterior cortical atrophy. Journal of the Neurological Sciences, 2017, 372, 459-463.	0.6	3

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73	Sixth nerve and superior division of third nerve palsy due to intracranial extension of multiple myeloma. A diagnostic challenge and differential diagnosis. Neurological Sciences, 2018, 39, 593-594.	1.9	3
74	Experiencing COVID19 pandemic and neurology: learning by the recent reports and by old literary or scientific descriptions. Neurological Sciences, 2020, 41, 1323-1327.	1.9	3
75	Paroxysmal supraventricular tachycardia in anti-musk Myasthenia gravis: A case report. Journal of the Neurological Sciences, 2016, 369, 250-251.	0.6	2
76	Demyelinating polyneuropathy in a case of antiâ€ŁGI1 encephalitis. Muscle and Nerve, 2017, 56, E2-E3.	2.2	2
77	Awareness of rare and genetic neurological diseases among italian neurologist. A national survey. Neurological Sciences, 2020, 41, 1567-1570.	1.9	2
78	Rare Neurologic Diseases and Neurological Sciences: a report for the celebration of the 2020 Rare Diseases Day. Neurological Sciences, 2020, 41, 491-495.	1.9	2
79	Co-occurrence of DMPK expansion and CLCN1 mutation in a patient with myotonia. Neurological Sciences, 2021, 42, 5365-5368.	1.9	2
80	Message from the Editor-in-Chief. Neurological Sciences, 2021, 42, 1-7.	1.9	2
81	Compound heterozygosity in the GALC gene in a late onset Iranian patient with spastic paraparesis, peripheral neuropathy and leukoencephalopathy. Neurological Sciences, 2017, 38, 1721-1722.	1.9	2
82	Cutaneous Sensory and Autonomic Small Fiber Neuropathy in HTRA1-Related Cerebral Small Vessel Disease. Journal of Neuropathology and Experimental Neurology, 2021, 80, 713-716.	1.7	2
83	Early/lateâ€life adversities and behavioural phenotypes: insight into metabolomics, genomics and connectomics. Journal of Intellectual Disability Research, 2016, 60, 833-834.	2.0	1
84	Clinical and MRI improvement in a case of progressive multifocal leukoencephalopathy. Neurological Sciences, 2017, 38, 1517-1519.	1.9	1
85	Advances in clinical neurology through the journal â€œNeurological Sciencesâ€•(2015â€“2016). Neurological Sciences, 2017, 38, 9-18.	1.9	1
86	Perilesional edema in brain cancer: Independent prognosticator or epiphenomenon of biomolecular signature?. Radiotherapy and Oncology, 2018, 129, 183-184.	0.6	1
87	A case of reversible cerebral vasoconstriction syndrome and cavernous hemangioma: just a coincidence?. Neurological Sciences, 2018, 39, 1989-1990.	1.9	1
88	The history of the first 40 years of Neurological Sciences. Neurological Sciences, 2019, 40, 2239-2250.	1.9	1
89	Message from the Editor-in-Chief Neurological Sciences is 40-year-old (founded in 1979)!. Neurological Sciences, 2019, 40, 5-11.	1.9	1
90	Scientific publishing in the COVID-19 era: successes and pitfalls. Neurological Sciences, 2020, 41, 1643-1645.	1.9	1

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91	Our Impact Factor 2015 is increased to 1.783: a comment from the Editor-in-Chief. Neurological Sciences, 2016, 37, 1189-1189.	1.9	0
92	C9ORF72 gene expansion in a patient with intellectual disability and psychiatric disease. Neurological Sciences, 2017, 38, 207-208.	1.9	0
93	Message from the editor in chief. Neurological Sciences, 2018, 39, 9-13.	1.9	0
94	News on the journal Neurological Sciences in 2017. Neurological Sciences, 2018, 39, 15-21.	1.9	0
95	Acknowledgments to the members of the Editorial Board. Neurological Sciences, 2018, 39, 397-397.	1.9	0
96	Replay to: Phenotypic spectrum of POLG1 mutations. Neurological Sciences, 2018, 39, 575-575.	1.9	0
97	Neurological sciences impact factor 2017 is increased to 2.285: a comment from the Editor in Chief. Neurological Sciences, 2018, 39, 1145-1146.	1.9	0
98	Message from the editor-in-chief, 2020. Neurological Sciences, 2020, 41, 5-9.	1.9	0
99	A case of painless neuralgic amyotrophy responsive to immunotherapy. Neurological Sciences, 2020, 41, 2297-2298.	1.9	0
100	More then 10Âyears as Editor-in-Chief of Neurological Sciences: greetings and acknowledgements. Neurological Sciences, 2021, 42, 4861-4863.	1.9	0
101	A case of spastic paraplegia type 11 mimicking a GM2-gangliosidosis. Neurological Sciences, 2022, 43, 2849-2852.	1.9	0