# Antonio Federico

## List of Publications by Year in Descending Order



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| 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 | A case of spastic paraplegia type 11 mimicking a GM2-gangliosidosis.. *Neurological Sciences*, **2022**, 43, 2849 | 3.5 | |
| 83 | Neurological music therapy during the COVID-19 outbreak: updates and future challenges.. *Neurological Sciences*, **2022**, 1 | 3.5 | 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 | 3.1 | 0 |
| 81 | 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 |
| 80 | Co-occurrence of DMPK expansion and CLCN1 mutation in a patient with myotonia. *Neurological Sciences*, **2021**, 42, 5365-5368 | 3.5 | 1 |
| 79 | 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 |
| 78 | Expanding the clinical and genetic spectrum of pathogenic variants in STIM1. *Muscle and Nerve*, **2021**, 64, 567-575 | 3.4 | 2 |
| 77 | Message from the Editor-in-Chief. *Neurological Sciences*, **2021**, 42, 1-7 | 3.5 | |
| 76 | A case of painless neuralgic amyotrophy responsive to immunotherapy. *Neurological Sciences*, **2020**, 41, 2297-2298 | 3.5 | |
| 75 | Eye movement changes in autosomal dominant spinocerebellar ataxias. *Neurological Sciences*, **2020** , 41, 1719-1734 | 3.5 | 5 |
| 74 | 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 |
| 73 | Awareness of rare and genetic neurological diseases among italian neurologist. A national survey. *Neurological Sciences*, **2020**, 41, 1567-1570 | 3.5 | 0 |
| 72 | HTRA1 expression profile and activity on TGF-β signaling in HTRA1 mutation carriers. *Journal of Cellular Physiology*, **2020**, 235, 7120-7127 | 7 | 7 |
| 71 | 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 |
| 70 | Message from the editor-in-chief, 2020. *Neurological Sciences*, **2020**, 41, 5-9 | 3.5 | |
| 69 | Changes in grey matter volume and functional connectivity in cluster headache versus migraine. *Brain Imaging and Behavior*, **2020**, 14, 496-504 | 4.1 | 9 |
| 68 | A case of pneumocephalus as complication of ozone therapy: diagnosis and treatment. *Neurological Sciences*, **2020**, 41, 481-483 | 3.5 | 3 |

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| 67 | The European Reference Network for Rare Neurological Diseases. *Frontiers in Neurology*, **2020**, 11, 616569 | 4.1 | 8 |
| 66 | Relevance of brain lesion location for cognition in vascular mild cognitive impairment. *NeuroImage: Clinical*, **2019**, 22, 101789 | 5.3 | 8 |
| 65 | 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 |
| 64 | 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 |
| 63 | Primary familial brain calcification caused by MYORG mutations in an Italian family. *Parkinsonism and Related Disorders*, **2019**, 67, 24-26 | 3.6 | 4 |
| 62 | Next-generation sequencing approach to hyperCKemia: A 2-year cohort study. *Neurology: Genetics*, **2019**, 5, e352 | 3.8 | 12 |
| 61 | 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 |
| 60 | 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 |
| 59 | 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 |
| 58 | Message from the editor in chief. *Neurological Sciences*, **2018**, 39, 9-13 | 3.5 | |
| 57 | Perilesional edema in brain cancer: Independent prognosticator or epiphenomenon of biomolecular signature?. *Radiotherapy and Oncology*, **2018**, 129, 183-184 | 5.3 | 1 |
| 56 | Replay to: Phenotypic spectrum of POLG1 mutations. *Neurological Sciences*, **2018**, 39, 575 | 3.5 | |
| 55 | 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 |
| 54 | 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 |
| 53 | A case of reversible cerebral vasoconstriction syndrome and cavernous hemangioma: just a coincidence?. *Neurological Sciences*, **2018**, 39, 1989-1990 | 3.5 | 1 |
| 52 | Anti-Saccades in Cerebellar Ataxias Reveal a Contribution of the Cerebellum in Executive Functions. *Frontiers in Neurology*, **2018**, 9, 274 | 4.1 | 10 |
| 51 | Location, number and factors associated with cerebral microbleeds in an Italian-British cohort of CADASIL patients. *PLoS ONE*, **2018**, 13, e0190878 | 3.7 | 18 |
| 50 | Imaging of the thymus in myotonic dystrophy type 1. *Neurological Sciences*, **2018**, 39, 347-351 | 3.5 | 7 |

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| 49 | Next Generation Molecular Diagnosis of Hereditary Spastic Paraplegias: An Italian Cross-Sectional Study. *Frontiers in Neurology*, **2018**, 9, 981 | 4.1 | 33 |
| 48 | Can untreated PKU patients escape from intellectual disability? A systematic review. *Orphanet Journal of Rare Diseases*, **2018**, 13, 149 | 4.2 | 21 |
| 47 | 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 |
| 46 | Demyelinating polyneuropathy in a case of anti-LGI1 encephalitis. *Muscle and Nerve*, **2017**, 56, E2-E3 | 3.4 | 1 |
| 45 | Novel POLG mutations and variable clinical phenotypes in 13 Italian patients. *Neurological Sciences*, **2017**, 38, 563-570 | 3.5 | 13 |
| 44 | 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 |
| 43 | SPG2 mimicking multiple sclerosis in a family identified using next generation sequencing. *Journal of the Neurological Sciences*, **2017**, 375, 198-202 | 3.2 | 9 |
| 42 | The role of dentate nuclei in human oculomotor control: insights from cerebrotendinous xanthomatosis. *Journal of Physiology*, **2017**, 595, 3607-3620 | 3.9 | 10 |
| 41 | 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 |
| 40 | Clinical and MRI improvement in a case of progressive multifocal leukoencephalopathy. *Neurological Sciences*, **2017**, 38, 1517-1519 | 3.5 | 1 |
| 39 | Vitamin D levels in cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL). *Neurological Sciences*, **2017**, 38, 1333-1336 | 3.5 | 1 |
| 38 | 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 |
| 37 | Eye movements in genetic parkinsonisms affecting the □synuclein, PARK9, and manganese network. *Clinical Neurophysiology*, **2017**, 128, 2450-2453 | 4.3 | 4 |
| 36 | Heterozygous mutations of HTRA1 gene in patients with familial cerebral small vessel disease. *CNS Neuroscience and Therapeutics*, **2017**, 23, 759-765 | 6.8 | 31 |
| 35 | Progression of oculomotor deficit in a patient with posterior cortical atrophy. *Journal of the Neurological Sciences*, **2017**, 372, 459-463 | 3.2 | 2 |
| 34 | C9ORF72 gene expansion in a patient with intellectual disability and psychiatric disease. *Neurological Sciences*, **2017**, 38, 207-208 | 3.5 | |
| 33 | WRN Mutation Update: Mutation Spectrum, Patient Registries, and Translational Prospects. *Human Mutation*, **2017**, 38, 7-15 | 4.7 | 56 |
| 32 | 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 |

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| 31 | Characteristic Eye Movements in Ataxia-Telangiectasia-Like Disorder: An Explanatory Hypothesis. *Frontiers in Neurology*, **2017**, 8, 596 | 4.1 | 8 |
| 30 | 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 |
| 29 | Evaluation of cholesterol metabolism in cerebrotendinous xanthomatosis. *Journal of Inherited Metabolic Disease*, **2016**, 39, 75-83 | 5.4 | 37 |
| 28 | Operationalizing mild cognitive impairment criteria in small vessel disease: the VMCI-Tuscany Study. *Alzheimer's and Dementia*, **2016**, 12, 407-18 | 1.2 | 26 |
| 27 | Hereditary diffuse leukoencephalopathy with axonal spheroids (HDLS): update on molecular genetics. *Neurological Sciences*, **2016**, 37, 1565-9 | 3.5 | 23 |
| 26 | Mitochondrial dysfunction in hereditary spastic paraparesis with mutations in DDHD1/SPG28. *Journal of the Neurological Sciences*, **2016**, 362, 287-91 | 3.2 | 15 |
| 25 | 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 |
| 24 | 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 |
| 23 | Paroxysmal supraventricular tachycardia in anti-musk Myasthenia gravis: A case report. *Journal of the Neurological Sciences*, **2016**, 369, 250-251 | 3.2 | 1 |
| 22 | Eye movement changes in mitochondrial neurogastrointestinal encephalomyopathy (MNGIE). *Journal of the Neurological Sciences*, **2015**, 350, 107-9 | 3.2 | 4 |
| 21 | 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 |
| 20 | 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 |
| 19 | 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 |
| 18 | Treatment of SPG5 with cholesterol-lowering drugs. *Journal of Neurology*, **2015**, 262, 2783-5 | 5.5 | 11 |
| 17 | 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 |
| 16 | Mitochondrial recessive ataxia syndrome: A neurological rarity not to be missed. *Journal of the Neurological Sciences*, **2015**, 349, 254-5 | 3.2 | 5 |
| 15 | 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 |
| 14 | Apoptosis and oxidative stress in neurodegenerative diseases. *Journal of Alzheimer's Disease*, **2014**, 42 Suppl 3, S125-52 | 4.3 | 337 |

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| 13 | Temporal lobe abnormalities in neurosyphilis. *Practical Neurology*, **2014**, 14, 449-50 | 2.4 | 8 |
| 12 | Update on several/certain adult-onset genetic leukoencephalopathies: clinical signs and molecular confirmation. *Journal of Alzheimerɪs Disease*, **2014**, 42 Suppl 3, S27-35 | 4.3 | 9 |
| 11 | 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 |
| 10 | Structural and functional brain changes beyond visual system in patients with advanced glaucoma. *PLoS ONE*, **2014**, 9, e105931 | 3.7 | 62 |
| 9 | Two novel HTRA1 mutations in a European CARASIL patient. *Neurology*, **2014**, 82, 898-900 | 6.5 | 42 |
| 8 | Effects of sapropterin on endothelium-dependent vasodilation in patients with CADASIL: a randomized controlled trial. *Stroke*, **2014**, 45, 2959-66 | 6.7 | 9 |
| 7 | A suspicion index for early diagnosis and treatment of cerebrotendinous xanthomatosis. *Journal of Inherited Metabolic Disease*, **2014**, 37, 421-9 | 5.4 | 72 |
| 6 | Hereditary cerebral small vessel diseases: a review. *Journal of the Neurological Sciences*, **2012**, 322, 25-30 | 3.2 | 62 |
| 5 | Mitochondria, oxidative stress and neurodegeneration. *Journal of the Neurological Sciences*, **2012**, 322, 254-62 | 3.2 | 524 |
| 4 | Italian neurology: past, present and future. *Functional Neurology*, **2011**, 26, 73-6 | 2.2 | 3 |
| 3 | Cerebrotendinous xanthomatosis: clinical manifestations, diagnostic criteria, pathogenesis, and therapy. *Journal of Child Neurology*, **2003**, 18, 633-8 | 2.5 | 63 |
| 2 | Cerebrotendinous xanthomatosis with predominant parkinsonian syndrome: further confirmation of the clinical heterogeneity. *Movement Disorders*, **2000**, 15, 1017-9 | 7 | 20 |
| 1 | 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 |