Anne M Connolly

List of Publications by Year
in descending order

100 papers	6,854 citations	66343 42 h-index	62596 80 g-index
103 all docs	103 docs citations	103 times ranked	7302 citing authors

#	Article	IF	Citations
1	The DNA replication FoSTeS/MMBIR mechanism can generate genomic, genic and exonic complex rearrangements in humans. Nature Genetics, 2009, 41, 849-853.	21.4	382
2	Ataluren treatment of patients with nonsense mutation dystrophinopathy. Muscle and Nerve, 2014, 50, 477-487.	2.2	357
3	Mitochondrial neurogastrointestinal encephalomyopathy: An autosomal recessive disorder due to thymidine phosphorylase mutations. Annals of Neurology, 2000, 47, 792-800.	5.3	324
4	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.	13.7	306
5	Mutational spectrum of DMD mutations in dystrophinopathy patients: application of modern diagnostic techniques to a large cohort. Human Mutation, 2009, 30, 1657-1666.	2.5	279
6	Brain-Derived Neurotrophic Factor and Autoantibodies to Neural Antigens in Sera of Children with Autistic Spectrum Disorders, Landau-Kleffner Syndrome, and Epilepsy. Biological Psychiatry, 2006, 59, 354-363.	1.3	260
7	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.	10.2	227
8	Serum autoantibodies to brain in Landau-Kleffner variant, autism, and other neurologic disorders. Journal of Pediatrics, 1999, 134, 607-613.	1.8	217
9	Course and outcome of acute cerebellar ataxia. Annals of Neurology, 1994, 35, 673-679.	5.3	207
10	Consensus Statement on Standard of Care for Congenital Muscular Dystrophies. Journal of Child Neurology, 2010, 25, 1559-1581.	1.4	200
11	Safety, Tolerability, and Efficacy of Viltolarsen in Boys With Duchenne Muscular Dystrophy Amenable to Exon 53 Skipping. JAMA Neurology, 2020, 77, 982.	9.0	169
12	Treatment Algorithm for Infants Diagnosed with Spinal Muscular Atrophy through Newborn Screening. Journal of Neuromuscular Diseases, 2018, 5, 145-158.	2.6	148
13	Consensus Statement on Standard of Care for Congenital Myopathies. Journal of Child Neurology, 2012, 27, 363-382.	1.4	147
14	Clinical and genetic characterization of manifesting carriers of DMD mutations. Neuromuscular Disorders, 2010, 20, 499-504.	0.6	136
15	Compositional analysis of muscle in boys with Duchenne muscular dystrophy using MR imaging. Skeletal Radiology, 2005, 34, 140-148.	2.0	132
16	High dose weekly oral prednisone improves strength in boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2002, 12, 917-925.	0.6	125
17	Serum autoantibodies in childhood opsoclonus-myoclonus syndrome: An analysis of antigenic targets in neural tissues. Journal of Pediatrics, 1997, 130, 878-884.	1.8	121
18	Three mouse models of muscular dystrophy: the natural history of strength and fatigue in dystrophin-, dystrophin/utrophin-, and laminin Î±2-deficient mice. Neuromuscular Disorders, 2001, 11, 703-712.	0.6	116

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19	Novel mutations expand the clinical spectrum of DYNC1H1 -associated spinal muscular atrophy. Neurology, 2015, 84, 668-679.	1.1	106
20	Childhood chronic inflammatory demyelinating neuropathies. Neurology, 1996, 47, 98-102.	1.1	104
21	Nonsense mutation-associated Becker muscular dystrophy: interplay between exon definition and splicing regulatory elements within the DMD gene. Human Mutation, 2011, 32, 299-308.	2.5	103
22	Impact of congenital talipes equinovarus etiology on treatment outcomes. Developmental Medicine and Child Neurology, 2008, 50, 498-502.	2.1	99
23	Childhood amyotrophic lateral sclerosis caused by excess sphingolipid synthesis. Nature Medicine, 2021, 27, 1197-1204.	30.7	96
24	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	5.3	93
25	Myosin binding protein C1: a novel gene for autosomal dominant distal arthrogryposis type 1. Human Molecular Genetics, 2010, 19, 1165-1173.	2.9	91
26	CINRG randomized controlled trial of creatine and glutamine in Duchenne muscular dystrophy. Annals of Neurology, 2005, 58, 151-155.	5.3	89
27	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.	2.6	89
28	Longitudinal pulmonary function testing outcome measures in Duchenne muscular dystrophy: Long-term natural history with and without glucocorticoids. Neuromuscular Disorders, 2018, 28, 897-909.	0.6	83
29	Gene Therapy for Spinal Muscular Atrophy: Safety and Early Outcomes. Pediatrics, 2020, 146, .	2.1	82
30	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.	0.6	79
31	Novel Mutations Widen the Phenotypic Spectrum of Slow Skeletal/Î²-Cardiac Myosin (MYH7) Distal Myopathy. Human Mutation, 2014, 35, 868-879.	2.5	79
32	Distribution of ten laminin chains in dystrophic and regenerating muscles. Neuromuscular Disorders, 1999, 9, 423-433.	0.6	77
33	Long-term treatment with eteplirsen in nonambulatory patients with Duchenne muscular dystrophy. Medicine (United States), 2019, 98, e15858.	1.0	61
34	Outcome reliability in nonâ€Ambulatory Boys/Men with duchenne muscular dystrophy. Muscle and Nerve, 2015, 51, 522-532.	2.2	60
35	Rapamycin nanoparticles target defective autophagy in muscular dystrophy to enhance both strength and cardiac function. FASEB Journal, 2014, 28, 2047-2061.	0.5	59
36	Weekly oral prednisolone improves survival and strength in malemdx mice. Muscle and Nerve, 2007, 35, 43-48.	2.2	57

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37	Quantitative ultrasound using backscatter analysis in Duchenne and Becker muscular dystrophy. Neuromuscular Disorders, 2010, 20, 805-809.	0.6	55
38	The care of patients with Duchenne, Becker, and other muscular dystrophies in the <scp>COVID</scp>â€19 pandemic. Muscle and Nerve, 2020, 62, 41-45.	2.2	54
39	Open-Label Evaluation of Eteplirsen in Patients with Duchenne Muscular Dystrophy Amenable to Exon 51 Skipping: PROMOVI Trial. Journal of Neuromuscular Diseases, 2021, 8, 989-1001.	2.6	50
40	Chronic inflammatory demyelinating polyneuropathy in childhood. Pediatric Neurology, 2001, 24, 177-182.	2.1	49
41	Electrical impedance myography in duchenne muscular dystrophy and healthy controls: A multicenter study of reliability and validity. Muscle and Nerve, 2015, 52, 592-597.	2.2	49
42	Primary ?-sarcoglycan deficiency responsive to immunosuppression over three years. , 1998, 21, 1549-1553.		46
43	Feasibility and Reproducibility of Echocardiographic Measures in Children with Muscular Dystrophies. Journal of the American Society of Echocardiography, 2015, 28, 999-1008.	2.8	45
44	<scp>MORC</scp>2 mutations cause axonal <scp>C</scp>harcotâ€“<scp>M</scp>arieâ€“<scp>T</scp>ooth disease with pyramidal signs. Annals of Neurology, 2016, 79, 419-427.	5.3	44
45	DMD Trp3X nonsense mutation associated with a founder effect in North American families with mild Becker muscular dystrophy. Neuromuscular Disorders, 2009, 19, 743-748.	0.6	43
46	Muscle ultrasound quantifies disease progression over time in infants and young boys with duchenne muscular dystrophy. Muscle and Nerve, 2015, 52, 334-338.	2.2	42
47	Pulmonary Endpoints in Duchenne Muscular Dystrophy. A Workshop Summary. American Journal of Respiratory and Critical Care Medicine, 2017, 196, 512-519.	5.6	39
48	CINRG pilot trial of coenzyme Q10 in steroidâ€treated duchenne muscular dystrophy. Muscle and Nerve, 2011, 44, 174-178.	2.2	38
49	Cardiac pathology exceeds skeletal muscle pathology in two cases of limbâ€girdle muscular dystrophy type 2I. Muscle and Nerve, 2009, 40, 883-889.	2.2	37
50	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.	2.1	36
51	Repeated intravenous cardiosphere-derived cell therapy in late-stage Duchenne muscular dystrophy (HOPE-2): a multicentre, randomised, double-blind, placebo-controlled, phase 2 trial. Lancet, The, 2022, 399, 1049-1058.	13.7	36
52	Poliomyelitis-like syndrome associated with Epstein-Barr virus infection. Pediatric Neurology, 1999, 20, 235-237.	2.1	35
53	Spinal Muscular Atrophy. Seminars in Pediatric Neurology, 2021, 37, 100878.	2.0	35
54	A recurrent de novo CTBP1 mutation is associated with developmental delay, hypotonia, ataxia, and tooth enamel defects. Neurogenetics, 2016, 17, 173-178.	1.4	32

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55	Twiceâ€weekly glucocorticosteroids in infants and young boys with Duchenne muscular dystrophy. Muscle and Nerve, 2019, 59, 650-657.	2.2	32
56	Sensitive Ultrasonic Detection of Dystrophic Skeletal Muscle in Patients with Duchenne Muscular Dystrophy Using an Entropy-Based Signal Receiver. Ultrasound in Medicine and Biology, 2007, 33, 1236-1243.	1.5	31
57	Newborn screening for Duchenne muscular dystrophy in China: follow-up diagnosis and subsequent treatment. World Journal of Pediatrics, 2017, 13, 197-201.	1.8	31
58	Spinal muscular atrophy care in the COVIDâ€19 pandemic era. Muscle and Nerve, 2020, 62, 46-49.	2.2	31
59	Complement 3 deficiency and oral prednisolone improve strength and prolong survival of laminin Î±2-deficient mice. Journal of Neuroimmunology, 2002, 127, 80-87.	2.3	29
60	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.	2.2	29
61	Reldesemtiv in Patients with Spinal Muscular Atrophy: a Phase 2 Hypothesis-Generating Study. Neurotherapeutics, 2021, 18, 1127-1136.	4.4	28
62	Skeletal Muscle Abnormalities and Genetic Factors Related to Vertical Talus. Clinical Orthopaedics and Related Research, 2011, 469, 1167-1174.	1.5	27
63	Testing preexisting antibodies prior to AAV gene transfer therapy: rationale, lessons and future considerations. Molecular Therapy - Methods and Clinical Development, 2022, 25, 74-83.	4.1	27
64	Congenital Myasthenic Syndrome: Presentation, Electrodiagnosis, and Muscle Biopsy. Journal of Child Neurology, 2004, 19, 175-182.	1.4	25
65	Evidence-Based Decision Support for Neurological Diagnosis Reduces Errors and Unnecessary Workup. Journal of Child Neurology, 2014, 29, 487-492.	1.4	25
66	A Combined Prospective and Retrospective Comparison of Long-Term Functional Outcomes Suggests Delayed Loss of Ambulation and Pulmonary Decline with Long-Term Eteplirsen Treatment. Journal of Neuromuscular Diseases, 2022, 9, 39-52.	2.6	24
67	Strength and corticosteroid responsiveness of mdx mice is unchanged by RAG2 gene knockout. Neuromuscular Disorders, 2007, 17, 376-384.	0.6	23
68	Sensitive Ultrasonic Delineation of Steroid Treatment in Living Dystrophic Mice with Energy-Based and Entropy-Based Radio Frequency Signal Processing. IEEE Transactions on Ultrasonics, Ferroelectrics, and Frequency Control, 2007, 54, 2291-2299.	3.0	23
69	Longitudinal natural history in young boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2019, 29, 857-862.	0.6	23
70	Progress in treatment and newborn screening for Duchenne muscular dystrophy and spinal muscular atrophy. World Journal of Pediatrics, 2019, 15, 219-225.	1.8	21
71	Management of Adrenal Insufficiency Risk After Long-term Systemic Glucocorticoid Therapy in Duchenne Muscular Dystrophy: Clinical Practice Recommendations. Journal of Neuromuscular Diseases, 2019, 6, 31-41.	2.6	20
72	Quantitative muscle ultrasound measures rapid declines over time in children with SMA type 1. Journal of the Neurological Sciences, 2015, 358, 178-182.	0.6	19

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73	Clinical Follow-Up for Duchenne Muscular Dystrophy Newborn Screening: A Proposal. Muscle and Nerve, 2016, 54, 186-191.	2.2	19
74	Diagnosis and management of adult hereditary cardio-neuromuscular disorders: A model for the multidisciplinary care of complex genetic disorders. Trends in Cardiovascular Medicine, 2017, 27, 51-58.	4.9	19
75	Medical management of eosinophilic meningitis following bovine graft duraplasty for Chiari malformation Type I repair. Journal of Neurosurgery: Pediatrics, 2013, 12, 357-359.	1.3	17
76	Therapeutic Approaches for Duchenne Muscular Dystrophy: Old and New. Seminars in Pediatric Neurology, 2021, 37, 100877.	2.0	17
77	Intron mutations and early transcription termination in Duchenne and Becker muscular dystrophy. Human Mutation, 2022, 43, 511-528.	2.5	16
78	Multi-Omics Identifies Circulating miRNA and Protein Biomarkers for Facioscapulohumeral Dystrophy. Journal of Personalized Medicine, 2020, 10, 236.	2.5	15
79	Natural History of Steroid-Treated Young Boys With Duchenne Muscular Dystrophy Using the NSAA, 100m, and Timed Functional Tests. Pediatric Neurology, 2020, 113, 15-20.	2.1	14
80	Selective response to rituximab in a young child with MuSK-associated myasthenia gravis. Neuromuscular Disorders, 2015, 25, 651-652.	0.6	13
81	Intramuscular blood flow in Duchenne and Becker Muscular Dystrophy: Quantitative power Doppler sonography relates to disease severity. Clinical Neurophysiology, 2020, 131, 1-5.	1.5	12
82	Time is muscle: A recommendation for early treatment for preterm infants with spinal muscular atrophy. Muscle and Nerve, 2021, 64, 153-155.	2.2	11
83	Liquid formulation of pentoxifylline is a poorly tolerated treatment for duchenne dystrophy. Muscle and Nerve, 2011, 44, 170-173.	2.2	10
84	Laboratory monitoring of nusinersen safety. Muscle and Nerve, 2021, 63, 902-905.	2.2	9
85	RAG2 gene knockout in mice causes fatigue. Muscle and Nerve, 2007, 36, 471-476.	2.2	7
86	Corticosteroids Can Reduce the Severity of Scoliosis in Duchenne Muscular Dystrophy. Journal of Bone and Joint Surgery - Series A, 2013, 95, e86-1-2.	3.0	6
87	PMP22 exon 4 deletion causes ER retention of PMP22 and a gainâ€ofâ€function allele in CMT1E. Annals of Clinical and Translational Neurology, 2017, 4, 236-245.	3.7	6
88	Medical management of muscle weakness in Duchenne muscular dystrophy. PLoS ONE, 2020, 15, e0240687.	2.5	6
89	Clinical and Molecular Spectrum Associated with COL6A3 c.7447A>G p.(Lys2483Glu) Variant: Elucidating its Role in Collagen VI-related Myopathies. Journal of Neuromuscular Diseases, 2021, 8, 633-645.	2.6	6
90	Meta-analyses of deflazacort versus prednisone/prednisolone in patients with nonsense mutation Duchenne muscular dystrophy. Journal of Comparative Effectiveness Research, 2021, 10, 1337-1347.	1.4	6

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91	Validity and Reliability of the Neuromuscular Gross Motor Outcome. Pediatric Neurology, 2021, 122, 21-26.	2.1	5
92	Mutation analysis in emery-dreifuss muscular dystrophy. Pediatric Neurology, 1999, 21, 456-459.	2.1	4
93	Tracking diaphragm movement by using ultrasound to assess its strength. Journal of Physiology, 2016, 594, 7147-7148.	2.9	3
94	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.	1.9	3
95	Pulse oral corticosteroids in pediatric chronic inflammatory demyelinating polyneuropathy. Muscle and Nerve, 2020, 62, 705-709.	2.2	3
96	The impact of genotype on outcomes in individuals with Duchenne muscular dystrophy: A systematic review. Muscle and Nerve, 2022, 65, 266-277.	2.2	1
97	Response to comments by G. VrbovÃ¡. Neuromuscular Disorders, 2002, 12, 609.	0.6	0
98	Congenital Myasthenic Syndrome: Presentation, Electrodiagnosis, and Muscle Biopsy. Journal of Child Neurology, 2004, 19, 175-182.	1.4	0
99	An evaluation of onasemnogene abeparvovec for Spinal Muscular Atrophy (SMN1). Expert Opinion on Orphan Drugs, 0, , .	0.8	0
100	Neuromuscular disease - Gene transfer for children. Journal of International Child Neurology Association, 2021, 1, .	0.0	0