Antonio Federico

List of Publications by Citations

84 papers	2,132 citations	20 h-index	44 g-index
105 ext. papers	2,612 ext. citations	3.6 avg, IF	5.12 L-index

#	Paper	IF	Citations
84	Mitochondria, oxidative stress and neurodegeneration. *Journal of the Neurological Sciences*, **2012**, 322, 254-62	3.2	524
83	Apoptosis and oxidative stress in neurodegenerative diseases. *Journal of Alzheimer's Disease*, **2014**, 42 Suppl 3, S125-52	4.3	337
82	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	11.4	126
81	A suspicion index for early diagnosis and treatment of cerebrotendinous xanthomatosis. *Journal of Inherited Metabolic Disease*, **2014**, 37, 421-9	5.4	72
80	Cerebrotendinous xanthomatosis: clinical manifestations, diagnostic criteria, pathogenesis, and therapy. *Journal of Child Neurology*, **2003**, 18, 633-8	2.5	63
79	Structural and functional brain changes beyond visual system in patients with advanced glaucoma. *PLoS ONE*, **2014**, 9, e105931	3.7	62
78	Hereditary cerebral small vessel diseases: a review. *Journal of the Neurological Sciences*, **2012**, 322, 25-30	3.2	62
77	WRN Mutation Update: Mutation Spectrum, Patient Registries, and Translational Prospects. *Human Mutation*, **2017**, 38, 7-15	4.7	56
76	The effect of NOTCH3 pathogenic variant position on CADASIL disease severity: NOTCH3 EGFr 1-6 pathogenic variant are associated with a more severe phenotype and lower survival compared with EGFr 7-34 pathogenic variant. *Genetics in Medicine*, **2019**, 21, 676-682	8.1	53
75	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	4.2	45
74	Two novel HTRA1 mutations in a European CARASIL patient. *Neurology*, **2014**, 82, 898-900	6.5	42
73	Evaluation of cholesterol metabolism in cerebrotendinous xanthomatosis. *Journal of Inherited Metabolic Disease*, **2016**, 39, 75-83	5.4	37
72	Next Generation Molecular Diagnosis of Hereditary Spastic Paraplegias: An Italian Cross-Sectional Study. *Frontiers in Neurology*, **2018**, 9, 981	4.1	33
71	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	5.5	31
70	Heterozygous mutations of HTRA1 gene in patients with familial cerebral small vessel disease. *CNS Neuroscience and Therapeutics*, **2017**, 23, 759-765	6.8	31
69	Operationalizing mild cognitive impairment criteria in small vessel disease: the VMCI-Tuscany Study. *Alzheimer's and Dementia*, **2016**, 12, 407-18	1.2	26
68	Hereditary diffuse leukoencephalopathy with axonal spheroids (HDLS): update on molecular genetics. *Neurological Sciences*, **2016**, 37, 1565-9	3.5	23

67	Can untreated PKU patients escape from intellectual disability? A systematic review. *Orphanet Journal of Rare Diseases*, **2018**, 13, 149	4.2	21
66	Cerebrotendinous xanthomatosis with predominant parkinsonian syndrome: further confirmation of the clinical heterogeneity. *Movement Disorders*, **2000**, 15, 1017-9	7	20
65	Primary cilium alterations and expression changes of Patched1 proteins in niemann-pick type C disease. *Journal of Cellular Physiology*, **2018**, 233, 663-672	7	18
64	Location, number and factors associated with cerebral microbleeds in an Italian-British cohort of CADASIL patients. *PLoS ONE*, **2018**, 13, e0190878	3.7	18
63	Schnyder corneal crystalline dystrophy: description of a new family with evidence of abnormal lipid storage in skin fibroblasts. *American Journal of Medical Genetics Part A*, **1998**, 75, 35-9		17
62	Mitochondrial dysfunction in hereditary spastic paraparesis with mutations in DDHD1/SPG28. *Journal of the Neurological Sciences*, **2016**, 362, 287-91	3.2	15
61	Double trouble? Progranulin mutation and C9ORF72 repeat expansion in a case of primary non-fluent aphasia. *Journal of the Neurological Sciences*, **2014**, 341, 176-8	3.2	15
60	The safety and effectiveness of chenodeoxycholic acid treatment in patients with cerebrotendinous xanthomatosis: two retrospective cohort studies. *Neurological Sciences*, **2020**, 41, 943-949	3.5	14
59	Novel POLG mutations and variable clinical phenotypes in 13 Italian patients. *Neurological Sciences*, **2017**, 38, 563-570	3.5	13
58	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	3.5	12
57	Next-generation sequencing approach to hyperCKemia: A 2-year cohort study. *Neurology: Genetics*, **2019**, 5, e352	3.8	12
56	Treatment of SPG5 with cholesterol-lowering drugs. *Journal of Neurology*, **2015**, 262, 2783-5	5.5	11
55	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, 319-22	4.3	11
54	The role of dentate nuclei in human oculomotor control: insights from cerebrotendinous xanthomatosis. *Journal of Physiology*, **2017**, 595, 3607-3620	3.9	10
53	Anti-Saccades in Cerebellar Ataxias Reveal a Contribution of the Cerebellum in Executive Functions. *Frontiers in Neurology*, **2018**, 9, 274	4.1	10
52	Exploring the role of music therapy in multiple sclerosis: brief updates from research to clinical practice. *Neurological Sciences*, **2019**, 40, 2277-2285	3.5	10
51	SPG2 mimicking multiple sclerosis in a family identified using next generation sequencing. *Journal of the Neurological Sciences*, **2017**, 375, 198-202	3.2	9
50	Update on several/certain adult-onset genetic leukoencephalopathies: clinical signs and molecular confirmation. *Journal of Alzheimer's Disease*, **2014**, 42 Suppl 3, S27-35	4.3	9

49	Circulating Biomarkers in Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy Patients. *Journal of Stroke and Cerebrovascular Diseases*, **2017**, 26, 823-833	2.8	9
48	Effects of sapropterin on endothelium-dependent vasodilation in patients with CADASIL: a randomized controlled trial. *Stroke*, **2014**, 45, 2959-66	6.7	9
47	Changes in grey matter volume and functional connectivity in cluster headache versus migraine. *Brain Imaging and Behavior*, **2020**, 14, 496-504	4.1	9
46	Relevance of brain lesion location for cognition in vascular mild cognitive impairment. *NeuroImage: Clinical*, **2019**, 22, 101789	5.3	8
45	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	3.2	8
44	Primary familial brain calcification with a novel SLC20A2 mutation: Analysis of PiT-2 expression and localization. *Journal of Cellular Physiology*, **2018**, 233, 2324-2331	7	8
43	Temporal lobe abnormalities in neurosyphilis. *Practical Neurology*, **2014**, 14, 449-50	2.4	8
42	Characteristic Eye Movements in Ataxia-Telangiectasia-Like Disorder: An Explanatory Hypothesis. *Frontiers in Neurology*, **2017**, 8, 596	4.1	8
41	Oculodentodigital dysplasia with massive brain calcification and a new mutation of GJA1 gene. *Journal of Alzheimer's Disease*, **2016**, 49, 27-30	4.3	8
40	The European Reference Network for Rare Neurological Diseases. *Frontiers in Neurology*, **2020**, 11, 616569	4.1	8
39	HTRA1 expression profile and activity on TGF-β signaling in HTRA1 mutation carriers. *Journal of Cellular Physiology*, **2020**, 235, 7120-7127	7	7
38	Application of the DSM-5 Criteria for Major Neurocognitive Disorder to Vascular MCI Patients. *Dementia and Geriatric Cognitive Disorders Extra*, **2018**, 8, 104-116	2.5	7
37	Imaging of the thymus in myotonic dystrophy type 1. *Neurological Sciences*, **2018**, 39, 347-351	3.5	7
36	Eye movement changes in autosomal dominant spinocerebellar ataxias. *Neurological Sciences*, **2020**, 41, 1719-1734	3.5	5
35	Mitochondrial recessive ataxia syndrome: A neurological rarity not to be missed. *Journal of the Neurological Sciences*, **2015**, 349, 254-5	3.2	5
34	Eye movements in genetic parkinsonisms affecting the α-synuclein, PARK9, and manganese network. *Clinical Neurophysiology*, **2017**, 128, 2450-2453	4.3	4
33	Eye movement changes in mitochondrial neurogastrointestinal encephalomyopathy (MNGIE). *Journal of the Neurological Sciences*, **2015**, 350, 107-9	3.2	4
32	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	3.4	4

31	Primary familial brain calcification caused by MYORG mutations in an Italian family. *Parkinsonism and Related Disorders*, **2019**, 67, 24-26	3.6	4
30	A case of 3243A>G mutation in mtDNA presenting as apparently idiopathic hyperCKemia. *Journal of the Neurological Sciences*, **2014**, 338, 232-4	3.2	4
29	Oxidative stress-induced apoptosis in peripheral blood lymphocytes from patients with POLG-related disorders. *Journal of the Neurological Sciences*, **2016**, 368, 359-68	3.2	4
28	Sporadic hereditary motor and sensory neuropathies: Advances in the diagnosis using next generation sequencing technology. *Journal of the Neurological Sciences*, **2015**, 359, 409-17	3.2	3
27	Tarlov's cyst as an underestimated cause of persistent genital arousal disorder: a case report and review. *Neurological Sciences*, **2020**, 41, 3337-3339	3.5	3
26	Italian neurology: past, present and future. *Functional Neurology*, **2011**, 26, 73-6	2.2	3
25	POLR3A variants in hereditary spastic paraparesis and ataxia: clinical, genetic, and neuroradiological findings in a cohort of Italian patients. *Neurological Sciences*, **2021**, 1	3.5	3
24	A case of pneumocephalus as complication of ozone therapy: diagnosis and treatment. *Neurological Sciences*, **2020**, 41, 481-483	3.5	3
23	Expert opinion on diagnosing, treating and managing patients with cerebrotendinous xanthomatosis (CTX): a modified Delphi study. *Orphanet Journal of Rare Diseases*, **2021**, 16, 353	4.2	3
22	Progression of oculomotor deficit in a patient with posterior cortical atrophy. *Journal of the Neurological Sciences*, **2017**, 372, 459-463	3.2	2
21	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	2.9	2
20	Expanding the clinical and genetic spectrum of pathogenic variants in STIM1. *Muscle and Nerve*, **2021**, 64, 567-575	3.4	2
19	Neurological music therapy during the COVID-19 outbreak: updates and future challenges.. *Neurological Sciences*, **2022**, 1	3.5	2
18	Demyelinating polyneuropathy in a case of anti-LGI1 encephalitis. *Muscle and Nerve*, **2017**, 56, E2-E3	3.4	1
17	Clinical and MRI improvement in a case of progressive multifocal leukoencephalopathy. *Neurological Sciences*, **2017**, 38, 1517-1519	3.5	1
16	Vitamin D levels in cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL). *Neurological Sciences*, **2017**, 38, 1333-1336	3.5	1
15	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	3.5	1
14	Perilesional edema in brain cancer: Independent prognosticator or epiphenomenon of biomolecular signature?. *Radiotherapy and Oncology*, **2018**, 129, 183-184	5.3	1

13	A case of reversible cerebral vasoconstriction syndrome and cavernous hemangioma: just a coincidence?. *Neurological Sciences*, **2018**, 39, 1989-1990	3.5	1
12	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	3.5	1
11	Paroxysmal supraventricular tachycardia in anti-musk Myasthenia gravis: A case report. *Journal of the Neurological Sciences*, **2016**, 369, 250-251	3.2	1
10	Co-occurrence of DMPK expansion and CLCN1 mutation in a patient with myotonia. *Neurological Sciences*, **2021**, 42, 5365-5368	3.5	1
9	Awareness of rare and genetic neurological diseases among italian neurologist. A national survey. *Neurological Sciences*, **2020**, 41, 1567-1570	3.5	0
8	Cutaneous Sensory and Autonomic Small Fiber Neuropathy in HTRA1-Related Cerebral Small Vessel Disease. *Journal of Neuropathology and Experimental Neurology*, **2021**, 80, 713-716	3.1	0
7	A case of painless neuralgic amyotrophy responsive to immunotherapy. *Neurological Sciences*, **2020**, 41, 2297-2298	3.5	
6	Message from the editor in chief. *Neurological Sciences*, **2018**, 39, 9-13	3.5	
5	Replay to: Phenotypic spectrum of POLG1 mutations. *Neurological Sciences*, **2018**, 39, 575	3.5	
4	C9ORF72 gene expansion in a patient with intellectual disability and psychiatric disease. *Neurological Sciences*, **2017**, 38, 207-208	3.5	
3	A case of spastic paraplegia type 11 mimicking a GM2-gangliosidosis.. *Neurological Sciences*, **2022**, 43, 2849	3.5	
2	Message from the editor-in-chief, 2020. *Neurological Sciences*, **2020**, 41, 5-9	3.5	
1	Message from the Editor-in-Chief. *Neurological Sciences*, **2021**, 42, 1-7	3.5	