Patrizia Formichi

List of Publications by Year
in descending order

81	2,828	257101 24	182168 51
papers	citations	h-index	g-index

81	81	81	5317
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.	0.3	621
2	Apoptosis and Oxidative Stress in Neurodegenerative Diseases. Journal of Alzheimer's Disease, 2014, 42, S125-S152.	1.2	467
3	Clinical and molecular diagnosis of cerebrotendinous xanthomatosis with a review of the mutations in the CYP27A1 gene. Neurological Sciences, 2006, 27, 143-149.	0.9	151
4	Ataxia with vitamin E deficiency: update of molecular diagnosis. Neurological Sciences, 2010, 31, 511-515.	0.9	119
5	A suspicion index for early diagnosis and treatment of cerebrotendinous xanthomatosis. Journal of Inherited Metabolic Disease, 2014, 37, 421-429.	1.7	109
6	The spectrum of mutations for CADASIL diagnosis. Neurological Sciences, 2005, 26, 117-124.	0.9	87
7	Cerebrotendinous Xanthomatosis. Journal of Child Neurology, 2003, 18, 633-638.	0.7	77
8	Hereditary cerebral small vessel diseases: A review. Journal of the Neurological Sciences, 2012, 322, 25-30.	0.3	76
9	Cerebrospinal fluid tau, AÃŸ, and phosphorylated tau protein for the diagnosis of Alzheimer's disease. Journal of Cellular Physiology, 2006, 208, 39-46.	2.0	66
10	Physiology and pathology of notch signalling system. Journal of Cellular Physiology, 2006, 207, 300-308.	2.0	65
11	Cerebrotendinous xanthomatosis: Heterogeneity of clinical phenotype with evidence of previously undescribed ophthalmological findings. Journal of Inherited Metabolic Disease, 2001, 24, 696-706.	1.7	50
12	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, 403-409.	1.8	42
13	Psychosine-induced apoptosis and cytokine activation in immune peripheral cells of Krabbe patients. Journal of Cellular Physiology, 2007, 212, 737-743.	2.0	42
14	The spectrum of mutations for the diagnosis of vanishing white matter disease. Neurological Sciences, 2006, 27, 271-277.	0.9	41
15	Tarlov cysts: clinical evaluation of an italian cohort of patients. Neurological Sciences, 2013, 34, 1679-1682.	0.9	41
16	Normalisation of serum cholestanol concentration in a patient with cerebrotendinous xanthomatosis by combined treatment with chenodeoxycholic acid, simvastatin and LDH apheresis. Neurological Sciences, 2004, 25, 185-191.	0.9	35
17	Lung involvement in Niemann-Pick disease type C1: improvement with bronchoalveolar lavage. Neurological Sciences, 2005, 26, 171-173.	0.9	32
18	Primary familial brain calcification: update on molecular genetics. Neurological Sciences, 2015, 36, 787-794.	0.9	31

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19	Human fibroblasts undergo oxidative stress-induced apoptosis without internucleosomal DNA fragmentation. Journal of Cellular Physiology, 2006, 208, 289-297.	2.0	29
20	Hereditary diffuse leukoencephalopathy with axonal spheroids: three patients with stroke-like presentation carrying new mutations in the CSF1R gene. Journal of Neurology, 2014, 261, 768-772.	1.8	27
21	Oxidative-stress-induced apoptosis in PBLs of two patients with Parkinson disease secondary to alpha-synuclein mutation. Journal of the Neurological Sciences, 2008, 267, 120-124.	0.3	26
22	Vitamin E serum levels in Rett syndrome. Journal of the Neurological Sciences, 1998, 156, 227-230.	0.3	25
23	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.	2.0	25
24	Hereditary diffuse leukoencephalopathy with axonal spheroids (HDLS): update on molecular genetics. Neurological Sciences, 2016, 37, 1565-1569.	0.9	25
25	Primary cilium alterations and expression changes of Patched1 proteins in niemannâ€pick type C disease. Journal of Cellular Physiology, 2018, 233, 663-672.	2.0	22
26	Typical pathological changes of CADASIL in the optic nerve. Neurological Sciences, 2005, 26, 271-274.	0.9	20
27	Homozygosity and severity of phenotypic presentation in a CADASIL family. Neurological Sciences, 2014, 35, 91-93.	0.9	20
28	Alzheimerâ€™s disease: the controversial approval of Aducanumab. Neurological Sciences, 2021, 42, 3069-3070.	0.9	20
29	Leukoencephalopathy as a rare complication of hepatitis C infection. Neurological Sciences, 2006, 27, 360-363.	0.9	19
30	A second MNGIE patient without typical mitochondrial skeletal muscle involvement. Neurological Sciences, 2010, 31, 491-494.	0.9	19
31	The Primrose syndrome with progressive neurological involvement and cerebral calcification. Journal of Neurology, 2002, 249, 1466-1468.	1.8	18
32	SPG2 mimicking multiple sclerosis in a family identified using next generation sequencing. Journal of the Neurological Sciences, 2017, 375, 198-202.	0.3	18
33	A novel heteroplasmic tRNALeu(CUN) mtDNA point mutation associated with chronic progressive external ophthalmoplegia. Biochemical and Biophysical Research Communications, 2005, 327, 675-678.	1.0	17
34	Cerebrolysin administration reduces oxidative stressâ€induced apoptosis in limphocytes from healthy individuals. Journal of Cellular and Molecular Medicine, 2012, 16, 2840-2843.	1.6	17
35	Novel POLG mutations and variable clinical phenotypes in 13 Italian patients. Neurological Sciences, 2017, 38, 563-570.	0.9	17
36	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, 885-887.	1.8	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.	1.1	16
38	Increased apoptotic response to 2-deoxy- d -ribose in ataxia-telangiectasia. Journal of the Neurological Sciences, 1996, 144, 128-134.	0.3	15
39	CSF Biomarkers Profile in CADASILâ€"A Model of Pure Vascular Dementia: Usefulness in Differential Diagnosis in the Dementia Disorder. International Journal of Alzheimer's Disease, 2010, 2010, 1-6.	1.1	15
40	Adult-onset phenylketonuria revealed by acute reversible dementia, prosopagnosia and parkinsonism. Journal of Neurology, 2014, 261, 2446-2448.	1.8	15
41	Increased lung surfactant phosphatidylcholine in patients affected by lysosomal storage diseases. Journal of Inherited Metabolic Disease, 2007, 30, 983-983.	1.7	14
42	Evaluating the human ongoing visual search performance by eye tracking application and sequencing tests. Computer Methods and Programs in Biomedicine, 2012, 107, 468-477.	2.6	13
43	Primary familial brain calcification with a novel SLC20A2 mutation: Analysis of PiTâ€2 expression and localization. Journal of Cellular Physiology, 2018, 233, 2324-2331.	2.0	13
44	Relevance of brain lesion location for cognition in vascular mild cognitive impairment. NeuroImage: Clinical, 2019, 22, 101789.	1.4	12
45	HTRA1 expression profile and activity on TGFâ€Î²2 signaling in HTRA1 mutation carriers. Journal of Cellular Physiology, 2020, 235, 7120-7127.	2.0	12
46	Ataxia with isolated vitamin E deficiency: a treatable neurologic disorder resembling Friedreich?s ataxia. Neurological Sciences, 2004, 25, 119-21.	0.9	11
47	High frequency of exon 10 mutations in the NOTCH3 gene in Italian CADASIL families: phenotypic peculiarities. Journal of Neurology, 2010, 257, 1039-1042.	1.8	10
48	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, 235-244.	3.9	10
49	Occurrence of ankylosing spondylitis and multiple sclerosis-like syndrome in a HLA-B27 positive patient. Neurological Sciences, 2009, 30, 329-332.	0.9	9
50	Altered apoptosis regulation in Kuforâ€"Rakeb syndrome patients with mutations in the ATP13A2 gene. Journal of Cellular and Molecular Medicine, 2012, 16, 1916-1923.	1.6	9
51	Hydroxychloroquine neuromyotoxicity: a case with rapid course and complete recovery. Neurological Sciences, 2015, 36, 2293-2294.	0.9	9
52	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.	0.9	9
53	A novel mutation producing premature termination codon at the OPA1 gene causes autosomal dominant optic atrophy. Journal of Neurology, 2006, 253, 672-673.	1.8	8
54	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.	0.3	8

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55	A new case of short-chain acyl-CoA dehydrogenase deficiency: clinical, biochemical, genetic and 1H-NMR spectroscopic studies. Neurological Sciences, 2007, 28, 328-330.	0.9	7
56	Vitamin E serum levels are normal in ataxia telangiectasia (Louis-Bar disease). Journal of the Neurological Sciences, 1996, 141, 114-116.	0.3	6
57	High frequency of OPA1 mutations causing high ADOA prevalence in southâ€eastern Sicily, Italy. Clinical Genetics, 2012, 82, 277-282.	1.0	6
58	Oxidative stress-induced apoptosis in peripheral blood lymphocytes from patients with POLG-related disorders. Journal of the Neurological Sciences, 2016, 368, 359-368.	0.3	6
59	Detection of Î²-A4 amyloid and its precursor protein in the muscle of a patient with juvenile neuronal ceroid lipofuscinosis (Spielmeyer-Vogt-SjÃ¶gren). Acta Neuropathologica, 1999, 98, 78-84.	3.9	5
60	Leberâ€™s Hereditary Optic Neuropathy associated with cocaine, ecstasy and telithromycin consumption. Journal of Neurology, 2007, 254, 255-256.	1.8	5
61	Oxidative stress-induced apoptosis in two patients with Alagille syndrome. Journal of the Neurological Sciences, 2011, 308, 49-56.	0.3	5
62	Human peripheral blood lymphocytes and fibroblasts as Notch3 expression models. Journal of Cellular Physiology, 2012, 227, 1771-1775.	2.0	5
63	From the liver to the brain: manganese matters. Neurological Sciences, 2014, 35, 521-522.	0.9	5
64	Sporadic PEO caused by a novel POLG variation and a Twinkle mutation: digenic inheritance?. Neurological Sciences, 2015, 36, 1713-1715.	0.9	5
65	Cerebellar ataxia associated with anti-glutamic acid decarboxylase antibodies: a case report. Neurological Sciences, 2019, 40, 1303-1306.	0.9	5
66	Video-Based Eye Tracking: Our Experience with Advanced Stimuli Design for Eye Tracking Software. Annals of the New York Academy of Sciences, 2005, 1039, 575-579.	1.8	4
67	Commentary to mitDNA research for the pathogenesis of mitochondrial disorders. Biochemical and Biophysical Research Communications, 2005, 336, 1003-1004.	1.0	4
68	Italian neurology: past, present and future. Functional Neurology, 2011, 26, 73-6.	1.3	4
69	Effects of cerebrolysin administration on oxidative stress-induced apoptosis in lymphocytes from CADASIL patients. Neurological Sciences, 2013, 34, 553-556.	0.9	3
70	Rita Levi-Montalcini, one of the most prominent Italian personalities of the twentieth century. Neurological Sciences, 2013, 34, 131-133.	0.9	2
71	Paroxysmal supraventricular tachycardia in anti-musk Myasthenia gravis: A case report. Journal of the Neurological Sciences, 2016, 369, 250-251.	0.3	2
72	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.	0.9	2

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73	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.	0.3	2
74	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.	1.3	2
75	Cerebrotendinous xanthomatosis: recurrence of the CYP27A1 mutation p.Arg479Cys in Sardinia. Neurological Sciences, 2014, 35, 1303-5.	0.9	1
76	Adult Alexander disease with de novo c.1193C>T heterozygous variant in GFAP gene. Neurological Sciences, 2016, 37, 143-145.	0.9	1
77	Early/lateâ€life adversities and behavioural phenotypes: insight into metabolomics, genomics and connectomics. Journal of Intellectual Disability Research, 2016, 60, 833-834.	1.2	1
78	Wilson?s disease with Leu492Ser mutation and arylsulfatase A pseudodeficiency: just a coincidence?. Neurological Sciences, 2004, 25, 18-20.	0.9	0
79	Progressive neurodegenerative syndrome associated with Langerhans cell histiocytosis: a rare condition that we have to consider in patients with sporadic spastic ataxia and diabetes insipidus. Neurological Sciences, 2012, 33, 489-490.	0.9	0
80	Tarlov Cysts: clinical evaluation of an Italian cohort of patients. Neurological Sciences, 2014, 35, 613-613.	0.9	0
81	News on the journal Neurological Sciences in 2017. Neurological Sciences, 2018, 39, 15-21.	0.9	0