# Darryl C De Vivo

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/1638652/publications.pdf

Version: 2024-02-01

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|---|---|---|---|
| 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 | Revised upper limb module in type II and III spinal muscular atrophy: 24-month changes. Neuromuscular Disorders, 2022, 32, 36-42. | 0.3 | 13 |
| 2 | Distribution of Weight, Stature and Growth Status in Children and Adolescents with Spinal Muscular Atrophy: An Observational Retrospective Study in the United States. Muscle and Nerve, 2022, , . | 1.0 | 2 |
| 3 | A randomized, doubleâ€blind trial of triheptanoin for drugâ€resistant epilepsy in glucose transporter 1 deficiency syndrome. Epilepsia, 2022, 63, 1748-1760. | 2.6 | 9 |
| 4 | Visual memory failure presages conversion to <scp>MELAS</scp> phenotype. Annals of Clinical and Translational Neurology, 2022, , . | 1.7 | 0 |
| 5 | Scientific rationale for a higher dose of nusinersen. Annals of Clinical and Translational Neurology, 2022, 9, 819-829. | 1.7 | 9 |
| 6 | JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. Genetics in Medicine, 2021, 23, 374-383. | 1.1 | 13 |
| 7 | Circulating markers of NADH-reductive stress correlate with mitochondrial disease severity. Journal of Clinical Investigation, 2021, 131, . | 3.9 | 95 |
| 8 | Reduction of Glut1 in the Neural Retina But Not the RPE Alleviates Polyol Accumulation and Normalizes Early Characteristics of Diabetic Retinopathy. Journal of Neuroscience, 2021, 41, 3275-3299. | 1.7 | 14 |
| 9 | Diminished muscle oxygen uptake and fatigue in spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2021, 8, 1086-1095. | 1.7 | 11 |
| 10 | Exploring triheptanoin as treatment for short chain enoyl CoA hydratase deficiency. Annals of Clinical and Translational Neurology, 2021, 8, 1151-1157. | 1.7 | 2 |
| 11 | Continuous Glucose Monitoring Facilitates Diazoxide Use in the Management of Glut1 Deficiency Syndrome. Journal of the Endocrine Society, 2021, 5, A698-A699. | 0.1 | 0 |
| 12 | Nusinersen in pediatric and adult patients with type III spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2021, 8, 1622-1634. | 1.7 | 27 |
| 13 | Treatment of infantile-onset spinal muscular atrophy with nusinersen: final report of a phase 2, open-label, multicentre, dose-escalation study. The Lancet Child and Adolescent Health, 2021, 5, 491-500. | 2.7 | 47 |
| 14 | Age related treatment effect in type II Spinal Muscular Atrophy pediatric patients treated with nusinersen. Neuromuscular Disorders, 2021, 31, 596-602. | 0.3 | 29 |
| 15 | Different trajectories in upper limb and gross motor function in spinal muscular atrophy. Muscle and Nerve, 2021, 64, 552-559. | 1.0 | 18 |
| 16 | Psychometric properties of the PEDI-CAT for children and youth with spinal muscular atrophy. Journal of Pediatric Rehabilitation Medicine, 2021, 14, 451-461. | 0.3 | 2 |
| 17 | Nusinersen Treatment in Adults With Spinal Muscular Atrophy. Neurology: Clinical Practice, 2021, 11, e317-e327. | 0.8 | 35 |
| 18 | Hypotoniaâ€“cystinuria 2p21 deletion syndrome: Intrafamilial variability of clinical expression. Annals of Clinical and Translational Neurology, 2021, 8, 2199-2204. | 1.7 | 3 |

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|---|---|---|---|
| 19 | 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 |
| 20 | Limitations of 6â€minute walk test reference values for spinal muscular atrophy. Muscle and Nerve, 2020, 61, 375-382. | 1.0 | 6 |
| 21 | Age and baseline values predict 12 and 24-month functional changes in type 2 SMA. Neuromuscular Disorders, 2020, 30, 756-764. | 0.3 | 25 |
| 22 | Neuroanatomical Models of Muscle Strength and Relationship to Ambulatory Function in Spinal Muscular Atrophy. Journal of Neuromuscular Diseases, 2020, 7, 459-466. | 1.1 | 0 |
| 23 | 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 |
| 24 | 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. | 1.1 | 17 |
| 25 | Glut1 Deficiency Syndrome (Glut1DS): State of the art in 2020 and recommendations of the international Glut1DS study group. Epilepsia Open, 2020, 5, 354-365. | 1.3 | 142 |
| 26 | Transient hyperreflexia. Neurology: Clinical Practice, 2020, 10, e66-e67. | 0.8 | 3 |
| 27 | Longitudinal natural history of type I spinal muscular atrophy: a critical review. Orphanet Journal of Rare Diseases, 2020, 15, 84. | 1.2 | 45 |
| 28 | VAC14 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 |
| 29 | Exploring mTOR inhibition as treatment for mitochondrial disease. Annals of Clinical and Translational Neurology, 2019, 6, 1877-1881. | 1.7 | 40 |
| 30 | Nusinersen improves walking distance and reduces fatigue in laterâ€onset spinal muscular atrophy. Muscle and Nerve, 2019, 60, 409-414. | 1.0 | 62 |
| 31 | Therapeutic strategies for glucose transporter 1 deficiency syndrome. Annals of Clinical and Translational Neurology, 2019, 6, 1923-1932. | 1.7 | 44 |
| 32 | 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 |
| 33 | Revised upper limb module for spinal muscular atrophy: 12â€‰month changes. Muscle and Nerve, 2019, 59, 426-430. | 1.0 | 61 |
| 34 | Nusinersen in later-onset spinal muscular atrophy. Neurology, 2019, 92, e2492-e2506. | 1.5 | 183 |
| 35 | Perceived Fatigue in Spinal Muscular Atrophy: A Pilot Study. Journal of Neuromuscular Diseases, 2019, 6, 109-117. | 1.1 | 30 |
| 36 | Nusinersen versus Sham Control in Later-Onset Spinal Muscular Atrophy. New England Journal of Medicine, 2018, 378, 625-635. | 13.9 | 977 |

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|---|---|---|---|
| 37 | Ambulatory function in spinal muscular atrophy: Age-related patterns of progression. PLoS ONE, 2018, 13, e0199657. | 1.1 | 65 |
| 38 | Evaluator Training and Reliability for SMA Global Nusinersen Trials1. Journal of Neuromuscular Diseases, 2018, 5, 159-166. | 1.1 | 36 |
| 39 | Brain microvasculature defects and Glut1 deficiency syndrome averted by early repletion of the glucose transporter-1 protein. Nature Communications, 2017, 8, 14152. | 5.8 | 91 |
| 40 | Content validity and clinical meaningfulness of the HFMSE in spinal muscular atrophy. BMC Neurology, 2017, 17, 39. | 0.8 | 102 |
| 41 | Paroxysmal eyeâ€“head movements in Glut1 deficiency syndrome. Neurology, 2017, 88, 1666-1673. | 1.5 | 43 |
| 42 | Gait assessment with solesound instrumented footwear in spinal muscular atrophy. Muscle and Nerve, 2017, 56, 230-236. | 1.0 | 20 |
| 43 | Revised upper limb module for spinal muscular atrophy: Development of a new module. Muscle and Nerve, 2017, 55, 869-874. | 1.0 | 166 |
| 44 | Revised Hammersmith Scale for spinal muscular atrophy: A SMA specific clinical outcome assessment tool. PLoS ONE, 2017, 12, e0172346. | 1.1 | 67 |
| 45 | Six-minute walk test is reliable and valid in spinal muscular atrophy. Muscle and Nerve, 2016, 54, 836-842. | 1.0 | 98 |
| 46 | Rasch analysis of the Pediatric Evaluation of Disability Inventoryâ€“computer adaptive test (PEDIâ€CAT) item bank for children and young adults with spinal muscular atrophy. Muscle and Nerve, 2016, 54, 1097-1107. | 1.0 | 17 |
| 47 | A De Novo Mutation in MTND6 Causes Generalized Dystonia in 2 Unrelated Children. Child Neurology Open, 2016, 3, 2329048X1562793. | 0.5 | 4 |
| 48 | Physical therapy services received by individuals with spinal muscular atrophy (SMA). Journal of Pediatric Rehabilitation Medicine, 2016, 9, 35-44. | 0.3 | 13 |
| 49 | Treatment of infantile-onset spinal muscular atrophy with nusinersen: a phase 2, open-label, dose-escalation study. Lancet, The, 2016, 388, 3017-3026. | 6.3 | 801 |
| 50 | Disruption of Glut1 in Hematopoietic Stem Cells Prevents Myelopoiesis and Enhanced Glucose Flux in Atheromatous Plaques of ApoE <sup>â^'/â^'</sup> Mice. Circulation Research, 2016, 118, 1062-1077. | 2.0 | 93 |
| 51 | Analysis of Gait Disturbance in Glut 1 Deficiency Syndrome. Journal of Child Neurology, 2016, 31, 1483-1488. | 0.7 | 2 |
| 52 | Maternal Calorie Restriction Causing Uteroplacental Insufficiency Differentially Affects Mammalian Placental Glucose and Leucine Transport Molecular Mechanisms. Endocrinology, 2016, 157, 4041-4054. | 1.4 | 25 |
| 53 | Developmental milestones in type I spinal muscular atrophy. Neuromuscular Disorders, 2016, 26, 754-759. | 0.3 | 96 |
| 54 | N-acetylaspartate supports the energetic demands of developmental myelination via oligodendroglial aspartoacylase. Neurobiology of Disease, 2016, 96, 323-334. | 2.1 | 28 |

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|---|---|---|---|
| 55 | Diagnosing Glucose Transporter 1 Deficiency at Initial Presentation Facilitates Early Treatment. Journal of Pediatrics, 2016, 171, 220-226. | 0.9 | 33 |
| 56 | Functional cellular analyses reveal energy metabolism defect and mitochondrial DNA depletion in a case of mitochondrial aconitase deficiency. Molecular Genetics and Metabolism, 2016, 118, 28-34. | 0.5 | 32 |
| 57 | Results from a phase 1 study of nusinersen (ISIS-SMN <sub>Rx</sub> ) in children with spinal muscular atrophy. Neurology, 2016, 86, 890-897. | 1.5 | 506 |
| 58 | Patterns of disease progression in type 2 and 3 SMA: Implications for clinical trials. Neuromuscular Disorders, 2016, 26, 126-131. | 0.3 | 142 |
| 59 | Mitochondrial Encephalomyopathy Lactic Acidosis and Stroke-Like Episodes (MELAS): A Case Report and Critical Reappraisal of Treatment Options. Pediatric Neurology, 2016, 56, 59-61. | 1.0 | 37 |
| 60 | Movement Disorders in Glucose Transporter Type 1 Deficiency. Journal of Pediatric Neurology, 2015, 13, 168-173. | 0.0 | 0 |
| 61 | Single-Blind, Randomized, Controlled Clinical Trial of Exercise in Ambulatory Spinal Muscular Atrophy: Why are the Results Negative?. Journal of Neuromuscular Diseases, 2015, 2, 463-470. | 1.1 | 33 |
| 62 | Old measures and new scores in spinal muscular atrophy patients. Muscle and Nerve, 2015, 52, 435-437. | 1.0 | 6 |
| 63 | Spinal muscular atrophy functional composite score: A functional measure in spinal muscular atrophy. Muscle and Nerve, 2015, 52, 942-947. | 1.0 | 26 |
| 64 | CoQ10 Deficiency Is Not a Common Finding in GLUT1 Deficiency Syndrome. JIMD Reports, 2015, 29, 47-52. | 0.7 | 7 |
| 65 | Long-Term Clinical Course of Glut1 Deficiency Syndrome. Journal of Child Neurology, 2015, 30, 160-169. | 0.7 | 86 |
| 66 | GLUT1 reductions exacerbate Alzheimer's disease vasculo-neuronal dysfunction and degeneration. Nature Neuroscience, 2015, 18, 521-530. | 7.1 | 496 |
| 67 | Topography of brain glucose hypometabolism and epileptic network in glucose transporter 1 deficiency. Epilepsy Research, 2015, 110, 206-215. | 0.8 | 31 |
| 68 | Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. Neuron, 2015, 88, 499-513. | 3.8 | 258 |
| 69 | 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 | 14 |
| 70 | Neurodegeneration in spinal muscular atrophy: from disease phenotype and animal models to therapeutic strategies and beyond. Future Neurology, 2014, 9, 49-65. | 0.9 | 44 |
| 71 | Long Survival in Patients With Leigh Syndrome and the m.10191T>C Mutation in MT-ND3. Journal of Child Neurology, 2014, 29, NP105-NP110. | 0.7 | 16 |
| 72 | Cerebral metabolic abnormalities in A3243G mitochondrial DNA mutation carriers. Neurology, 2014, 82, 798-805. | 1.5 | 39 |

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|---|---|---|---|
| 73 | Observational study of spinal muscular atrophy type I and implications for clinical trials. Neurology, 2014, 83, 810-817. | 1.5 | 367 |
| 74 | 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 |
| 75 | Acute hyperglycemia produces transient improvement in glucose transporter type 1 deficiency. Annals of Neurology, 2010, 67, 31-40. | 2.8 | 33 |
| 76 | Adiposity is increased among high-functioning, non-ambulatory patients with spinal muscular atrophy. Neuromuscular Disorders, 2010, 20, 448-452. | 0.3 | 47 |
| 77 | Protean Phenotypic Features of the A3243G Mitochondrial DNA Mutation. Archives of Neurology, 2009, 66, 85-91. | 4.9 | 53 |
| 78 | Increased fat mass and high incidence of overweight despite low body mass index in patients with spinal muscular atrophy. Neuromuscular Disorders, 2009, 19, 391-396. | 0.3 | 74 |
| 79 | Glucose transporter protein syndromes. International Review of Neurobiology, 2002, 51, 259-IN10. | 0.9 | 25 |
| 80 | Glucose transporter 1 deficiency syndrome and other glycolytic defects. Journal of Child Neurology, 2002, 17 Suppl 3, 3S15-23; discussion 3S24-5. | 0.7 | 28 |
| 81 | Selective deficits in verbal working memory associated with a known genetic etiology: The neuropsychological profile of Duchenne muscular dystrophy. Journal of the International Neuropsychological Society, 2001, 7, 45-54. | 1.2 | 101 |
| 82 | Cerebral energy failure. Current Neurology and Neuroscience Reports, 2001, 1, 203-204. | 2.0 | 5 |
| 83 | Navajo neurohepatopathy: A mitochondrial DNA depletion syndrome?. Hepatology, 2001, 34, 116-120. | 3.6 | 57 |
| 84 | Glucose Transporter Type 1 Deficiency Syndrome (Glut1DS): Methylxanthines Potentiate GLUT1 Haploinsufficiency In Vitro. Pediatric Research, 2001, 50, 254-260. | 1.1 | 58 |
| 85 | Mutational analysis of GLUT1 (SLC2A1) in Glut-1 Deficiency Syndrome. Human Mutation, 2000, 16, 224-231. | 1.1 | 143 |
| 86 | Defective glucose transport across brain tissue barriers: a newly recognized neurological syndrome. Neurochemical Research, 1999, 24, 587-594. | 1.6 | 100 |
| 87 | Solving the COX Puzzle. Annals of Neurology, 1999, 46, 142-143. | 2.8 | 2 |
| 88 | Erythrocyte 3-O-methyl-D-glucose uptake assay for diagnosis of glucose-transporter-protein syndrome. Journal of Clinical Laboratory Analysis, 1999, 13, 116-121. | 0.9 | 86 |
| 89 | Erythrocyte 3-O-methyl-D-glucose uptake assay for diagnosis of glucose-transporter-protein syndrome. , 1999, 13, 116. | | 1 |
| 90 | GLUT1-Deficiency: Barbiturates Potentiate Haploinsufficiency in Vitro. Pediatric Research, 1999, 46, 677-677. | 1.1 | 59 |

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|---|---|---|---|
| 91 | Leigh syndrome: Historical perspective and clinical variations. BioFactors, 1998, 7, 269-271. | 2.6 | 15 |
| 92 | l-Carnitine Supplementation in Childhood Epilepsy: Current Perspectives. Epilepsia, 1998, 39, 1216-1225. | 2.6 | 172 |
| 93 | GLUT-1 deficiency syndrome caused by haploinsufficiency of the blood-brain barrier hexose carrier. Nature Genetics, 1998, 18, 188-191. | 9.4 | 349 |
| 94 | The Ketogenic Diet Revisited: Back to the Future. Epilepsia, 1997, 38, 743-749. | 2.6 | 124 |
| 95 | Deficient Muscle Carnitine Transport in Primary Carnitine Deficiency. Pediatric Research, 1997, 42, 583-587. | 1.1 | 38 |
| 96 | Clinical Heterogeneity Associated with the Mitochondrial DNA T8993C Point Mutation. Pediatric Research, 1996, 39, 914-917. | 1.1 | 56 |
| 97 | Pyruvate dehydrogenase deficiency: Molecular basis for intrafamilial heterogeneity. Annals of Neurology, 1994, 36, 83-89. | 2.8 | 27 |
| 98 | Cerebrospinal fluid polyamines: Biochemical markers of malignant childhood brain tumors. Annals of Neurology, 1986, 19, 360-364. | 2.8 | 13 |
| 99 | Cholesterol and phospholipids in cultured skin fibroblasts from patients with dystonia. Annals of Neurology, 1984, 16, 250-252. | 2.8 | 8 |
| 100 | Mitochondrial myopathy, encephalopathy, lactic acidosis, and strokelike episodes: A distinctive clinical syndrome. Annals of Neurology, 1984, 16, 481-488. | 2.8 | 1,128 |