Xin-Ming Shen

List of Publications by Year
in descending order

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60 papers	3,211 citations	172207 29 h-index	155451 55 g-index
61 all docs	61 docs citations	61 times ranked	2650 citing authors

#	Article	IF	Citations
1	Congenital myasthenic syndromes: pathogenesis, diagnosis, and treatment. Lancet Neurology, The, 2015, 14, 420-434.	4.9	413
2	Rapsyn Mutations in Humans Cause Endplate Acetylcholine-Receptor Deficiency and Myasthenic Syndrome. American Journal of Human Genetics, 2002, 70, 875-885.	2.6	221
3	Sleuthing molecular targets for neurological diseases at the neuromuscular junction. Nature Reviews Neuroscience, 2003, 4, 339-352.	4.9	212
4	Myasthenic syndrome caused by mutation of the SCN4A sodium channel. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 7377-7382.	3.3	176
5	Congenital Myasthenic Syndromes due to Heteroallelic Nonsense/Missense Mutations in the Acetylcholine Receptor Â Subunit Gene: Identification and Functional Characterization of Six New Mutations. Human Molecular Genetics, 1997, 6, 753-766.	1.4	164
6	The spectrum of mutations causing end-plate acetylcholinesterase deficiency. Annals of Neurology, 2000, 47, 162-170.	2.8	123
7	Acetylcholine receptor M3 domain: stereochemical and volume contributions to channel gating. Nature Neuroscience, 1999, 2, 226-233.	7.1	119
8	Dokâ€7 myasthenia: Phenotypic and molecular genetic studies in 16 patients. Annals of Neurology, 2008, 64, 71-87.	2.8	117
9	Mode Switching Kinetics Produced by a Naturally Occurring Mutation in the Cytoplasmic Loop of the Human Acetylcholine Receptor Îµ Subunit. Neuron, 1998, 20, 575-588.	3.8	109
10	Myopathy, Myasthenic Syndrome, and Epidermolysis Bullosa Simplex Due to Plectin Deficiency. Journal of Neuropathology and Experimental Neurology, 1999, 58, 832-846.	0.9	104
11	Mutant SNAP25B causes myasthenia, cortical hyperexcitability, ataxia, and intellectual disability. Neurology, 2014, 83, 2247-2255.	1.5	102
12	LRP4 third Î²-propeller domain mutations cause novel congenital myasthenia by compromising agrin-mediated MuSK signaling in a position-specific manner. Human Molecular Genetics, 2014, 23, 1856-1868.	1.4	96
13	What Have We Learned from the Congenital Myasthenic Syndromes. Journal of Molecular Neuroscience, 2010, 40, 143-153.	1.1	82
14	Mutation causing severe myasthenia reveals functional asymmetry of AChR signature cystine loops in agonist binding and gating. Journal of Clinical Investigation, 2003, 111, 497-505.	3.9	68
15	hnRNP H enhances skipping of a nonfunctional exon P3A in CHRNA1 and a mutation disrupting its binding causes congenital myasthenic syndrome. Human Molecular Genetics, 2008, 17, 4022-4035.	1.4	54
16	GFPT1-myasthenia. Neurology, 2013, 81, 370-378.	1.5	54
17	PREPL deficiency with or without cystinuria causes a novel myasthenic syndrome. Neurology, 2014, 82, 1254-1260.	1.5	52
18	Loss of MUNC13-1 function causes microcephaly, cortical hyperexcitability, and fatal myasthenia. Neurology: Genetics, 2016, 2, e105.	0.9	50

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19	Congenital myastheniaâ€“related AChR Î´ subunit mutation interferes with intersubunit communication essential for channel gating. Journal of Clinical Investigation, 2008, 118, 1867-1876.	3.9	50
20	Congenital Myasthenic Syndromes: Multiple Molecular Targets at the Neuromuscular Junction. Annals of the New York Academy of Sciences, 2003, 998, 138-160.	1.8	49
21	DPAGT1 myasthenia and myopathy. Neurology, 2014, 82, 1822-1830.	1.5	48
22	Urine-Derived Compound Evokes Membrane Responses in Mouse Vomeronasal Receptor Neurons. Journal of Neurophysiology, 1997, 77, 2856-2862.	0.9	47
23	Slow-channel mutation in acetylcholine receptor Î±M4 domain and its efficient knockdown. Annals of Neurology, 2006, 60, 128-136.	2.8	44
24	Functional consequences and structural interpretation of mutations of human choline acetyltransferase. Human Mutation, 2011, 32, 1259-1267.	1.1	43
25	Beneficial effect of albuterol in congenital myasthenic syndrome with epsilonâ€subunit mutations. Muscle and Nerve, 2011, 44, 289-291.	1.0	42
26	Impaired Synaptic Development, Maintenance, and Neuromuscular Transmission in LRP4-Related Myasthenia. JAMA Neurology, 2015, 72, 889.	4.5	41
27	Endplate structure and parameters of neuromuscular transmission in sporadic centronuclear myopathy associated with myasthenia. Neuromuscular Disorders, 2011, 21, 387-395.	0.3	39
28	Novel synaptobrevinâ€1 mutation causes fatal congenital myasthenic syndrome. Annals of Clinical and Translational Neurology, 2017, 4, 130-138.	1.7	38
29	Congenital myasthenic syndromes in adult neurology clinic. Neurology, 2018, 91, e1770-e1777.	1.5	36
30	A frameshifting mutation in CHRNE unmasks skipping of the preceding exon. Human Molecular Genetics, 2003, 12, 3055-3066.	1.4	29
31	Subunit-specific contribution to agonist binding and channel gating revealed by inherited mutation in muscle acetylcholine receptor M3-M4 linker. Brain, 2004, 128, 345-355.	3.7	29
32	Further Observations in Congenital Myasthenic Syndromes. Annals of the New York Academy of Sciences, 2008, 1132, 104-113.	1.8	28
33	Congenital myasthenic syndromes in Turkey: Clinical clues and prognosis with long term follow-up. Neuromuscular Disorders, 2018, 28, 315-322.	0.3	28
34	Highly fatal fast-channel syndrome caused by AChR É› subunit mutation at the agonist binding site. Neurology, 2012, 79, 449-454.	1.5	27
35	New horizons for congenital myasthenic syndromes. Annals of the New York Academy of Sciences, 2012, 1275, 54-62.	1.8	24
36	Electrophysiological and Biochemical Responses of Mouse Vorneronasal Receptor Cells to Urine-derived Compounds: Possible Mechanism of Action. Chemical Senses, 1998, 23, 483-489.	1.1	23

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37	Myasthenic syndrome AChRÎ± C-loop mutant disrupts initiation of channel gating. Journal of Clinical Investigation, 2012, 122, 2613-2621.	3.9	23
38	The Spectrum of Congenital Myasthenic Syndromes. Molecular Neurobiology, 2002, 26, 347-367.	1.9	21
39	Congenital myasthenic syndromes: A diverse array of molecular targets. Journal of Neurocytology, 2003, 32, 1017-1037.	1.6	20
40	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.	1.1	19
41	Limb girdle muscular dystrophy D3 HNRNPDL related in a Chinese family with distal muscle weakness caused by a mutation in the prion-like domain. Journal of Neurology, 2019, 266, 498-506.	1.8	19
42	Congenital myasthenic syndrome in Japan: Ethnically unique mutations in muscle nicotinic acetylcholine receptor subunits. Neuromuscular Disorders, 2015, 25, 60-69.	0.3	18
43	Atypical familial amyotrophic lateral sclerosis with initial symptoms of pain or tremor in a Chinese family harboring VAPB-P56S mutation. Journal of Neurology, 2016, 263, 263-268.	1.8	18
44	Mechanistic Diversity Underlying Fast Channel Congenital Myasthenic Syndromes. Annals of the New York Academy of Sciences, 2003, 998, 128-137.	1.8	13
45	A homozygous mutation in GMPPB leads to centronuclear myopathy with combined pre- and postsynaptic defects of neuromuscular transmission. Neuromuscular Disorders, 2019, 29, 614-617.	0.3	11
46	Investigation of Congenital Myasthenia Reveals Functional Asymmetry of Invariant Acetylcholine Receptor (AChR) Cys-loop Aspartates. Journal of Biological Chemistry, 2016, 291, 3291-3301.	1.6	10
47	Monoclonal gammopathy with both nemaline myopathy and amyloid myopathy. Neuromuscular Disorders, 2017, 27, 942-946.	0.3	10
48	Mutations causing congenital myasthenia reveal principal coupling pathway in the acetylcholine receptor Îµ-subunit. JCI Insight, 2018, 3, .	2.3	8
49	The unfolding landscape of the congenital myasthenic syndromes. Annals of the New York Academy of Sciences, 2018, 1413, 25-34.	1.8	7
50	Slowâ€channel myasthenia due to novel mutation in M2 domain of AChR delta subunit. Annals of Clinical and Translational Neurology, 2019, 6, 2066-2078.	1.7	7
51	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.	1.8	6
52	Determinants of the repetitive-CMAP occurrence and therapy efficacy in slow-channel myasthenia. Neurology, 2020, 95, e2781-e2793.	1.5	5
53	A novel fast-channel myasthenia caused by mutation in Î² subunit of AChR reveals subunit-specific contribution of the intracellular M1-M2 linker to channel gating. Experimental Neurology, 2020, 331, 113375.	2.0	5
54	Presence of antibodies against low-density lipoprotein receptor-related protein 4 and impairment of neuromuscular junction in a Chinese cohort of amyotrophic lateral sclerosis. Chinese Medical Journal, 2019, 132, 1487-1489.	0.9	4

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55	Missense Mutations of Codon 116 in the SOD1 Gene Cause Rapid Progressive Familial ALS and Predict Short Viability With PMA Phenotype. Frontiers in Genetics, 2021, 12, 776831.	1.1	2
56	LRP4 myasthenia: Investigation of a second kinship reveals impaired development and maintenance of the neuromuscular junction. Neuromuscular Disorders, 2015, 25, S186-S187.	0.3	1
57	A unique case of dysferlinopathy with a large-segment duplication mutation who experienced rapid deterioration after small-dosage corticosteroid treatment. Journal of the Neurological Sciences, 2017, 376, 35-37.	0.3	1
58	CYP3A5*3 polymorphism and age affect tacrolimus blood trough concentration in myasthenia gravis patients. Journal of Neuroimmunology, 2021, 355, 577571.	1.1	1
59	Charcotâ€“Marieâ€“Tooth Disease With Episodic Rhabdomyolysis Due to Two Novel Mutations in the Î² Subunit of Mitochondrial Trifunctional Protein and Effective Response to Modified Diet Therapy. Frontiers in Neurology, 2021, 12, 694966.	1.1	1
60	G.P.11.11 Myasthenic syndrome due to defects in rapsyn: Clinical and molecular findings in 39 patients. Neuromuscular Disorders, 2009, 19, 626-627.	0.3	0