Maria Teresa Dotti

List of Publications by Year
in descending order

94 papers	2,491 citations	218677 26 h-index	243625 44 g-index
94 all docs	94 docs citations	94 times ranked	3740 citing authors

#	Article	IF	Citations
1	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
2	MECP2 mutation in male patients with nonâ€specific Xâ€linked mental retardation. FEBS Letters, 2000, 481, 285-288.	2.8	208
3	A suspicion index for early diagnosis and treatment of cerebrotendinous xanthomatosis. Journal of Inherited Metabolic Disease, 2014, 37, 421-429.	3.6	109
4	Primary familial brain calcification: Genetic analysis and clinical spectrum. Movement Disorders, 2014, 29, 1691-1695.	3.9	95
5	Cerebrotendinous Xanthomatosis. Journal of Child Neurology, 2003, 18, 633-638.	1.4	77
6	Hereditary cerebral small vessel diseases: A review. Journal of the Neurological Sciences, 2012, 322, 25-30.	0.6	76
7	The Cerebral Autosomal-Dominant Arteriopathy With Subcortical Infarcts and Leukoencephalopathy (CADASIL) Scale. Stroke, 2012, 43, 2871-2876.	2.0	68
8	CADASIL in central Italy: a retrospective clinical and genetic study in 229 patients. Journal of Neurology, 2015, 262, 134-141.	3.6	67
9	Physiology and pathology of notch signalling system. Journal of Cellular Physiology, 2006, 207, 300-308.	4.1	65
10	Next Generation Molecular Diagnosis of Hereditary Spastic Paraplegias: An Italian Cross-Sectional Study. Frontiers in Neurology, 2018, 9, 981.	2.4	64
11	Two novel HTRA1 mutations in a European CARASIL patient. Neurology, 2014, 82, 898-900.	1.1	53
12	A Novel NOTCH3 Frameshift Deletion and Mitochondrial Abnormalities in a Patient With CADASIL. Archives of Neurology, 2004, 61, 942.	4.5	52
13	Evaluation of cholesterol metabolism in cerebrotendinous xanthomatosis. Journal of Inherited Metabolic Disease, 2016, 39, 75-83.	3.6	52
14	Narcolepsy is a common phenotype in HSAN IE and ADCA-DN. Brain, 2014, 137, 1643-1655.	7.6	49
15	Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE): Position paper on diagnosis, prognosis, and treatment by the <scp>MNGIE</scp> International Network. Journal of Inherited Metabolic Disease, 2021, 44, 376-387.	3.6	47
16	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
17	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
18	Progression of Brain Atrophy in Spinocerebellar Ataxia Type 2: A Longitudinal Tensor-Based Morphometry Study. PLoS ONE, 2014, 9, e89410.	2.5	41

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19	Cerebrotendinous xanthomatosis: evidence of lipomatous hypertrophy of the atrial septum. Journal of Neurology, 1998, 245, 723-726.	3.6	39
20	Polyneuropathy in cerebrotendinous xanthomatosis and response to treatment with chenodeoxycholic acid. Journal of Neurology, 2013, 260, 268-274.	3.6	35
21	Operationalizing mild cognitive impairment criteria in small vessel disease: the VMCI-Tuscany Study. , 2016, 12, 407-418.		34
22	Location, number and factors associated with cerebral microbleeds in an Italian-British cohort of CADASIL patients. PLoS ONE, 2018, 13, e0190878.	2.5	33
23	A pathogenic mutation on exon 21 of the NOTCH3 gene causing CADASIL in an octogenarian paucisymptomatic patient. Journal of the Neurological Sciences, 2008, 267, 170-173.	0.6	32
24	Primary familial brain calcification: update on molecular genetics. Neurological Sciences, 2015, 36, 787-794.	1.9	31
25	Neurological involvement and quadricuspid aortic valve in a patient with Ehlers-Danlos syndrome. Journal of Neurology, 1999, 246, 612-613.	3.6	30
26	Nonsense mutations in alpha-II spectrin in three families with juvenile onset hereditary motor neuropathy. Brain, 2019, 142, 2605-2616.	7.6	29
27	Clinical relevance and neurophysiological correlates of spasticity in cerebrotendinous xanthomatosis. Journal of Neurology, 2011, 258, 783-790.	3.6	28
28	Plasma Levels of Asymmetric Dimethylarginine in Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarct and Leukoencephalopathy. Cerebrovascular Diseases, 2008, 26, 636-640.	1.7	27
29	Cerebrotendinous xanthomatosis with predominant parkinsonian syndrome: Further confirmation of the clinical heterogeneity. Movement Disorders, 2000, 15, 1017-1019.	3.9	26
30	Risk and Determinants of Dementia in Patients with Mild Cognitive Impairment and Brain Subcortical Vascular Changes: A Study of Clinical, Neuroimaging, and Biological Markersâ€"The VMCI-Tuscany Study: Rationale, Design, and Methodology. International Journal of Alzheimer's Disease, 2012, 2012, 1-7.	2.0	26
31	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
32	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.	4.1	25
33	Mitochondrial dysfunction in hereditary spastic paraparesis with mutations in DDHD1/SPG28. Journal of the Neurological Sciences, 2016, 362, 287-291.	0.6	24
34	Peripheral neuropathy in CADASIL. Journal of Neurology, 2005, 252, 1206-1209.	3.6	23
35	Liver transplant reverses biochemical imbalance in mitochondrial neurogastrointestinal encephalomyopathy. Mitochondrion, 2017, 34, 101-102.	3.4	23
36	AARS2-related ovarioleukodystrophy: Clinical and neuroimaging features of three new cases. Acta Neurologica Scandinavica, 2018, 138, 278-283.	2.1	22

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37	Cerebellum and neuropsychiatric disorders: insights from ARSACS. Neurological Sciences, 2014, 35, 95-97.	1.9	21
38	A second MNGIE patient without typical mitochondrial skeletal muscle involvement. Neurological Sciences, 2010, 31, 491-494.	1.9	19
39	First report of a pathogenic mutation on exon 24 of the NOTCH3 gene in a CADASIL family. Journal of Neurology, 2011, 258, 1632-1636.	3.6	19
40	Clinical and biochemical improvement following HSCT in a patient with MNGIE: 1-year follow-up. Journal of Neurology, 2012, 259, 1985-1987.	3.6	18
41	Cerebral Mitochondrial Microangiopathy Leads to Leukoencephalopathy in Mitochondrial Neurogastrointestinal Encephalopathy. American Journal of Neuroradiology, 2018, 39, 427-434.	2.4	18
42	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
43	Treatment of SPG5 with cholesterol-lowering drugs. Journal of Neurology, 2015, 262, 2783-2785.	3.6	17
44	Novel POLG mutations and variable clinical phenotypes in 13 Italian patients. Neurological Sciences, 2017, 38, 563-570.	1.9	17
45	Liver transplantation in mitochondrial neurogastrointestinal encephalomyopathy (MNGIE): clinical long-term follow-up and pathogenic implications. Journal of Neurology, 2020, 267, 3702-3710.	3.6	17
46	Effects of Sapropterin on Endothelium-Dependent Vasodilation in Patients With CADASIL. Stroke, 2014, 45, 2959-2966.	2.0	16
47	The first deep intronic mutation in the NOTCH3 gene in a family with late-onset CADASIL. Neurobiology of Aging, 2013, 34, 2234.e9-2234.e12.	3.1	15
48	The analysis of myotonia congenita mutations discloses functional clusters of amino acids within the CBS2 domain and the C-terminal peptide of the ClC-1 channel. Human Mutation, 2018, 39, 1273-1283.	2.5	15
49	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
50	Lower medulla hypoplasia in Friedreich ataxia: MR Imaging confirmation 140Âyears later. Journal of Neurology, 2017, 264, 1526-1528.	3.6	14
51	Cerebrotendinous xanthomatosis with progressive cerebellar vacuolation. Neuroradiology, 2012, 54, 649-651.	2.2	13
52	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
53	Anti-Saccades in Cerebellar Ataxias Reveal a Contribution of the Cerebellum in Executive Functions. Frontiers in Neurology, 2018, 9, 274.	2.4	13
54	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

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55	Hereditary spastic paraplegia type 5: a potentially treatable disorder of cholesterol metabolism. Journal of Neurology, 2014, 261, 617-619.	3.6	12
56	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
57	Relevance of brain lesion location for cognition in vascular mild cognitive impairment. NeuroImage: Clinical, 2019, 22, 101789.	2.7	12
58	HTRA1 expression profile and activity on TGFâ€î² signaling in HTRA1 mutation carriers. Journal of Cellular Physiology, 2020, 235, 7120-7127.	4.1	12
59	NGS in Hereditary Ataxia: When Rare Becomes Frequent. International Journal of Molecular Sciences, 2021, 22, 8490.	4.1	12
60	Lithium neurotoxicity mimicking rapidly progressive dementia. Journal of Neurology, 2013, 260, 1152-1154.	3.6	11
61	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
62	Neuromuscular excitability changes produced by sustained voluntary contraction and response to mexiletine in myotonia congenita. Neurophysiologie Clinique, 2017, 47, 247-252.	2.2	11
63	Case Report: Early Treatment With Chenodeoxycholic Acid in Cerebrotendinous Xanthomatosis Presenting as Neonatal Cholestasis. Frontiers in Pediatrics, 2020, 8, 382.	1.9	11
64	Eye movement changes in autosomal dominant spinocerebellar ataxias. Neurological Sciences, 2020, 41, 1719-1734.	1.9	11
65	Autonomic Dysfunction in Mental Retardation and Spastic Paraparesis With MECP2 Mutation. Journal of Child Neurology, 2004, 19, 964-966.	1.4	9
66	Evidence of enteric angiopathy and neuromuscular hypoxia in patients with mitochondrial neurogastrointestinal encephalomyopathy. American Journal of Physiology - Renal Physiology, 2021, 320, G768-G779.	3.4	9
67	An infantile case of Alexander disease unusual for its MRI features and a GFAP allele carrying both the p.Arg79His mutation and the p.Glu223Gln coding variant. Journal of Neurology, 2009, 256, 679-682.	3.6	8
68	A new thymidine phosphorylase mutation causing elongation of the protein underlies mitochondrial neurogastrointestinal encephalomyopathy. Journal of Neurology, 2012, 259, 172-174.	3.6	8
69	Temporal lobe abnormalities in neurosyphilis. Practical Neurology, 2014, 14, 449-450.	1.1	8
70	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
71	Imaging of the thymus in myotonic dystrophy type 1. Neurological Sciences, 2018, 39, 347-351.	1.9	8
72	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

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73	A new missense mutation in caveolin-3 gene causes rippling muscle disease. Journal of the Neurological Sciences, 2006, 243, 61-64.	0.6	7
74	Bloodâ€“brain barrier permeability in a patient with Labrune syndrome due to SNORD118 mutations. European Journal of Neurology, 2018, 25, e86-e87.	3.3	7
75	Episodic ataxia and severe infantile phenotype in spinocerebellar ataxia type 14: expansion of the phenotype and novel mutations. Journal of Neurology, 2022, 269, 1476-1484.	3.6	7
76	Mitochondrial recessive ataxia syndrome: A neurological rarity not to be missed. Journal of the Neurological Sciences, 2015, 349, 254-255.	0.6	6
77	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
78	Primary familial brain calcification caused by MYORG mutations in an Italian family. Parkinsonism and Related Disorders, 2019, 67, 24-26.	2.2	6
79	Ataxia with oculomotor apraxia type 2: not always an easy diagnosis. Neurological Sciences, 2015, 36, 1505-1507.	1.9	5
80	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
81	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
82	First report of an Iraqi Kurdish CADASIL patient. Neurological Sciences, 2011, 32, 359-360.	1.9	4
83	Cerebellar hypometabolism with normal structural findings in Cerebrotendinous xanthomatosis. A case report. Clinical Neurology and Neurosurgery, 2015, 139, 221-223.	1.4	4
84	Nicolaidesâ€“Baraitser syndrome: defining a phenotype. Journal of Neurology, 2016, 263, 1659-1660.	3.6	4
85	Neuroendocrine lung cancer in a patient with limbic encephalopathy due to anti-Hu antibodies: A rare association not to be missed. Journal of the Neurological Sciences, 2017, 378, 91-93.	0.6	4
86	Multi-imaging study in a patient with cerebrotendinous xanthomatosis: radiology, clinic and pathology correlation of a rare condition. BJR|case Reports, 2020, 6, 20190047.	0.2	4
87	Vitamin D levels in cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL). Neurological Sciences, 2017, 38, 1333-1336.	1.9	3
88	SPG5 siblings with different phenotypes showing reduction of 27-hydroxycholesterol after simvastatin-ezetimibe treatment. Journal of the Neurological Sciences, 2017, 383, 39-41.	0.6	3
89	Brachial plexopathy due to breast cancer metastases: electrophysiological and imaging findings. Neurological Sciences, 2018, 39, 1503-1505.	1.9	2
90	Cerebrotendinous xanthomatosis: recurrence of the CYP27A1 mutation p.Arg479Cys in Sardinia. Neurological Sciences, 2014, 35, 1303-5.	1.9	1

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91	Challenges and resources in adult life with Joubert syndrome: issues from an international classification of functioning (ICF) perspective. Disability and Rehabilitation, 2021, , 1-8.	1.8	1
92	Clinical aspects of genetic forms of small vessel disease. , 0, , 283-297.		0
93	C9ORF72 gene expansion in a patient with intellectual disability and psychiatric disease. Neurological Sciences, 2017, 38, 207-208.	1.9	0
94	Cerebrotendinous Xanthomatosis. , 2008, , 999-1009.		0