

Darryl C De Vivo

List of Publications by Year in descending order

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Version: 2024-02-01

100
papers

9,138
citations

81743

39
h-index

40881

93
g-index

101
all docs

101
docs citations

101
times ranked

8365
citing authors

#	ARTICLE	IF	CITATIONS
1	Mitochondrial myopathy, encephalopathy, lactic acidosis, and strokelike episodes: A distinctive clinical syndrome. <i>Annals of Neurology</i> , 1984, 16, 481-488.	2.8	1,128
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	Treatment of infantile-onset spinal muscular atrophy with nusinersen: a phase 2, open-label, dose-escalation study. <i>Lancet, The</i> , 2016, 388, 3017-3026.	6.3	801
4	Results from a phase 1 study of nusinersen (ISIS-SMN _{Rx}) in children with spinal muscular atrophy. <i>Neurology</i> , 2016, 86, 890-897.	1.5	506
5	GLUT1 reductions exacerbate Alzheimer's disease vasculo-neuronal dysfunction and degeneration. <i>Nature Neuroscience</i> , 2015, 18, 521-530.	7.1	496
6	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
7	Observational study of spinal muscular atrophy type I and implications for clinical trials. <i>Neurology</i> , 2014, 83, 810-817.	1.5	367
8	GLUT-1 deficiency syndrome caused by haploinsufficiency of the blood-brain barrier hexose carrier. <i>Nature Genetics</i> , 1998, 18, 188-191.	9.4	349
9	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. <i>Neuron</i> , 2015, 88, 499-513.	3.8	258
10	Nusinersen in later-onset spinal muscular atrophy. <i>Neurology</i> , 2019, 92, e2492-e2506.	1.5	183
11	l-Carnitine Supplementation in Childhood Epilepsy: Current Perspectives. <i>Epilepsia</i> , 1998, 39, 1216-1225.	2.6	172
12	Revised upper limb module for spinal muscular atrophy: Development of a new module. <i>Muscle and Nerve</i> , 2017, 55, 869-874.	1.0	166
13	Mutational analysis of GLUT1 (SLC2A1) in Glut-1 Deficiency Syndrome. <i>Human Mutation</i> , 2000, 16, 224-231.	1.1	143
14	Patterns of disease progression in type 2 and 3 SMA: Implications for clinical trials. <i>Neuromuscular Disorders</i> , 2016, 26, 126-131.	0.3	142
15	Glut1 Deficiency Syndrome (Glut1DS): State of the art in 2020 and recommendations of the international Glut1DS study group. <i>Epilepsia Open</i> , 2020, 5, 354-365.	1.3	142
16	The Ketogenic Diet Revisited: Back to the Future. <i>Epilepsia</i> , 1997, 38, 743-749.	2.6	124
17	Content validity and clinical meaningfulness of the HFMSE in spinal muscular atrophy. <i>BMC Neurology</i> , 2017, 17, 39.	0.8	102
18	Selective deficits in verbal working memory associated with a known genetic etiology: The neuropsychological profile of Duchenne muscular dystrophy. <i>Journal of the International Neuropsychological Society</i> , 2001, 7, 45-54.	1.2	101

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19	Defective glucose transport across brain tissue barriers: a newly recognized neurological syndrome. <i>Neurochemical Research</i> , 1999, 24, 587-594.	1.6	100
20	Six-minute walk test is reliable and valid in spinal muscular atrophy. <i>Muscle and Nerve</i> , 2016, 54, 836-842.	1.0	98
21	Developmental milestones in type I spinal muscular atrophy. <i>Neuromuscular Disorders</i> , 2016, 26, 754-759.	0.3	96
22	Circulating markers of NADH-reductive stress correlate with mitochondrial disease severity. <i>Journal of Clinical Investigation</i> , 2021, 131, .	3.9	95
23	Disruption of Glut1 in Hematopoietic Stem Cells Prevents Myelopoiesis and Enhanced Glucose Flux in Atheromatous Plaques of ApoE ^{-/-} Mice. <i>Circulation Research</i> , 2016, 118, 1062-1077.	2.0	93
24	Brain microvasculature defects and Glut1 deficiency syndrome averted by early repletion of the glucose transporter-1 protein. <i>Nature Communications</i> , 2017, 8, 14152.	5.8	91
25	Erythrocyte 3-O-methyl-D-glucose uptake assay for diagnosis of glucose-transporter-protein syndrome. <i>Journal of Clinical Laboratory Analysis</i> , 1999, 13, 116-121.	0.9	86
26	Long-Term Clinical Course of Glut1 Deficiency Syndrome. <i>Journal of Child Neurology</i> , 2015, 30, 160-169.	0.7	86
27	Increased fat mass and high incidence of overweight despite low body mass index in patients with spinal muscular atrophy. <i>Neuromuscular Disorders</i> , 2009, 19, 391-396.	0.3	74
28	Revised Hammersmith Scale for spinal muscular atrophy: A SMA specific clinical outcome assessment tool. <i>PLoS ONE</i> , 2017, 12, e0172346.	1.1	67
29	Ambulatory function in spinal muscular atrophy: Age-related patterns of progression. <i>PLoS ONE</i> , 2018, 13, e0199657.	1.1	65
30	Nusinersen improves walking distance and reduces fatigue in later-onset spinal muscular atrophy. <i>Muscle and Nerve</i> , 2019, 60, 409-414.	1.0	62
31	Revised upper limb module for spinal muscular atrophy: 12-month changes. <i>Muscle and Nerve</i> , 2019, 59, 426-430.	1.0	61
32	GLUT1-Deficiency: Barbiturates Potentiate Haploinsufficiency in Vitro. <i>Pediatric Research</i> , 1999, 46, 677-677.	1.1	59
33	Glucose Transporter Type 1 Deficiency Syndrome (Glut1DS): Methylxanthines Potentiate GLUT1 Haploinsufficiency In Vitro. <i>Pediatric Research</i> , 2001, 50, 254-260.	1.1	58
34	Navajo neurohepatopathy: A mitochondrial DNA depletion syndrome?. <i>Hepatology</i> , 2001, 34, 116-120.	3.6	57
35	Clinical Heterogeneity Associated with the Mitochondrial DNA T8993C Point Mutation. <i>Pediatric Research</i> , 1996, 39, 914-917.	1.1	56
36	Protean Phenotypic Features of the A3243G Mitochondrial DNA Mutation. <i>Archives of Neurology</i> , 2009, 66, 85-91.	4.9	53

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37	Adiposity is increased among high-functioning, non-ambulatory patients with spinal muscular atrophy. <i>Neuromuscular Disorders</i> , 2010, 20, 448-452.	0.3	47
38	Treatment of infantile-onset spinal muscular atrophy with nusinersen: final report of a phase 2, open-label, multicentre, dose-escalation study. <i>The Lancet Child and Adolescent Health</i> , 2021, 5, 491-500.	2.7	47
39	Longitudinal natural history of type I spinal muscular atrophy: a critical review. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 84.	1.2	45
40	Neurodegeneration in spinal muscular atrophy: from disease phenotype and animal models to therapeutic strategies and beyond. <i>Future Neurology</i> , 2014, 9, 49-65.	0.9	44
41	Therapeutic strategies for glucose transporter 1 deficiency syndrome. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1923-1932.	1.7	44
42	Paroxysmal eye-head movements in Glut1 deficiency syndrome. <i>Neurology</i> , 2017, 88, 1666-1673.	1.5	43
43	Exploring mTOR inhibition as treatment for mitochondrial disease. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1877-1881.	1.7	40
44	Cerebral metabolic abnormalities in A3243G mitochondrial DNA mutation carriers. <i>Neurology</i> , 2014, 82, 798-805.	1.5	39
45	Deficient Muscle Carnitine Transport in Primary Carnitine Deficiency. <i>Pediatric Research</i> , 1997, 42, 583-587.	1.1	38
46	Mitochondrial Encephalomyopathy Lactic Acidosis and Stroke-Like Episodes (MELAS): A Case Report and Critical Reappraisal of Treatment Options. <i>Pediatric Neurology</i> , 2016, 56, 59-61.	1.0	37
47	Evaluator Training and Reliability for SMA Global Nusinersen Trials1. <i>Journal of Neuromuscular Diseases</i> , 2018, 5, 159-166.	1.1	36
48	Nusinersen Treatment in Adults With Spinal Muscular Atrophy. <i>Neurology: Clinical Practice</i> , 2021, 11, e317-e327.	0.8	35
49	Acute hyperglycemia produces transient improvement in glucose transporter type 1 deficiency. <i>Annals of Neurology</i> , 2010, 67, 31-40.	2.8	33
50	Single-Blind, Randomized, Controlled Clinical Trial of Exercise in Ambulatory Spinal Muscular Atrophy: Why are the Results Negative?. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, 463-470.	1.1	33
51	Diagnosing Glucose Transporter 1 Deficiency at Initial Presentation Facilitates Early Treatment. <i>Journal of Pediatrics</i> , 2016, 171, 220-226.	0.9	33
52	Functional cellular analyses reveal energy metabolism defect and mitochondrial DNA depletion in a case of mitochondrial aconitase deficiency. <i>Molecular Genetics and Metabolism</i> , 2016, 118, 28-34.	0.5	32
53	Topography of brain glucose hypometabolism and epileptic network in glucose transporter 1 deficiency. <i>Epilepsy Research</i> , 2015, 110, 206-215.	0.8	31
54	Perceived Fatigue in Spinal Muscular Atrophy: A Pilot Study. <i>Journal of Neuromuscular Diseases</i> , 2019, 6, 109-117.	1.1	30

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55	Age related treatment effect in type II Spinal Muscular Atrophy pediatric patients treated with nusinersen. <i>Neuromuscular Disorders</i> , 2021, 31, 596-602.	0.3	29
56	N-acetylaspartate supports the energetic demands of developmental myelination via oligodendroglial aspartoacylase. <i>Neurobiology of Disease</i> , 2016, 96, 323-334.	2.1	28
57	Glucose transporter 1 deficiency syndrome and other glycolytic defects. <i>Journal of Child Neurology</i> , 2002, 17 Suppl 3, 3S15-23; discussion 3S24-5.	0.7	28
58	Pyruvate dehydrogenase deficiency: Molecular basis for intrafamilial heterogeneity. <i>Annals of Neurology</i> , 1994, 36, 83-89.	2.8	27
59	Nusinersen in pediatric and adult patients with type III spinal muscular atrophy. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 1622-1634.	1.7	27
60	Spinal muscular atrophy functional composite score: A functional measure in spinal muscular atrophy. <i>Muscle and Nerve</i> , 2015, 52, 942-947.	1.0	26
61	Glucose transporter protein syndromes. <i>International Review of Neurobiology</i> , 2002, 51, 259-IN10.	0.9	25
62	Maternal Calorie Restriction Causing Uteroplacental Insufficiency Differentially Affects Mammalian Placental Glucose and Leucine Transport Molecular Mechanisms. <i>Endocrinology</i> , 2016, 157, 4041-4054.	1.4	25
63	Age and baseline values predict 12 and 24-month functional changes in type 2 SMA. <i>Neuromuscular Disorders</i> , 2020, 30, 756-764.	0.3	25
64	Gait assessment with solesound instrumented footwear in spinal muscular atrophy. <i>Muscle and Nerve</i> , 2017, 56, 230-236.	1.0	20
65	Different trajectories in upper limb and gross motor function in spinal muscular atrophy. <i>Muscle and Nerve</i> , 2021, 64, 552-559.	1.0	18
66	Rasch analysis of the Pediatric Evaluation of Disability Inventoryâ€œcomputer adaptive test (PEDIâ€œCAT) item bank for children and young adults with spinal muscular atrophy. <i>Muscle and Nerve</i> , 2016, 54, 1097-1107.	1.0	17
67	RESTORE: A Prospective Multinational Registry of Patients with Genetically Confirmed Spinal Muscular Atrophy - Rationale and Study Design. <i>Journal of Neuromuscular Diseases</i> , 2020, 7, 145-152.	1.1	17
68	Long Survival in Patients With Leigh Syndrome and the m.10191T>C Mutation in <i>MT-ND3</i> . <i>Journal of Child Neurology</i> , 2014, 29, NP105-NP110.	0.7	16
69	Leigh syndrome: Historical perspective and clinical variations. <i>BioFactors</i> , 1998, 7, 269-271.	2.6	15
70	A Randomized, Controlled Clinical Trial of Exercise in Patients with Spinal Muscular Atrophy: Methods and Baseline Characteristics. <i>Journal of Neuromuscular Diseases</i> , 2014, 1, 151-161.	1.1	14
71	Reduction of Glut1 in the Neural Retina But Not the RPE Alleviates Polyol Accumulation and Normalizes Early Characteristics of Diabetic Retinopathy. <i>Journal of Neuroscience</i> , 2021, 41, 3275-3299.	1.7	14
72	Cerebrospinal fluid polyamines: Biochemical markers of malignant childhood brain tumors. <i>Annals of Neurology</i> , 1986, 19, 360-364.	2.8	13

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73	Physical therapy services received by individuals with spinal muscular atrophy (SMA). Journal of Pediatric Rehabilitation Medicine, 2016, 9, 35-44.	0.3	13
74	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. Genetics in Medicine, 2021, 23, 374-383.	1.1	13
75	Revised upper limb module in type II and III spinal muscular atrophy: 24-month changes. Neuromuscular Disorders, 2022, 32, 36-42.	0.3	13
76	Diminished muscle oxygen uptake and fatigue in spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2021, 8, 1086-1095.	1.7	11
77	<i>VAC14</i> syndrome in two siblings with retinitis pigmentosa and neurodegeneration with brain iron accumulation. Journal of Physical Education and Sports Management, 2019, 5, a003715.	0.5	10
78	A randomized, double-blind trial of triheptanoin for drug-resistant epilepsy in glucose transporter 1 deficiency syndrome. Epilepsia, 2022, 63, 1748-1760.	2.6	9
79	Scientific rationale for a higher dose of nusinersen. Annals of Clinical and Translational Neurology, 2022, 9, 819-829.	1.7	9
80	Cholesterol and phospholipids in cultured skin fibroblasts from patients with dystonia. Annals of Neurology, 1984, 16, 250-252.	2.8	8
81	Exploring diazoxide and continuous glucose monitoring as treatment for Glut1 deficiency syndrome. Annals of Clinical and Translational Neurology, 2021, 8, 2205-2209.	1.7	8
82	CoQ10 Deficiency Is Not a Common Finding in GLUT1 Deficiency Syndrome. JIMD Reports, 2015, 29, 47-52.	0.7	7
83	Scoliosis Surgery Significantly Impacts Motor Abilities in Higher-functioning Individuals with Spinal Muscular Atrophy1. Journal of Neuromuscular Diseases, 2020, 7, 183-192.	1.1	7
84	Old measures and new scores in spinal muscular atrophy patients. Muscle and Nerve, 2015, 52, 435-437.	1.0	6
85	Limitations of 6-minute walk test reference values for spinal muscular atrophy. Muscle and Nerve, 2020, 61, 375-382.	1.0	6
86	A Randomized, Controlled Clinical Trial of Exercise in Patients with Spinal Muscular Atrophy: Methods and Baseline Characteristics. Journal of Neuromuscular Diseases, 2014, 1, 151-161.	1.1	6
87	Cerebral energy failure. Current Neurology and Neuroscience Reports, 2001, 1, 203-204.	2.0	5
88	A De Novo Mutation in MTND6 Causes Generalized Dystonia in 2 Unrelated Children. Child Neurology Open, 2016, 3, 2329048X1562793.	0.5	4
89	Transient hyperreflexia. Neurology: Clinical Practice, 2020, 10, e66-e67.	0.8	3
90	Hypotonia and cystinuria <i>2p21</i> deletion syndrome: Intrafamilial variability of clinical expression. Annals of Clinical and Translational Neurology, 2021, 8, 2199-2204.	1.7	3

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91	Solving the COX Puzzle. <i>Annals of Neurology</i> , 1999, 46, 142-143.	2.8	2
92	Analysis of Gait Disturbance in Glut 1 Deficiency Syndrome. <i>Journal of Child Neurology</i> , 2016, 31, 1483-1488.	0.7	2
93	Exploring triheptanoin as treatment for short chain enoyl CoA hydratase deficiency. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 1151-1157.	1.7	2
94	Psychometric properties of the PEDI-CAT for children and youth with spinal muscular atrophy. <i>Journal of Pediatric Rehabilitation Medicine</i> , 2021, 14, 451-461.	0.3	2
95	Distribution of Weight, Stature and Growth Status in Children and Adolescents with Spinal Muscular Atrophy: An Observational Retrospective Study in the United States. <i>Muscle and Nerve</i> , 2022, , .	1.0	2
96	Erythrocyte 3-O-methyl-D-glucose uptake assay for diagnosis of glucose-transporter-protein syndrome. , 1999, 13, 116.		1
97	Movement Disorders in Glucose Transporter Type 1 Deficiency. <i>Journal of Pediatric Neurology</i> , 2015, 13, 168-173.	0.0	0
98	Neuroanatomical Models of Muscle Strength and Relationship to Ambulatory Function in Spinal Muscular Atrophy. <i>Journal of Neuromuscular Diseases</i> , 2020, 7, 459-466.	1.1	0
99	Continuous Glucose Monitoring Facilitates Diazoxide Use in the Management of Glut1 Deficiency Syndrome. <i>Journal of the Endocrine Society</i> , 2021, 5, A698-A699.	0.1	0
100	Visual memory failure presages conversion to <scp>MELAS</scp> phenotype. <i>Annals of Clinical and Translational Neurology</i> , 2022, , .	1.7	0