Roberto Massa

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/1208277/publications.pdf

Version: 2024-02-01

109	2,719	147726	214721
papers	citations	31 h-index	47 g-index

112	112	112	3727
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	SPATACSIN mutations cause autosomal recessive juvenile amyotrophic lateral sclerosis. Brain, 2010, 133, 591-598.	3.7	227
2	Diagnosis, treatment and follow-up of the carpal tunnel syndrome: a review. Neurological Sciences, 2010, 31, 243-252.	0.9	136
3	Randomized comparison of awake nonresectional versus nonawake resectional lung volume reduction surgery. Journal of Thoracic and Cardiovascular Surgery, 2012, 143, 47-54.e1.	0.4	112
4	Loss and renewal of thick myofilaments in glucocorticoid-treated rat soleus after denervation and reinnervation. Muscle and Nerve, 1992, 15, 1290-1298.	1.0	108
5	ALS5/SPG11/KIAA1840mutations cause autosomal recessive axonal Charcotâ€“Marieâ€“Tooth disease. Brain, 2016, 139, 73-85.	3.7	80
6	Congenital myopathies: clinical phenotypes and new diagnostic tools. Italian Journal of Pediatrics, 2017, 43, 101.	1.0	80
7	Sleep disorders in adult-onset myotonic dystrophy type 1: a controlled polysomnographic study. European Journal of Neurology, 2011, 18, 1139-1145.	1.7	72
8	MYH7-related myopathies: clinical, histopathological and imaging findings in a cohort of Italian patients. Orphanet Journal of Rare Diseases, 2016, 11, 91.	1.2	70
9	Familial Inclusion Body Myositis Among Kurdish-Iranian Jews. Archives of Neurology, 1991, 48, 519-522.	4.9	63
10	Non-neural phenotype of spinal and bulbar muscular atrophy: results from a large cohort of Italian patients. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 810-816.	0.9	59
11	Measuring quality of life impairment in skeletal muscle channelopathies. European Journal of Neurology, 2012, 19, 1470-1476.	1.7	57
12	Screening of ARHSP-TCC patients expands the spectrum ofSPG11mutations and includes a large scale gene deletion. Human Mutation, 2009, 30, E500-E519.	1.1	53
13	Differential features of muscle fiber atrophy in osteoporosis and osteoarthritis. Osteoporosis International, 2013, 24, 1095-1100.	1.3	53
14	LOPED study: looking for an early diagnosis in a late-onset Pompe disease high-risk population. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, jnnp-2014-310164.	0.9	50
15	Validation of plasma microRNAs as biomarkers for myotonic dystrophy type 1. Scientific Reports, 2016, 6, 38174.	1.6	49
16	Neutral Lipid Storage Diseases: clinical/genetic features and natural history in a large cohort of Italian patients. Orphanet Journal of Rare Diseases, 2017, 12, 90.	1.2	49
17	Preferential central nucleation of type 2 myofibers is an invariable feature of myotonic dystrophy type 2. Muscle and Nerve, 2008, 38, 1405-1411.	1.0	48
18	Italian validation of INQoL, a quality of life questionnaire for adults with muscle diseases. European Journal of Neurology, 2010, 17, 1178-1187.	1.7	48

#	Article	IF	Citations
19	Risk Prediction for Clinical Phenotype in Myotonic Dystrophy Type 1: Data from 2,650 Patients. Genetic Testing and Molecular Biomarkers, 2007, 11, 84-90.	1.7	46
20	Long-term outcome of thoracoscopic extended thymectomy for nonthymomatous myasthenia gravisâ˜†â˜†â˜†. European Journal of Cardio-thoracic Surgery, 2009, 36, 164-169.	0.6	45
21	Complete loss of the DNAJB6 G/F domain and novel missense mutations cause distal-onset DNAJB6 myopathy. Acta Neuropathologica Communications, 2015, 3, 44.	2.4	45
22	Effect of the [CCTG]n repeat expansion on ZNF9 expression in myotonic dystrophy type II (DM2). Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2006, 1762, 329-334.	1.8	44
23	Nitrergic 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. Neuroscience, 2000, 99, 627-642.	1.1	41
24	Identification and characterization of 5â€² CCG interruptions in complex DMPK expanded alleles. European Journal of Human Genetics, 2017, 25, 257-261.	1.4	38
25	Italian recommendations for the diagnosis and treatment of myasthenia gravis. Neurological Sciences, 2019, 40, 1111-1124.	0.9	38
26	Sleep disorders in myotonic dystrophy type 2: a controlled polysomnographic study and selfâ€reported questionnaires. European Journal of Neurology, 2014, 21, 929-934.	1.7	37
27	An Age-Standardized Prevalence Estimate and a Sex and Age Distribution of Myotonic Dystrophy Types 1 and 2 in the Rome Province, Italy. Neuroepidemiology, 2016, 46, 191-197.	1.1	37
28	Glucocorticoids and immunosup pressants do not change the prevalence of necrosis and regeneration in mdx skeletal muscles. Muscle and Nerve, 1991, 14, 771-774.	1.0	35
29	Late-onset MNGIE without peripheral neuropathy due to incomplete loss of thymidine phosphorylase activity. Neuromuscular Disorders, 2009, 19, 837-840.	0.3	34
30	Hyper-CK-emia as the sole manifestation of myotonic dystrophy type 2. Muscle and Nerve, 2005, 31, 764-767.	1.0	33
31	Peripheral nerve extracellular matrix remodeling in Charcot-Marie-Tooth type I disease. Acta Neuropathologica, 2002, 104, 287-296.	3.9	32
32	Increased risk of tumor in DM1 is not related to exposure to common lifestyle risk factors. Journal of Neurology, 2016, 263, 492-498.	1.8	32
33	Prevalence and predictor factors of respiratory impairment in a large cohort of patients with Myotonic Dystrophy type 1 (DM1): A retrospective, cross sectional study. Journal of the Neurological Sciences, 2019, 399, 118-124.	0.3	31
34	Association of HLA-DQB1*05:02 and DRB1*16 Alleles with Late-Onset, Nonthymomatous, AChR-Ab-Positive Myasthenia Gravis. Autoimmune Diseases, 2012, 2012, 1-3.	2.7	30
35	Intracellular localization and isoform expression of the voltage-dependent anion channel (VDAC) in normal and dystrophic skeletal muscle. Journal of Muscle Research and Cell Motility, 2000, 21, 433-442.	0.9	27
36	Adult polyglucosan body disease: Proton magnetic resonance spectroscopy of the brain and novel mutation in the GBE1 gene. Muscle and Nerve, 2008, 37, 530-536.	1.0	27

#	Article	IF	Citations
37	Recurrent hyperCKemia with normal muscle biopsy in a pediatric patient with neuromyelitis optica. Neurology, 2012, 79, 1182-1184.	1.5	27
38	Vitamin D deficiency in myotonic dystrophy type 1. Journal of Neurology, 2013, 260, 2330-2334.	1.8	24
39	Migraine-like disorder segregating with mtDNA 14484 Leber hereditary optic neuropathy mutation. Neurology, 2003, 60, 717-719.	1.5	23
40	Sleep-Wake Cycle and Daytime Sleepiness in the Myotonic Dystrophies. Journal of Neurodegenerative Diseases, 2013, 2013, 1-13.	1.1	23
41	Sleep disorders in spinal and bulbar muscular atrophy (Kennedyâ€™s disease): a controlled polysomnographic and self-reported questionnaires study. Journal of Neurology, 2014, 261, 889-893.	1.8	21
42	Overexpression of ErbB2 and ErbB3 receptors in Schwann cells of patients with Charcotâ€“Marieâ€“Tooth disease type 1A. Muscle and Nerve, 2006, 33, 342-349.	1.0	20
43	Aberrant splicing and expression of the non muscle myosin heavy-chain gene MYH14 in DM1 muscle tissues. Neurobiology of Disease, 2012, 45, 264-271.	2.1	20
44	Upper motor neuron involvement in X-linked recessive bulbospinal muscular atrophy. Clinical Neurophysiology, 2007, 118, 262-268.	0.7	19
45	Comparative Sleep Disturbances in Myotonic Dystrophy Types 1 and 2. Current Neurology and Neuroscience Reports, 2018, 18, 102.	2.0	19
46	Myofibrillar disruption in the rabbit soleus muscle after one-week hindlimb suspension. Muscle and Nerve, 1991, 14, 358-369.	1.0	18
47	Inverse correlation between VEGF and soluble VEGF receptor 2 in POEMS with AIDP responsive to intravenous immunoglobulin. Muscle and Nerve, 2010, 42, 445-448.	1.0	18
48	Cutaneous features of myotonic dystrophy types 1 and 2: Implication of premature aging and vitamin D homeostasis. Neuromuscular Disorders, 2017, 27, 163-169.	0.3	18
49	Sudomotor skin responses to brain stimulation do not depend on nerve sensory fiber functionality. Electroencephalography and Clinical Neurophysiology - Evoked Potentials, 1993, 89, 447-451.	2.0	17
50	Subclinical autonomic dysfunction in spinobulbar muscular atrophy (Kennedy disease). Muscle and Nerve, 2011, 44, 737-740.	1.0	17
51	NADPH-Diaphorase Neurons Contacting the Cerebrospinal Fluid in the Ventricles of Rat Brain. Journal of Cerebral Blood Flow and Metabolism, 1996, 16, 517-522.	2.4	15
52	The myotonic dystrophy type 2 (DM2) gene product zinc finger protein 9 (ZNF9) is associated with sarcomeres and normally localized in DM2 patients' muscles. Neuropathology and Applied Neurobiology, 2010, 36, 275-284.	1.8	15
53	A novel LITAF/SIMPLE mutation within a family with a demyelinating form of Charcotâ€“Marieâ€“Tooth disease. Journal of the Neurological Sciences, 2014, 343, 183-186.	0.3	15
54	Muscle MRI in neutral lipid storage disease (NLSD). Journal of Neurology, 2017, 264, 1334-1342.	1.8	15

#	Article	IF	Citations
55	A mobile app for patients with Pompe disease and its possible clinical applications. Neuromuscular Disorders, 2018, 28, 471-475.	0.3	15
56	Morphological Changes in the Sciatic Nerve of Diabetic Rats Treated with Low Molecular Weight Heparin OP 2123/Parnaparin. Journal of Veterinary Medicine Series C: Anatomia Histologia Embryologia, 2002, 31, 193-197.	0.3	14
57	Thymomatous myasthenia gravis: novel association with HLA DQB1*05:01 and strengthened evidence of high clinical and serological severity. Journal of Neurology, 2019, 266, 982-989.	1.8	14
58	Subacute demyelinating polyneuropathy in B-cell lymphoma with IgM antibodies against glycolipid GD1b. Neurological Sciences, 2005, 26, 355-357.	0.9	13
59	Partial block of glycolysis in late-onset phosphofructokinase deficiency myopathy. Acta Neuropathologica, 1996, 91, 322-329.	3.9	12
60	Early subclinical cochlear dysfunction in myotonic dystrophy type 1. European Journal of Neurology, 2011, 18, 1412-1416.	1.7	12
61	Expanded [CCTG]n repetitions are not associated with abnormal methylation at the CNBP locus in myotonic dystrophy type 2 (DM2) patients. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 917-924.	1.8	12
62	Immunocytochemical localization of vinculin in muscle and nerve. Muscle and Nerve, 1995, 18, 1277-1284.	1.0	11
63	Delayed focal involvement of upper motor neurons in the Madras pattern of motor neuron disease. Electroencephalography and Clinical Neurophysiology - Electromyography and Motor Control, 1998, 109, 523-526.	1.4	11
64	Hereditary spastic paraplegia: a novel mutation and expansion of the phenotype variability in SPG10: TableÂ1. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 702-704.	0.9	11
65	Brain MR diffusion tensor imaging in Kennedyâ€™s disease. Neuroradiology Journal, 2015, 28, 126-132.	0.6	11
66	Neutral lipidâ€storage disease with myopathy and extended phenotype with novel PNPLA2 mutation. Muscle and Nerve, 2016, 53, 644-648.	1.0	11
67	Minimalist thoracoscopic resection of thymoma associated with myasthenia gravis. Journal of Thoracic and Cardiovascular Surgery, 2017, 154, 1463-1465.	0.4	11
68	Active muscle length reduction progressively damages soleus in hindlimb-suspended rabbits. Muscle and Nerve, 1992, 15, 1002-1015.	1.0	10
69	Epileptic activity following 10 minute cerebral ischemia in Mongolian gerbils: An electrophysiological study. Neuroscience Letters, 1990, 112, 48-53.	1.0	9
70	Hashimoto's encephalopathy presenting with musical hallucinosis. Journal of Neurology, 2003, 250, 627-628.	1.8	9
71	Validation of Motor Outcome Measures in Myotonic Dystrophy Type 2. Frontiers in Neurology, 2020, 11, 306.	1.1	9
72	ATTRv in Lazio-Italy: A High-Prevalence Region in a Non-Endemic Country. Genes, 2021, 12, 829.	1.0	9

#	Article	IF	Citations
73	Towards clinical outcome measures in myotonic dystrophy type 2: a systematic review. Current Opinion in Neurology, 2018, 31, 599-609.	1.8	8
74	NADPH diaphorase activity is inhibited by EDTA in neurons but not in choroid plexus epithelium. Neuroscience Letters, 1993, 158, 101-104.	1.0	7
75	A case of fulminant subacute sclerosing panencephalitis presenting with acute myoclonic-astatic epilepsy. Annali Dell'Istituto Superiore Di Sanita, 2017, 53, 167-169.	0.2	7
76	Myoimaging in the NGS era: the discovery of a novel mutation in MYH7 in a family with distal myopathy and core-like features â€“ a case report. BMC Medical Genetics, 2016, 17, 25.	2.1	6
77	Neuromuscular transmission abnormalities in myotonic dystrophy type 1: A neurophysiological study. Clinical Neurology and Neurosurgery, 2016, 150, 84-88.	0.6	6
78	Dopamine denervation induces neurotensin immunoreactivity in GABA-parvalbumin striatal neurons. Synapse, 2001, 41, 360-362.	0.6	5
79	Prognostic evaluation of brainstem hematomas: the role of CT scan and brainstem auditory evoked potentials. Acta Neurologica Scandinavica, 2009, 70, 396-406.	1.0	5
80	Validation of the Nine Hole Peg Test as a measure of dexterity in myotonic dystrophy type 1. Neuromuscular Disorders, 2018, 28, 947-951.	0.3	5
81	Assessment of self-reported and objective daytime sleepiness in adult-onset myotonic dystrophy type 1. Journal of Clinical Sleep Medicine, 2021, 17, 2383-2391.	1.4	5
82	The actigraphic documentation of circadian sleep-wake rhythm dysregulation in myotonic dystrophy type 1. Sleep Medicine, 2021, 88, 134-139.	0.8	5
83	Restless legs syndrome and daytime sleepiness are prominent in myotonic dystrophy type 2. Neurology, 2014, 83, 572-573.	1.5	4
84	Aquaporin 4 expression in human skeletal muscle fiber types. Muscle and Nerve, 2018, 57, 856-858.	1.0	4
85	Cochlear Dysfunction Is a Frequent Feature of Facioscapulohumeral Muscular Dystrophy Type 1 (FSHD1). Otology and Neurotology, 2021, 42, 18-23.	0.7	4
86	Adrenomyeloneuropathy partially responsive to steroid pulse therapy. Neurological Sciences, 2002, 23, 141-142.	0.9	3
87	Neurofibromatous neuropathy: An ultrastructural study. Ultrastructural Pathology, 2018, 42, 312-316.	0.4	3
88	Generation and Neuronal Differentiation of hiPSCs From Patients With Myotonic Dystrophy Type 2. Frontiers in Physiology, 2018, 9, 967.	1.3	3
89	A 14-Year Italian Experience in DM2 Genetic Testing: Frequency and Distribution of Normal and Premutated CNBP Alleles. Frontiers in Genetics, 2021, 12, 668094.	1.1	3
90	Epileptic activity following cerebral ischemia in mongolian gerbils is depressed by CPP, a competitive antagonist of the receptor. Neuroscience Letters, 1991, 129, 306-310.	1.0	2

#	Article	IF	Citations
91	Intestinal pseudobstruction as presenting event of fatal cerivastatininduced myopathy. Journal of Neurology, 2003, 250, 887-888.	1.8	2
92	Periodic acid-Schiff staining on resin muscle sections: Improvement in the histological diagnosis of late-onset Pompe disease. Muscle and Nerve, 2012, 45, 611-612.	1.0	2
93	Paucisymptomatic Marchiafavaâ€“Bignami disease with relevant diffusion-weighted MRI lesions. International Journal of Neuroscience, 2013, 123, 738-740.	0.8	2
94	Design of a multiplex ligation-dependent probe amplification assay for SLC20A2: identification of two novel deletions in primary familial brain calcification. Journal of Human Genetics, 2019, 64, 1083-1090.	1.1	2
95	How to capture activities of daily living in myotonic dystrophy type 2?. Neuromuscular Disorders, 2020, 30, 796-806.	0.3	2
96	Movement disorders in primary central nervous system lymphoma: two unreported cases and a review of literature. Neurological Sciences, 2021, 42, 905-910.	0.9	2
97	Dystrophin is not essential for the integrity of the cytoskeleton. Acta Neuropathologica, 1994, 87, 377-384.	3.9	2
98	Sural Nerve Without Nerve Fibers in Leprous Neuropathy. Archives of Neurology, 2002, 59, 306.	4.9	1
99	Spasticity as an ictal pattern due to excitotoxic upper motor neuron damage. Epilepsy and Behavior, 2012, 25, 397-400.	0.9	1
100	Late-Onset Pompe Disease with Nemaline Bodies. Case Reports in Neurological Medicine, 2018, 2018, 1-5.	0.3	1
101	Objective Assessment of Walking Impairments in Myotonic Dystrophy by Means of a Wearable Technology and a Novel Severity Index. Electronics (Switzerland), 2021, 10, 708.	1.8	1
102	Muscle phosphofructokinase deficiency. Neurology, 1997, 49, 899-899.	1.5	0
103	G.P.14.12 Myotonic dystrophy unlinked to DM1 and DM2 mutations in three siblings. Neuromuscular Disorders, 2007, 17, 857.	0.3	0
104	G.P.3.02 Late-onset MNGIE without peripheral neuropathy due to incomplete loss of thymidine phosphorylase activity. Neuromuscular Disorders, 2009, 19, 561-562.	0.3	0
105	Clinical and molecular features of a large cohort of Italian McArdle patients. Neuromuscular Disorders, 2015, 25, S219.	0.3	0
106	Reply to the letter entitled â€œPredictors of respiratory impairment in patients with myotonic dystrophy type 1â€• Journal of the Neurological Sciences, 2019, 403, 166-167.	0.3	0
107	Nodular morphea in a patient with Steinert disease. Giornale Italiano Di Dermatologia E Venereologia, 2019, 154, 209-210.	0.8	0
108	RESPONSE TO LETTER TO THE EDITOR â€œAUDITORY DYSFUNCTION IN FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY TYPE 1: BEYOND THE INNER EAR INVOLVEMENTâ€•BY GHELLER ET AL. Otology and Neurotology, 2022, 43, e392-e393.	0.7	0

#	Article	IF	Citations
109	A clinical and epidemiological prevalence study on Friedreichâ€™s Ataxia in Latium, Italy.. Neuroepidemiology, 0, , .	1.1	0