Payam Mohassel

List of Publications by Year
in descending order

42 papers	1,572 citations	361045 20 h-index	315357 38 g-index
48 all docs	48 docs citations	48 times ranked	3213 citing authors

#	Article	IF	Citations
1	Cross-sectional Neuromuscular Phenotyping Study of Patients With Arhinia With SMCHD1 Variants. Neurology, 2022, 98, .	1.5	3
2	Heterozygous frameshift variants in HNRNPA2B1 cause early-onset oculopharyngeal muscular dystrophy. Nature Communications, 2022, 13, 2306.	5.8	20
3	New Insights into the Neuromyogenic Spectrum of a Gain of Function Mutation in SPTLC1. Genes, 2022, 13, 893.	1.0	2
4	Slowly Progressive Limb-Girdle Weakness and HyperCKemia â€“ Limb Girdle Muscular Dystrophy or Anti-3-Hydroxy-3-Methylglutaryl-CoA-Reductase-Myopathy?. Journal of Neuromuscular Diseases, 2022, , 1-8.	1.1	2
5	A form of muscular dystrophy associated with pathogenic variants in JAG2. American Journal of Human Genetics, 2021, 108, 840-856.	2.6	15
6	Pathogenic variants in TNNC2 cause congenital myopathy due to an impaired force response to calcium. Journal of Clinical Investigation, 2021, 131, .	3.9	11
7	Childhood amyotrophic lateral sclerosis caused by excess sphingolipid synthesis. Nature Medicine, 2021, 27, 1197-1204.	15.2	96
8	Transcriptome analysis of collagen VIâ€related muscular dystrophy muscle biopsies. Annals of Clinical and Translational Neurology, 2021, 8, 2184-2198.	1.7	10
9	Biallelic loss of function variants in <scp>SYT2</scp> cause a treatable congenital onset presynaptic myasthenic syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2272-2283.	0.7	20
10	Collagen VIÎ±2 chain deficiency causes trabecular bone loss by potentially promoting osteoclast differentiation through enhanced TNFÎ± signaling. Scientific Reports, 2020, 10, 13749.	1.6	13
11	Cardiac MRI identifies valvular and myocardial disease in a subset of ANO5-related muscular dystrophy patients. Neuromuscular Disorders, 2020, 30, 742-749.	0.3	4
12	Hypoglycemia in patients with congenital muscle disease. BMC Pediatrics, 2020, 20, 57.	0.7	7
13	SPTLC1 Mutations Associated with Early Onset Amyotrophic Lateral Sclerosis. FASEB Journal, 2020, 34, 1-1.	0.2	2
14	Genetic regulatory variation in populations informs transcriptome analysis in rare disease. Science, 2019, 366, 351-356.	6.0	99
15	Dominant collagen XII mutations cause a distal myopathy. Annals of Clinical and Translational Neurology, 2019, 6, 1980-1988.	1.7	24
16	Longitudinal changes in clinical outcome measures in COL6-related dystrophies and LAMA2-related dystrophies. Neurology, 2019, 93, e1932-e1943.	1.5	23
17	MSTO1 mutations cause mtDNA depletion, manifesting as muscular dystrophy with cerebellar involvement. Acta Neuropathologica, 2019, 138, 1013-1031.	3.9	31
18	Identification of a Novel Deep Intronic Mutation in CAPN3 Presenting a Promising Target for Therapeutic Splice Modulation. Journal of Neuromuscular Diseases, 2019, 6, 475-483.	1.1	6

#	Article	IF	Citations
19	Anti-HMGCR myopathy may resemble limb-girdle muscular dystrophy. Neurology: Neuroimmunology and NeuroInflammation, 2019, 6, e523.	3.1	66
20	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	2.8	93
21	Anti-HMGCR Myopathy. Journal of Neuromuscular Diseases, 2018, 5, 11-20.	1.1	101
22	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	3.5	86
23	Extracellular matrix-driven congenital muscular dystrophies. Matrix Biology, 2018, 71-72, 188-204.	1.5	44
24	Antiâ€“3â€hydroxyâ€3â€methylglutarylâ€coenzyme a reductase necrotizing myopathy masquerading as a muscular dystrophy in a child. Muscle and Nerve, 2017, 56, 1177-1181.	1.0	30
25	De novo missense variants inHECW2are associated with neurodevelopmental delay and hypotonia. Journal of Medical Genetics, 2017, 54, 84-86.	1.5	46
26	Common and variable clinical, histological, and imaging findings of recessive RYR1-related centronuclear myopathy patients. Neuromuscular Disorders, 2017, 27, 975-985.	0.3	34
27	HSP and deafness. Neurology: Genetics, 2017, 3, e151.	0.9	5
28	Cytoplasmic body pathology in severe ACTA1 -related myopathy in the absence of typical nemaline rods. Neuromuscular Disorders, 2017, 27, 531-536.	0.3	15
29	P4HA1 mutations cause a unique congenital disorder of connective tissue involving tendon, bone, muscle and the eye. Human Molecular Genetics, 2017, 26, 2207-2217.	1.4	37
30	A defect in myoblast fusion underlies Carey-Fineman-Ziter syndrome. Nature Communications, 2017, 8, 16077.	5.8	72
31	Congenital, Limb Girdle and Other Muscular Dystrophies. , 2017, , 1112-1122.		1
32	Novel De Novo Mutations in KIF1A as a Cause of Hereditary Spastic Paraplegia With Progressive Central Nervous System Involvement. Journal of Child Neurology, 2016, 31, 1114-1119.	0.7	44
33	Development of a subset of forelimb muscles and their attachment sites requires the ulnar-mammary syndrome geneTbx3. DMM Disease Models and Mechanisms, 2016, 9, 1257-1269.	1.2	38
34	Development of a subset of forelimb muscles and their attachment sites requires the ulnar-mammary syndrome gene Tbx3. Development (Cambridge), 2016, 143, e1.1-e1.1.	1.2	2
35	Neurophysiology Simplified for Imagers. Seminars in Musculoskeletal Radiology, 2015, 19, 112-120.	0.4	2
36	Expression of the Dermatomyositis Autoantigen Transcription Intermediary Factor 1Î³ in Regenerating Muscle. Arthritis and Rheumatology, 2015, 67, 266-272.	2.9	42

#	Article	IF	Citations
37	Limb-girdle Muscular Dystrophies. , 2015, , 635-666.		1
38	Statinâ€associated autoimmune myopathy and antiâ€HMGCR autoantibodies. Muscle and Nerve, 2013, 48, 477-483.	1.0	115
39	Anterior spinal artery syndrome presenting as cervical myelopathy in a patient with subclavian steal syndrome. Neurology: Clinical Practice, 2013, 3, 358-360.	0.8	4
40	The spectrum of statin myopathy. Current Opinion in Rheumatology, 2013, 25, 747-752.	2.0	57
41	Fish consumption, long-chain omega-3 fatty acids and risk of cognitive decline or Alzheimer disease: a complex association. Nature Reviews Neurology, 2009, 5, 140-152.	4.9	240
42	Attribution of original contribution in large datasets in the era of multiâ€omic studies. Annals of Clinical and Translational Neurology, 0, , .	1.7	0