Peter Hackman

List of Publications by Year
in descending order

33 papers	1,823 citations	394421 19 h-index	377865 34 g-index
34 all docs	34 docs citations	34 times ranked	4626 citing authors

#	Article	IF	Citations
1	Congenital asymmetric distal myopathy with hemifacial weakness caused by a heterozygous large de novo mosaic deletion in nebulin. Neuromuscular Disorders, 2021, 31, 539-545.	0.6	9
2	Out-of-Frame Mutations in ACTN2 Last Exon Cause a Dominant Distal Myopathy With Facial Weakness. Neurology: Genetics, 2021, 7, e619.	1.9	7
3	Dominant Distal Myopathy 3 (MPD3) Caused by a Deletion in the HNRNPA1 Gene. Neurology: Genetics, 2021, 7, e632.	1.9	7
4	Panorama of the distal myopathies. Acta Myologica, 2020, 39, 245-265.	1.5	22
5	Expanding the importance of HMERF titinopathy: new mutations and clinical aspects. Journal of Neurology, 2019, 266, 680-690.	3.6	31
6	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	5.3	93
7	The complexity of titin splicing pattern in human adult skeletal muscles. Skeletal Muscle, 2018, 8, 11.	4.2	65
8	TIA1 variant drives myodegeneration in multisystem proteinopathy with SQSTM1 mutations. Journal of Clinical Investigation, 2018, 128, 1164-1177.	8.2	75
9	219th ENMC International Workshop Titinopathies International database of titin mutations and phenotypes, Heemskerk, The Netherlands, 29 Aprilâ€“1 May 2016. Neuromuscular Disorders, 2017, 27, 396-407.	0.6	29
10	A â€˜second truncationâ€™ in TTN causes early onset recessive muscular dystrophy. Neuromuscular Disorders, 2017, 27, 1009-1017.	0.6	18
11	Copy number variation analysis increases the diagnostic yield in muscle diseases. Neurology: Genetics, 2017, 3, e204.	1.9	17
12	SIRT6 polymorphism rs117385980 is associated with longevity and healthy aging in Finnish men. BMC Medical Genetics, 2017, 18, 41.	2.1	21
13	Targeted Next-Generation Sequencing Reveals Novel TTN Mutations Causing Recessive Distal Titinopathy. Molecular Neurobiology, 2017, 54, 7212-7223.	4.0	38
14	A novel FLNC frameshift and an OBSCN variant in a family with distal muscular dystrophy. PLoS ONE, 2017, 12, e0186642.	2.5	29
15	Increasing Role of Titin Mutations in Neuromuscular Disorders. Journal of Neuromuscular Diseases, 2016, 3, 293-308.	2.6	120
16	Homozygosity of the Dominant Myotilin c.179C>T (p.Ser60Phe) Mutation Causes a More Severe and Proximal Muscular Dystrophy. Journal of Neuromuscular Diseases, 2016, 3, 275-281.	2.6	9
17	Mutations in HSPB8 causing a new phenotype of distal myopathy and motor neuropathy. Neurology, 2016, 86, 391-398.	1.1	107
18	Targeted next-generation sequencing assay for detection of mutations in primary myopathies. Neuromuscular Disorders, 2016, 26, 7-15.	0.6	87

#	Article	IF	Citations
19	Re-evaluation of the phenotype caused by the commonMATR3p.Ser85Cys mutation in a new family. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 448-450.	1.9	24
20	Adult onset limb-girdle muscular dystrophy â€" A recessive titinopathy masquerading as myositis. Journal of the Neurological Sciences, 2015, 351, 120-123.	0.6	12
21	SQSTM1 splice site mutation in distal myopathy with rimmed vacuoles. Neurology, 2015, 85, 665-674.	1.1	74
22	Novel mutations in DNAJB6 gene cause a very severe early-onset limb-girdle muscular dystrophy 1D disease. Neuromuscular Disorders, 2015, 25, 835-842.	0.6	35
23	Differential Isoform Expression and Selective Muscle Involvement in Muscular Dystrophies. American Journal of Pathology, 2015, 185, 2833-2842.	3.8	12
24	Isolated semitendinosus involvement in the initial stages of limb-girdle muscular dystrophy 2L. Neuromuscular Disorders, 2014, 24, 1118-1119.	0.6	9
25	Mutation screening of SEMA3A and SEMA7A in patients with congenital hypogonadotropic hypogonadism. Pediatric Research, 2014, 75, 641-644.	2.3	64
26	Abnormal Splicing of NEDD4 in Myotonic Dystrophy Type 2. American Journal of Pathology, 2014, 184, 2322-2332.	3.8	16
27	Gene Expression Profiling in Tibial Muscular Dystrophy Reveals Unfolded Protein Response and Altered Autophagy. PLoS ONE, 2014, 9, e90819.	2.5	16
28	Welander distal myopathy is caused by a mutation in the RNAâ€binding protein TIA1. Annals of Neurology, 2013, 73, 500-509.	5.3	118
29	Four new Finnish families with LGMD1D; refinement of the clinical phenotype and the linked 7q36 locus. Neuromuscular Disorders, 2011, 21, 338-344.	0.6	22
30	Mutations in the N-terminal Actin-Binding Domain of Filamin C Cause a Distal Myopathy. American Journal of Human Genetics, 2011, 88, 729-740.	6.2	124
31	Truncating mutations in C-terminal titin may cause more severe tibial muscular dystrophy (TMD). Neuromuscular Disorders, 2008, 18, 922-928.	0.6	87
32	Linkage to two separate loci in a family with a novel distal myopathy phenotype (MPD3). Neuromuscular Disorders, 2004, 14, 183-187.	0.6	17
33	Tibial Muscular Dystrophy Is a Titinopathy Caused by Mutations in TTN, the Gene Encoding the Giant Skeletal-Muscle Protein Titin. American Journal of Human Genetics, 2002, 71, 492-500.	6.2	408