Nicholas E Johnson

List of Publications by Year
in descending order

66	1,648	279798 23	315739 38
papers	citations	h-index	g-index

68	68	68	1711
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Patient-reported impact of symptoms in myotonic dystrophy type 1 (PRISM-1). Neurology, 2012, 79, 348-357.	1.1	164
2	Review of the Diagnosis and Treatment of Periodic Paralysis. Muscle and Nerve, 2018, 57, 522-530.	2.2	157
3	Anti-NMDA receptor encephalitis causing prolonged nonconvulsive status epilepticus. Neurology, 2010, 75, 1480-1482.	1.1	125
4	Quality-of-life in Charcotâ€“Marieâ€“Tooth disease: The patientâ€™s perspective. Neuromuscular Disorders, 2014, 24, 1018-1023.	0.6	71
5	Population-Based Prevalence of Myotonic Dystrophy Type 1 Using Genetic Analysis of Statewide Blood Screening Program. Neurology, 2021, 96, e1045-e1053.	1.1	66
6	p53 convergently activates Dux/DUX4 in embryonic stem cells and in facioscapulohumeral muscular dystrophy cell models. Nature Genetics, 2021, 53, 1207-1220.	21.4	59
7	Estimating prevalence for limb-girdle muscular dystrophy based on public sequencing databases. Genetics in Medicine, 2019, 21, 2512-2520.	2.4	56
8	Patient Reported Impact of Symptoms in Spinal Muscular Atrophy (PRISM-SMA). Neurology, 2018, 91, e1206-e1214.	1.1	55
9	Myotonic Dystrophy Health Index: Initial evaluation of a diseaseâ€specific outcome measure. Muscle and Nerve, 2014, 49, 906-914.	2.2	53
10	Patientâ€identified disease burden in facioscapulohumeral muscular dystrophy. Muscle and Nerve, 2012, 46, 948-950.	2.2	50
11	Patient-Reported Impact of Symptoms in Myotonic Dystrophy Type 2 (PRISM-2). Neurology, 2015, 85, 2136-2146.	1.1	44
12	Patient-Reported Symptoms in Facioscapulohumeral Muscular Dystrophy (PRISM-FSHD). Neurology, 2019, 93, e1180-e1192.	1.1	43
13	Parentâ€reported multiâ€national study of the impact of congenital and childhood onset myotonic dystrophy. Developmental Medicine and Child Neurology, 2016, 58, 698-705.	2.1	41
14	Myotonic Dystrophy: From Bench to Bedside. Seminars in Neurology, 2012, 32, 246-254.	1.4	39
15	Myotonic dystrophy health index: Correlations with clinical tests and patient function. Muscle and Nerve, 2016, 53, 183-190.	2.2	38
16	The Impact of Pregnancy on Myotonic Dystrophy: A Registry-Based Study. Journal of Neuromuscular Diseases, 2015, 2, 447-452.	2.6	33
17	Disease burden and functional outcomes in congenital myotonic dystrophy. Neurology, 2016, 87, 160-167.	1.1	33
18	Consensus-based care recommendations for congenital and childhood-onset myotonic dystrophy type 1. Neurology: Clinical Practice, 2019, 9, 443-454.	1.6	32

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19	Education Research: Neurology training reassessed. Neurology, 2012, 79, 1831-1834.	1.1	28
20	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
21	The Impact of Congenital and Childhood Myotonic Dystrophy on Quality of Life. Journal of Child Neurology, 2014, 29, 983-986.	1.4	27
22	Mexiletine in Myotonic Dystrophy Type 1. Neurology, 2021, 96, e228-e240.	1.1	27
23	A population-based survey of risk for cancer in individuals diagnosed with myotonic dystrophy. Muscle and Nerve, 2016, 54, 783-785.	2.2	25
24	Effects of weakness of orofacial muscles on swallowing and communication in FSHD. Neurology, 2019, 92, e957-e963.	1.1	25
25	The <scp>C</scp>harcotâ€“<scp>M</scp>arieâ€“<scp>T</scp>ooth <scp>H</scp>ealth <scp>I</scp>ndex: Evaluation of a Patientâ€Reported Outcome. Annals of Neurology, 2018, 84, 225-233.	5.3	24
26	Relative risks for comorbidities associated with myotonic dystrophy: A populationâ€based analysis. Muscle and Nerve, 2015, 52, 659-661.	2.2	23
27	Repeat-associated RNA structure and aberrant splicing. Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms, 2019, 1862, 194405.	1.9	23
28	Visualization of the diaphragm muscle with ultrasound improves diagnostic accuracy of phrenic nerve conduction studies. Muscle and Nerve, 2014, 49, 669-675.	2.2	21
29	Orofacial strength, dysarthria, and dysphagia in congenital myotonic dystrophy. Muscle and Nerve, 2018, 58, 413-417.	2.2	20
30	Prospective study of muscle cramps in Charcotâ€Marieâ€Tooth disease. Muscle and Nerve, 2015, 51, 485-488.	2.2	18
31	The Spinal Muscular Atrophy Health Index: A novel outcome for measuring how a patient feels and functions. Muscle and Nerve, 2021, 63, 837-844.	2.2	16
32	Patient Identification of the Symptomatic Impact of Charcotâ€“Marieâ€“Tooth Disease Type 1A. Journal of Clinical Neuromuscular Disease, 2013, 15, 19-23.	0.7	15
33	Limbâ€girdle muscular dystrophy: A perspective from adult patients on what matters most. Muscle and Nerve, 2019, 60, 419-424.	2.2	15
34	Physical function and mobility in children with congenital myotonic dystrophy. Muscle and Nerve, 2017, 56, 224-229.	2.2	14
35	Disease course and therapeutic approach in dermatomyositis: A four-center retrospective study of 100 patients. Neuromuscular Disorders, 2015, 25, 625-631.	0.6	12
36	Phenotypic diversity in an international Cure VCP Disease registry. Orphanet Journal of Rare Diseases, 2020, 15, 267.	2.7	11

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37	Myotonic dystrophy patient preferences in patientâ€reported outcome measures. Muscle and Nerve, 2018, 58, 49-55.	2.2	10
38	What Matters Most: A Perspective From Adult Spinal Muscular Atrophy Patients. Journal of Neuromuscular Diseases, 2016, 3, 425-429.	2.6	9
39	Association of genetic mutations and loss of ambulation in childhoodâ€onset dystrophinopathy. Muscle and Nerve, 2021, 63, 181-191.	2.2	9
40	Whole-exome sequencing in neurologic practice. Neurology: Genetics, 2015, 1, e37.	1.9	8
41	Patientâ€reported study of the impact of pediatricâ€onset myotonic dystrophy. Muscle and Nerve, 2019, 60, 392-399.	2.2	8
42	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
43	FSHD1 or FSHD2: That is the question. Neurology, 2019, 92, 881-882.	1.1	6
44	A patientâ€focused survey to assess the effects of the <scp>COVID</scp>â€19 pandemic and social guidelines on people with muscular dystrophy. Muscle and Nerve, 2021, 64, 321-327.	2.2	6
45	<scp>12â€Month</scp> progression of motor and functional outcomes in congenital myotonic dystrophy. Muscle and Nerve, 2021, 63, 384-391.	2.2	5
46	Longâ€term efficacy and safety of dichlorphenamide for treatment of primary periodic paralysis. Muscle and Nerve, 2021, 64, 342-346.	2.2	5
47	Evaluation of effects of continued corticosteroid treatment on cardiac and pulmonary function in nonâ€ambulatory males with Duchenne muscular dystrophy from MD STAR net. Muscle and Nerve, 2022, , .	2.2	5
48	Gender difference in clinical conditions among hospitalized adults with myotonic dystrophy. Muscle and Nerve, 2019, 59, 348-353.	2.2	4
49	Understanding the Perseverance of the Muscular Dystrophy Community One-Year into the COVID-19 Pandemic. Journal of Neuromuscular Diseases, 2022, 9, 517-523.	2.6	4
50	Teaching Video Neuro Images : Trapezius myotonia percussion sign in myotonic dystrophy type 2. Neurology, 2013, 80, e251.	1.1	3
51	Validation of the Italian version of the <scp>Charcotâ€Marieâ€Tooth</scp> Health Index. Journal of the Peripheral Nervous System, 2020, 25, 292-296.	3.1	3
52	High throughput screening for expanded CTG repeats in myotonic dystrophy type 1 using melt curve analysis. Molecular Genetics & Genomic Medicine, 2021, 9, e1619.	1.2	3
53	Characteristics of Clinical Trial Participants with Duchenne Muscular Dystrophy: Data from the Muscular Dystrophy Surveillance, Tracking, and Research Network (MD STARnet). Children, 2021, 8, 835.	1.5	3
54	Patient reported quality of life in limb girdle muscular dystrophy. Neuromuscular Disorders, 2022, 32, 57-64.	0.6	3

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55	Patient-Centered Therapy Development for Myotonic Dystrophy: Report of the Myotonic Dystrophy Foundationâ€“Sponsored Workshop. Therapeutic Innovation and Regulatory Science, 2017, 51, 516-522.	1.6	2
56	Modified dynamic gait index and limits of stability in myotonic dystrophy type 1. Muscle and Nerve, 2018, 58, 694-699.	2.2	2
57	A Phenotypic Description of Congenital Myotonic Dystrophy using PhenoStacks. Journal of Neuromuscular Diseases, 2019, 6, 341-347.	2.6	2
58	Body composition in patients with congenital myotonic dystrophy. Muscle and Nerve, 2019, 60, 176-179.	2.2	2
59	Neurologic Therapeutics in 2035. Neurology, 2021, 97, 1121-1127.	1.1	2
60	The difficulties and importance of research in rare genetic diseases. Muscle and Nerve, 2018, 57, 520-521.	2.2	1
61	Myopathy in the York Platelet Syndrome: An Underrecognized Complication. Case Reports in Pathology, 2018, 2018, 1-3.	0.3	1
62	A Qualitative Approach to Health Related Quality-of-Life in Congenital Muscular Dystrophy. Journal of Neuromuscular Diseases, 2018, 5, 251-255.	2.6	1
63	Evolving genetic heterogeneity of facioscapulohumeral muscular dystrophy. Neurology, 2020, 94, 1011-1012.	1.1	1
64	A populationâ€based study of scoliosis among males diagnosed with a dystrophinopathy identified by the Muscular Dystrophy Surveillance, Tracking, and Research Network (<scp>MD STARnet</scp>). Muscle and Nerve, 2022, 65, 193-202.	2.2	1
65	Neurology Advocacy 2.0. JAMA Neurology, 2016, 73, 151.	9.0	0
66	Muscle at the junction. Neurology, 2019, 92, 591-592.	1.1	0