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	Molecular and clinical genetics of mitochondrial diseases due toPOLGmutations. Human Mutation, 2008, 29, E150-E172.	2.5	256
2	Diseased Skeletal Muscle. Journal of the American College of Cardiology, 2011, 58, 1819-1824.	2.8	244
3	Rapsyn Mutations in Humans Cause Endplate Acetylcholine-Receptor Deficiency and Myasthenic Syndrome. American Journal of Human Genetics, 2002, 70, 875-885.	6.2	221
4	Clinical Features and Treatment Outcomes of Necrotizing Autoimmune Myopathy. JAMA Neurology, 2015, 72, 996.	9.0	174
5	Polymerase Gamma 1 Mutations. Neurologist, 2010, 16, 84-91.	0.7	153
6	Mitochondrial DNA polymerase Â mutations: an ever expanding molecular and clinical spectrum. Journal of Medical Genetics, 2011, 48, 669-681.	3.2	140
7	The spectrum of mutations causing end-plate acetylcholinesterase deficiency. Annals of Neurology, 2000, 47, 162-170.	5.3	123
8	Dokâ€7 myasthenia: Phenotypic and molecular genetic studies in 16 patients. Annals of Neurology, 2008, 64, 71-87.	5.3	117
9	Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE)-like phenotype: an expanded clinical spectrum of POLG1 mutations. Journal of Neurology, 2012, 259, 862-868.	3.6	112
10	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
11	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
12	Untangling the complexity of limbâ€girdle muscular dystrophies. Muscle and Nerve, 2018, 58, 167-177.	2.2	76
13	Diagnosis of mitochondrial myopathies. Molecular Genetics and Metabolism, 2013, 110, 35-41.	1.1	74
14	Sensory ataxic neuropathy with ophthalmoparesis caused by POLG mutations. Neuromuscular Disorders, 2008, 18, 626-632.	0.6	71
15	Autoimmune Aquaporin-4 Myopathy in Neuromyelitis Optica Spectrum. JAMA Neurology, 2014, 71, 1025.	9.0	68
16	Myasthenic syndrome due to defects in rapsyn. Neurology, 2009, 73, 228-235.	1.1	65
17	Diagnosis and Management of Immune-Mediated Myopathies. Mayo Clinic Proceedings, 2017, 92, 826-837.	3.0	60
18	Amyloidosis and exercise intolerance in ANO5 muscular dystrophy. Neuromuscular Disorders, 2012, 22, 13-15.	0.6	52

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19	<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
20	Immune checkpoint inhibitor-associated myopathy: a clinicoseropathologically distinct myopathy. Brain Communications, 2020, 2, fcaa181.	3.3	51
21	Loss of MUNC13-1 function causes microcephaly, cortical hyperexcitability, and fatal myasthenia. Neurology: Genetics, 2016, 2, e105.	1.9	50
22	Absent, unrecognized, and minimal myotonic discharges in myotonic dystrophy type 2. Muscle and Nerve, 2010, 41, 758-762.	2.2	45
23	Characterization of isolated amyloid myopathy. European Journal of Neurology, 2017, 24, 1437-1445.	3.3	45
24	Sporadic late-onset nemaline myopathy. Neurology, 2019, 93, e298-e305.	1.1	45
25	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
26	Endplate structure and parameters of neuromuscular transmission in sporadic centronuclear myopathy associated with myasthenia. Neuromuscular Disorders, 2011, 21, 387-395.	0.6	39
27	Mitochondrial disorder with OPA1 mutation lacking optic atrophy. Mitochondrion, 2009, 9, 279-281.	3.4	37
28	Congenital myasthenic syndromes in adult neurology clinic. Neurology, 2018, 91, e1770-e1777.	1.1	36
29	Sporadic late onset nemaline myopathy responsive to IVIg and immunotherapy. Muscle and Nerve, 2010, 41, 272-276.	2.2	34
30	Camptocormia as presenting manifestation of a spectrum of myopathic disorders. Muscle and Nerve, 2015, 52, 1008-1012.	2.2	34
31	Mitochondrial dynamics. Neurology, 2012, 78, 1612-1619.	1.1	32
32	Clinical and laboratory findings of 21 patients with radiation-induced myopathy. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 152-158.	1.9	32
33	Autosomal dominant calpainopathy due to heterozygous CAPN3 C.643_663del21. Muscle and Nerve, 2018, 57, 679-683.	2.2	32
34	POLG-related disorders. Neurology, 2011, 77, 1847-1852.	1.1	31
35	Epidemiology and Natural History of Inclusion Body Myositis. Neurology, 2021, 96, e2653-e2661.	1.1	31
36	Hereditary myopathies with early respiratory insufficiency in adults. Muscle and Nerve, 2017, 56, 881-886.	2.2	30

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37	Novel POLG Splice Site Mutation and Optic Atrophy. Archives of Neurology, 2011, 68, 806-11.	4.5	29
38	Rhabdomyolysis featuring muscular dystrophies. Journal of the Neurological Sciences, 2016, 361, 29-33.	0.6	29
39	Clinical spectrum of valosin containing protein (VCP)â€opathy. Muscle and Nerve, 2016, 54, 94-99.	2.2	28
40	Myopathy With SQSTM1 and TIA1 Variants: Clinical and Pathological Features. Frontiers in Neurology, 2018, 9, 147.	2.4	28
41	Novel ANO5 homozygous microdeletion causing myalgia and unprovoked rhabdomyolysis in an Arabic man. Muscle and Nerve, 2014, 50, 610-613.	2.2	27
42	The unfolding spectrum of inherited distal myopathies. Muscle and Nerve, 2019, 59, 283-294.	2.2	27
43	Transthyretin amyloidosis: Putting myopathy on the map. Muscle and Nerve, 2020, 61, 95-100.	2.2	27
44	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
45	Myotonic dystrophy type 2 with focal asymmetric muscle weakness and no electrical myotonia. Muscle and Nerve, 2009, 39, 383-385.	2.2	22
46	DNAJB6 myopathy: A vacuolar myopathy with childhood onset. Muscle and Nerve, 2014, 49, 607-610.	2.2	22
47	Congenital myopathies in the adult neuromuscular clinic. Neurology: Genetics, 2019, 5, e341.	1.9	22
48	Exercise intolerance due to cytochrome b mutation. Muscle and Nerve, 2010, 42, 136-140.	2.2	19
49	Sporadic inclusion body myositis presenting with severe camptocormia. Journal of Clinical Neuroscience, 2013, 20, 1628-1629.	1.5	19
50	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
51	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
52	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
53	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
54	Autosomal dominant distal myopathy due to a novel ACTA1 mutation. Neuromuscular Disorders, 2017, 27, 742-746.	0.6	18

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55	Rhabdomyolysis and fluctuating asymptomatic hyperCKemia associated with CACNA1S variant. European Journal of Neurology, 2018, 25, 417-419.	3.3	18
56	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
57	Novel congenital myasthenic syndromes associated with defects in quantal release. Neurology, 2006, 66, 1223-1229.	1.1	17
58	Fuchs' Endothelial Corneal Dystrophy in Patients With Myotonic Dystrophy, Type 1. , 2018, 59, 3053.		17
59	A form of muscular dystrophy associated with pathogenic variants in JAG2. American Journal of Human Genetics, 2021, 108, 840-856.	6.2	15
60	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
61	Distal myopathy with coexisting heterozygous TIA1 and MYH7 Variants. Neuromuscular Disorders, 2016, 26, 511-515.	0.6	14
62	RYR1 causing distal myopathy. Molecular Genetics & Genomic Medicine, 2017, 5, 800-804.	1.2	14
63	Muscle Biopsy and Electromyography Correlation. Frontiers in Neurology, 2018, 9, 839.	2.4	14
64	Improving accuracy of myasthenia gravis autoantibody testing by reflex algorithm. Neurology, 2020, 95, e3002-e3011.	1.1	14
65	AnesthÃ©sie et dystrophie myotonique de type 2: une sÃ©rie de cas. Canadian Journal of Anaesthesia, 2010, 57, 248-255.	1.6	13
66	Orthostatic Tremor, Progressive External Ophthalmoplegia, and Twinkle. JAMA Neurology, 2013, 70, 1429.	9.0	13
67	Cardiac and Respiratory Complications of Necrotizing Autoimmune Myopathy. Mayo Clinic Proceedings, 2020, 95, 2144-2149.	3.0	13
68	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
69	Myotonia associated with caveolinâ€3 mutation. Muscle and Nerve, 2012, 45, 897-900.	2.2	12
70	Neuromuscular transmission defects in myopathies: Rare but worth searching for. Muscle and Nerve, 2019, 59, 475-478.	2.2	12
71	Immuneâ€mediated necrotizing myopathy: Unusual presentations of a treatable disease. Muscle and Nerve, 2021, 64, 734-739.	2.2	12
72	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

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73	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
74	Myopathies with finger flexor weakness: Not only inclusionâ€body myositis. Muscle and Nerve, 2020, 62, 445-454.	2.2	10
75	Myopathies featuring early or prominent dysphagia. Muscle and Nerve, 2020, 62, 344-350.	2.2	10
76	Electrically Active Immune-mediated Rippling Muscle Disease Preceding Breast Cancer. Neurologist, 2012, 18, 155-158.	0.7	9
77	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
78	Filamentous tangles with nemaline rods in MYH2 myopathy: a novel phenotype. Acta Neuropathologica Communications, 2021, 9, 79.	5.2	9
79	Diagnostic modelling and therapeutic monitoring of immune-mediated necrotizing myopathy: role of electrical myotonia. Brain Communications, 2020, 2, fcaa191.	3.3	9
80	Myopathy during treatment with the antianginal drug ranolazine. Journal of the Neurological Sciences, 2014, 347, 380-382.	0.6	8
81	Signal recognition particle immunoglobulin g detected incidentally associates with autoimmune myopathy. Muscle and Nerve, 2016, 53, 925-932.	2.2	8
82	Rapsyn congenital myasthenic syndrome worsened by fluoxetine. Muscle and Nerve, 2017, 55, 131-135.	2.2	8
83	Myofibrillar myopathy due to dominant LMNA mutations: A report of 2 cases. Muscle and Nerve, 2018, 57, E124-E126.	2.2	8
84	Cardiac Involvement in Facioscapulohumeral Muscular Dystrophy (FSHD). Frontiers in Neurology, 2021, 12, 668180.	2.4	8
85	Guidelines for genetic testing of muscle and neuromuscular junction disorders. Muscle and Nerve, 2021, 64, 255-269.	2.2	8
86	Incidence and prevalence of immuneâ€mediated necrotizing myopathy in adults in Olmsted County, Minnesota. Muscle and Nerve, 2022, 65, 541-546.	2.2	8
87	A novel ACTA1 mutation causing progressive facioscapuloperoneal myopathy in an adult. Journal of Clinical Neuroscience, 2018, 53, 261-262.	1.5	7
88	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
89	Congenital Vocal Cord Paralysis and Late-Onset Limb-Girdle Weakness in MuSKâ€“Congenital Myasthenic Syndrome. Frontiers in Neurology, 2019, 10, 1300.	2.4	7
90	Intramuscular interstitial amyloid deposition does not impact anoctaminopathyâ€5 phenotype. Muscle and Nerve, 2019, 59, 133-137.	2.2	7

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91	Clinical Utility of Striational Antibodies in Paraneoplastic and Myasthenia Gravis Paraneoplastic Panels. Neurology, 2021, , 10.1212/WNL.0000000000012050.	1.1	7
92	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
93	Coexistence of DMPK gene expansion and CLCN1 missense mutation in the same patient. Neurogenetics, 2014, 15, 213-214.	1.4	5
94	A tropomyosin-receptor kinase-fused gene mutation associates with vacuolar myopathy. Neurology: Genetics, 2018, 4, e287.	1.9	5
95	ACTA1-myopathy with prominent finger flexor weakness and rimmed vacuoles. Neuromuscular Disorders, 2019, 29, 388-391.	0.6	5
96	Paraneoplastic encephalopathy: an unusual presenting feature of bladder cancer metastasis. BMJ Case Reports, 2015, 2015, bcr2014208913-bcr2014208913.	0.5	5
97	Cancer and immune-mediated necrotizing myopathy: a longitudinal referral case-controlled outcomes evaluation. Rheumatology, 2022, 62, 281-289.	1.9	5
98	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
99	Pearls & Oy-sters: HyperCKemia with limb-girdle weakness. Neurology, 2014, 83, e209-12.	1.1	3
100	<scp>GYG1</scp>: A distal myopathy with polyglucosan bodies. JIMD Reports, 2020, 55, 88-90.	1.5	3
101	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
102	Expanding Phenotypic Spectrum ofNKX2-1â€“Related Disordersâ€”Mitochondrial and Immunologic Dysfunction. JAMA Neurology, 2016, 73, 237.	9.0	2
103	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
104	Mitochondrial DNA Multiple Deletion Syndromes, Autosomal Dominant and Recessive (POLG, POLG2,) Tj ETQq0 0 0 rgBT /Overlock 10 T	1	
105	Novel OPA1 mutation featuring spastic paraparesis and intestinal dysmotility. Molecular Genetics and Metabolism Reports, 2014, 1, 443-445.	1.1	1
106	An adult with a rare form of congenital fiber type disproportion. Muscle and Nerve, 2018, 57, E97-E99.	2.2	1
107	Pompe Disease Could Mimic Exam Findings of Amyloidosis: Two Rare Diagnoses Bona Fide. Case Reports in Hematology, 2018, 2018, 1-4.	0.4	1
108	Necrotizing autoimmune myopathy with tubular aggregates. Neurology, 2019, 93, 313-314.	1.1	1

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109	Neuromuscular transmission defects in myopathies. Muscle and Nerve, 2019, 60, E8-E9.	2.2	1
110	Interstitial amyloidosis in sporadic inclusion body myositis. Muscle and Nerve, 2021, 64, 590-594.	2.2	1
111	Abdominal wall muscle fatty replacement and enlargement in autosomal dominant calpainopathy-3. Neuromuscular Disorders, 2022, 32, 84-85.	0.6	1
112	Glycogen accumulation in GNE myopathy. Neuromuscular Disorders, 2022, 32, 774-775.	0.6	1
113	Reply to: Myotonic disorder without myotonia?. Muscle and Nerve, 2009, 40, 1073-1073.	2.2	0
114	Clinical Reasoning: A 38-year-old woman with childhood-onset weakness. Neurology, 2014, 83, e81-4.	1.1	0
115	Clinical Reasoning: A 51-year-old woman with weakness and stiff neck. Neurology, 2015, 85, e32-6.	1.1	0
116	Reply. Muscle and Nerve, 2016, 54, 343-344.	2.2	0
117	PRES leading to the diagnosis of McArdle disease. Journal of Clinical Neuroscience, 2017, 46, 62-64.	1.5	0
118	Neurology: Genetics Year in Review. Neurology: Genetics, 2021, 7, e556.	1.9	0
119	Reply to: Atypical presentations of immuneâ€mediated necrotizing myopathy: Clues and caveats. Muscle and Nerve, 2022, 65, .	2.2	0