Patrizia Formichi

List of Publications by Year in Descending Order

75 papers	2,230 citations	22 h-index	46 g-index
81 ext. papers	2,560 ext. citations	4.1 avg, IF	4.84 L-index

#	Paper	IF	Citations
75	Fibroblast growth factor 21 and grow differentiation factor 15 are sensitive biomarkers of mitochondrial diseases due to mitochondrial transfer-RNA mutations and mitochondrial DNA deletions. *Neurological Sciences*, **2020**, 41, 3653-3662	3.5	6
74	HTRA1 expression profile and activity on TGF-β signaling in HTRA1 mutation carriers. *Journal of Cellular Physiology*, **2020**, 235, 7120-7127	7	7
73	Changes in grey matter volume and functional connectivity in cluster headache versus migraine. *Brain Imaging and Behavior*, **2020**, 14, 496-504	4.1	9
72	Relevance of brain lesion location for cognition in vascular mild cognitive impairment. *NeuroImage: Clinical*, **2019**, 22, 101789	5.3	8
71	Cerebellar ataxia associated with anti-glutamic acid decarboxylase antibodies: a case report. *Neurological Sciences*, **2019**, 40, 1303-1306	3.5	3
70	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
69	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
68	Novel POLG mutations and variable clinical phenotypes in 13 Italian patients. *Neurological Sciences*, **2017**, 38, 563-570	3.5	13
67	SPG2 mimicking multiple sclerosis in a family identified using next generation sequencing. *Journal of the Neurological Sciences*, **2017**, 375, 198-202	3.2	9
66	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
65	Hereditary diffuse leukoencephalopathy with axonal spheroids (HDLS): update on molecular genetics. *Neurological Sciences*, **2016**, 37, 1565-9	3.5	23
64	Adult Alexander disease with de novo c.1193C>T heterozygous variant in GFAP gene. *Neurological Sciences*, **2016**, 37, 143-145	3.5	0
63	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
62	Paroxysmal supraventricular tachycardia in anti-musk Myasthenia gravis: A case report. *Journal of the Neurological Sciences*, **2016**, 369, 250-251	3.2	1
61	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
60	Sporadic PEO caused by a novel POLG variation and a Twinkle mutation: digenic inheritance?. *Neurological Sciences*, **2015**, 36, 1713-5	3.5	2
59	Hydroxychloroquine neuromyotoxicity: a case with rapid course and complete recovery. *Neurological Sciences*, **2015**, 36, 2293-4	3.5	5

58	Primary familial brain calcification: update on molecular genetics. *Neurological Sciences*, **2015**, 36, 787-94	3.5	25
57	Cerebrotendinous xanthomatosis: recurrence of the CYP27A1 mutation p.Arg479Cys in Sardinia. *Neurological Sciences*, **2014**, 35, 1303-5	3.5	1
56	Hereditary diffuse leukoencephalopathy with axonal spheroids: three patients with stroke-like presentation carrying new mutations in the CSF1R gene. *Journal of Neurology*, **2014**, 261, 768-72	5.5	24
55	From the liver to the brain: manganese matters: focus on cirrhosis-related Parkinsonism. *Neurological Sciences*, **2014**, 35, 521-2	3.5	5
54	Tarlov cysts: clinical evaluation of an Italian cohort of patients. *Neurological Sciences*, **2014**, 35, 613	3.5	
53	Apoptosis and oxidative stress in neurodegenerative diseases. *Journal of Alzheimer's Disease*, **2014**, 42 Suppl 3, S125-52	4.3	337
52	Adult-onset phenylketonuria revealed by acute reversible dementia, prosopagnosia and parkinsonism. *Journal of Neurology*, **2014**, 261, 2446-8	5.5	7
51	A suspicion index for early diagnosis and treatment of cerebrotendinous xanthomatosis. *Journal of Inherited Metabolic Disease*, **2014**, 37, 421-9	5.4	72
50	Homozygosity and severity of phenotypic presentation in a CADASIL family. *Neurological Sciences*, **2014**, 35, 91-3	3.5	17
49	Effects of cerebrolysin administration on oxidative stress-induced apoptosis in lymphocytes from CADASIL patients. *Neurological Sciences*, **2013**, 34, 553-6	3.5	2
48	Tarlov cysts: clinical evaluation of an italian cohort of patients. *Neurological Sciences*, **2013**, 34, 1679-82	3.5	27
47	Human peripheral blood lymphocytes and fibroblasts as Notch3 expression models. *Journal of Cellular Physiology*, **2012**, 227, 1771-5	7	4
46	High frequency of OPA1 mutations causing high ADOA prevalence in south-eastern Sicily, Italy. *Clinical Genetics*, **2012**, 82, 277-82	4	2
45	Hereditary cerebral small vessel diseases: a review. *Journal of the Neurological Sciences*, **2012**, 322, 25-30	3.2	62
44	Evaluating gaze control on a multi-target sequencing task: the distribution of fixations is evidence of exploration optimisation. *Computers in Biology and Medicine*, **2012**, 42, 235-44	7	8
43	Evaluating the human ongoing visual search performance by eye tracking application and sequencing tests. *Computer Methods and Programs in Biomedicine*, **2012**, 107, 468-77	6.9	9
42	Mitochondria, oxidative stress and neurodegeneration. *Journal of the Neurological Sciences*, **2012**, 322, 254-62	3.2	524
41	Altered apoptosis regulation in Kufor-Rakeb syndrome patients with mutations in the ATP13A2 gene. *Journal of Cellular and Molecular Medicine*, **2012**, 16, 1916-23	5.6	8

40	Cerebrolysin administration reduces oxidative stress-induced apoptosis in lymphocytes from healthy individuals. *Journal of Cellular and Molecular Medicine*, **2012**, 16, 2840-3	5.6	13
39	Oxidative stress-induced apoptosis in two patients with Alagille syndrome. *Journal of the Neurological Sciences*, **2011**, 308, 49-56	3.2	4
38	Italian neurology: past, present and future. *Functional Neurology*, **2011**, 26, 73-6	2.2	3
37	CSF Biomarkers Profile in CADASIL-A Model of Pure Vascular Dementia: Usefulness in Differential Diagnosis in the Dementia Disorder. *International Journal of Alzheimer's Disease*, **2010**, 2010,	3.7	12
36	High frequency of exon 10 mutations in the NOTCH3 gene in Italian CADASIL families: phenotypic peculiarities. *Journal of Neurology*, **2010**, 257, 1039-42	5.5	9
35	A second MNGIE patient without typical mitochondrial skeletal muscle involvement. *Neurological Sciences*, **2010**, 31, 491-4	3.5	17
34	Ataxia with vitamin E deficiency: update of molecular diagnosis. *Neurological Sciences*, **2010**, 31, 511-5	3.5	90
33	Apoptosis in CADASIL: an in vitro study of lymphocytes and fibroblasts from a cohort of Italian patients. *Journal of Cellular Physiology*, **2009**, 219, 494-502	7	20
32	Occurrence of ankylosing spondylitis and multiple sclerosis-like syndrome in a HLA-B27 positive patient. *Neurological Sciences*, **2009**, 30, 329-32	3.5	8
31	Oxidative-stress-induced apoptosis in PBLs of two patients with Parkinson disease secondary to alpha-synuclein mutation. *Journal of the Neurological Sciences*, **2008**, 267, 120-4	3.2	25
30	Psychosine-induced apoptosis and cytokine activation in immune peripheral cells of Krabbe patients. *Journal of Cellular Physiology*, **2007**, 212, 737-43	7	39
29	Increased lung surfactant phosphatidylcholine in patients affected by lysosomal storage diseases. *Journal of Inherited Metabolic Disease*, **2007**, 30, 983	5.4	12
28	Leber's hereditary optic neuropathy associated with cocaine, ecstasy and telithromycin consumption. *Journal of Neurology*, **2007**, 254, 255-6	5.5	4
27	A new case of short-chain acyl-CoA dehydrogenase deficiency: clinical, biochemical, genetic and (1)H-NMR spectroscopic studies. *Neurological Sciences*, **2007**, 28, 328-30	3.5	6
26	Physiology and pathology of notch signalling system. *Journal of Cellular Physiology*, **2006**, 207, 300-8	7	53
25	Cerebrospinal fluid tau, A beta, and phosphorylated tau protein for the diagnosis of Alzheimer's disease. *Journal of Cellular Physiology*, **2006**, 208, 39-46	7	46
24	Human fibroblasts undergo oxidative stress-induced apoptosis without internucleosomal DNA fragmentation. *Journal of Cellular Physiology*, **2006**, 208, 289-97	7	26
23	A novel mutation producing premature termination codon at the OPA1 gene causes autosomal dominant optic atrophy. *Journal of Neurology*, **2006**, 253, 672-3	5.5	5

22	Clinical and molecular diagnosis of cerebrotendinous xanthomatosis with a review of the mutations in the CYP27A1 gene. *Neurological Sciences*, **2006**, 27, 143-9	3.5	122
21	The spectrum of mutations for the diagnosis of vanishing white matter disease. *Neurological Sciences*, **2006**, 27, 271-7	3.5	36
20	Leukoencephalopathy as a rare complication of hepatitis C infection. *Neurological Sciences*, **2006**, 27, 360-3	3.5	17
19	A novel heteroplasmic tRNA(Leu(CUN)) mtDNA point mutation associated with chronic progressive external ophthalmoplegia. *Biochemical and Biophysical Research Communications*, **2005**, 327, 675-8	3.4	16
18	Commentary to mitDNA research for the pathogenesis of mitochondrial disorders. *Biochemical and Biophysical Research Communications*, **2005**, 336, 1003-4	3.4	4
17	Video-based eye tracking: our experience with Advanced Stimuli Design for Eye Tracking software. *Annals of the New York Academy of Sciences*, **2005**, 1039, 575-9	6.5	1
16	The spectrum of mutations for CADASIL diagnosis. *Neurological Sciences*, **2005**, 26, 117-24	3.5	79
15	Lung involvement in Niemann-Pick disease type C1: improvement with bronchoalveolar lavage. *Neurological Sciences*, **2005**, 26, 171-3	3.5	27
14	Typical pathological changes of CADASIL in the optic nerve. *Neurological Sciences*, **2005**, 26, 271-4	3.5	17
13	Wilson's disease with Leu492Ser mutation and arylsulfatase A pseudodeficiency: just a coincidence?. *Neurological Sciences*, **2004**, 25, 18-20	3.5	
12	Normalisation of serum cholestanol concentration in a patient with cerebrotendinous xanthomatosis by combined treatment with chenodeoxycholic acid, simvastatin and LDL apheresis. *Neurological Sciences*, **2004**, 25, 185-91	3.5	29
11	Cerebrotendinous xanthomatosis: clinical manifestations, diagnostic criteria, pathogenesis, and therapy. *Journal of Child Neurology*, **2003**, 18, 633-8	2.5	63
10	The Primrose syndrome with progressive neurological involvement and cerebral calcification. *Journal of Neurology*, **2002**, 249, 1466-8	5.5	15
9	Severe metabolic abnormalities in the white matter of patients with vacuolating megalencephalic leukoencephalopathy with subcortical cysts. A proton MR spectroscopic imaging study. *Journal of Neurology*, **2001**, 248, 403-9	5.5	36
8	Cerebrotendinous xanthomatosis: heterogeneity of clinical phenotype with evidence of previously undescribed ophthalmological findings. *Journal of Inherited Metabolic Disease*, **2001**, 24, 696-706	5.4	39
7	The Siena experience on rare neurological diseases: diagnosis, therapy and research model for investigations of central and peripheral nervous systems and muscle. *Functional Neurology*, **2001**, 16, 263-9	2.2	2
6	Heteroplasmy of the A3243G transition of mitochondrial tRNA(Leu(UUR)) in a MELAS case and in a 25-week-old miscarried fetus. *Journal of Neurology*, **2000**, 247, 885-7	5.5	13
5	Detection of beta-A4 amyloid and its precursor protein in the muscle of a patient with juvenile neuronal ceroid lipofuscinosis (Spielmeyer-Vogt-Sjögren). *Acta Neuropathologica*, **1999**, 98, 78-84	14.3	4

4	Vitamin E serum levels in Rett syndrome. *Journal of the Neurological Sciences*, **1998**, 156, 227-30	3.2	21
3	Association in the same patient of autosomal dominant progressive external ophthalmoplegia with multiple mtDNA deletions and X-linked ichthyosis: clinical, biochemical, histological, submicroscopic and molecular genetic study. *Journal of Submicroscopic Cytology and Pathology*, **1998**, 30, 521-6		2
2	Increased apoptotic response to 2-deoxy-D-ribose in ataxia-telangiectasia. *Journal of the Neurological Sciences*, **1996**, 144, 128-34	3.2	13
1	Vitamin E serum levels are normal in ataxia telangiectasia (Louis-Bar disease). *Journal of the Neurological Sciences*, **1996**, 141, 114-6	3.2	5