

Ricardo A Maselli

List of Publications by Year in descending order

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Version: 2024-02-01

52
papers

1,531
citations

279798

23
h-index

330143

37
g-index

52
all docs

52
docs citations

52
times ranked

1312
citing authors

#	ARTICLE	IF	CITATIONS
1	Neuromuscular transmission in amyotrophic lateral sclerosis. <i>Muscle and Nerve</i> , 1993, 16, 1193-1203.	2.2	118
2	Mutations in MUSK causing congenital myasthenic syndrome impair MuSKâ€Dok-7 interaction. <i>Human Molecular Genetics</i> , 2010, 19, 2370-2379.	2.9	86
3	LG2 agrin mutation causing severe congenital myasthenic syndrome mimics functional characteristics of non-neural (zâˆ) agrin. <i>Human Genetics</i> , 2012, 131, 1123-1135.	3.8	86
4	Sympathetic skin response in diabetic neuropathy. <i>Muscle and Nerve</i> , 1987, 10, 711-716.	2.2	84
5	Sympathetic skin responses are decreased and lymphocyte betaâ€adrenergic receptors are increased in progressive multiple sclerosis. <i>Annals of Neurology</i> , 1990, 27, 366-372.	5.3	84
6	Comparison of sympathetic skin response with quantitative sudomotor axon reflex test in diabetic neuropathy. <i>Muscle and Nerve</i> , 1989, 12, 420-423.	2.2	78
7	Choline acetyltransferase mutations in myasthenic syndrome due to deficient acetylcholine resynthesis. <i>Muscle and Nerve</i> , 2003, 27, 180-187.	2.2	74
8	Defective fast inactivation recovery of <scp>N</scp>a_v1.4 in congenital myasthenic syndrome. <i>Annals of Neurology</i> , 2015, 77, 840-850.	5.3	55
9	Analysis of the organophosphate-induced electromyographic response to repetitive nerve stimulation: Paradoxical response to edrophonium andD-Tubocurarine. <i>Muscle and Nerve</i> , 1991, 14, 1182-1188.	2.2	51
10	Botulism. <i>Muscle and Nerve</i> , 2000, 23, 1137-1144.	2.2	51
11	Cluster of wound botulism in California: Clinical, electrophysiologic, and pathologic study. , 1997, 20, 1284-1295.		48
12	Variable phenotypes associated with mutations in <i>DOK7</i>. <i>Muscle and Nerve</i> , 2008, 37, 448-456.	2.2	46
13	Identification of pathogenic mutations in the human rapsyn gene. <i>Journal of Human Genetics</i> , 2003, 48, 204-207.	2.3	43
14	Rapsyn mutations in myasthenic syndrome due to impaired receptor clustering. <i>Muscle and Nerve</i> , 2003, 28, 293-301.	2.2	40
15	Single fiber electromyography of extraocular muscles: A sensitive method for the diagnosis of ocular myasthenia gravis. <i>Muscle and Nerve</i> , 1995, 18, 943-947.	2.2	38
16	Analysis of anticholinesterase-induced neuromuscular transmission failure. <i>Muscle and Nerve</i> , 1993, 16, 548-553.	2.2	37
17	Acute Severe Animal Model of Antiâ€Muscle-Specific Kinase Myasthenia. <i>Archives of Neurology</i> , 2012, 69, 453.	4.5	37
18	Anconeus muscle: A human muscle preparation suitable for in-vitro microelectrode studies. <i>Muscle and Nerve</i> , 1991, 14, 1189-1192.	2.2	33

#	ARTICLE	IF	CITATIONS
19	Neuromuscular transmission as a function of motor unit size in patients with prior polymyolitis. <i>Muscle and Nerve</i> , 1992, 15, 648-655.	2.2	32
20	Focal caspase activation underlies the endplate myopathy in slow-channel syndrome. <i>Annals of Neurology</i> , 2004, 55, 347-352.	5.3	30
21	COOH-terminal collagen Q (COLQ) mutants causing human deficiency of endplate acetylcholinesterase impair the interaction of ColQ with proteins of the basal lamina. <i>Human Genetics</i> , 2014, 133, 599-616.	3.8	29
22	Presynaptic congenital myasthenic syndrome with a homozygous sequence variant in <i>LAMA5</i> combines myopia, facial tics, and failure of neuromuscular transmission. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2240-2245.	1.2	29
23	Pathogenesis of Human Botulism. <i>Annals of the New York Academy of Sciences</i> , 1998, 841, 122-129.	3.8	28
24	Editorial by concerned physicians: Unintended effect of the orphan drug act on the potential cost of 3,4-diaminopyridine. <i>Muscle and Nerve</i> , 2016, 53, 165-168.	2.2	24
25	Synaptic basal lamina-associated congenital myasthenic syndromes. <i>Annals of the New York Academy of Sciences</i> , 2012, 1275, 36-48.	3.8	23
26	Stimulated single-fiber electromyography in wound botulism. , 1996, 19, 1171-1173.		22
27	Edrophonium: An aid in the diagnosis of acute organophosphate poisoning. <i>Annals of Neurology</i> , 1986, 19, 508-510.	5.3	20
28	Choline Acetyltransferase Mutations Causing Congenital Myasthenic Syndrome: Molecular Findings and Genotype-Phenotype Correlations. <i>Human Mutation</i> , 2015, 36, 881-893.	2.5	20
29	In vitro microelectrode study of neuromuscular transmission in a case of botulism. <i>Muscle and Nerve</i> , 1992, 15, 273-276.	2.2	18
30	Congenital Myasthenic Syndrome due to DOK7 mutations in a family from Chile. <i>European Journal of Translational Myology</i> , 2017, 27, 6832.	1.7	14
31	Recessive congenital myasthenic syndrome caused by a homozygous mutation in SYT2 altering a highly conserved C-terminal amino acid sequence. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1744-1749.	1.2	14
32	Function and Ultrastructure of the Neuromuscular Junction in Post-Polio Syndrome. <i>Annals of the New York Academy of Sciences</i> , 1995, 753, 129-137.	3.8	13
33	High Throughput Genetic Analysis of Congenital Myasthenic Syndromes Using Resequencing Microarrays. <i>PLoS ONE</i> , 2007, 2, e918.	2.5	13
34	Electrodiagnosis of Disorders of Neuromuscular Transmission. <i>Annals of the New York Academy of Sciences</i> , 1998, 841, 696-711.	3.8	10
35	Single motor unit H-reflex in motor neuron disorders. <i>Muscle and Nerve</i> , 1992, 15, 656-660.	2.2	9
36	Analysis of Neuromuscular Transmission Failure Induced by Anticholinesterases. <i>Annals of the New York Academy of Sciences</i> , 1993, 681, 402-404.	3.8	9

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37	Presynaptic congenital myasthenic syndrome with altered synaptic vesicle homeostasis linked to compound heterozygous sequence variants in <i>RPH3A</i> . <i>Molecular Genetics & Genomic Medicine</i> , 2018, 6, 434-440.	1.2	9
38	Antibodies to AChR, Synapse-organizing Proteins, Titin, and Other Muscle Proteins in Morvan's Fibrillary Chorea. <i>Annals of the New York Academy of Sciences</i> , 1998, 841, 522-524.	3.8	8
39	Effect of Inherited Abnormalities of Calcium Regulation on Human Neuromuscular Transmission. <i>Annals of the New York Academy of Sciences</i> , 2003, 998, 18-28.	3.8	8
40	Common founder effect of rapsyn N88K studied using intragenic markers. <i>Journal of Human Genetics</i> , 2004, 49, 366-369.	2.3	8
41	Dominant and recessive congenital myasthenic syndromes caused by SYT2 mutations. <i>Muscle and Nerve</i> , 2021, 64, 219-224.	2.2	8
42	End-plate electromyography: Use of spectral analysis of end-plate noise. , 1997, 20, 52-58.		7
43	Effect of 3,4-diaminopyridine at the murine neuromuscular junction. <i>Muscle and Nerve</i> , 2017, 55, 223-231.	2.2	7
44	A presynaptic congenital myasthenic syndrome attributed to a homozygous sequence variant in <i>LAMA5</i> . <i>Annals of the New York Academy of Sciences</i> , 2018, 1413, 119-125.	3.8	7
45	Pathogenic effects of agrin V1727F mutation are isoform specific and decrease its expression and affinity for HSPGs and LRP4. <i>Human Molecular Genetics</i> , 2019, 28, 2648-2658.	2.9	7
46	Protection of human muscle acetylcholinesterase from soman by pyridostigmine bromide. <i>Muscle and Nerve</i> , 2011, 43, 591-595.	2.2	6
47	Axonal neuropathy in female carriers of the fragile X premutation with fragile X-associated tremor ataxia syndrome. <i>Muscle and Nerve</i> , 2015, 52, 234-239.	2.2	5
48	Letters to the editor. <i>Muscle and Nerve</i> , 1992, 15, 1369-1375.	2.2	2
49	Phenotypic Differences in 2 Unrelated Cases Carrying Identical DOK7 Mutations. <i>Journal of Clinical Neuromuscular Disease</i> , 2019, 21, 30-34.	0.7	2
50	Atypical Case of POEMS Presented as Demyelinating Polyneuropathy With Motor Conduction Block. <i>Journal of Clinical Neuromuscular Disease</i> , 2021, 23, 116-118.	0.7	1
51	Letters to the editor. <i>Muscle and Nerve</i> , 1993, 16, 562-570.	2.2	0
52	Rhodamine-labeled Alpha-Bungarotoxin Allows Visualization of End Plates in Congenital End Plate Acetylcholinesterase Deficiency (CEAD). <i>Annals of the New York Academy of Sciences</i> , 1998, 841, 207-209.	3.8	0