Ricardo A Maselli

List of Publications by Year
in descending order

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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	Dominant and recessive congenital myasthenic syndromes caused by SYT2 mutations. Muscle and Nerve, 2021, 64, 219-224.	2.2	8
2	Atypical Case of POEMS Presented as Demyelinating Polyneuropathy With Motor Conduction Block. Journal of Clinical Neuromuscular Disease, 2021, 23, 116-118.	0.7	1
3	Recessive congenital myasthenic syndrome caused by a homozygous mutation inSYT2altering a highly conserved Câ€terminal amino acid sequence. American Journal of Medical Genetics, Part A, 2020, 182, 1744-1749.	1.2	14
4	Pathogenic effects of agrin V1727F mutation are isoform specific and decrease its expression and affinity for HSPGs and LRP4. Human Molecular Genetics, 2019, 28, 2648-2658.	2.9	7
5	Phenotypic Differences in 2 Unrelated Cases Carrying Identical DOK7 Mutations. Journal of Clinical Neuromuscular Disease, 2019, 21, 30-34.	0.7	2
6	Presynaptic congenital myasthenic syndrome with altered synaptic vesicle homeostasis linked to compound heterozygous sequence variants in <scp>RPH</scp>3A. Molecular Genetics & Genomic Medicine, 2018, 6, 434-440.	1.2	9
7	A presynaptic congenital myasthenic syndrome attributed to a homozygous sequence variant in LAMA5. Annals of the New York Academy of Sciences, 2018, 1413, 119-125.	3.8	7
8	Effect of 3,4â€diaminopyridine at the murine neuromuscular junction. Muscle and Nerve, 2017, 55, 223-231.	2.2	7
9	Presynaptic congenital myasthenic syndrome with a homozygous sequence variant in LAMA5 combines myopia, facial tics, and failure of neuromuscular transmission. American Journal of Medical Genetics, Part A, 2017, 173, 2240-2245.	1.2	29
10	Congenital Myasthenic Syndrome due to DOK7 mutations in a family from Chile. European Journal of Translational Myology, 2017, 27, 6832.	1.7	14
11	Editorial by concerned physicians: Unintended effect of the orphan drug act on the potential cost of 3,4-diaminopyridine. Muscle and Nerve, 2016, 53, 165-168.	2.2	24
12	Axonal neuropathy in female carriers of the fragile X premutation with fragile xâ€“associated tremor ataxia syndrome. Muscle and Nerve, 2015, 52, 234-239.	2.2	5
13	Choline Acetyltransferase Mutations Causing Congenital Myasthenic Syndrome: Molecular Findings and Genotype-Phenotype Correlations. Human Mutation, 2015, 36, 881-893.	2.5	20
14	Defective fast inactivation recovery of <scp>N</scp>a_v1.4 in congenital myasthenic syndrome. Annals of Neurology, 2015, 77, 840-850.	5.3	55
15	COOH-terminal collagen Q (COLQ) mutants causing human deficiency of endplate acetylcholinesterase impair the interaction of ColQ with proteins of the basal lamina. Human Genetics, 2014, 133, 599-616.	3.8	29
16	Acute Severe Animal Model of Antiâ€“Muscle-Specific Kinase Myasthenia. Archives of Neurology, 2012, 69, 453.	4.5	37
17	Synaptic basal laminaâ€“associated congenital myasthenic syndromes. Annals of the New York Academy of Sciences, 2012, 1275, 36-48.	3.8	23
18	LG2 agrin mutation causing severe congenital myasthenic syndrome mimics functional characteristics of non-neural (zâ^') agrin. Human Genetics, 2012, 131, 1123-1135.	3.8	86

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19	Protection of human muscle acetylcholinesterase from soman by pyridostigmine bromide. Muscle and Nerve, 2011, 43, 591-595.	2.2	6
20	Mutations in MUSK causing congenital myasthenic syndrome impair MuSKâ€“Dok-7 interaction. Human Molecular Genetics, 2010, 19, 2370-2379.	2.9	86
21	Variable phenotypes associated with mutations in DOK7. Muscle and Nerve, 2008, 37, 448-456.	2.2	46
22	High Throughput Genetic Analysis of Congenital Myasthenic Syndromes Using Resequencing Microarrays. PLoS ONE, 2007, 2, e918.	2.5	13
23	Common founder effect of rapsyn N88K studied using intragenic markers. Journal of Human Genetics, 2004, 49, 366-369.	2.3	8
24	Focal caspase activation underlies the endplate myopathy in slow-channel syndrome. Annals of Neurology, 2004, 55, 347-352.	5.3	30
25	Effect of Inherited Abnormalities of Calcium Regulation on Human Neuromuscular Transmission. Annals of the New York Academy of Sciences, 2003, 998, 18-28.	3.8	8
26	Identification of pathogenic mutations in the human rapsyn gene. Journal of Human Genetics, 2003, 48, 204-207.	2.3	43
27	Choline acetyltransferase mutations in myasthenic syndrome due to deficient acetylcholine resynthesis. Muscle and Nerve, 2003, 27, 180-187.	2.2	74
28	Rapsyn mutations in myasthenic syndrome due to impaired receptor clustering. Muscle and Nerve, 2003, 28, 293-301.	2.2	40
29	Botulism. Muscle and Nerve, 2000, 23, 1137-1144.	2.2	51
30	Pathogenesis of Human Botulism. Annals of the New York Academy of Sciences, 1998, 841, 122-129.	3.8	28
31	Rhodamine-labeled Alpha-Bungarotoxin Allows Visualization of End Plates in Congenital End Plate Acetylcholinesterase Deficiency (CEAD). Annals of the New York Academy of Sciences, 1998, 841, 207-209.	3.8	0
32	Antibodies to AChR, Synapse-organizing Proteins, Titin, and Other Muscle Proteins in Morvan's Fibrillary Choreaa. Annals of the New York Academy of Sciences, 1998, 841, 522-524.	3.8	8
33	Electrodiagnosis of Disorders of Neuromuscular Transmission. Annals of the New York Academy of Sciences, 1998, 841, 696-711.	3.8	10
34	End-plate electromyography: Use of spectral analysis of end-plate noise. , 1997, 20, 52-58.		7
35	Cluster of wound botulism in California: Clinical, electrophysiologic, and pathologic study. , 1997, 20, 1284-1295.		48
36	Stimulated single-fiber electromyography in wound botulism. , 1996, 19, 1171-1173.		22

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37	Single fiber electromyography of extraocular muscles: A sensitive method for the diagnosis of ocular myasthenia gravis. Muscle and Nerve, 1995, 18, 943-947.	2.2	38
38	Function and Ultrastructure of the Neuromuscular Junction in Post-Polio Syndrome. Annals of the New York Academy of Sciences, 1995, 753, 129-137.	3.8	13
39	Analysis of anticholinesterase-induced neuromuscular transmission failure. Muscle and Nerve, 1993, 16, 548-553.	2.2	37
40	Letters to the editor. Muscle and Nerve, 1993, 16, 562-570.	2.2	0
41	Neuromuscular transmission in amyotrophic lateral sclerosis. Muscle and Nerve, 1993, 16, 1193-1203.	2.2	118
42	Analysis of Neuromuscular Transmission Failure Induced by Anticholinesterases. Annals of the New York Academy of Sciences, 1993, 681, 402-404.	3.8	9
43	In vitro microelectrode study of neuromuscular transmission in a case of botulism. Muscle and Nerve, 1992, 15, 273-276.	2.2	18
44	Neuromuscular transmission as a function of motor unit size in patients with prior polimyelitis. Muscle and Nerve, 1992, 15, 648-655.	2.2	32
45	Single motor unit H-reflex in motor neuron disorders. Muscle and Nerve, 1992, 15, 656-660.	2.2	9
46	Letters to the editor. Muscle and Nerve, 1992, 15, 1369-1375.	2.2	2
47	Analysis of the organophosphate-induced electromyographic response to repetitive nerve stimulation: Paradoxical response to edrophonium andD-Tubocurarine. Muscle and Nerve, 1991, 14, 1182-1188.	2.2	51
48	Anconeus muscle: A human muscle preparation suitable for in-vitro microelectrode studies. Muscle and Nerve, 1991, 14, 1189-1192.	2.2	33
49	Sympathetic skin responses are decreased and lymphocyte betaâ€adrenergic receptors are increased in progressive multiple sclerosis. Annals of Neurology, 1990, 27, 366-372.	5.3	84
50	Comparison of sympathetic skin response with quantitative sudomotor axon reflex test in diabetic neuropathy. Muscle and Nerve, 1989, 12, 420-423.	2.2	78
51	Sympathetic skin response in diabetic neuropathy. Muscle and Nerve, 1987, 10, 711-716.	2.2	84
52	Edrophonium: An aid in the diagnosis of acute organophosphate poisoning. Annals of Neurology, 1986, 19, 508-510.	5.3	20