Basil T Darras

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3131286/publications.pdf

Version: 2024-02-01

197 papers	11,961 citations	30047 54 h-index	30058 103 g-index
198 all docs	198 docs citations	198 times ranked	8262 citing authors

#	Article	IF	Citations
1	Nusinersen versus Sham Control in Infantile-Onset Spinal Muscular Atrophy. New England Journal of Medicine, 2017, 377, 1723-1732.	13.9	1,533
2	Nusinersen versus Sham Control in Later-Onset Spinal Muscular Atrophy. New England Journal of Medicine, 2018, 378, 625-635.	13.9	977
3	Results from a phase 1 study of nusinersen (ISIS-SMN _{Rx}) in children with spinal muscular atrophy. Neurology, 2016, 86, 890-897.	1.5	506
4	Observational study of spinal muscular atrophy type I and implications for clinical trials. Neurology, 2014, 83, 810-817.	1.5	367
5	Ataluren in patients with nonsense mutation Duchenne muscular dystrophy (ACT DMD): a multicentre, randomised, double-blind, placebo-controlled, phase 3 trial. Lancet, The, 2017, 390, 1489-1498.	6.3	365
6	Natural history of infantileâ€onset spinal muscular atrophy. Annals of Neurology, 2017, 82, 883-891.	2.8	276
7	An expanded version of the Hammersmith Functional Motor Scale for SMA II and III patients. Neuromuscular Disorders, 2007, 17, 693-697.	0.3	245
8	Risdiplam in Type 1 Spinal Muscular Atrophy. New England Journal of Medicine, 2021, 384, 915-923.	13.9	229
9	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
10	Nemaline Myopathy with Minicores Caused by Mutation of the CFL2 Gene Encoding the Skeletal Muscle Actinâ€“Binding Protein, Cofilin-2. American Journal of Human Genetics, 2007, 80, 162-167.	2.6	213
11	Prospective cohort study of spinal muscular atrophy types 2 and 3. Neurology, 2012, 79, 1889-1897.	1.5	207
12	Nusinersen in later-onset spinal muscular atrophy. Neurology, 2019, 92, e2492-e2506.	1.5	183
13	Revised upper limb module for spinal muscular atrophy: Development of a new module. Muscle and Nerve, 2017, 55, 869-874.	1.0	166
14	Treatment Algorithm for Infants Diagnosed with Spinal Muscular Atrophy through Newborn Screening. Journal of Neuromuscular Diseases, 2018, 5, 145-158.	1.1	148
15	Identification of KLHL41 Mutations Implicates BTB-Kelch-Mediated Ubiquitination as an Alternate Pathway to Myofibrillar Disruption in Nemaline Myopathy. American Journal of Human Genetics, 2013, 93, 1108-1117.	2.6	147
16	Validation of the Expanded Hammersmith Functional Motor Scale in Spinal Muscular Atrophy Type II and III. Journal of Child Neurology, 2011, 26, 1499-1507.	0.7	143
17	Spinal Muscular Atrophy: A Clinical and Research Update. Pediatric Neurology, 2012, 46, 1-12.	1.0	143
18	Observational Study of Spinal Muscular Atrophy Type 2 and 3. Archives of Neurology, 2011, 68, 779.	4.9	142

#	Article	IF	Citations
19	Patterns of disease progression in type 2 and 3 SMA: Implications for clinical trials. Neuromuscular Disorders, 2016, 26, 126-131.	0.3	142
20	Neurofilament as a potential biomarker for spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2019, 6, 932-944.	1.7	137
21	Risdiplam-Treated Infants with Type 1 Spinal Muscular Atrophy versus Historical Controls. New England Journal of Medicine, 2021, 385, 427-435.	13.9	137
22	Spinal Muscular Atrophies. Pediatric Clinics of North America, 2015, 62, 743-766.	0.9	119
23	Mutation spectrum in the large GTPase dynamin 2, and genotype-phenotype correlation in autosomal dominant centronuclear myopathy. Human Mutation, 2012, 33, 949-959.	1.1	115
24	Association of Duchenne Muscular Dystrophy With Autism Spectrum Disorder. Journal of Child Neurology, 2005, 20, 790-795.	0.7	111
25	Juvenile myasthenia gravis. Muscle and Nerve, 2009, 39, 423-431.	1.0	110
26	Novel and recurrent mutations in lamin A/C in patients with Emery-Dreifuss muscular dystrophy. American Journal of Medical Genetics Part A, 2001, 102, 359-367.	2.4	109
27	Validation of the Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP) Tj ETQq1 1 0.784314 rgBT /Overl	0.3	106
28	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
29	Content validity and clinical meaningfulness of the HFMSE in spinal muscular atrophy. BMC Neurology, 2017, 17, 39.	0.8	102
30	Developmental milestones in type I spinal muscular atrophy. Neuromuscular Disorders, 2016, 26, 754-759.	0.3	96
31	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
32	Characterizing spinal muscular atrophy with electrical impedance myography. Muscle and Nerve, 2010, 42, 915-921.	1.0	88
33	Overturning the Paradigm of Spinal Muscular Atrophy as Just a Motor Neuron Disease. Pediatric Neurology, 2020, 109, 12-19.	1.0	85
34	Assessing spinal muscular atrophy with quantitative ultrasound. Neurology, 2010, 75, 526-531.	1.5	82
35	Motor and cognitive assessment of infants and young boys with Duchenne Muscular Dystrophy: results from the Muscular Dystrophy Association DMD Clinical Research Network. Neuromuscular Disorders, 2013, 23, 529-539.	0.3	79
36	Posterior Spinal Fusion for Scoliosis in Duchenne Muscular Dystrophy Diminishes the Rate of Respiratory Decline. Spine, 2007, 32, 459-465.	1.0	76

#	Article	IF	Citations
37	The sensitivity of exome sequencing in identifying pathogenic mutations for LGMD in the United States. Journal of Human Genetics, 2017, 62, 243-252.	1.1	73
38	Prenatal Diagnosis and Detection of Carriers with DNA Probes in Duchenne's Muscular Dystrophy. New England Journal of Medicine, 1987, 316, 985-992.	13.9	71
39	Minimal training is required to reliably perform quantitative ultrasound of muscle. Muscle and Nerve, 2014, 50, 124-128.	1.0	70
40	An Integrated Safety Analysis of Infants and Children with Symptomatic Spinal Muscular Atrophy (SMA) Treated with Nusinersen in Seven Clinical Trials. CNS Drugs, 2019, 33, 919-932.	2.7	69
41	Mutations in the satellite cell gene MEGF10 cause a recessive congenital myopathy with minicores. Neurogenetics, 2012, 13, 115-124.	0.7	68
42	Revised Hammersmith Scale for spinal muscular atrophy: A SMA specific clinical outcome assessment tool. PLoS ONE, 2017, 12, e0172346.	1.1	67
43	X-linked myotubular myopathy. Neurology, 2019, 92, e1852-e1867.	1.5	66
44	Ambulatory function in spinal muscular atrophy: Age-related patterns of progression. PLoS ONE, 2018, 13, e0199657.	1.1	65
45	Comparison of Plasmapheresis and Intravenous Immunoglobulin as Maintenance Therapies for Juvenile Myasthenia Gravis. JAMA Neurology, 2014, 71, 575.	4.5	64
46	Serum Transaminase Levels in Boys With Duchenne and Becker Muscular Dystrophy. Pediatrics, 2011, 127, e132-e136.	1.0	63
47	The Longitudinal Course of Cardiomyopathy in Friedreichâ€™s Ataxia During Childhood. Pediatric Cardiology, 2009, 30, 306-310.	0.6	62
48	Childhood chronic inflammatory demyelinating polyradiculoneuropathy: Combined analysis of a large cohort and eleven published series. Neuromuscular Disorders, 2013, 23, 103-111.	0.3	62
49	Nusinersen improves walking distance and reduces fatigue in lateâ€onset spinal muscular atrophy. Muscle and Nerve, 2019, 60, 409-414.	1.0	62
50	Quantitative muscle ultrasound detects disease progression in Duchenne muscular dystrophy. Annals of Neurology, 2017, 81, 633-640.	2.8	61
51	Revised upper limb module for spinal muscular atrophy: 12â€‰month changes. Muscle and Nerve, 2019, 59, 426-430.	1.0	61
52	Outcome reliability in nonâ€Ambulatory Boys/Men with duchenne muscular dystrophy. Muscle and Nerve, 2015, 51, 522-532.	1.0	60
53	Electrical impedance myography in spinal muscular atrophy: A longitudinal study. Muscle and Nerve, 2012, 45, 642-647.	1.0	57
54	Inherited Myopathies and Muscular Dystrophies. Seminars in Neurology, 2008, 28, 250-259.	0.5	56

#	Article	IF	Citations
55	Developing standardized corticosteroid treatment for Duchenne muscular dystrophy. Contemporary Clinical Trials, 2017, 58, 34-39.	0.8	56
56	Quantitative muscle ultrasound in Duchenne muscular dystrophy: A comparison of techniques. Muscle and Nerve, 2015, 51, 207-213.	1.0	55
57	Association of Plastin 3 Expression With Disease Severity in Spinal Muscular Atrophy Only in Postpubertal Females. Archives of Neurology, 2010, 67, 1252-6.	4.9	53
58	Molecular genetics of Duchenne and Becker muscular dystrophy. Journal of Pediatrics, 1990, 117, 1-15.	0.9	52
59	Electrical impedance myography for assessment of Duchenne muscular dystrophy. Annals of Neurology, 2017, 81, 622-632.	2.8	52
60	Metabolic myopathies: a clinical approach; part I. Pediatric Neurology, 2000, 22, 87-97.	1.0	51
61	Cross-sectional Evaluation of Electrical Impedance Myography and Quantitative Ultrasound for the Assessment of Duchenne Muscular Dystrophy in a Clinical Trial Setting. Pediatric Neurology, 2014, 51, 88-92.	1.0	50
62	Diagnosis of pediatric neuromuscular disorders in the era of DNA analysis. Pediatric Neurology, 2000, 23, 289-300.	1.0	49
63	Adiposity is increased among high-functioning, non-ambulatory patients with spinal muscular atrophy. Neuromuscular Disorders, 2010, 20, 448-452.	0.3	47
64	Clinical and genetic characterization of AP4B1â€associated SPG47. American Journal of Medical Genetics, Part A, 2018, 176, 311-318.	0.7	47
65	Systemic nature of spinal muscular atrophy revealed by studying insurance claims. PLoS ONE, 2019, 14, e0213680.	1.1	47
66	Longitudinal natural history of type I spinal muscular atrophy: a critical review. Orphanet Journal of Rare Diseases, 2020, 15, 84.	1.2	45
67	Non-5q spinal muscular atrophies. Neurology, 2011, 77, 312-314.	1.5	43
68	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
69	Deflazacort versus prednisone/prednisolone for maintaining motor function and delaying loss of ambulation: A post HOC analysis from the ACT DMD trial. Muscle and Nerve, 2018, 58, 639-645.	1.0	42
70	Frequency of p53 Tumor Suppressor Gene Mutations in Human Primary Brain Tumors. Neurosurgery, 1993, 33, 824-831.	0.6	42
71	A Randomized, Double-Blind Trial of Lisinopril and Losartan for the Treatment of Cardiomyopathy in Duchenne Muscular Dystrophy. PLOS Currents, 2013, 5, .	1.4	42
72	Meta-analyses of ataluren randomized controlled trials in nonsense mutation Duchenne muscular dystrophy. Journal of Comparative Effectiveness Research, 2020, 9, 973-984.	0.6	41

#	Article	IF	Citations
73	Deflazacort vs prednisone treatment for Duchenne muscular dystrophy: A metaâ€analysis of disease progression rates in recent multicenter clinical trials. Muscle and Nerve, 2020, 61, 26-35.	1.0	40
74	SMA-MAP: A Plasma Protein Panel for Spinal Muscular Atrophy. PLoS ONE, 2013, 8, e60113.	1.1	40
75	Optimizing electrical impedance myography measurements by using a multifrequency ratio: A study in Duchenne muscular dystrophy. Clinical Neurophysiology, 2015, 126, 202-208.	0.7	39
76	Congenital Myasthenic Syndrome With Episodic Apnea. Pediatric Neurology, 2009, 41, 42-45.	1.0	38
77	Analysis of extracellular mRNA in human urine reveals splice variant biomarkers of muscular dystrophies. Nature Communications, 2018, 9, 3906.	5.8	38
78	Multifocal slowing of nerve conduction in metachromatic leukodystrophy. Muscle and Nerve, 2004, 29, 531-536.	1.0	37
79	One Year Outcome of Boys With Duchenne Muscular Dystrophy Using the Bayley-III Scales of Infant and Toddler Development. Pediatric Neurology, 2014, 50, 557-563.	1.0	36
80	Dystrophinopathies. Seminars in Neurology, 2015, 35, 369-384.	0.5	36
81	Evaluator Training and Reliability for SMA Global Nusinersen Trials1. Journal of Neuromuscular Diseases, 2018, 5, 159-166.	1.1	36
82	Respiratory Trajectories in Type 2 and 3 Spinal Muscular Atrophy in the iSMAC Cohort Study. Neurology, 2021, 96, e587-e599.	1.5	36
83	Nusinersen Treatment in Adults With Spinal Muscular Atrophy. Neurology: Clinical Practice, 2021, 11, e317-e327.	0.8	35
84	Frequency of p53 Tumor Suppressor Gene Mutations in Human Primary Brain Tumors. Neurosurgery, 1993, 33, 824-831.	0.6	34
85	The motor neuron response to SMN1 deficiency in spinal muscular atrophy. Muscle and Nerve, 2014, 49, 636-644.	1.0	34
86	Clinical Variability in Spinal Muscular Atrophy Type <scp>III</scp>. Annals of Neurology, 2020, 88, 1109-1117.	2.8	34
87	Clinical trials in spinal muscular atrophy. Current Opinion in Pediatrics, 2007, 19, 675-679.	1.0	32
88	Exome sequencing identifies a novel SMCHD1 mutation in facioscapulohumeral muscular dystrophy 2. Neuromuscular Disorders, 2013, 23, 975-980.	0.3	32
89	Diagnostic value of electromyography and muscle biopsy in arthrogryposis multiplex congenita. Annals of Neurology, 2003, 54, 790-795.	2.8	31
90	Clinical trial readiness in non-ambulatory boys and men with duchenne muscular dystrophy: MDA-DMD network follow-up. Muscle and Nerve, 2016, 54, 681-689.	1.0	29

#	Article	IF	Citations
91	Development of an academic disease registry for spinal muscular atrophy. Neuromuscular Disorders, 2019, 29, 794-799.	0.3	29
92	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. Brain, 2020, 143, 2929-2944.	3.7	29
93	Age related treatment effect in type II Spinal Muscular Atrophy pediatric patients treated with nusinersen. Neuromuscular Disorders, 2021, 31, 596-602.	0.3	29
94	Reldesemtiv in Patients with Spinal Muscular Atrophy: a Phase 2 Hypothesis-Generating Study. Neurotherapeutics, 2021, 18, 1127-1136.	2.1	28
95	Thalidomide neuropathy in childhood. Neuromuscular Disorders, 2005, 15, 172-176.	0.3	27
96	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
97	Compound heterozygosity of predicted loss-of-function DESvariants in a family with recessive desminopathy. BMC Medical Genetics, 2013, 14, 68.	2.1	26
98	Spinal muscular atrophy functional composite score: A functional measure in spinal muscular atrophy. Muscle and Nerve, 2015, 52, 942-947.	1.0	26
99	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
100	Thigh Muscle Volume Measured by Magnetic Resonance Imaging Is Stable Over a 6-Month Interval in Spinal Muscular Atrophy. Journal of Child Neurology, 2011, 26, 1252-1259.	0.7	25
101	Acute care pediatric electromyography. Muscle and Nerve, 2000, 23, S53-S62.	1.0	24
102	Autoimmune Neuromuscular Disorders in Childhood. Current Treatment Options in Neurology, 2011, 13, 590-607.	0.7	24
103	Machine learning algorithms to classify spinal muscular atrophy subtypes. Neurology, 2012, 79, 358-364.	1.5	23
104	Clinical correlates of charcotâ€"marieâ€"tooth disease in patients with pes cavus deformities. Muscle and Nerve, 2013, 47, 488-492.	1.0	22
105	Spinal Muscular Atrophies. , 2015, , 117-145.		22
106	Gain and loss of abilities in type II SMA: A 12-month natural history study. Neuromuscular Disorders, 2020, 30, 765-771.	0.3	22
107	Inefficient dystrophin expression after cord blood transplantation in Duchenne muscular dystrophy. Muscle and Nerve, 2010, 41, 746-750.	1.0	21
108	Lambert-Eaton Syndrome, an Unrecognized Treatable Pediatric Neuromuscular Disorder: Three Patients and Literature Review. Pediatric Neurology, 2014, 50, 11-17.	1.0	21

#	Article	IF	Citations
109	Precious SMA natural history data. Neurology, 2018, 91, 337-339.	1.5	21
110	Comprehensive nutritional and metabolic assessment in patients with spinal muscular atrophy: Opportunity for an individualized approach. Neuromuscular Disorders, 2018, 28, 512-519.	0.3	20
111	Muscle Volume Estimation by Magnetic Resonance Imaging in Spinal Muscular Atrophy. Journal of Child Neurology, 2011, 26, 309-317.	0.7	18
112	Quantitative Evaluation of Lower Extremity Joint Contractures in Spinal Muscular Atrophy: Implications for Motor Function. Pediatric Physical Therapy, 2018, 30, 209-215.	0.3	18
113	Clinical, neuroimaging, and molecular spectrum of TECPR2 â€associated hereditary sensory and autonomic neuropathy with intellectual disability. Human Mutation, 2021, 42, 762-776.	1.1	18
114	Dysphagia Phenotypes in Spinal Muscular Atrophy: The Past, Present, and Promise for the Future. American Journal of Speech-Language Pathology, 2021, 30, 1008-1022.	0.9	18
115	Different trajectories in upper limb and gross motor function in spinal muscular atrophy. Muscle and Nerve, 2021, 64, 552-559.	1.0	18
116	Referral and diagnostic trends in pediatric electromyography in the molecular era. Muscle and Nerve, 2014, 50, 244-249.	1.0	17
117	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
118	Muscle compression improves reliability of ultrasound echo intensity. Muscle and Nerve, 2018, 57, 423-429.	1.0	17
119	Current advances in drug development in spinal muscular atrophy. Current Opinion in Pediatrics, 2013, 25, 682-688.	1.0	16
120	Genetic Disorders Affecting the Motor Neuron. , 2017, , 1057-1064.		16
121	Exploring the relationship between electrical impedance myography and quantitative ultrasound parameters in Duchenne muscular dystrophy. Clinical Neurophysiology, 2019, 130, 515-520.	0.7	16
122	Massachusettsâ€™ Findings from Statewide Newborn Screening for Spinal Muscular Atrophy. International Journal of Neonatal Screening, 2021, 7, 26.	1.2	16
123	Assessing spinal muscular atrophy with quantitative ultrasound. Neurology, 2011, 76, 933-934.	1.5	15
124	Novel mutation in CNTNAP1 results in congenital hypomyelinating neuropathy. Muscle and Nerve, 2017, 55, 761-765.	1.0	15
125	Homozygous TRPV4 mutation causes congenital distal spinal muscular atrophy and arthrogryposis. Neurology: Genetics, 2019, 5, e312.	0.9	15
126	Exclusion of the gastrin-releasing peptide receptor (GRPR) locus as a candidate gene for Rett syndrome. , 1998, 78, 173-175.		14

#	Article	IF	Citations
127	Electrical impedance myography for reducing sample size in Duchenne muscular dystrophy trials. Annals of Clinical and Translational Neurology, 2020, 7, 4-14.	1.7	14
128	Automated DNA mutation detection using universal conditions direct sequencing: application to ten muscular dystrophy genes. BMC Genetics, 2009, 10, 66.	2.7	13
129	The Spectrum of Myotonic and Myopathic Disorders in a Pediatric Electromyography Laboratory Over 12 Years. Pediatric Neurology, 2012, 47, 97-100.	1.0	13
130	Inter-session reliability of electrical impedance myography in children in a clinical trial setting. Clinical Neurophysiology, 2015, 126, 1790-1796.	0.7	13
131	Physical therapy services received by individuals with spinal muscular atrophy (SMA). Journal of Pediatric Rehabilitation Medicine, 2016, 9, 35-44.	0.3	13
132	Revised upper limb module in type II and III spinal muscular atrophy: 24-month changes. Neuromuscular Disorders, 2022, 32, 36-42.	0.3	13
133	Electrophysiologic Evidence for Anterior Horn Cell Disease in Amyoplasia. Pediatric Neurology, 2010, 43, 142-147.	1.0	12
134	Pediatric sciatic neuropathy associated with neoplasms. Muscle and Nerve, 2011, 43, 183-188.	1.0	12
135	Composite Biomarkers for Assessing Duchenne Muscular Dystrophy: An Initial Assessment. Pediatric Neurology, 2015, 52, 202-205.	1.0	12
136	Quantitative Ultrasound Assessment of Duchenne Muscular Dystrophy Using Edge Detection Analysis. Journal of Ultrasound in Medicine, 2016, 35, 1889-1897.	0.8	12
137	Longitudinal Patterns of Thalidomide Neuropathy in Children and Adolescents. Journal of Pediatrics, 2016, 178, 227-232.	0.9	12
138	Loss of electrical anisotropy is an unrecognized feature of dystrophic muscle that may serve as a convenient index of disease status. Clinical Neurophysiology, 2016, 127, 3546-3551.	0.7	12
139	Spectrum of Nondystrophic Skeletal Muscle Channelopathies in Children. Pediatric Neurology, 2017, 70, 26-33.	1.0	12
140	Spectrum of Neuromuscular Disorders With HyperCKemia From a Tertiary Care Pediatric Neuromuscular Center. Journal of Child Neurology, 2018, 33, 389-396.	0.7	12
141	Acute Neuromuscular Disorders in the Pediatric Intensive Care Unit. Journal of Child Neurology, 2020, 35, 17-24.	0.7	12
142	A search for X-chromosome uniparental disomy and DNA rearrangements in the rett syndrome. Brain and Development, 1992, 14, 273-275.	0.6	11
143	Electrophysiologic features of fibular neuropathy in childhood and adolescence. Muscle and Nerve, 2017, 55, 693-697.	1.0	11
144	Recruitment & retention program for the NeuroNEXT SMA Biomarker Study: Super Babies for SMA!. Contemporary Clinical Trials Communications, 2018, 11, 113-119.	0.5	11

#	Article	IF	Citations
145	Neuromuscular problems of the critically ill neonate and child. Seminars in Pediatric Neurology, 2004, 11, 147-168.	1.0	10
146	Child Neurology: Past, present, and future. Neurology, 2010, 74, e17-9.	1.5	10
147	A slowly progressive form of limbâ€girdle muscular dystrophy type 2C associated with founder mutation in the SGCG gene in Puerto Rican Hispanics. Molecular Genetics & Genomic Medicine, 2015, 3, 92-98.	0.6	10
148	Mitochondrial Membrane Proteinâ€“Associated Neurodegeneration Mimicking Juvenile Amyotrophic LateralÂSclerosis. Pediatric Neurology, 2016, 64, 83-86.	1.0	10
149	Yeo and Darras: Extraneuronal Phenotypes of Spinal Muscular Atrophy. Annals of Neurology, 2021, 89, 24-26.	2.8	10
150	Diagnostic capabilities of nanopore longâ€read sequencing in muscular dystrophy. Annals of Clinical and Translational Neurology, 2022, 9, 1302-1309.	1.7	10
151	Dystrophinopathies. , 2015, , 551-592.		9
152	Identification of a pathogenic mutation in ATP2A1 via in silico analysis of exome data for cryptic aberrant splice sites. Molecular Genetics & Genomic Medicine, 2019, 7, e552.	0.6	9
153	The Value of Imaging and Composition-Based Biomarkers in Duchenne Muscular Dystrophy Clinical Trials. Neurotherapeutics, 2020, 17, 142-152.	2.1	9
154	A novel homozygous splice-site mutation in the SPTBN4 gene causes axonal neuropathy without intellectual disability. European Journal of Medical Genetics, 2020, 63, 103826.	0.7	9
155	Putting the patient first: The validity and value of surface-based electrical impedance myography techniques. Clinical Neurophysiology, 2021, 132, 1752-1753.	0.7	9
156	Ethical Perspectives on Treatment Options with Spinal Muscular Atrophy Patients. Annals of Neurology, 2022, 91, 305-316.	2.8	9
157	Identification of a novel truncating mutation (S171X) in the Emerin gene in five members of a caucasian American family with Emery-Dreifuss muscular dystrophy. Human Mutation, 2000, 16, 94-94.	1.1	7
158	Functional Mixed-Effects Modeling of Longitudinal Duchenne Muscular Dystrophy Electrical Impedance Myography Data Using State-Space Approach. IEEE Transactions on Biomedical Engineering, 2019, 66, 1761-1768.	2.5	7
159	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
160	Multicenter Consensus Approach to Evaluation of Neonatal Hypotonia in the Genomic Era: A Review. JAMA Neurology, 2022, 79, 405.	4.5	7
161	Assessing electrical impedance alterations in spinal muscular atrophy via the finite element method. , 2011, 2011, 1871-4.		6
162	Old measures and new scores in spinal muscular atrophy patients. Muscle and Nerve, 2015, 52, 435-437.	1.0	6

#	Article	IF	Citations
163	Medical management of muscle weakness in Duchenne muscular dystrophy. PLoS ONE, 2020, 15, e0240687.	1.1	6
164	Seven-Year Experience From the National Institute of Neurological Disorders and Strokeâ€“Supported Network for Excellence in Neuroscience Clinical Trials. JAMA Neurology, 2020, 77, 755.	4.5	6
165	Meta-analyses of deflazacort versus prednisone/prednisolone in patients with nonsense mutation Duchenne muscular dystrophy. Journal of Comparative Effectiveness Research, 2021, 10, 1337-1347.	0.6	6
166	Pediatric monomelic amyotrophy: Evidence for poliomyelitis in vulnerable populations. Muscle and Nerve, 2009, 40, 860-863.	1.0	5
167	Electrophysiologic Features of Radial Neuropathy in Childhood and Adolescence. Pediatric Neurology, 2018, 81, 14-18.	1.0	5
168	Nusinersen for Patients With Spinal Muscular Atrophy: 1415 Doses via an Interdisciplinary Institutional Approach. Pediatric Neurology, 2022, 132, 33-40.	1.0	5
169	Electrophysiologic features of ulnar neuropathy in childhood and adolescence. Clinical Neurophysiology, 2017, 128, 751-755.	0.7	4
170	Health related quality of life in young, steroid-naÃ¯ve boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2021, 31, 1161-1168.	0.3	4
171	Exclusion of growth factor gene mutations as a common cause of Sotos syndrome. American Journal of Medical Genetics Part A, 2001, 98, 101-102.	2.4	3
172	Predicting hearing loss in facioscapulohumeral muscular dystrophy. Neurology, 2013, 81, 1370-1371.	1.5	3
173	Force-controlled ultrasound to measure passive mechanical properties of muscle in Duchenne muscular dystrophy. , 2016, 2016, 2865-2868.		3
174	Muscle Involvement and Restricted Disorders. , 2018, , 922-970.e15.		3
175	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
176	More can be less: SMN1 gene duplications are associated with sporadic ALS. Neurology, 2012, 78, 770-771.	1.5	2
177	NeuroNEXT is at your service. Annals of Neurology, 2017, 82, 857-858.	2.8	2
178	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
179	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
180	Child Neurology Residency Training in Neuromuscular Disorders. Seminars in Pediatric Neurology, 2011, 18, 116-119.	1.0	1

#	Article	IF	Citations
181	Teaching Neuro Images : Characteristic phenotype of Ullrich congenital muscular dystrophy. Neurology, 2013, 81, e44-5.	1.5	1
182	Neuromuscular Problems of the Critically Ill Neonate and Child. , 2015, , 885-903.		1
183	X-linked myotubular myopathy. Neurology, 2017, 89, 1316-1317.	1.5	1
184	[Editorial] Spinal muscular atrophy, pediatric virology and gene therapy: A challenge of modern weakness and hope. Experimental and Therapeutic Medicine, 2018, 15, 3671-3672.	0.8	1
185	Onasemnogene Abeparvovec Gene-Replacement Therapy (GRT) for Spinal Muscular Atrophy Type 1 (SMA1): Pivotal Phase 3 Study (STR1VE) Update. , 2019, 50, .		1
186	Dystrophinopathies. , 2020, , 413-436.		1
187	A tale of two diseases: spinal muscular atrophy and Pompe disease. The Lancet Child and Adolescent Health, 2022, 6, 2-3.	2.7	1
188	Reply to: The 4â€Copy Conundrum in the Treatment of Infants with Spinal Muscular Atrophy. Annals of Neurology, 2022, 91, 892-892.	2.8	1
189	Peripheral Neuropathies in Infants and Children: Polyneuropathies, Mononeuropathies, Plexopathies, and Radiculopathies. , 2005, , 2707-2753.		0
190	Neuromuscular disorders. Current Opinion in Pediatrics, 2013, 25, 674-675.	1.0	0
191	Reply. Muscle and Nerve, 2014, 50, 458-459.	1.0	0
192	Congenital myopathies. Neurology, 2015, 84, 15-16.	1.5	0
193	Levels Above Lower Motor Neuron to Neuromuscular Junction. , 2018, , 887-921.e11.		0
194	Response to â€œThe Spectrum of Neuromuscular Disorders Admitted to a Pediatric Intensive Care Unit Is Broader Than Anticipatedâ€• Journal of Child Neurology, 2020, 35, 302-303.	0.7	0
195	Leveraging Natural History Data in One- and Two-Arm Hierarchical Bayesian Studies of Rare Disease Progression. Statistics in Biosciences, 0, , 1.	0.6	0
196	Motor Neuron Disease. , 2017, , 199-220.		0
197	131â€... Pooled safety data from the risdiplam clinical trial development programme. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, A142.1-A142.	0.9	0