Monica Sciacco

List of Publications by Year
in descending order

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101 papers	5,130 citations	94269 37 h-index	91712 69 g-index
102 all docs	102 docs citations	102 times ranked	5741 citing authors

#	Article	IF	Citations
1	Ataluren treatment of patients with nonsense mutation dystrophinopathy. Muscle and Nerve, 2014, 50, 477-487.	1.0	357
2	Distribution of wild-type and common deletion forms of mtDNA in normal and respiration-deficient muscle fibers from patients with mitochondrial myopathy. Human Molecular Genetics, 1994, 3, 13-19.	1.4	310
3	Cytochromec Oxidase subunit I microdeletion in a patient with motor neuron disease. Annals of Neurology, 1998, 43, 110-116.	2.8	251
4	Atypical clinical presentations associated with the MELAS mutation at position 3243 of human mitochondrial DNA. Neuromuscular Disorders, 1993, 3, 43-50.	0.3	219
5	Loss-of-function mutations in MGME1 impair mtDNA replication and cause multisystemic mitochondrial disease. Nature Genetics, 2013, 45, 214-219.	9.4	198
6	[43]Cytochemistry and immunocytochemistry of mitochondria in tissue sections. Methods in Enzymology, 1996, 264, 509-521.	0.4	174
7	Muscle Coenzyme Q10 Level in Statin-Related Myopathy. Archives of Neurology, 2005, 62, 1709.	4.9	158
8	Phenotypic heterogeneity of the 8344A>G mtDNA â€œMERRFâ€•mutation. Neurology, 2013, 80, 2049-2054.	1.5	157
9	Clinical manifestations of mitochondria1 DNA depletion. Neurology, 1998, 50, 1783-1790.	1.5	136
10	Genotype and phenotype characterization in a large dystrophinopathic cohort with extended follow-up. Journal of Neurology, 2011, 258, 1610-1623.	1.8	134
11	Mitochondrial Respiratory Chain Dysfunction in Muscle From Patients With Amyotrophic Lateral Sclerosis. Archives of Neurology, 2010, 67, 849-54.	4.9	122
12	The m.3243A>G mitochondrial DNA mutation and related phenotypes. A matter of gender?. Journal of Neurology, 2014, 261, 504-510.	1.8	119
13	Mutations in DNA2 Link Progressive Myopathy to Mitochondrial DNA Instability. American Journal of Human Genetics, 2013, 92, 293-300.	2.6	115
14	Guillain-BarrÃ© syndrome associated with high titers of anti-GM1 antibodies. Journal of the Neurological Sciences, 1992, 109, 200-206.	0.3	113
15	Multiple mitochondrial DNA deletions in sporadic inclusion body myositis: A study of 56 patients. Annals of Neurology, 1996, 39, 789-795.	2.8	107
16	Impaired Muscle Mitochondrial Biogenesis and Myogenesis in Spinal Muscular Atrophy. JAMA Neurology, 2015, 72, 666.	4.5	106
17	Transcriptomic profiling of TK2 deficient human skeletal muscle suggests a role for the p53 signalling pathway and identifies growth and differentiation factor-15 as a potential novel biomarker for mitochondrial myopathies. BMC Genomics, 2014, 15, 91.	1.2	104
18	Retrospective study of a large population of patients with asymptomatic or minimally symptomatic raised serum creatine kinase levels. Journal of Neurology, 2002, 249, 305-311.	1.8	100

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19	Partial depletion and multiple deletions of muscle mtDNA in familial MNGIE syndrome. Neurology, 1998, 51, 1086-1092.	1.5	96
20	The italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. Muscle and Nerve, 2017, 55, 55-68.	1.0	86
21	A novel missense adenine nucleotide translocator-1 gene mutation in a Greek adPEO family. Neurology, 2001, 57, 2295-2298.	1.5	85
22	Desmin and Vimentin as markers of regeneration in muscle diseases. Acta Neuropathologica, 1992, 85, 88-92.	3.9	81
23	Next-generation sequencing reveals DGUOK mutations in adult patients with mitochondrial DNA multiple deletions. Brain, 2012, 135, 3404-3415.	3.7	81
24	A missense mutation in the mitochondrial ND5 gene associated with a Leigh-MELAS overlap syndrome. Neurology, 2003, 60, 1857-1861.	1.5	68
25	Redefining phenotypes associated with mitochondrial DNA single deletion. Journal of Neurology, 2015, 262, 1301-1309.	1.8	68
26	New Morphological Approaches to the Study of Mitochondrial Encephalomyopathies. Brain Pathology, 1992, 2, 113-119.	2.1	67
27	Linear mtDNA fragments and unusual mtDNA rearrangements associated with pathological deficiency of MGME1 exonuclease. Human Molecular Genetics, 2014, 23, 6147-6162.	1.4	64
28	New Mutations in TK2 Gene Associated With Mitochondrial DNA Depletion. Pediatric Neurology, 2006, 34, 177-185.	1.0	63
29	Remarkable infidelity of polymerase Î³A associated with mutations in POLG1 exonuclease domain. Neurology, 2003, 61, 903-908.	1.5	60
30	Biallelic C1QBP Mutations Cause Severe Neonatal-, Childhood-, or Later-Onset Cardiomyopathy Associated with Combined Respiratory-Chain Deficiencies. American Journal of Human Genetics, 2017, 101, 525-538.	2.6	58
31	Frequency and characterisation of anoctamin 5 mutations in a cohort of Italian limb-girdle muscular dystrophy patients. Neuromuscular Disorders, 2012, 22, 934-943.	0.3	53
32	Decrease of nerve Na+,K+-ATPase activity in the pathogenesis of human diabetic neuropathy. Journal of the Neurological Sciences, 1993, 120, 159-167.	0.3	52
33	Low anaerobic threshold and increased skeletal muscle lactate production in subjects with Huntington's disease. Movement Disorders, 2011, 26, 130-137.	2.2	52
34	Retrospective study of a large population of patients affected with mitochondrial disorders: clinical, morphological and molecular genetic evaluation. Journal of Neurology, 2001, 248, 778-788.	1.8	45
35	Evidence and age-related distribution of mtDNA D-loop point mutations in skeletal muscle from healthy subjects and mitochondrial patients. Journal of the Neurological Sciences, 2002, 202, 85-91.	0.3	42
36	Nutritional Challenges in Duchenne Muscular Dystrophy. Nutrients, 2017, 9, 594.	1.7	41

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37	High mutational burden in the mtDNA control region from aged muscles: a single-fiber study. Neurobiology of Aging, 2003, 24, 829-838.	1.5	40
38	Histologic muscular history in steroid-treated and untreated patients with Duchenne dystrophy. Neurology, 2015, 85, 1886-1893.	1.5	39
39	â€œMitochondrial neuropathiesâ€: A survey from the large cohort of the Italian Network. Neuromuscular Disorders, 2016, 26, 272-276.	0.3	37
40	Lack of apoptosis in mitochondrial encephalomyopathies. Neurology, 2001, 56, 1070-1074.	1.5	36
41	Clinical, morphological and immunological evaluation of six patients with dysferlin deficiency. Acta Neuropathologica, 2003, 105, 537-542.	3.9	33
42	A mitochondrial tRNA ^{His} gene mutation causing pigmentary retinopathy and neurosensorial deafness. Neurology, 2003, 60, 1200-1203.	1.5	33
43	Phenotype-genotype correlations in skeletal muscle of patients with mtDNA deletions. Muscle and Nerve, 1995, 18, S150-S153.	1.0	32
44	Clinical and molecular characterization of a cohort of patients with novel nucleotide alterations of the Dystrophin gene detected by direct sequencing. BMC Medical Genetics, 2011, 12, 37.	2.1	32
45	Revisiting mitochondrial ocular myopathies: a study from the Italian Network. Journal of Neurology, 2017, 264, 1777-1784.	1.8	32
46	Familial mtDNA T8993C transition causing both the NARP and the MILS phenotype in the same generation. Journal of Neurology, 2003, 250, 1498-1500.	1.8	28
47	A novel mitochondrial tRNA Ile point mutation in chronic progressive external ophthalmoplegia. Journal of Neurology, 1998, 245, 755-758.	1.8	27
48	POLG1 mutations and stroke like episodes: a distinct clinical entity rather than an atypical MELAS syndrome. BMC Neurology, 2013, 13, 8.	0.8	26
49	Sarcoglycan deficiency in a large Italian population of myopathic patients. Acta Neuropathologica, 1998, 96, 509-514.	3.9	25
50	Mitochondrial disease heterogeneity: a prognostic challenge. Acta Myologica, 2014, 33, 86-93.	1.5	25
51	Estimating the impact of COVID-19 pandemic on services provided by Italian Neuromuscular Centers: an Italian Association of Myology survey of the acute phase. Acta Myologica, 2020, 39, 57-66.	1.5	24
52	Effects of shortâ€toâ€long term enzyme replacement therapy (ERT) on skeletal muscle tissue in late onset Pompe disease (LOPD). Neuropathology and Applied Neurobiology, 2018, 44, 449-462.	1.8	23
53	Asymptomatic familial hyperCKemia associated with desmin accumulation in skeletal muscle. Journal of the Neurological Sciences, 1996, 140, 132-136.	0.3	22
54	Follow-up of a large population of asymptomatic/oligosymptomatic hyperckemic subjects. Journal of Neurology, 2006, 253, 1399-1403.	1.8	22

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55	Sural nerve immunoreactivity for nerve growth factor receptor in a case of localized hypertrophic neuropathy. Acta Neuropathologica, 1992, 83, 547-553.	3.9	21
56	Study of mitochondrial DNA depletion in muscle by single-fiber polymerase chain reaction. , 1998, 21, 1374-1381.		21
57	Women with pregnancy-related polymyositis and high serum CK levels in the newborn. Neurology, 2002, 58, 482-484.	1.5	21
58	A collection of 33 novel human mtDNA homoplasmic variants. Human Mutation, 2002, 20, 409-409.	1.1	19
59	Mitochondrial A12308G polymorphism affects clinical features in patients with single mtDNA macrodeletion. European Journal of Human Genetics, 2003, 11, 896-898.	1.4	19
60	Unusual adult-onset Leigh syndrome presentation due to the mitochondrial m.9176T>C mutation. Biochemical and Biophysical Research Communications, 2011, 412, 245-248.	1.0	19
61	Clinical and molecular features of an infant patient affected by Leigh Disease associated to m.14459G > A mitochondrial DNA mutation: a case report. BMC Neurology, 2011, 11, 85.	0.8	18
62	Longitudinal follow-up and muscle MRI pattern of two siblings with polyglucosan body myopathy due to glycogenin-1 mutation. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 797-800.	0.9	17
63	Can Intestinal Pseudo-Obstruction Drive Recurrent Stroke-Like Episodes in Late-Onset MELAS Syndrome? A Case Report and Review of the Literature. Frontiers in Neurology, 2019, 10, 38.	1.1	17
64	Steroid-responsive Hashimoto encephalopathy mimicking Creutzfeldtâ€“Jakob disease. Neurological Sciences, 2011, 32, 719-722.	0.9	16
65	Long Survival in Patients With Leigh Syndrome and the m.10191T>C Mutation in <i>MT-ND3</i>. Journal of Child Neurology, 2014, 29, NP105-NP110.	0.7	16
66	Paucity of deleted mitochondrial DNAs in brain regions of Huntington's disease patients. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 1995, 1271, 229-233.	1.8	15
67	A case of CPT deficiency, homoplasmic mtDNA mutation and ragged red fibers at muscle biopsy. Journal of the Neurological Sciences, 2005, 239, 21-24.	0.3	15
68	Appearance and localization of dystrophin in normal human fetal muscle. International Journal of Developmental Neuroscience, 1991, 9, 607-612.	0.7	14
69	Adult Polyglucosan Body Disease: Clinical and histological heterogeneity of a large Italian family. Neuromuscular Disorders, 2015, 25, 423-428.	0.3	14
70	Lack of apoptosis in patients with progressive external ophthalmoplegia and mutated adenine nucleotide translocator-1 gene. Muscle and Nerve, 2002, 26, 265-269.	1.0	12
71	Schizophreniform Disorder with Cerebrospinal Fluid PCR Positivity for Herpes Simplex Virus Type 1. European Neurology, 2003, 50, 182-183.	0.6	12
72	Tyr78Phe Transthyretin Mutation with Predominant Motor Neuropathy as the Initial Presentation. Case Reports in Neurology, 2011, 3, 62-68.	0.3	12

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73	Purkinje cell COX deficiency and mtDNA depletion in an animal model of spinocerebellar ataxia type 1. Journal of Neuroscience Research, 2018, 96, 1576-1585.	1.3	12
74	Muscle histological changes in a large cohort of patients affected with Becker muscular dystrophy. Acta Neuropathologica Communications, 2022, 10, 48.	2.4	11
75	Value of insoluble PABPN1 accumulation in the diagnosis of oculopharyngeal muscular dystrophy. European Journal of Neurology, 2020, 27, 709-715.	1.7	10
76	MYH2 myopathy, a new case expands the clinical and pathological spectrum of the recessive form. Molecular Genetics & Genomic Medicine, 2020, 8, e1320.	0.6	10
77	Mitochondrial-DNA nucleotides G4298A and T10010C as pathogenic mutations: the confirmation in two new cases. Mitochondrion, 2004, 3, 279-283.	1.6	8
78	Aphasic and visual aura with increased vasogenic leakage: An atypical migrainosus status. Journal of the Neurological Sciences, 2009, 285, 227-229.	0.3	8
79	Movement Disorders in Children with a Mitochondrial Disease: A Cross-Sectional Survey from the Nationwide Italian Collaborative Network of Mitochondrial Diseases. Journal of Clinical Medicine, 2021, 10, 2063.	1.0	8
80	Immune-mediated necrotizing myopathy due to statins exposure. Acta Myologica, 2018, 37, 257-262.	1.5	8
81	Lack of anionic phospholipid calcium binding sites in duchenne muscular dystrophy. Muscle and Nerve, 1992, 15, 325-331.	1.0	7
82	Immunolocalization of heat shock proteins in ragged-red fibers of patients with mitochondrial encephalomyopathies. Neuromuscular Disorders, 1993, 3, 71-76.	0.3	7
83	Two novel mutations in PEO1 (Twinkle) gene associated with chronic external ophthalmoplegia. Journal of the Neurological Sciences, 2011, 308, 173-176.	0.3	7
84	Impact of <scp>COVIDâ€19</scp> on the quality of life of patients with neuromuscular disorders in the <scp>L</scp>ombardy area, <scp>I</scp>taly. Muscle and Nerve, 2021, 64, 474-482.	1.0	7
85	Genetic defects are common in myopathies with tubular aggregates. Annals of Clinical and Translational Neurology, 2022, 9, 4-15.	1.7	7
86	TYMP Variants Result in Late-Onset Mitochondrial Myopathy With Altered Muscle Mitochondrial DNA Homeostasis. Frontiers in Genetics, 2020, 11, 860.	1.1	6
87	IgD Multiple Myeloma Paraproteinemia as a Cause of Myositis. Neurology Research International, 2010, 2010, 1-3.	0.5	5
88	Coexistence of VHL Disease and CPT2 Deficiency: A Case Report. Cancer Research and Treatment, 2016, 48, 1438-1442.	1.3	5
89	The novel mitochondrial tRNAAsn gene mutation m.5709T>C produces ophthalmoparesis and respiratory impairment. European Journal of Human Genetics, 2012, 20, 357-360.	1.4	4
90	Non-alcoholic beriberi, Wernicke encephalopathy and long-term eating disorder: case report and a mini-review. Eating and Weight Disorders, 2021, 26, 729-732.	1.2	4

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91	Severe polyneuropathy in a patient with Churg-Strauss syndrome. Journal of the Peripheral Nervous System, 2000, 5, 106-110.	1.4	3
92	A case report with the peculiar concomitance of 2 different genetic syndromes. Medicine (United) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	0.4	3
93	Reply: DGUOK recessive mutations in patients with CPEO, mitochondrial myopathy, parkinsonism and mtDNA deletions. Brain, 2018, 141, e4-e4.	3.7	3
94	Early Findings in Neonatal Cases of RYR1â€"Related Congenital Myopathies. Frontiers in Neurology, 2021, 12, 664618.	1.1	3
95	G.P.251. Neuromuscular Disorders, 2014, 24, 892.	0.3	2
96	CACNA1S mutation associated with a case of juvenile-onset congenital myopathy. Journal of the Neurological Sciences, 2021, 431, 120047.	0.3	2
97	Anionic phospholipids calcium binding sites in Duchenne and murine X-linked muscular dystrophy. Muscle and Nerve, 1994, 17, 485-488.	1.0	1
98	The m.12316G>A mutation in the mitochondrial tRNALeu(CUN) gene is associated with mitochondrial myopathy and respiratory impairment. Journal of the Neurological Sciences, 2010, 292, 107-110.	0.3	1
99	O.24 Loss of function of MGME1, a novel player in mitochondrial DNA replication, causes a distinct autosomal recessive mitochondrial disorder. Neuromuscular Disorders, 2013, 23, 852.	0.3	1
100	Mitochondria: Muscle Morphology. , 2019, , 19-40.		0
101	A young male with walking difficulties and subacute brainstem dysfunction: Adult-onset Leigh syndrome. Journal of the Neurological Sciences, 2021, 429, 119363.	0.3	0