Perry B Shieh

List of Publications by Year
in descending order

50 papers	4,248 citations	236925 25 h-index	197818 49 g-index
50 all docs	50 docs citations	50 times ranked	6640 citing authors

#	Article	IF	Citations
1	Nusinersen versus Sham Control in Later-Onset Spinal Muscular Atrophy. New England Journal of Medicine, 2018, 378, 625-635.	27.0	977
2	Clinical Exome Sequencing for Genetic Identification of Rare Mendelian Disorders. JAMA - Journal of the American Medical Association, 2014, 312, 1880.	7.4	842
3	Ataluren in patients with nonsense mutation Duchenne muscular dystrophy (ACT DMD): a multicentre, randomised, double-blind, placebo-controlled, phase 3 trial. Lancet, The, 2017, 390, 1489-1498.	13.7	365
4	Natural history of infantileâ€onset spinal muscular atrophy. Annals of Neurology, 2017, 82, 883-891.	5.3	276
5	Onasemnogene abeparvovec gene therapy for symptomatic infantile-onset spinal muscular atrophy in patients with two copies of SMN2 (STR1VE): an open-label, single-arm, multicentre, phase 3 trial. Lancet Neurology, The, 2021, 20, 284-293.	10.2	227
6	Recessive truncating titin gene, TTN , mutations presenting as centronuclear myopathy. Neurology, 2013, 81, 1205-1214.	1.1	177
7	Review of the Diagnosis and Treatment of Periodic Paralysis. Muscle and Nerve, 2018, 57, 522-530.	2.2	157
8	Diagnostic utility of transcriptome sequencing for rare Mendelian diseases. Genetics in Medicine, 2020, 22, 490-499.	2.4	136
9	Baseline results of the Neuro<scp>NEXT</scp> spinal muscular atrophy infant biomarker study. Annals of Clinical and Translational Neurology, 2016, 3, 132-145.	3.7	106
10	Safety, tolerability, and pharmacokinetics of casimersen in patients with <scp>D</scp>uchenne muscular dystrophy amenable to exon <scp>45</scp> skipping: A randomized, doubleâ€blind, placeboâ€controlled, doseâ€titration trial. Muscle and Nerve, 2021, 64, 285-292.	2.2	83
11	Emerging Strategies in the Treatment of Duchenne Muscular Dystrophy. Neurotherapeutics, 2018, 15, 840-848.	4.4	73
12	Muscular Dystrophies and Other Genetic Myopathies. Neurologic Clinics, 2013, 31, 1009-1029.	1.8	66
13	Safety and efficacy of nusinersen in spinal muscular atrophy: The <scp>EMBRACE</scp> study. Muscle and Nerve, 2021, 63, 668-677.	2.2	56
14	The care of patients with Duchenne, Becker, and other muscular dystrophies in the <scp>COVID</scp>â€19 pandemic. Muscle and Nerve, 2020, 62, 41-45.	2.2	54
15	Randomized, placebo-controlled trials of dichlorphenamide in periodic paralysis. Neurology, 2016, 86, 1408-1416.	1.1	53
16	Amifampridine phosphate (Firdapse^{Â®}) is effective and safe in a phase 3 clinical trial in LEMS. Muscle and Nerve, 2016, 53, 717-725.	2.2	51
17	Open-Label Evaluation of Eteplirsen in Patients with Duchenne Muscular Dystrophy Amenable to Exon 51 Skipping: PROMOVI Trial. Journal of Neuromuscular Diseases, 2021, 8, 989-1001.	2.6	50
18	Effect of Different Corticosteroid Dosing Regimens on Clinical Outcomes in Boys With Duchenne Muscular Dystrophy. JAMA - Journal of the American Medical Association, 2022, 327, 1456.	7.4	43

#	Article	IF	Citations
19	Deflazacort versus prednisone/prednisolone for maintaining motor function and delaying loss of ambulation: A post HOC analysis from the ACT DMD trial. Muscle and Nerve, 2018, 58, 639-645.	2.2	42
20	Rare variants in SQSTM1 and VCP genes and risk of sporadic inclusion body myositis. Neurobiology of Aging, 2016, 47, 218.e1-218.e9.	3.1	40
21	Deflazacort vs prednisone treatment for Duchenne muscular dystrophy: A metaâ€analysis of disease progression rates in recent multicenter clinical trials. Muscle and Nerve, 2020, 61, 26-35.	2.2	40
22	Limb-girdle muscular dystrophy 2H and the role of TRIM32. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2011, 101, 125-133.	1.8	35
23	Duchenne muscular dystrophy. Current Opinion in Neurology, 2015, 28, 542-546.	3.6	28
24	Clinical trial readiness to solve barriers to drug development in FSHD (ReSolve): protocol of a large, international, multi-center prospective study. BMC Neurology, 2019, 19, 224.	1.8	28
25	Congenital Myasthenic Syndromes. Neurologic Clinics, 2018, 36, 367-378.	1.8	27
26	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
27	Myasthenic congenital myopathy from recessive mutations at a single residue in Na _V 1.4. Neurology, 2019, 92, e1405-e1415.	1.1	24
28	Amifampridine Phosphate (Firdapse) Is Effective in a Confirmatory Phase 3 Clinical Trial in LEMS. Journal of Clinical Neuromuscular Disease, 2019, 20, 111-119.	0.7	17
29	RESTORE: A Prospective Multinational Registry of Patients with Genetically Confirmed Spinal Muscular Atrophy - Rationale and Study Design. Journal of Neuromuscular Diseases, 2020, 7, 145-152.	2.6	17
30	The effects of an intronic polymorphism in TOMM40 and APOE genotypes in sporadic inclusion body myositis. Neurobiology of Aging, 2015, 36, 1766.e1-1766.e3.	3.1	16
31	Genotypeâ€“phenotype correlations and novel molecular insights into the DHX30-associated neurodevelopmental disorders. Genome Medicine, 2021, 13, 90.	8.2	16
32	Metabolic Myopathies. Seminars in Neurology, 2015, 35, 385-397.	1.4	14
33	Incorporating Spinal Muscular Atrophy Analysis by Next-Generation Sequencing into a Comprehensive Multigene Panel for Neuromuscular Disorders. Genetic Testing and Molecular Biomarkers, 2020, 24, 616-624.	0.7	12
34	Recruitment & retention program for the NeuroNEXT SMA Biomarker Study: Super Babies for SMA!. Contemporary Clinical Trials Communications, 2018, 11, 113-119.	1.1	11
35	Congenital myasthenic syndrome caused by a frameshift insertion mutation in GFPT1. Neurology: Genetics, 2020, 6, e468.	1.9	8
36	Randomized phase 2 study of <scp>ACE</scp>â€083, a <scp>muscleâ€promoting</scp> agent, in facioscapulohumeral muscular dystrophy. Muscle and Nerve, 2022, 66, 50-62.	2.2	8

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37	Large in-frame 5â€² deletions in DMD associated with mild Duchenne muscular dystrophy: Two case reports and a review of the literature. Neuromuscular Disorders, 2019, 29, 863-873.	0.6	6
38	Myopathy associated with homozygous PYROXD1 pathogenic variants detected by genome sequencing. Neuropathology, 2020, 40, 302-307.	1.2	6
39	Meta-analyses of deflazacort versus prednisone/prednisolone in patients with nonsense mutation Duchenne muscular dystrophy. Journal of Comparative Effectiveness Research, 2021, 10, 1337-1347.	1.4	6
40	Hearing and Vision Loss in an Older Man. JAMA Neurology, 2018, 75, 1439.	9.0	5
41	Longâ€term efficacy and safety of dichlorphenamide for treatment of primary periodic paralysis. Muscle and Nerve, 2021, 64, 342-346.	2.2	5
42	Action Potential-Evoked Calcium Release Is Impaired in Single Skeletal Muscle Fibers from Heart Failure Patients. PLoS ONE, 2014, 9, e109309.	2.5	4
43	Reverse fiber type disproportion: A distinct metabolic myopathy. Muscle and Nerve, 2016, 54, 86-93.	2.2	3
44	Exercise responses in patients with chronically high creatine kinase levels. Muscle and Nerve, 2017, 56, 264-270.	2.2	3
45	066â€…Avxs-101 gene-replacement therapy (GRT) for spinal muscular atrophy type 1 (SMA1): pivotal phase 3 study (STR1VE) update. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, A22.1-A22.	1.9	3
46	Myelopathy in a patient with leukodystrophy due to CSF1R mutation. Neurology: Genetics, 2019, 5, e376.	1.9	3
47	Advances in the Genetic Testing of Neuromuscular Diseases. Neurologic Clinics, 2020, 38, 519-528.	1.8	3
48	Secondary membranous nephropathy in a patient with myasthenia gravis without thymic disease, and partial remission induced by adrenocorticotropic hormone therapy. SAGE Open Medical Case Reports, 2019, 7, 2050313X1986976.	0.3	1
49	Quantitative Muscle Analysis in Facioscapulohumeral Muscular Dystrophy Using <scp>Wholeâ€Body Fatâ€Referenced MRI</scp> : Protocol Development, Multicenter Feasibility, and Repeatability. Muscle and Nerve, 2022, , .	2.2	1
50	Facial diplegia with paresthesia associated with anti-GD1a antibodies. Baylor University Medical Center Proceedings, 2022, 35, 1-2.	0.5	0