

Nancy L Kuntz

List of Publications by Year in descending order

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Version: 2024-02-01

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	Nusinersen versus Sham Control in Infantile-Onset Spinal Muscular Atrophy. <i>New England Journal of Medicine</i> , 2017, 377, 1723-1732.	13.9	1,533
2	Nusinersen versus Sham Control in Later-Onset Spinal Muscular Atrophy. <i>New England Journal of Medicine</i> , 2018, 378, 625-635.	13.9	977
3	International consensus guidance for management of myasthenia gravis. <i>Neurology</i> , 2016, 87, 419-425.	1.5	736
4	Nusinersen initiated in infants during the presymptomatic stage of spinal muscular atrophy: Interim efficacy and safety results from the Phase 2 NURTURE study. <i>Neuromuscular Disorders</i> , 2019, 29, 842-856.	0.3	401
5	Long-term effects of glucocorticoids on function, quality of life, and survival in patients with Duchenne muscular dystrophy: a prospective cohort study. <i>Lancet, The</i> , 2018, 391, 451-461.	6.3	306
6	Natural history of infantile-onset spinal muscular atrophy. <i>Annals of Neurology</i> , 2017, 82, 883-891.	2.8	276
7	International Consensus Guidance for Management of Myasthenia Gravis. <i>Neurology</i> , 2021, 96, 114-122.	1.5	272
8	Onasemnogene abeparvovec gene therapy for symptomatic infantile-onset spinal muscular atrophy in patients with two copies of SMN2 (STRIVE): an open-label, single-arm, multicentre, phase 3 trial. <i>Lancet Neurology, The</i> , 2021, 20, 284-293.	4.9	227
9	Treatment Algorithm for Infants Diagnosed with Spinal Muscular Atrophy through Newborn Screening. <i>Journal of Neuromuscular Diseases</i> , 2018, 5, 145-158.	1.1	148
10	Longitudinal study of intraneural perineurioma—a benign, focal hypertrophic neuropathy of youth. <i>Brain</i> , 2009, 132, 2265-2276.	3.7	125
11	Pediatric multiple sclerosis. <i>Nature Reviews Neurology</i> , 2009, 5, 621-631.	4.9	124
12	Review Article: Nerve Conduction Studies in Infants and Children. <i>Journal of Child Neurology</i> , 1986, 1, 19-26.	0.7	110
13	Baseline results of the NeuroNEXT spinal muscular atrophy infant biomarker study. <i>Annals of Clinical and Translational Neurology</i> , 2016, 3, 132-145.	1.7	106
14	Postural Orthostatic Tachycardia Syndrome: A Clinical Review. <i>Pediatric Neurology</i> , 2010, 42, 77-85.	1.0	104
15	Successful treatment of refractory myasthenia gravis using rituximab: a pediatric case report. <i>Journal of Pediatrics</i> , 2003, 143, 674-677.	0.9	101
16	Multiple Sclerosis Therapies in Pediatric Patients With Refractory Multiple Sclerosis. <i>Archives of Neurology</i> , 2011, 68, 437.	4.9	101
17	Revised Recommendations for the Treatment of Infants Diagnosed with Spinal Muscular Atrophy Via Newborn Screening Who Have 4 Copies of SMN2. <i>Journal of Neuromuscular Diseases</i> , 2020, 7, 97-100.	1.1	89
18	Management of Pediatric Central Nervous System Demyelinating Disorders: Consensus of United States Neurologists. <i>Journal of Child Neurology</i> , 2011, 26, 675-682.	0.7	85

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19	The DcpS inhibitor RG3039 improves survival, function and motor unit pathologies in two SMA mouse models. <i>Human Molecular Genetics</i> , 2013, 22, 4084-4101.	1.4	78
20	Association Study of Exon Variants in the NF- κ B and TGF β 2 Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy. <i>American Journal of Human Genetics</i> , 2016, 99, 1163-1171.	2.6	71
21	Phase IIa trial in Duchenne muscular dystrophy shows vamorolone is a first-in-class dissociative steroidal anti-inflammatory drug. <i>Pharmacological Research</i> , 2018, 136, 140-150.	3.1	69
22	Vamorolone trial in Duchenne muscular dystrophy shows dose-related improvement of muscle function. <i>Neurology</i> , 2019, 93, e1312-e1323.	1.5	64
23	Re: "Moving Forward After Two Deaths in a Gene Therapy Trial of Myotubular Myopathy" by Wilson and Flotte. <i>Human Gene Therapy</i> , 2020, 31, 787-787.	1.4	60
24	Neuronal Voltage-Gated Potassium Channel Complex Autoimmunity in Children. <i>Pediatric Neurology</i> , 2011, 44, 275-281.	1.0	56
25	Developing standardized corticosteroid treatment for Duchenne muscular dystrophy. <i>Contemporary Clinical Trials</i> , 2017, 58, 34-39.	0.8	56
26	A multicenter, retrospective medical record review of X-linked myotubular myopathy: The recensus study. <i>Muscle and Nerve</i> , 2018, 57, 550-560.	1.0	54
27	Pediatric Multiple Sclerosis. <i>Neurologic Clinics</i> , 2011, 29, 481-505.	0.8	53
28	Combination molecular therapies for type 1 spinal muscular atrophy. <i>Muscle and Nerve</i> , 2020, 62, 550-554.	1.0	51
29	Effect of Different Corticosteroid Dosing Regimens on Clinical Outcomes in Boys With Duchenne Muscular Dystrophy. <i>JAMA - Journal of the American Medical Association</i> , 2022, 327, 1456.	3.8	43
30	Pupillometry in congenital central hypoventilation syndrome (CCHS): quantitative evidence of autonomic nervous system dysregulation. <i>Pediatric Research</i> , 2012, 71, 280-285.	1.1	41
31	Efficacy and safety of vamorolone in Duchenne muscular dystrophy: An 18-month interim analysis of a non-randomized open-label extension study. <i>PLoS Medicine</i> , 2020, 17, e1003222.	3.9	41
32	The electrophysiologic profile of Dejerine-Sottas disease (HMSN III). <i>Muscle and Nerve</i> , 1990, 13, 586-592.	1.0	40
33	Muscle specific kinase autoimmune myasthenia gravis in children: A case series. <i>Neuromuscular Disorders</i> , 2013, 23, 874-882.	0.3	38
34	Antibody response to common viruses and human leukocyte antigen-DRB1 in pediatric multiple sclerosis. <i>Multiple Sclerosis Journal</i> , 2013, 19, 891-895.	1.4	32
35	Twice-weekly glucocorticosteroids in infants and young boys with Duchenne muscular dystrophy. <i>Muscle and Nerve</i> , 2019, 59, 650-657.	1.0	32
36	Pulsed glucocorticoids enhance dystrophic muscle performance through epigenetic-metabolic reprogramming. <i>JCI Insight</i> , 2019, 4, .	2.3	32

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37	Spinal muscular atrophy care in the COVID-19 pandemic era. <i>Muscle and Nerve</i> , 2020, 62, 46-49.	1.0	31
38	Efficacy and Safety of Vamorolone in Duchenne Muscular Dystrophy. <i>JAMA Network Open</i> , 2022, 5, e2144178.	2.8	31
39	Orthostatic Heart Rate and Blood Pressure in Adolescents: Reference Ranges. <i>Journal of Child Neurology</i> , 2010, 25, 1210-1215.	0.7	28
40	Reldesemtiv in Patients with Spinal Muscular Atrophy: a Phase 2 Hypothesis-Generating Study. <i>Neurotherapeutics</i> , 2021, 18, 1127-1136.	2.1	28
41	The instability of the BTB-KELCH protein Gigaxonin causes Giant Axonal Neuropathy and constitutes a new penetrant and specific diagnostic test. <i>Acta Neuropathologica Communications</i> , 2014, 2, 47.	2.4	26
42	A checklist for clinical trials in rare disease: obstacles and anticipatory actions—lessons learned from the FOR-DMD trial. <i>Trials</i> , 2018, 19, 291.	0.7	26
43	Moderate exercise improves function and increases adiponectin in the mdx mouse model of muscular dystrophy. <i>Scientific Reports</i> , 2019, 9, 5770.	1.6	26
44	Treatment of multiple sclerosis in children and adolescents. <i>Expert Opinion on Pharmacotherapy</i> , 2010, 11, 505-520.	0.9	24
45	Mortality and respiratory support in X-linked myotubular myopathy: a RECENSUS retrospective analysis. <i>Archives of Disease in Childhood</i> , 2020, 105, 332-338.	1.0	24
46	Pathogenic Variants in the Myosin Chaperone UNC-45B Cause Progressive Myopathy with Eccentric Cores. <i>American Journal of Human Genetics</i> , 2020, 107, 1078-1095.	2.6	24
47	Two siblings with limb-girdle muscular dystrophy type 2E responsive to deflazacort. <i>Neuromuscular Disorders</i> , 2010, 20, 122-124.	0.3	20
48	Diagnostic Criteria for Pediatric Multiple Sclerosis. <i>Current Neurology and Neuroscience Reports</i> , 2013, 13, 354.	2.0	19
49	An expanded access program of risdiplam for patients with Type 1 or 2 spinal muscular atrophy. <i>Annals of Clinical and Translational Neurology</i> , 2022, 9, 810-818.	1.7	18
50	Utility and practice of electrodiagnostic testing in the pediatric population: An AANEM consensus statement. <i>Muscle and Nerve</i> , 2020, 61, 143-155.	1.0	14
51	The <sc>CINRG</sc> Becker Natural History Study: Baseline characteristics. <i>Muscle and Nerve</i> , 2020, 62, 369-376.	1.0	14
52	Antiphospholipid Syndrome With Catastrophic Bleeding and Recurrent Ischemic Strokes as Initial Presentation of Systemic Lupus Erythematosus. <i>Journal of Pediatric Hematology/Oncology</i> , 2005, 27, 403-407.	0.3	13
53	Spasmodic Muscle Cramps and Weakness as Presenting Symptoms in Wilson Disease. <i>Pediatrics</i> , 2013, 132, e1039-e1042.	1.0	12
54	242nd ENMC International Workshop: Diagnosis and management of juvenile myasthenia gravis Hoofddorp, the Netherlands, 1-3 March 2019. <i>Neuromuscular Disorders</i> , 2020, 30, 254-264.	0.3	12

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55	Cerebellar Mutism in Acute Disseminating Encephalomyelitis. <i>Pediatric Neurology</i> , 2014, 50, 511-514.	1.0	11
56	Recruitment & retention program for the NeuroNEXT SMA Biomarker Study: Super Babies for SMA!. <i>Contemporary Clinical Trials Communications</i> , 2018, 11, 113-119.	0.5	11
57	Liquid formulation of pentoxifylline is a poorly tolerated treatment for duchenne dystrophy. <i>Muscle and Nerve</i> , 2011, 44, 170-173.	1.0	10
58	Localized Aquadynia Responsive to Clonidine in a 13-Year-Old Girl. <i>Pediatric Dermatology</i> , 2010, 27, 646-649.	0.5	9
59	Laboratory Evaluation of Pediatric Autonomic Disorders. <i>Seminars in Pediatric Neurology</i> , 2013, 20, 35-43.	1.0	8
60	Restoration of Nusinersen Levels Following Treatment Interruption in People With Spinal Muscular Atrophy: Simulations Based on a Population Pharmacokinetic Model. <i>CNS Drugs</i> , 2022, 36, 181-190.	2.7	6
61	Hereditary Neuropathy with Liability to Pressure Palsies. <i>Pediatric Neurology Briefs</i> , 2015, 29, 83.	0.2	5
62	INCEPTUS Natural History, Run-in Study for Gene Replacement Clinical Trial in X-Linked Myotubular Myopathy. <i>Journal of Neuromuscular Diseases</i> , 2022, 9, 503-516.	1.1	5
63	Quantitative MRI analysis in children with multiple sclerosis: a multicenter feasibility pilot study. <i>BMC Neurology</i> , 2013, 13, 173.	0.8	4
64	Delay in Diagnosis of Duchenne Muscular Dystrophy. <i>Pediatric Neurology Briefs</i> , 2015, 29, 5.	0.2	4
65	Health related quality of life in young, steroid-naïve boys with Duchenne muscular dystrophy. <i>Neuromuscular Disorders</i> , 2021, 31, 1161-1168.	0.3	4
66	Novel methods of imaging and analysis for the thermoregulatory sweat test. <i>Journal of Applied Physiology</i> , 2018, 125, 755-762.	1.2	3
67	Onasemnogene AAVS1 gene-replacement therapy (GRT) for spinal muscular atrophy type 1 (SMA1): pivotal phase 3 study (STRIVE) update. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2019, 90, A22.1-A22.	0.9	3
68	Congenital Central Hypoventilation Syndrome (CCHS) and PHOX2B Mutations. , 2012, , 445-449.		2
69	Peripheral Nerve Disorders in the Neonate. <i>NeoReviews</i> , 2016, 17, e719-e728.	0.4	2
70	Transforaminal Intrathecal Access for Injection of Nusinersen in Adult and Pediatric Patients with Spinal Muscular Atrophy. <i>Journal of Pediatric Neurology</i> , 2020, 18, 088-094.	0.0	2
71	Diagnosis and Evaluation of Small Fiber Peripheral Neuropathy in Children. , 2017, , 265-280.		2
72	Differential diagnosis of multiple sclerosis and acquired central nervous system demyelinating disorders in children and adolescents. , 2011, , 58-74.		1

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73	Difficulties classifying myasthenia gravis in the pediatric surgical literature. Journal of Pediatric Surgery, 2020, 55, 1679.	0.8	1
74	Clinical Neurophysiology of Pediatric Polyneuropathies. , 2006, , 645-686.		0
75	Clinical Neurophysiology of the Motor Unit in Infants and Children. , 2006, , 130-145.		0
76	Diagnosis and treatment of peripheral nerve lesions in children. Paediatrics and Child Health (United) Tj ETQq0 0 0 ggBT /Overlock 10 Tf	0.2	0
77	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
78	Autonomic Nervous System Dysregulation in Monozygous Twins With Nephropathic Cystinosis. Kidney International Reports, 2018, 3, 1489-1496.	0.4	0
79	Spinal muscular atrophy. Neurology, 2020, 95, 11-12.	1.5	0
80	Autonomic Testing in Childhood. , 2006, , 687-712.		0
81	Muscle Disorders in Children: Neurophysiologic Contributions to Diagnosis and Management. , 2006, , 747-762.		0
82	Axonal Damage in Pediatric Multiple Sclerosis. Pediatric Neurology Briefs, 2015, 29, 34.	0.2	0
83	Outcome Measures for COL6 and LAMA2-Related Dystrophies. Pediatric Neurology Briefs, 2020, 34, 15.	0.2	0
84	Neuromuscular disease - Gene transfer for children. Journal of International Child Neurology Association, 2021, 1, .	0.0	0