Margherita Milone

List of Publications by Year
in descending order

119 papers	3,766 citations	159585 30 h-index	149698 56 g-index
119 all docs	119 docs citations	119 times ranked	3991 citing authors

#	Article	IF	Citations
1	Abdominal wall muscle fatty replacement and enlargement in autosomal dominant calpainopathy-3. Neuromuscular Disorders, 2022, 32, 84-85.	0.6	1
2	Development of a standard of care for patients with valosin-containing protein associated multisystem proteinopathy. Orphanet Journal of Rare Diseases, 2022, 17, 23.	2.7	19
3	Incidence and prevalence of immuneâ€mediated necrotizing myopathy in adults in Olmsted County, Minnesota. Muscle and Nerve, 2022, 65, 541-546.	2.2	8
4	A novel missense HNRNPA1 variant in the PY-NLS domain in a patient with late-onset distal myopathy. Neuromuscular Disorders, 2022, 32, 521-526.	0.6	3
5	Cancer and immune-mediated necrotizing myopathy: a longitudinal referral case-controlled outcomes evaluation. Rheumatology, 2022, 62, 281-289.	1.9	5
6	Reply to: Atypical presentations of immuneâ€mediated necrotizing myopathy: Clues and caveats. Muscle and Nerve, 2022, 65, .	2.2	0
7	Distal spinal muscular atrophy featured by predominant calf muscle involvement in VRK1 associated disease â€“ Case series and review. Neuromuscular Disorders, 2022, 32, 527-532.	0.6	2
8	Glycogen accumulation in GNE myopathy. Neuromuscular Disorders, 2022, 32, 774-775.	0.6	1
9	Neurology: Genetics Year in Review. Neurology: Genetics, 2021, 7, e556.	1.9	0
10	Epidemiology and Natural History of Inclusion Body Myositis. Neurology, 2021, 96, e2653-e2661.	1.1	31
11	Filamentous tangles with nemaline rods in MYH2 myopathy: a novel phenotype. Acta Neuropathologica Communications, 2021, 9, 79.	5.2	9
12	Clinical Utility of Striational Antibodies in Paraneoplastic and Myasthenia Gravis Paraneoplastic Panels. Neurology, 2021, , 10.1212/WNL.0000000000012050.	1.1	7
13	A form of muscular dystrophy associated with pathogenic variants in JAG2. American Journal of Human Genetics, 2021, 108, 840-856.	6.2	15
14	Cardiac Involvement in Facioscapulohumeral Muscular Dystrophy (FSHD). Frontiers in Neurology, 2021, 12, 668180.	2.4	8
15	Guidelines for genetic testing of muscle and neuromuscular junction disorders. Muscle and Nerve, 2021, 64, 255-269.	2.2	8
16	Interstitial amyloidosis in sporadic inclusion body myositis. Muscle and Nerve, 2021, 64, 590-594.	2.2	1
17	Antiâ€cN1A antibodies do not correlate with specific clinical, electromyographic, or pathological findings in sporadic inclusion body myositis. Muscle and Nerve, 2021, 63, 490-496.	2.2	19
18	Immuneâ€mediated necrotizing myopathy: Unusual presentations of a treatable disease. Muscle and Nerve, 2021, 64, 734-739.	2.2	12

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19	Transthyretin amyloidosis: Putting myopathy on the map. Muscle and Nerve, 2020, 61, 95-100.	2.2	27
20	<scp>GYG1</scp>: A distal myopathy with polyglucosan bodies. JIMD Reports, 2020, 55, 88-90.	1.5	3
21	Cardiac and Respiratory Complications of Necrotizing Autoimmune Myopathy. Mayo Clinic Proceedings, 2020, 95, 2144-2149.	3.0	13
22	Improving accuracy of myasthenia gravis autoantibody testing by reflex algorithm. Neurology, 2020, 95, e3002-e3011.	1.1	14
23	Immune checkpoint inhibitor-associated myopathy: a clinicoseropathologically distinct myopathy. Brain Communications, 2020, 2, fcaa181.	3.3	51
24	Myopathies with finger flexor weakness: Not only inclusionâ€body myositis. Muscle and Nerve, 2020, 62, 445-454.	2.2	10
25	Myopathies featuring early or prominent dysphagia. Muscle and Nerve, 2020, 62, 344-350.	2.2	10
26	A novel heterozygous mutation in the C-terminal region of HSPB8 leads to limb-girdle rimmed vacuolar myopathy. Neuromuscular Disorders, 2020, 30, 236-240.	0.6	13
27	Diagnostic modelling and therapeutic monitoring of immune-mediated necrotizing myopathy: role of electrical myotonia. Brain Communications, 2020, 2, fcaa191.	3.3	9
28	Neuromuscular transmission defects in myopathies: Rare but worth searching for. Muscle and Nerve, 2019, 59, 475-478.	2.2	12
29	Necrotizing autoimmune myopathy with tubular aggregates. Neurology, 2019, 93, 313-314.	1.1	1
30	A homozygous mutation in GMPPB leads to centronuclear myopathy with combined pre- and postsynaptic defects of neuromuscular transmission. Neuromuscular Disorders, 2019, 29, 614-617.	0.6	11
31	Slowâ€channel myasthenia due to novel mutation in M2 domain of AChR delta subunit. Annals of Clinical and Translational Neurology, 2019, 6, 2066-2078.	3.7	7
32	Congenital myopathy with a novel SELN missense mutation and the challenge to differentiate it from congenital muscular dystrophy. Journal of Clinical Neuroscience, 2019, 62, 238-239.	1.5	4
33	Congenital myopathies in the adult neuromuscular clinic. Neurology: Genetics, 2019, 5, e341.	1.9	22
34	Sporadic late-onset nemaline myopathy. Neurology, 2019, 93, e298-e305.	1.1	45
35	Neuromuscular transmission defects in myopathies. Muscle and Nerve, 2019, 60, E8-E9.	2.2	1
36	ACTA1-myopathy with prominent finger flexor weakness and rimmed vacuoles. Neuromuscular Disorders, 2019, 29, 388-391.	0.6	5

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37	The Electrophysiology of Presynaptic Congenital Myasthenic Syndromes With and Without Facilitation: From Electrodiagnostic Findings to Molecular Mechanisms. Frontiers in Neurology, 2019, 10, 257.	2.4	9
38	Congenital Vocal Cord Paralysis and Late-Onset Limb-Girdle Weakness in MuSKâ€“Congenital Myasthenic Syndrome. Frontiers in Neurology, 2019, 10, 1300.	2.4	7
39	The unfolding spectrum of inherited distal myopathies. Muscle and Nerve, 2019, 59, 283-294.	2.2	27
40	Intramuscular interstitial amyloid deposition does not impact anoctaminopathyâ€5 phenotype. Muscle and Nerve, 2019, 59, 133-137.	2.2	7
41	Untangling the complexity of limbâ€girdle muscular dystrophies. Muscle and Nerve, 2018, 58, 167-177.	2.2	76
42	Autosomal dominant calpainopathy due to heterozygous CAPN3 C.643_663del21. Muscle and Nerve, 2018, 57, 679-683.	2.2	32
43	An adult with a rare form of congenital fiber type disproportion. Muscle and Nerve, 2018, 57, E97-E99.	2.2	1
44	Myofibrillar myopathy due to dominant LMNA mutations: A report of 2 cases. Muscle and Nerve, 2018, 57, E124-E126.	2.2	8
45	Rhabdomyolysis and fluctuating asymptomatic hyperCKemia associated with CACNA1S variant. European Journal of Neurology, 2018, 25, 417-419.	3.3	18
46	A tropomyosin-receptor kinase-fused gene mutation associates with vacuolar myopathy. Neurology: Genetics, 2018, 4, e287.	1.9	5
47	Pompe Disease Could Mimic Exam Findings of Amyloidosis: Two Rare Diagnoses Bona Fide. Case Reports in Hematology, 2018, 2018, 1-4.	0.4	1
48	Congenital myasthenic syndromes in adult neurology clinic. Neurology, 2018, 91, e1770-e1777.	1.1	36
49	Muscle Biopsy and Electromyography Correlation. Frontiers in Neurology, 2018, 9, 839.	2.4	14
50	Fuchs' Endothelial Corneal Dystrophy in Patients With Myotonic Dystrophy, Type 1. , 2018, 59, 3053.		17
51	Myopathy With SQSTM1 and TIA1 Variants: Clinical and Pathological Features. Frontiers in Neurology, 2018, 9, 147.	2.4	28
52	A novel ACTA1 mutation causing progressive facioscapuloperoneal myopathy in an adult. Journal of Clinical Neuroscience, 2018, 53, 261-262.	1.5	7
53	Hereditary myopathies with early respiratory insufficiency in adults. Muscle and Nerve, 2017, 56, 881-886.	2.2	30
54	Diagnosis and Management of Immune-Mediated Myopathies. Mayo Clinic Proceedings, 2017, 92, 826-837.	3.0	60

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55	Autosomal dominant distal myopathy due to a novel ACTA1 mutation. Neuromuscular Disorders, 2017, 27, 742-746.	0.6	18
56	PRES leading to the diagnosis of McArdle disease. Journal of Clinical Neuroscience, 2017, 46, 62-64.	1.5	0
57	Characterization of isolated amyloid myopathy. European Journal of Neurology, 2017, 24, 1437-1445.	3.3	45
58	RYR1 causing distal myopathy. Molecular Genetics & Genomic Medicine, 2017, 5, 800-804.	1.2	14
59	Rapsyn congenital myasthenic syndrome worsened by fluoxetine. Muscle and Nerve, 2017, 55, 131-135.	2.2	8
60	Reply. Muscle and Nerve, 2016, 54, 343-344.	2.2	0
61	Adult-onset respiratory insufficiency, scoliosis, and distal joint hyperlaxity in patients with multiminicore disease due to novel Megf10 mutations. Muscle and Nerve, 2016, 53, 984-988.	2.2	18
62	Clinical and Electrophysiological Findings in Hereditary Inclusion Body Myopathy Compared With Sporadic Inclusion Body Myositis. Journal of Clinical Neuromuscular Disease, 2016, 17, 190-196.	0.7	14
63	Distal myopathy with coexisting heterozygous TIA1 and MYH7 Variants. Neuromuscular Disorders, 2016, 26, 511-515.	0.6	14
64	Mutations Causing Slow-Channel Myasthenia Reveal That a Valine Ring in the Channel Pore of Muscle AChR is Optimized for Stabilizing Channel Gating. Human Mutation, 2016, 37, 1051-1059.	2.5	19
65	Loss of MUNC13-1 function causes microcephaly, cortical hyperexcitability, and fatal myasthenia. Neurology: Genetics, 2016, 2, e105.	1.9	50
66	Clinical spectrum of valosin containing protein (VCP)â€opathy. Muscle and Nerve, 2016, 54, 94-99.	2.2	28
67	Rhabdomyolysis featuring muscular dystrophies. Journal of the Neurological Sciences, 2016, 361, 29-33.	0.6	29
68	Signal recognition particle immunoglobulin g detected incidentally associates with autoimmune myopathy. Muscle and Nerve, 2016, 53, 925-932.	2.2	8
69	Expanding Phenotypic Spectrum ofNKX2-1â€“Related Disordersâ€”Mitochondrial and Immunologic Dysfunction. JAMA Neurology, 2016, 73, 237.	9.0	2
70	Camptocormia as presenting manifestation of a spectrum of myopathic disorders. Muscle and Nerve, 2015, 52, 1008-1012.	2.2	34
71	Clinical Reasoning: A 51-year-old woman with weakness and stiff neck. Neurology, 2015, 85, e32-6.	1.1	0
72	Clinical Features and Treatment Outcomes of Necrotizing Autoimmune Myopathy. JAMA Neurology, 2015, 72, 996.	9.0	174

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73	Clinical and laboratory findings of 21 patients with radiation-induced myopathy. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 152-158.	1.9	32
74	Paraneoplastic encephalopathy: an unusual presenting feature of bladder cancer metastasis. BMJ Case Reports, 2015, 2015, bcr2014208913-bcr2014208913.	0.5	5
75	Pearls & Oy-sters: HyperCKemia with limb-girdle weakness. Neurology, 2014, 83, e209-12.	1.1	3
76	A novel VCP mutation underlies scapuloperoneal muscular dystrophy and dropped head syndrome featuring lobulated fibers. Muscle and Nerve, 2014, 50, 295-299.	2.2	23
77	DNAJB6 myopathy: A vacuolar myopathy with childhood onset. Muscle and Nerve, 2014, 49, 607-610.	2.2	22
78	Myopathy during treatment with the antianginal drug ranolazine. Journal of the Neurological Sciences, 2014, 347, 380-382.	0.6	8
79	Novel ANO5 homozygous microdeletion causing myalgia and unprovoked rhabdomyolysis in an Arabic man. Muscle and Nerve, 2014, 50, 610-613.	2.2	27
80	Clinical Reasoning: A 38-year-old woman with childhood-onset weakness. Neurology, 2014, 83, e81-4.	1.1	0
81	Novel OPA1 mutation featuring spastic paraparesis and intestinal dysmotility. Molecular Genetics and Metabolism Reports, 2014, 1, 443-445.	1.1	1
82	Autoimmune Aquaporin-4 Myopathy in Neuromyelitis Optica Spectrum. JAMA Neurology, 2014, 71, 1025.	9.0	68
83	Coexistence of DMPK gene expansion and CLCN1 missense mutation in the same patient. Neurogenetics, 2014, 15, 213-214.	1.4	5
84	<scp>ANO</scp>5â€<scp>m</scp>uscular dystrophy: clinical, pathological and molecular findings. European Journal of Neurology, 2013, 20, 1383-1389.	3.3	51
85	Diagnosis of mitochondrial myopathies. Molecular Genetics and Metabolism, 2013, 110, 35-41.	1.1	74
86	Mitochondrial DNA Multiple Deletion Syndromes, Autosomal Dominant and Recessive (POLG, POLG2,) Tj ETQq0 0 0 rgBT /Overlock 10 T	1	
87	Comprehensive next-generation sequence analyses of the entire mitochondrial genome reveal new insights into the molecular diagnosis of mitochondrial DNA disorders. Genetics in Medicine, 2013, 15, 388-394.	2.4	106
88	Sporadic inclusion body myositis presenting with severe camptocormia. Journal of Clinical Neuroscience, 2013, 20, 1628-1629.	1.5	19
89	Orthostatic Tremor, Progressive External Ophthalmoplegia, and Twinkle. JAMA Neurology, 2013, 70, 1429.	9.0	13
90	Electrically Active Immune-mediated Rippling Muscle Disease Preceding Breast Cancer. Neurologist, 2012, 18, 155-158.	0.7	9

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91	Amyloidosis and exercise intolerance in ANO5 muscular dystrophy. Neuromuscular Disorders, 2012, 22, 13-15.	0.6	52
92	Mitochondrial myopathy due to novel missense mutation in the cytochrome c oxidase 1 gene. Journal of the Neurological Sciences, 2012, 319, 158-163.	0.6	11
93	Myotonia associated with caveolinâ€3 mutation. Muscle and Nerve, 2012, 45, 897-900.	2.2	12
94	Mitochondrial dynamics. Neurology, 2012, 78, 1612-1619.	1.1	32
95	Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE)-like phenotype: an expanded clinical spectrum of POLG1 mutations. Journal of Neurology, 2012, 259, 862-868.	3.6	112
96	Diseased Skeletal Muscle. Journal of the American College of Cardiology, 2011, 58, 1819-1824.	2.8	244
97	Endplate structure and parameters of neuromuscular transmission in sporadic centronuclear myopathy associated with myasthenia. Neuromuscular Disorders, 2011, 21, 387-395.	0.6	39
98	Mitochondrial DNA polymerase Â mutations: an ever expanding molecular and clinical spectrum. Journal of Medical Genetics, 2011, 48, 669-681.	3.2	140
99	Novel POLG Splice Site Mutation and Optic Atrophy. Archives of Neurology, 2011, 68, 806-11.	4.5	29
100	POLG-related disorders. Neurology, 2011, 77, 1847-1852.	1.1	31
101	Sporadic late onset nemaline myopathy responsive to IVIg and immunotherapy. Muscle and Nerve, 2010, 41, 272-276.	2.2	34
102	AnesthÃ©sie et dystrophie myotonique de type 2: une sÃ©rie de cas. Canadian Journal of Anaesthesia, 2010, 57, 248-255.	1.6	13
103	Absent, unrecognized, and minimal myotonic discharges in myotonic dystrophy type 2. Muscle and Nerve, 2010, 41, 758-762.	2.2	45
104	Exercise intolerance due to cytochrome b mutation. Muscle and Nerve, 2010, 42, 136-140.	2.2	19
105	Polymerase Gamma 1 Mutations. Neurologist, 2010, 16, 84-91.	0.7	153
106	Myasthenic syndrome due to defects in rapsyn. Neurology, 2009, 73, 228-235.	1.1	65
107	Myotonic dystrophy type 2 with focal asymmetric muscle weakness and no electrical myotonia. Muscle and Nerve, 2009, 39, 383-385.	2.2	22
108	Reply to: Myotonic disorder without myotonia?. Muscle and Nerve, 2009, 40, 1073-1073.	2.2	0

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109	Mitochondrial disorder with OPA1 mutation lacking optic atrophy. Mitochondrion, 2009, 9, 279-281.	3.4	37
110	Molecular and clinical genetics of mitochondrial diseases due toPOLGmutations. Human Mutation, 2008, 29, E150-E172.	2.5	256
111	Dokâ€7 myasthenia: Phenotypic and molecular genetic studies in 16 patients. Annals of Neurology, 2008, 64, 71-87.	5.3	117
112	Sensory ataxic neuropathy with ophthalmoparesis caused by POLG mutations. Neuromuscular Disorders, 2008, 18, 626-632.	0.6	71
113	Novel congenital myasthenic syndromes associated with defects in quantal release. Neurology, 2006, 66, 1223-1229.	1.1	17
114	Rapsyn Mutations in Humans Cause Endplate Acetylcholine-Receptor Deficiency and Myasthenic Syndrome. American Journal of Human Genetics, 2002, 70, 875-885.	6.2	221
115	The spectrum of mutations causing end-plate acetylcholinesterase deficiency. Annals of Neurology, 2000, 47, 162-170.	5.3	123
116	Fundamental Gating Mechanism of Nicotinic Receptor Channel Revealed by Mutation Causing a Congenital Myasthenic Syndrome. Journal of General Physiology, 2000, 116, 449-462.	1.9	77
117	Congenital Myasthenic Syndromes: New Insights from Molecular Genetic and Patch-Clamp Studiesa. Annals of the New York Academy of Sciences, 1998, 841, 140-156.	3.8	17
118	Congenital Myasthenic Syndrome Caused by Novel Loss-of-Function Mutations in the Human AChR e Subunit Genea. Annals of the New York Academy of Sciences, 1998, 841, 184-188.	3.8	6
119	Patch-clamp analysis of the properties of acetylcholine receptor channels at the normal human endplate. Muscle and Nerve, 1994, 17, 1364-1369.	2.2	39