

Roberto Massa

List of Publications by Year in descending order

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109
papers

2,719
citations

147726

31
h-index

214721

47
g-index

112
all docs

112
docs citations

112
times ranked

3727
citing authors

#	ARTICLE	IF	CITATIONS
1	RESPONSE TO LETTER TO THE EDITOR "AUDITORY DYSFUNCTION IN FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY TYPE 1: BEYOND THE INNER EAR INVOLVEMENT" BY GHELLER ET AL. <i>Otology and Neurotology</i> , 2022, 43, e392-e393.	0.7	0
2	Objective Assessment of Walking Impairments in Myotonic Dystrophy by Means of a Wearable Technology and a Novel Severity Index. <i>Electronics (Switzerland)</i> , 2021, 10, 708.	1.8	1
3	ATTRv in Lazio-Italy: A High-Prevalence Region in a Non-Endemic Country. <i>Genes</i> , 2021, 12, 829.	1.0	9
4	A 14-Year Italian Experience in DM2 Genetic Testing: Frequency and Distribution of Normal and Premutated CNBP Alleles. <i>Frontiers in Genetics</i> , 2021, 12, 668094.	1.1	3
5	Assessment of self-reported and objective daytime sleepiness in adult-onset myotonic dystrophy type 1. <i>Journal of Clinical Sleep Medicine</i> , 2021, 17, 2383-2391.	1.4	5
6	Movement disorders in primary central nervous system lymphoma: two unreported cases and a review of literature. <i>Neurological Sciences</i> , 2021, 42, 905-910.	0.9	2
7	The actigraphic documentation of circadian sleep-wake rhythm dysregulation in myotonic dystrophy type 1. <i>Sleep Medicine</i> , 2021, 88, 134-139.	0.8	5
8	Cochlear Dysfunction Is a Frequent Feature of Facioscapulohumeral Muscular Dystrophy Type 1 (FSHD1). <i>Otology and Neurotology</i> , 2021, 42, 18-23.	0.7	4
9	How to capture activities of daily living in myotonic dystrophy type 2?. <i>Neuromuscular Disorders</i> , 2020, 30, 796-806.	0.3	2
10	Validation of Motor Outcome Measures in Myotonic Dystrophy Type 2. <i>Frontiers in Neurology</i> , 2020, 11, 306.	1.1	9
11	Design of a multiplex ligation-dependent probe amplification assay for SLC20A2: identification of two novel deletions in primary familial brain calcification. <i>Journal of Human Genetics</i> , 2019, 64, 1083-1090.	1.1	2
12	Reply to the letter entitled "Predictors of respiratory impairment in patients with myotonic dystrophy type 1". <i>Journal of the Neurological Sciences</i> , 2019, 403, 166-167.	0.3	0
13	Prevalence and predictor factors of respiratory impairment in a large cohort of patients with Myotonic Dystrophy type 1 (DM1): A retrospective, cross sectional study. <i>Journal of the Neurological Sciences</i> , 2019, 399, 118-124.	0.3	31
14	Thymomatous myasthenia gravis: novel association with HLA DQB1*05:01 and strengthened evidence of high clinical and serological severity. <i>Journal of Neurology</i> , 2019, 266, 982-989.	1.8	14
15	Italian recommendations for the diagnosis and treatment of myasthenia gravis. <i>Neurological Sciences</i> , 2019, 40, 1111-1124.	0.9	38
16	Nodular morphea in a patient with Steinert disease. <i>Giornale Italiano Di Dermatologia E Venereologia</i> , 2019, 154, 209-210.	0.8	0
17	A mobile app for patients with Pompe disease and its possible clinical applications. <i>Neuromuscular Disorders</i> , 2018, 28, 471-475.	0.3	15
18	Expanded [CCTG] _n repetitions are not associated with abnormal methylation at the CNBP locus in myotonic dystrophy type 2 (DM2) patients. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2018, 1864, 917-924.	1.8	12

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19	Neurofibromatous neuropathy: An ultrastructural study. <i>Ultrastructural Pathology</i> , 2018, 42, 312-316.	0.4	3
20	Aquaporin 4 expression in human skeletal muscle fiber types. <i>Muscle and Nerve</i> , 2018, 57, 856-858.	1.0	4
21	Late-Onset Pompe Disease with Nemaline Bodies. <i>Case Reports in Neurological Medicine</i> , 2018, 2018, 1-5.	0.3	1
22	Towards clinical outcome measures in myotonic dystrophy type 2: a systematic review. <i>Current Opinion in Neurology</i> , 2018, 31, 599-609.	1.8	8
23	Comparative Sleep Disturbances in Myotonic Dystrophy Types 1 and 2. <i>Current Neurology and Neuroscience Reports</i> , 2018, 18, 102.	2.0	19
24	Validation of the Nine Hole Peg Test as a measure of dexterity in myotonic dystrophy type 1. <i>Neuromuscular Disorders</i> , 2018, 28, 947-951.	0.3	5
25	Generation and Neuronal Differentiation of hiPSCs From Patients With Myotonic Dystrophy Type 2. <i>Frontiers in Physiology</i> , 2018, 9, 967.	1.3	3
26	Muscle MRI in neutral lipid storage disease (NLS). <i>Journal of Neurology</i> , 2017, 264, 1334-1342.	1.8	15
27	Minimalist thoracoscopic resection of thymoma associated with myasthenia gravis. <i>Journal of Thoracic and Cardiovascular Surgery</i> , 2017, 154, 1463-1465.	0.4	11
28	Neutral Lipid Storage Diseases: clinical/genetic features and natural history in a large cohort of Italian patients. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 90.	1.2	49
29	Cutaneous features of myotonic dystrophy types 1 and 2: Implication of premature aging and vitamin D homeostasis. <i>Neuromuscular Disorders</i> , 2017, 27, 163-169.	0.3	18
30	Identification and characterization of 5â€² CCG interruptions in complex DMPK expanded alleles. <i>European Journal of Human Genetics</i> , 2017, 25, 257-261.	1.4	38
31	Congenital myopathies: clinical phenotypes and new diagnostic tools. <i>Italian Journal of Pediatrics</i> , 2017, 43, 101.	1.0	80
32	A case of fulminant subacute sclerosing panencephalitis presenting with acute myoclonic-astatic epilepsy. <i>Annali Dell'Istituto Superiore Di Sanita</i> , 2017, 53, 167-169.	0.2	7
33	LOPED study: looking for an early diagnosis in a late-onset Pompe disease high-risk population. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, jnnp-2014-310164.	0.9	50
34	Neutral lipid storage disease with myopathy and extended phenotype with novel <i>PNPLA2</i> mutation. <i>Muscle and Nerve</i> , 2016, 53, 644-648.	1.0	11
35	Validation of plasma microRNAs as biomarkers for myotonic dystrophy type 1. <i>Scientific Reports</i> , 2016, 6, 38174.	1.6	49
36	MYH7-related myopathies: clinical, histopathological and imaging findings in a cohort of Italian patients. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 91.	1.2	70

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37	Myoimaging in the NGS era: the discovery of a novel mutation in MYH7 in a family with distal myopathy and core-like features – a case report. <i>BMC Medical Genetics</i> , 2016, 17, 25.	2.1	6
38	Neuromuscular transmission abnormalities in myotonic dystrophy type 1: A neurophysiological study. <i>Clinical Neurology and Neurosurgery</i> , 2016, 150, 84-88.	0.6	6
39	Increased risk of tumor in DM1 is not related to exposure to common lifestyle risk factors. <i>Journal of Neurology</i> , 2016, 263, 492-498.	1.8	32
40	Non-neural phenotype of spinal and bulbar muscular atrophy: results from a large cohort of Italian patients. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, 810-816.	0.9	59
41	An Age-Standardized Prevalence Estimate and a Sex and Age Distribution of Myotonic Dystrophy Types 1 and 2 in the Rome Province, Italy. <i>Neuroepidemiology</i> , 2016, 46, 191-197.	1.1	37
42	ALS5/SPG11/ <i>KIAA1840</i> mutations cause autosomal recessive axonal Charcot-Marie-Tooth disease. <i>Brain</i> , 2016, 139, 73-85.	3.7	80
43	Hereditary spastic paraplegia: a novel mutation and expansion of the phenotype variability in SPG10: Table A1. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2015, 86, 702-704.	0.9	11
44	Complete loss of the DNAJB6 G/F domain and novel missense mutations cause distal-onset DNAJB6 myopathy. <i>Acta Neuropathologica Communications</i> , 2015, 3, 44.	2.4	45
45	Brain MR diffusion tensor imaging in Kennedy's disease. <i>Neuroradiology Journal</i> , 2015, 28, 126-132.	0.6	11
46	Clinical and molecular features of a large cohort of Italian McArdle patients. <i>Neuromuscular Disorders</i> , 2015, 25, S219.	0.3	0
47	Sleep disorders in myotonic dystrophy type 2: a controlled polysomnographic study and self-reported questionnaires. <i>European Journal of Neurology</i> , 2014, 21, 929-934.	1.7	37
48	Restless legs syndrome and daytime sleepiness are prominent in myotonic dystrophy type 2. <i>Neurology</i> , 2014, 83, 572-573.	1.5	4
49	Sleep disorders in spinal and bulbar muscular atrophy (Kennedy's disease): a controlled polysomnographic and self-reported questionnaires study. <i>Journal of Neurology</i> , 2014, 261, 889-893.	1.8	21
50	A novel LITAF/SIMPLE mutation within a family with a demyelinating form of Charcot-Marie-Tooth disease. <i>Journal of the Neurological Sciences</i> , 2014, 343, 183-186.	0.3	15
51	Differential features of muscle fiber atrophy in osteoporosis and osteoarthritis. <i>Osteoporosis International</i> , 2013, 24, 1095-1100.	1.3	53
52	Vitamin D deficiency in myotonic dystrophy type 1. <i>Journal of Neurology</i> , 2013, 260, 2330-2334.	1.8	24
53	Paucisymptomatic Marchiafava-Bignami disease with relevant diffusion-weighted MRI lesions. <i>International Journal of Neuroscience</i> , 2013, 123, 738-740.	0.8	2
54	Sleep-Wake Cycle and Daytime Sleepiness in the Myotonic Dystrophies. <i>Journal of Neurodegenerative Diseases</i> , 2013, 2013, 1-13.	1.1	23

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55	Recurrent hyperCKemia with normal muscle biopsy in a pediatric patient with neuromyelitis optica. <i>Neurology</i> , 2012, 79, 1182-1184.	1.5	27
56	Association of HLA-DQB1*05:02 and DRB1*16 Alleles with Late-Onset, Nonthymomatous, AChR-Ab-Positive Myasthenia Gravis. <i>Autoimmune Diseases</i> , 2012, 2012, 1-3.	2.7	30
57	Spasticity as an ictal pattern due to excitotoxic upper motor neuron damage. <i>Epilepsy and Behavior</i> , 2012, 25, 397-400.	0.9	1
58	Periodic acid-Schiff staining on resin muscle sections: Improvement in the histological diagnosis of late-onset Pompe disease. <i>Muscle and Nerve</i> , 2012, 45, 611-612.	1.0	2
59	Randomized comparison of awake nonresectional versus nonawake resectional lung volume reduction surgery. <i>Journal of Thoracic and Cardiovascular Surgery</i> , 2012, 143, 47-54.e1.	0.4	112
60	Aberrant splicing and expression of the non muscle myosin heavy-chain gene MYH14 in DM1 muscle tissues. <i>Neurobiology of Disease</i> , 2012, 45, 264-271.	2.1	20
61	Measuring quality of life impairment in skeletal muscle channelopathies. <i>European Journal of Neurology</i> , 2012, 19, 1470-1476.	1.7	57
62	Sleep disorders in adult-onset myotonic dystrophy type 1: a controlled polysomnographic study. <i>European Journal of Neurology</i> , 2011, 18, 1139-1145.	1.7	72
63	Early subclinical cochlear dysfunction in myotonic dystrophy type 1. <i>European Journal of Neurology</i> , 2011, 18, 1412-1416.	1.7	12
64	Subclinical autonomic dysfunction in spinobulbar muscular atrophy (Kennedy disease). <i>Muscle and Nerve</i> , 2011, 44, 737-740.	1.0	17
65	Diagnosis, treatment and follow-up of the carpal tunnel syndrome: a review. <i>Neurological Sciences</i> , 2010, 31, 243-252.	0.9	136
66	Inverse correlation between VEGF and soluble VEGF receptor 2 in POEMS with AIDP responsive to intravenous immunoglobulin. <i>Muscle and Nerve</i> , 2010, 42, 445-448.	1.0	18
67	Italian validation of INQoL, a quality of life questionnaire for adults with muscle diseases. <i>European Journal of Neurology</i> , 2010, 17, 1178-1187.	1.7	48
68	The myotonic dystrophy type 2 (<i>DM2</i>) gene product zinc finger protein 9 (ZNF9) is associated with sarcomeres and normally localized in DM2 patients' muscles. <i>Neuropathology and Applied Neurobiology</i> , 2010, 36, 275-284.	1.8	15
69	SPATACSIN mutations cause autosomal recessive juvenile amyotrophic lateral sclerosis. <i>Brain</i> , 2010, 133, 591-598.	3.7	227
70	Long-term outcome of thoracoscopic extended thymectomy for nonthymomatous myasthenia gravis. <i>European Journal of Cardio-thoracic Surgery</i> , 2009, 36, 164-169.	0.6	45
71	Prognostic evaluation of brainstem hematomas: the role of CT scan and brainstem auditory evoked potentials. <i>Acta Neurologica Scandinavica</i> , 2009, 70, 396-406.	1.0	5
72	Screening of ARHSP-TCC patients expands the spectrum of <i>SPG11</i> mutations and includes a large scale gene deletion. <i>Human Mutation</i> , 2009, 30, E500-E519.	1.1	53

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73	G.P.3.02 Late-onset MNGIE without peripheral neuropathy due to incomplete loss of thymidine phosphorylase activity. <i>Neuromuscular Disorders</i> , 2009, 19, 561-562.	0.3	0
74	Late-onset MNGIE without peripheral neuropathy due to incomplete loss of thymidine phosphorylase activity. <i>Neuromuscular Disorders</i> , 2009, 19, 837-840.	0.3	34
75	Adult polyglucosan body disease: Proton magnetic resonance spectroscopy of the brain and novel mutation in the <i>GBE1</i> gene. <i>Muscle and Nerve</i> , 2008, 37, 530-536.	1.0	27
76	Preferential central nucleation of type 2 myofibers is an invariable feature of myotonic dystrophy type 2. <i>Muscle and Nerve</i> , 2008, 38, 1405-1411.	1.0	48
77	Risk Prediction for Clinical Phenotype in Myotonic Dystrophy Type 1: Data from 2,650 Patients. <i>Genetic Testing and Molecular Biomarkers</i> , 2007, 11, 84-90.	1.7	46
78	G.P.14.12 Myotonic dystrophy unlinked to DM1 and DM2 mutations in three siblings. <i>Neuromuscular Disorders</i> , 2007, 17, 857.	0.3	0
79	Upper motor neuron involvement in X-linked recessive bulbospinal muscular atrophy. <i>Clinical Neurophysiology</i> , 2007, 118, 262-268.	0.7	19
80	Effect of the [CCTG] _n repeat expansion on ZNF9 expression in myotonic dystrophy type II (DM2). <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2006, 1762, 329-334.	1.8	44
81	Overexpression of ErbB2 and ErbB3 receptors in Schwann cells of patients with Charcot-Marie-Tooth disease type 1A. <i>Muscle and Nerve</i> , 2006, 33, 342-349.	1.0	20
82	Hyper-CK-emia as the sole manifestation of myotonic dystrophy type 2. <i>Muscle and Nerve</i> , 2005, 31, 764-767.	1.0	33
83	Subacute demyelinating polyneuropathy in B-cell lymphoma with IgM antibodies against glycolipid GD1b. <i>Neurological Sciences</i> , 2005, 26, 355-357.	0.9	13
84	Hashimoto's encephalopathy presenting with musical hallucinosis. <i>Journal of Neurology</i> , 2003, 250, 627-628.	1.8	9
85	Intestinal pseudobstruction as presenting event of fatal cerivastatin-induced myopathy. <i>Journal of Neurology</i> , 2003, 250, 887-888.	1.8	2
86	Migraine-like disorder segregating with mtDNA 14484 Leber hereditary optic neuropathy mutation. <i>Neurology</i> , 2003, 60, 717-719.	1.5	23
87	Sural Nerve Without Nerve Fibers in Leprous Neuropathy. <i>Archives of Neurology</i> , 2002, 59, 306.	4.9	1
88	Adrenomyeloneuropathy partially responsive to steroid pulse therapy. <i>Neurological Sciences</i> , 2002, 23, 141-142.	0.9	3
89	Peripheral nerve extracellular matrix remodeling in Charcot-Marie-Tooth type I disease. <i>Acta Neuropathologica</i> , 2002, 104, 287-296.	3.9	32
90	Morphological Changes in the Sciatic Nerve of Diabetic Rats Treated with Low Molecular Weight Heparin OP 2123/Parnaparin. <i>Journal of Veterinary Medicine Series C: Anatomia Histologia Embryologia</i> , 2002, 31, 193-197.	0.3	14

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91	Dopamine denervation induces neurotensin immunoreactivity in GABA-parvalbumin striatal neurons. <i>Synapse</i> , 2001, 41, 360-362.	0.6	5
92	Intracellular localization and isoform expression of the voltage-dependent anion channel (VDAC) in normal and dystrophic skeletal muscle. <i>Journal of Muscle Research and Cell Motility</i> , 2000, 21, 433-442.	0.9	27
93	Nitroergic neurons make synapses on dual-input dendritic spines of neurons in the cerebral cortex and the striatum of the rat: implication for a postsynaptic action of nitric oxide. <i>Neuroscience</i> , 2000, 99, 627-642.	1.1	41
94	Delayed focal involvement of upper motor neurons in the Madras pattern of motor neuron disease. <i>Electroencephalography and Clinical Neurophysiology - Electromyography and Motor Control</i> , 1998, 109, 523-526.	1.4	11
95	Muscle phosphofructokinase deficiency. <i>Neurology</i> , 1997, 49, 899-899.	1.5	0
96	Partial block of glycolysis in late-onset phosphofructokinase deficiency myopathy. <i>Acta Neuropathologica</i> , 1996, 91, 322-329.	3.9	12
97	NADPH-Diaphorase Neurons Contacting the Cerebrospinal Fluid in the Ventricles of Rat Brain. <i>Journal of Cerebral Blood Flow and Metabolism</i> , 1996, 16, 517-522.	2.4	15
98	Immunocytochemical localization of vinculin in muscle and nerve. <i>Muscle and Nerve</i> , 1995, 18, 1277-1284.	1.0	11
99	Dystrophin is not essential for the integrity of the cytoskeleton. <i>Acta Neuropathologica</i> , 1994, 87, 377-384.	3.9	2
100	NADPH diaphorase activity is inhibited by EDTA in neurons but not in choroid plexus epithelium. <i>Neuroscience Letters</i> , 1993, 158, 101-104.	1.0	7
101	Sudomotor skin responses to brain stimulation do not depend on nerve sensory fiber functionality. <i>Electroencephalography and Clinical Neurophysiology - Evoked Potentials</i> , 1993, 89, 447-451.	2.0	17
102	Active muscle length reduction progressively damages soleus in hindlimb-suspended rabbits. <i>Muscle and Nerve</i> , 1992, 15, 1002-1015.	1.0	10
103	Loss and renewal of thick myofilaments in glucocorticoid-treated rat soleus after denervation and reinnervation. <i>Muscle and Nerve</i> , 1992, 15, 1290-1298.	1.0	108
104	Epileptic activity following cerebral ischemia in mongolian gerbils is depressed by CPP, a competitive antagonist of the receptor. <i>Neuroscience Letters</i> , 1991, 129, 306-310.	1.0	2
105	Myofibrillar disruption in the rabbit soleus muscle after one-week hindlimb suspension. <i>Muscle and Nerve</i> , 1991, 14, 358-369.	1.0	18
106	Glucocorticoids and immunosuppressants do not change the prevalence of necrosis and regeneration in mdx skeletal muscles. <i>Muscle and Nerve</i> , 1991, 14, 771-774.	1.0	35
107	Familial Inclusion Body Myositis Among Kurdish-Iranian Jews. <i>Archives of Neurology</i> , 1991, 48, 519-522.	4.9	63
108	Epileptic activity following 10 minute cerebral ischemia in Mongolian gerbils: An electrophysiological study. <i>Neuroscience Letters</i> , 1990, 112, 48-53.	1.0	9

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109	A clinical and epidemiological prevalence study on Friedreich's Ataxia in Latium, Italy.. Neuroepidemiology, 0, , .	1.1	0