Nancy L Kuntz

List of Publications by Year
in descending order

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84 papers	7,218 citations	126708 33 h-index	85405 71 g-index
86 all docs	86 docs citations	86 times ranked	5945 citing authors

#	Article	IF	Citations
1	Efficacy and Safety of Vamorolone in Duchenne Muscular Dystrophy. JAMA Network Open, 2022, 5, e2144178.	2.8	31
2	Restoration of Nusinersen Levels Following Treatment Interruption in People With Spinal Muscular Atrophy: Simulations Based on a Population Pharmacokinetic Model. CNS Drugs, 2022, 36, 181-190.	2.7	6
3	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.	3.8	43
4	An expanded access program of risdiplam for patients with Type 1 or 2 spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2022, 9, 810-818.	1.7	18
5	INCEPTUS Natural History, Run-in Study for Gene Replacement Clinical Trial in X-Linked Myotubular Myopathy. Journal of Neuromuscular Diseases, 2022, 9, 503-516.	1.1	5
6	International Consensus Guidance for Management of Myasthenia Gravis. Neurology, 2021, 96, 114-122.	1.5	272
7	Reldesemtiv in Patients with Spinal Muscular Atrophy: a Phase 2 Hypothesis-Generating Study. Neurotherapeutics, 2021, 18, 1127-1136.	2.1	28
8	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.	4.9	227
9	Health related quality of life in young, steroid-naÃ¯ve boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2021, 31, 1161-1168.	0.3	4
10	Neuromuscular disease - Gene transfer for children. Journal of International Child Neurology Association, 2021, 1, .	0.0	0
11	Mortality and respiratory support in X-linked myotubular myopathy: a RECENSUS retrospective analysis. Archives of Disease in Childhood, 2020, 105, 332-338.	1.0	24
12	Utility and practice of electrodiagnostic testing in the pediatric population: An AANEM consensus statement. Muscle and Nerve, 2020, 61, 143-155.	1.0	14
13	Pathogenic Variants in the Myosin Chaperone UNC-45B Cause Progressive Myopathy with Eccentric Cores. American Journal of Human Genetics, 2020, 107, 1078-1095.	2.6	24
14	Combination molecular therapies for type 1 spinal muscular atrophy. Muscle and Nerve, 2020, 62, 550-554.	1.0	51
15	Difficulties classifying myasthenia gravis in the pediatric surgical literature. Journal of Pediatric Surgery, 2020, 55, 1679.	0.8	1
16	Re: â€œMoving Forward After Two Deaths in a Gene Therapy Trial of Myotubular Myopathyâ€•by Wilson and Flotte. Human Gene Therapy, 2020, 31, 787-787.	1.4	60
17	Efficacy and safety of vamorolone in Duchenne muscular dystrophy:ÂAn 18-month interim analysis of a non-randomized open-label extension study. PLoS Medicine, 2020, 17, e1003222.	3.9	41
18	The <scp>CINRG</scp> Becker Natural History Study: Baseline characteristics. Muscle and Nerve, 2020, 62, 369-376.	1.0	14

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19	Spinal muscular atrophy. Neurology, 2020, 95, 11-12.	1.5	0
20	Transforaminal Intrathecal Access for Injection of Nusinersen in Adult and Pediatric Patients with Spinal Muscular Atrophy. Journal of Pediatric Neurology, 2020, 18, 088-094.	0.0	2
21	242nd ENMC International Workshop: Diagnosis and management of juvenile myasthenia gravis Hoofddorp, the Netherlands, 1â€“3 March 2019. Neuromuscular Disorders, 2020, 30, 254-264.	0.3	12
22	Spinal muscular atrophy care in the COVIDâ€19 pandemic era. Muscle and Nerve, 2020, 62, 46-49.	1.0	31
23	Revised Recommendations for the Treatment of Infants Diagnosed with Spinal Muscular Atrophy Via Newborn Screening Who Have 4 Copies of SMN2. Journal of Neuromuscular Diseases, 2020, 7, 97-100.	1.1	89
24	Outcome Measures for COL6 and LAMA2-Related Dystrophies. Pediatric Neurology Briefs, 2020, 34, 15.	0.2	0
25	Nusinersen initiated in infants during the presymptomatic stage of spinal muscular atrophy: Interim efficacy and safety results from the Phase 2 NURTURE study. Neuromuscular Disorders, 2019, 29, 842-856.	0.3	401
26	Twiceâ€weekly glucocorticosteroids in infants and young boys with Duchenne muscular dystrophy. Muscle and Nerve, 2019, 59, 650-657.	1.0	32
27	Moderate exercise improves function and increases adiponectin in the mdx mouse model of muscular dystrophy. Scientific Reports, 2019, 9, 5770.	1.6	26
28	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.	0.9	3
29	Vamorolone trial in Duchenne muscular dystrophy shows dose-related improvement of muscle function. Neurology, 2019, 93, e1312-e1323.	1.5	64
30	Pulsed glucocorticoids enhance dystrophic muscle performance through epigenetic-metabolic reprogramming. JCI Insight, 2019, 4, .	2.3	32
31	Treatment Algorithm for Infants Diagnosed with Spinal Muscular Atrophy through Newborn Screening. Journal of Neuromuscular Diseases, 2018, 5, 145-158.	1.1	148
32	Nusinersen versus Sham Control in Later-Onset Spinal Muscular Atrophy. New England Journal of Medicine, 2018, 378, 625-635.	13.9	977
33	Long-term effects of glucocorticoids on function, quality of life, and survival in patients with Duchenne muscular dystrophy: a prospective cohort study. Lancet, The, 2018, 391, 451-461.	6.3	306
34	A multicenter, retrospective medical record review of Xâ€linked myotubular myopathy: The recensus study. Muscle and Nerve, 2018, 57, 550-560.	1.0	54
35	Autonomic Nervous System Dysregulation in Monozygous Twins With Nephropathic Cystinosis. Kidney International Reports, 2018, 3, 1489-1496.	0.4	0
36	Phase IIa trial in Duchenne muscular dystrophy shows vamorolone is a first-in-class dissociative steroidal anti-inflammatory drug. Pharmacological Research, 2018, 136, 140-150.	3.1	69

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37	A checklist for clinical trials in rare disease: obstacles and anticipatory actionsâ€"lessons learned from the FOR-DMD trial. Trials, 2018, 19, 291.	0.7	26
38	Recruitment & retention program for the NeuroNEXT SMA Biomarker Study: Super Babies for SMA!. Contemporary Clinical Trials Communications, 2018, 11, 113-119.	0.5	11
39	Novel methods of imaging and analysis for the thermoregulatory sweat test. Journal of Applied Physiology, 2018, 125, 755-762.	1.2	3
40	Developing standardized corticosteroid treatment for Duchenne muscular dystrophy. Contemporary Clinical Trials, 2017, 58, 34-39.	0.8	56
41	Nusinersen versus Sham Control in Infantile-Onset Spinal Muscular Atrophy. New England Journal of Medicine, 2017, 377, 1723-1732.	13.9	1,533
42	Natural history of infantileâ€onset spinal muscular atrophy. Annals of Neurology, 2017, 82, 883-891.	2.8	276
43	Diagnosis and Evaluation of Small Fiber Peripheral Neuropathy in Children. , 2017, , 265-280.		2
44	Peripheral Nerve Disorders in the Neonate. NeoReviews, 2016, 17, e719-e728.	0.4	2
45	Baseline results of the Neuro<scp>NEXT</scp> spinal muscular atrophy infant biomarker study. Annals of Clinical and Translational Neurology, 2016, 3, 132-145.	1.7	106
46	Association Study of Exon Variants in the NF-Î²B and TGFÎ² Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy. American Journal of Human Genetics, 2016, 99, 1163-1171.	2.6	71
47	International consensus guidance for management of myasthenia gravis. Neurology, 2016, 87, 419-425.	1.5	736
48	Delay in Diagnosis of Duchenne Muscular Dystrophy. Pediatric Neurology Briefs, 2015, 29, 5.	0.2	4
49	Hereditary Neuropathy with Liability to Pressure Palsies. Pediatric Neurology Briefs, 2015, 29, 83.	0.2	5
50	Axonal Damage in Pediatric Multiple Sclerosis. Pediatric Neurology Briefs, 2015, 29, 34.	0.2	0
51	Cerebellar Mutism in Acute Disseminating Encephalomyelitis. Pediatric Neurology, 2014, 50, 511-514.	1.0	11
52	The instability of the BTB-KELCH protein Gigaxonin causes Giant Axonal Neuropathy and constitutes a new penetrant and specific diagnostic test. Acta Neuropathologica Communications, 2014, 2, 47.	2.4	26
53	Laboratory Evaluation of Pediatric Autonomic Disorders. Seminars in Pediatric Neurology, 2013, 20, 35-43.	1.0	8
54	Quantitative MRI analysis in children with multiple sclerosis: a multicenter feasibility pilot study. BMC Neurology, 2013, 13, 173.	0.8	4

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55	Muscle specific kinase autoimmune myasthenia gravis in children: A case series. Neuromuscular Disorders, 2013, 23, 874-882.	0.3	38
56	Diagnostic Criteria for Pediatric Multiple Sclerosis. Current Neurology and Neuroscience Reports, 2013, 13, 354.	2.0	19
57	The DcpS inhibitor RG3039 improves survival, function and motor unit pathologies in two SMA mouse models. Human Molecular Genetics, 2013, 22, 4084-4101.	1.4	78
58	Spasmodic Muscle Cramps and Weakness as Presenting Symptoms in Wilson Disease. Pediatrics, 2013, 132, e1039-e1042.	1.0	12
59	Antibody response to common viruses and human leukocyte antigen-DRB1 in pediatric multiple sclerosis. Multiple Sclerosis Journal, 2013, 19, 891-895.	1.4	32
60	Unusual Case of Relapsing and Remitting Rash With Progressive Motor Sensory Neuropathy and Spinal Cord Atrophy. Journal of Child Neurology, 2012, 27, 225-228.	0.7	0
61	Pupillometry in congenital central hypoventilation syndrome (CCHS): quantitative evidence of autonomic nervous system dysregulation. Pediatric Research, 2012, 71, 280-285.	1.1	41
62	Congenital Central Hypoventilation Syndrome (CCHS) and PHOX2B Mutations. , 2012, , 445-449.		2
63	Management of Pediatric Central Nervous System Demyelinating Disorders: Consensus of United States Neurologists. Journal of Child Neurology, 2011, 26, 675-682.	0.7	85
64	Pediatric Multiple Sclerosis. Neurologic Clinics, 2011, 29, 481-505.	0.8	53
65	Neuronal Voltage-Gated Potassium Channel Complex Autoimmunity in Children. Pediatric Neurology, 2011, 44, 275-281.	1.0	56
66	Differential diagnosis of multiple sclerosis and acquired central nervous system demyelinating disorders in children and adolescents. , 2011, , 58-74.		1
67	Liquid formulation of pentoxifylline is a poorly tolerated treatment for duchenne dystrophy. Muscle and Nerve, 2011, 44, 170-173.	1.0	10
68	Multiple Sclerosis Therapies in Pediatric Patients With Refractory Multiple Sclerosis. Archives of Neurology, 2011, 68, 437.	4.9	101
69	Localized Aquadynia Responsive to Clonidine in a 13-Year-Old Girl. Pediatric Dermatology, 2010, 27, 646-649.	0.5	9
70	Orthostatic Heart Rate and Blood Pressure in Adolescents: Reference Ranges. Journal of Child Neurology, 2010, 25, 1210-1215.	0.7	28
71	Postural Orthostatic Tachycardia Syndrome: A Clinical Review. Pediatric Neurology, 2010, 42, 77-85.	1.0	104
72	Two siblings with limb-girdle muscular dystrophy type 2E responsive to deflazacort. Neuromuscular Disorders, 2010, 20, 122-124.	0.3	20

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73	Treatment of multiple sclerosis in children and adolescents. Expert Opinion on Pharmacotherapy, 2010, 11, 505-520.	0.9	24
74	Longitudinal study of intraneural perineurioma--a benign, focal hypertrophic neuropathy of youth. Brain, 2009, 132, 2265-2276.	3.7	125
75	Pediatric multiple sclerosis. Nature Reviews Neurology, 2009, 5, 621-631.	4.9	124
76	Diagnosis and treatment of peripheral nerve lesions in children. Paediatrics and Child Health (United) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	0.2	0
77	Clinical Neurophysiology of Pediatric Polyneuropathies. , 2006, , 645-686.		0
78	Clinical Neurophysiology of the Motor Unit in Infants and Children. , 2006, , 130-145.		0
79	Autonomic Testing in Childhood. , 2006, , 687-712.		0
80	Muscle Disorders in Children: Neurophysiologic Contributions to Diagnosis and Management. , 2006, , 747-762.		0
81	Antiphospholipid Syndrome With Catastrophic Bleeding and Recurrent Ischemic Strokes as Initial Presentation of Systemic Lupus Erythematosus. Journal of Pediatric Hematology/Oncology, 2005, 27, 403-407.	0.3	13
82	Successful treatment of refractory myasthenia gravis using rituximab: a pediatric case report. Journal of Pediatrics, 2003, 143, 674-677.	0.9	101
83	The electrophysiologic profile of Dejerine-Sottas disease (HMSN III). Muscle and Nerve, 1990, 13, 586-592.	1.0	40
84	Review Article: Nerve Conduction Studies in Infants and Children. Journal of Child Neurology, 1986, 1, 19-26.	0.7	110