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	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
2	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
3	Increasing Role of Titin Mutations in Neuromuscular Disorders. Journal of Neuromuscular Diseases, 2016, 3, 293-308.	2.6	120
4	Welander distal myopathy is caused by a mutation in the RNAâ€binding protein TIA1. Annals of Neurology, 2013, 73, 500-509.	5.3	118
5	Mutations in HSPB8 causing a new phenotype of distal myopathy and motor neuropathy. Neurology, 2016, 86, 391-398.	1.1	107
6	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	5.3	93
7	Truncating mutations in C-terminal titin may cause more severe tibial muscular dystrophy (TMD). Neuromuscular Disorders, 2008, 18, 922-928.	0.6	87
8	Targeted next-generation sequencing assay for detection of mutations in primary myopathies. Neuromuscular Disorders, 2016, 26, 7-15.	0.6	87
9	TIA1 variant drives myodegeneration in multisystem proteinopathy with SQSTM1 mutations. Journal of Clinical Investigation, 2018, 128, 1164-1177.	8.2	75
10	SQSTM1 splice site mutation in distal myopathy with rimmed vacuoles. Neurology, 2015, 85, 665-674.	1.1	74
11	The complexity of titin splicing pattern in human adult skeletal muscles. Skeletal Muscle, 2018, 8, 11.	4.2	65
12	Mutation screening of SEMA3A and SEMA7A in patients with congenital hypogonadotropic hypogonadism. Pediatric Research, 2014, 75, 641-644.	2.3	64
13	Targeted Next-Generation Sequencing Reveals Novel TTN Mutations Causing Recessive Distal Titinopathy. Molecular Neurobiology, 2017, 54, 7212-7223.	4.0	38
14	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
15	Expanding the importance of HMERF titinopathy: new mutations and clinical aspects. Journal of Neurology, 2019, 266, 680-690.	3.6	31
16	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
17	A novel FLNC frameshift and an OBSCN variant in a family with distal muscular dystrophy. PLoS ONE, 2017, 12, e0186642.	2.5	29
18	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

#	Article	IF	Citations
19	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
20	Panorama of the distal myopathies. Acta Myologica, 2020, 39, 245-265.	1.5	22
21	SIRT6 polymorphism rs117385980 is associated with longevity and healthy aging in Finnish men. BMC Medical Genetics, 2017, 18, 41.	2.1	21
22	A â€˜second truncationâ€™ in TTN causes early onset recessive muscular dystrophy. Neuromuscular Disorders, 2017, 27, 1009-1017.	0.6	18
23	Linkage to two separate loci in a family with a novel distal myopathy phenotype (MPD3). Neuromuscular Disorders, 2004, 14, 183-187.	0.6	17
24	Copy number variation analysis increases the diagnostic yield in muscle diseases. Neurology: Genetics, 2017, 3, e204.	1.9	17
25	Abnormal Splicing of NEDD4 in Myotonic Dystrophy Type 2. American Journal of Pathology, 2014, 184, 2322-2332.	3.8	16
26	Gene Expression Profiling in Tibial Muscular Dystrophy Reveals Unfolded Protein Response and Altered Autophagy. PLoS ONE, 2014, 9, e90819.	2.5	16
27	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
28	Differential Isoform Expression and Selective Muscle Involvement in Muscular Dystrophies. American Journal of Pathology, 2015, 185, 2833-2842.	3.8	12
29	Isolated semitendinosus involvement in the initial stages of limb-girdle muscular dystrophy 2L. Neuromuscular Disorders, 2014, 24, 1118-1119.	0.6	9
30	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
31	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
32	Out-of-Frame Mutations in ACTN2 Last Exon Cause a Dominant Distal Myopathy With Facial Weakness. Neurology: Genetics, 2021, 7, e619.	1.9	7
33	Dominant Distal Myopathy 3 (MPD3) Caused by a Deletion in the HNRNPA1 Gene. Neurology: Genetics, 2021, 7, e632.	1.9	7