

# Kevin M Flanigan

## List of Publications by Year in descending order

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156  
papers

11,370  
citations

25034

57  
h-index

30922

102  
g-index

169  
all docs

169  
docs citations

169  
times ranked

10530  
citing authors

#	ARTICLE	IF	CITATIONS
1	Evidence-based path to newborn screening for duchenne muscular dystrophy. Annals of Neurology, 2012, 71, 304-313.	5.3	633
2	Eteplirsen for the treatment of Duchenne muscular dystrophy. Annals of Neurology, 2013, 74, 637-647.	5.3	630
3	The TREAT-NMD DMD Global Database: Analysis of More than 7,000 Duchenne Muscular Dystrophy Mutations. Human Mutation, 2015, 36, 395-402.	2.5	507
4	Distinctive patterns of microRNA expression in primary muscular disorders. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 17016-17021.	7.1	458
5	A phase I/II trial of MYO-201 in adult subjects with muscular dystrophy. Annals of Neurology, 2008, 63, 561-571.	5.3	407
6	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.	13.7	365
7	Ataluren treatment of patients with nonsense mutation dystrophinopathy. Muscle and Nerve, 2014, 50, 477-487.	2.2	357
8	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
9	Spinocerebellar Ataxia Type 31 Is Associated with Inserted Penta-Nucleotide Repeats Containing (TGGA)n. American Journal of Human Genetics, 2009, 85, 544-557.	6.2	260
10	Gentamicin-induced readthrough of stop codons in duchenne muscular dystrophy. Annals of Neurology, 2010, 67, 771-780.	5.3	238
11	Sequence specificity of aminoglycoside-induced stop codon readthrough: Potential implications for treatment of Duchenne muscular dystrophy. Annals of Neurology, 2000, 48, 164-169.	5.3	233
12	Duchenne and Becker Muscular Dystrophies. Neurologic Clinics, 2014, 32, 671-688.	1.8	227
13	Sustained alpha-sarcoglycan gene expression after gene transfer in limb-girdle muscular dystrophy, type 2D. Annals of Neurology, 2010, 68, 629-638.	5.3	214
14	LTBP4 genotype predicts age of ambulatory loss in duchenne muscular dystrophy. Annals of Neurology, 2013, 73, 481-488.	5.3	202
15	Phase 2a Study of Ataluren-Mediated Dystrophin Production in Patients with Nonsense Mutation Duchenne Muscular Dystrophy. PLoS ONE, 2013, 8, e81302.	2.5	201
16	A Phase 1/2a Follistatin Gene Therapy Trial for Becker Muscular Dystrophy. Molecular Therapy, 2015, 23, 192-201.	8.2	193
17	Expression profiling of FSHD muscle supports a defect in specific stages of myogenic differentiation. Human Molecular Genetics, 2003, 12, 2895-2907.	2.9	191
18	Rapid Direct Sequence Analysis of the Dystrophin Gene. American Journal of Human Genetics, 2003, 72, 931-939.	6.2	178

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19	Selenoprotein N is required for ryanodine receptor calcium release channel activity in human and zebrafish muscle. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2008, 105, 12485-12490.	7.1	166
20	Clinical and electrophysiologic features of CMT2A with mutations in the mitofusin 2 gene. <i>Neurology</i> , 2005, 65, 197-204.	1.1	160
21	Muscle MRI in Ullrich congenital muscular dystrophy and Bethlem myopathy. <i>Neuromuscular Disorders</i> , 2005, 15, 303-310.	0.6	154
22	Canine models of Duchenne muscular dystrophy and their use in therapeutic strategies. <i>Mammalian Genome</i> , 2012, 23, 85-108.	2.2	140
23	Effects of Angiotensin-Converting Enzyme Inhibitors and/or Beta Blockers on the Cardiomyopathy in Duchenne Muscular Dystrophy. <i>American Journal of Cardiology</i> , 2012, 110, 98-102.	1.6	137
24	Analysis of Dystrophin Deletion Mutations Predicts Age of Cardiomyopathy Onset in Becker Muscular Dystrophy. <i>Circulation: Cardiovascular Genetics</i> , 2009, 2, 544-551.	5.1	136
25	Clinical and genetic characterization of manifesting carriers of DMD mutations. <i>Neuromuscular Disorders</i> , 2010, 20, 499-504.	0.6	136
26	Proteomic identification of FHL1 as the protein mutated in human reducing body myopathy. <i>Journal of Clinical Investigation</i> , 2008, 118, 904-12.	8.2	126
27	Clinical Outcomes in Duchenne Muscular Dystrophy: A Study of 5345 Patients from the TREAT-NMD DMD Global Database. <i>Journal of Neuromuscular Diseases</i> , 2017, 4, 293-306.	2.6	125
28	Translation from a DMD exon 5 IRES results in a functional dystrophin isoform that attenuates dystrophinopathy in humans and mice. <i>Nature Medicine</i> , 2014, 20, 992-1000.	30.7	113
29	Congenital muscular dystrophy with rigid spine syndrome: A clinical, pathological, radiological, and genetic study. <i>Annals of Neurology</i> , 2000, 47, 152-161.	5.3	111
30	Readthrough of dystrophin stop codon mutations induced by aminoglycosides. <i>Annals of Neurology</i> , 2004, 55, 422-426.	5.3	103
31	Nonsense mutation-associated Becker muscular dystrophy: interplay between exon definition and splicing regulatory elements within the DMD gene. <i>Human Mutation</i> , 2011, 32, 299-308.	2.5	103
32	RNA Interference Inhibits DUX4-induced Muscle Toxicity In Vivo: Implications for a Targeted FSHD Therapy. <i>Molecular Therapy</i> , 2012, 20, 1417-1423.	8.2	101
33	Expanding the clinical, pathological and MRI phenotype of DNM2-related centronuclear myopathy. <i>Neuromuscular Disorders</i> , 2010, 20, 229-237.	0.6	100
34	<i>EPG5</i>-related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. <i>Brain</i> , 2016, 139, 765-781.	7.6	99
35	Anti-Dystrophin T Cell Responses in Duchenne Muscular Dystrophy: Prevalence and a Glucocorticoid Treatment Effect. <i>Human Gene Therapy</i> , 2013, 24, 797-806.	2.7	97
36	<i>DMD</i> pseudoexon mutations: splicing efficiency, phenotype, and potential therapy. <i>Annals of Neurology</i> , 2008, 63, 81-89.	5.3	95

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37	The TREAT-NMD Duchenne Muscular Dystrophy Registries: Conception, Design, and Utilization by Industry and Academia. <i>Human Mutation</i> , 2013, 34, 1449-1457.	2.5	94
38	Genetic characterization of a large, historically significant Utah kindred with facioscapulohumeral dystrophy. <i>Neuromuscular Disorders</i> , 2001, 11, 525-529.	0.6	91
39	Mitochondrial fusion and function in Charcot-Marie-Tooth type 2A patient fibroblasts with mitofusin 2 mutations. <i>Experimental Neurology</i> , 2008, 211, 115-127.	4.1	88
40	Clinical, histological and genetic characterization of reducing body myopathy caused by mutations in FHL1. <i>Brain</i> , 2009, 132, 452-464.	7.6	88
41	Dexmedetomidine and ketamine sedation for muscle biopsies in patients with Duchenne muscular dystrophy. <i>Paediatric Anaesthesia</i> , 2014, 24, 851-856.	1.1	88
42	Mitochondrial DNA depletion syndrome due to mutations in the RRM2B gene. <i>Neuromuscular Disorders</i> , 2008, 18, 453-459.	0.6	87
43	Recoding elements located adjacent to a subset of eukaryal selenocysteine-specifying UGA codons. <i>EMBO Journal</i> , 2005, 24, 1596-1607.	7.8	84
44	Utility of cystatin C to monitor renal function in duchenne muscular dystrophy. <i>Muscle and Nerve</i> , 2009, 40, 438-442.	2.2	84
45	Follistatin Gene Therapy for Sporadic Inclusion Body Myositis Improves Functional Outcomes. <i>Molecular Therapy</i> , 2017, 25, 870-879.	8.2	84
46	Motor and cognitive assessment of infants and young boys with Duchenne Muscular Dystrophy: results from the Muscular Dystrophy Association DMD Clinical Research Network. <i>Neuromuscular Disorders</i> , 2013, 23, 529-539.	0.6	79
47	Position of Glycine Substitutions in the Triple Helix of COL6A1, COL6A2, and COL6A3 is Correlated with Severity and Mode of Inheritance in Collagen VI Myopathies. <i>Human Mutation</i> , 2013, 34, 1558-1567.	2.5	79
48	Feasibility and Safety of Systemic rAAV9-hNAGLU Delivery for Treating Mucopolysaccharidosis IIIB: Toxicology, Biodistribution, and Immunological Assessments in Primates. <i>Human Gene Therapy Clinical Development</i> , 2014, 25, 72-84.	3.1	79
49	Dystrophin quantification. <i>Neurology</i> , 2014, 83, 2062-2069.	1.1	73
50	The Muscular Dystrophies. <i>Seminars in Neurology</i> , 2012, 32, 255-263.	1.4	71
51	Genetics and Emerging Treatments for Duchenne and Becker Muscular Dystrophy. <i>Pediatric Clinics of North America</i> , 2015, 62, 723-742.	1.8	71
52	Association Study of Exon Variants in the NF- $\kappa$ B and TGF $\beta$ 2 Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy. <i>American Journal of Human Genetics</i> , 2016, 99, 1163-1171.	6.2	71
53	Update in Duchenne and Becker muscular dystrophy. <i>Current Opinion in Neurology</i> , 2019, 32, 722-727.	3.6	71
54	DMD exon 1 truncating point mutations: Amelioration of phenotype by alternative translation initiation in exon 6. <i>Human Mutation</i> , 2009, 30, 633-640.	2.5	66

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55	A mutation in the <i>SEPN1</i> selenocysteine redefinition element (SRE) reduces selenocysteine incorporation and leads to <i>SEPN1</i> -related myopathy. <i>Human Mutation</i> , 2009, 30, 411-416.	2.5	62
56	Pharmacokinetics and safety of single doses of drisapersen in non-ambulant subjects with Duchenne muscular dystrophy: Results of a double-blind randomized clinical trial. <i>Neuromuscular Disorders</i> , 2014, 24, 16-24.	0.6	62
57	Outcome reliability in non-ambulatory Boys/Men with duchenne muscular dystrophy. <i>Muscle and Nerve</i> , 2015, 51, 522-532.	2.2	60
58	Developing standardized corticosteroid treatment for Duchenne muscular dystrophy. <i>Contemporary Clinical Trials</i> , 2017, 58, 34-39.	1.8	56
59	Long-range genomic regulators of <i>THBS1</i> and <i>LTBP4</i> modify disease severity in duchenne muscular dystrophy. <i>Annals of Neurology</i> , 2018, 84, 234-245.	5.3	53
60	DMD Trp3X nonsense mutation associated with a founder effect in North American families with mild Becker muscular dystrophy. <i>Neuromuscular Disorders</i> , 2009, 19, 743-748.	0.6	43
61	Cryptic MHC class I-binding peptides are revealed by aminoglycoside-induced stop codon read-through into the 3' UTR. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, 5670-5675.	7.1	43
62	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.	7.4	43
63	A Randomized, Double-Blind Trial of Lisinopril and Losartan for the Treatment of Cardiomyopathy in Duchenne Muscular Dystrophy. <i>PLOS Currents</i> , 2013, 5, .	1.4	42
64	Meta-analyses of ataluren randomized controlled trials in nonsense mutation Duchenne muscular dystrophy. <i>Journal of Comparative Effectiveness Research</i> , 2020, 9, 973-984.	1.4	41
65	How a patient advocacy group developed the first proposed draft guidance document for industry for submission to the U.S. Food and Drug Administration. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 82.	2.7	39
66	Clinical phenotypes as predictors of the outcome of skipping around <i>DMD</i> exon 45. <i>Annals of Neurology</i> , 2015, 77, 668-674.	5.3	38
67	Localization of the giant axonal neuropathy gene to chromosome 16q24. <i>Annals of Neurology</i> , 1998, 43, 143-148.	5.3	37
68	Targeted Exon Skipping to Correct Exon Duplications in the Dystrophin Gene. <i>Molecular Therapy - Nucleic Acids</i> , 2014, 3, e155.	5.1	37
69	One Year Outcome of Boys With Duchenne Muscular Dystrophy Using the Bayley-III Scales of Infant and Toddler Development. <i>Pediatric Neurology</i> , 2014, 50, 557-563.	2.1	36
70	Exome sequencing identifies a DNAJB6 mutation in a family with dominantly-inherited limb-girdle muscular dystrophy. <i>Neuromuscular Disorders</i> , 2014, 24, 431-435.	0.6	35
71	Abnormal expression of mu-crystallin in facioscapulohumeral muscular dystrophy. <i>Experimental Neurology</i> , 2007, 205, 583-586.	4.1	34
72	Identification of New Dystroglycan Complexes in Skeletal Muscle. <i>PLoS ONE</i> , 2013, 8, e73224.	2.5	34

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73	Twice-weekly glucocorticosteroids in infants and young boys with Duchenne muscular dystrophy. <i>Muscle and Nerve</i> , 2019, 59, 650-657.	2.2	32
74	Differential Prevalence of Antibodies Against Adeno-Associated Virus in Healthy Children and Patients with Mucopolysaccharidosis III: Perspective for AAV-Mediated Gene Therapy. <i>Human Gene Therapy Clinical Development</i> , 2017, 28, 187-196.	3.1	31
75	204th ENMC International Workshop on Biomarkers in Duchenne Muscular Dystrophy 24-26 January 2014, Naarden, The Netherlands. <i>Neuromuscular Disorders</i> , 2015, 25, 184-198.	0.6	30
76	Prenatal diagnosis of Ullrich congenital muscular dystrophy using haplotype analysis and collagen VI immunocytochemistry. <i>Prenatal Diagnosis</i> , 2004, 24, 440-444.	2.3	29
77	Clinical trial readiness in non-ambulatory boys and men with duchenne muscular dystrophy: MDA-DMD network follow-up. <i>Muscle and Nerve</i> , 2016, 54, 681-689.	2.2	29
78	Impaired regeneration in calpain-3 null muscle is associated with perturbations in mTORC1 signaling and defective mitochondrial biogenesis. <i>Skeletal Muscle</i> , 2017, 7, 27.	4.2	29
79	Lack of Toxicity in Nonhuman Primates Receiving Clinically Relevant Doses of an AAV9.U7snRNA Vector Designed to Induce <i>DMD</i> Exon 2 Skipping. <i>Human Gene Therapy</i> , 2021, 32, 882-894.	2.7	29
80	Dp412e: a novel human embryonic dystrophin isoform induced by BMP4 in early differentiated cells. <i>Skeletal Muscle</i> , 2015, 5, 40.	4.2	28
81	Placebo-controlled Phase 2 Trial of Drisapersen for Duchenne Muscular Dystrophy. <i>Annals of Clinical and Translational Neurology</i> , 2018, 5, 913-926.	3.7	28
82	Diagnostic Utility of Whole Exome Sequencing in the Neuromuscular Clinic. <i>Neuropediatrics</i> , 2019, 50, 096-102.	0.6	28
83	Variants in MED12L, encoding a subunit of the mediator kinase module, are responsible for intellectual disability associated with transcriptional defect. <i>Genetics in Medicine</i> , 2019, 21, 2713-2722.	2.4	28
84	The first exon duplication mouse model of Duchenne muscular dystrophy: A tool for therapeutic development. <i>Neuromuscular Disorders</i> , 2015, 25, 827-834.	0.6	27
85	Comparison of Serum rAAV Serotype-Specific Antibodies in Patients with Duchenne Muscular Dystrophy, Becker Muscular Dystrophy, Inclusion Body Myositis, or GNE Myopathy. <i>Human Gene Therapy</i> , 2017, 28, 737-746.	2.7	27
86	Efficient Skipping of Single Exon Duplications in DMD Patient-Derived Cell Lines Using an Antisense Oligonucleotide Approach. <i>Journal of Neuromuscular Diseases</i> , 2017, 4, 199-207.	2.6	27
87	Low-level dystrophin expression attenuating the dystrophinopathy phenotype. <i>Neuromuscular Disorders</i> , 2018, 28, 116-121.	0.6	27
88	Update in the Mucopolysaccharidoses. <i>Seminars in Pediatric Neurology</i> , 2021, 37, 100874.	2.0	27
89	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.	1.6	26
90	Sarcolemmal reorganization in facioscapulohumeral muscular dystrophy. <i>Annals of Neurology</i> , 2006, 59, 289-297.	5.3	25

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91	Knee extensor strength exhibits potential to predict function in sporadic inclusion-body myositis. <i>Muscle and Nerve</i> , 2012, 45, 163-168.	2.2	25
92	Proof of Concept of the Ability of the Kinect to Quantify Upper Extremity Function in Dystrophinopathy. <i>PLOS Currents</i> , 2013, 5, .	1.4	25
93	The ZZ Domain of Dystrophin in DMD: Making Sense of Missense Mutations. <i>Human Mutation</i> , 2014, 35, 257-264.	2.5	23
94	AGE-RELATED BIOLOGY AND DISEASES OF MUSCLE AND NERVE. <i>Neurologic Clinics</i> , 1998, 16, 659-669.	1.8	22
95	The TREAT-NMD advisory committee for therapeutics (TACT): an innovative de-risking model to foster orphan drug development. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 49.	2.7	21
96	Pre-clinical dose-escalation studies establish a therapeutic range for U7snRNA-mediated DMD exon 2 skipping. <i>Molecular Therapy - Methods and Clinical Development</i> , 2021, 21, 325-340.	4.1	21
97	A Practical Approach to Molecular Diagnostic Testing in Neuromuscular Diseases. <i>Physical Medicine and Rehabilitation Clinics of North America</i> , 2012, 23, 589-608.	1.3	19
98	A Comparative Study of N-glycolylneuraminic Acid (Neu5Gc) and Cytotoxic T Cell (CT) Carbohydrate Expression in Normal and Dystrophin-Deficient Dog and Human Skeletal Muscle. <i>PLoS ONE</i> , 2014, 9, e88226.	2.5	19
99	Recurrent central nervous system white matter changes in charcot-Marie-Tooth type X disease. <i>Muscle and Nerve</i> , 2014, 49, 451-454.	2.2	19
100	A GLP-Compliant Toxicology and Biodistribution Study: Systemic Delivery of an rAAV9 Vector for the Treatment of Mucopolysaccharidosis IIIB. <i>Human Gene Therapy Clinical Development</i> , 2015, 26, 228-242.	3.1	19
101	Clinical Phenotypes of DMD Exon 51 Skip Equivalent Deletions: A Systematic Review. <i>Journal of Neuromuscular Diseases</i> , 2020, 7, 217-229.	2.6	18
102	The Genotypic and Phenotypic Spectrum of <i>BICD2</i> Variants in Spinal Muscular Atrophy. <i>Annals of Neurology</i> , 2020, 87, 487-496.	5.3	18
103	Early-progressive dilated cardiomyopathy in a family with Becker muscular dystrophy related to a novel frameshift mutation in the dystrophin gene exon 27. <i>Journal of Human Genetics</i> , 2015, 60, 151-155.	2.3	17
104	Correlation of knee strength to functional outcomes in becker muscular dystrophy. <i>Muscle and Nerve</i> , 2013, 47, 550-554.	2.2	16
105	The 100-meter timed test: Normative data in healthy males and comparative pilot outcome data for use in Duchenne muscular dystrophy clinical trials. <i>Neuromuscular Disorders</i> , 2017, 27, 452-457.	0.6	16
106	Intron mutations and early transcription termination in Duchenne and Becker muscular dystrophy. <i>Human Mutation</i> , 2022, 43, 511-528.	2.5	16
107	Cerebral proton magnetic resonance spectroscopy of a patient with giant axonal neuropathy. <i>Brain and Development</i> , 2003, 25, 45-50.	1.1	15
108	Dystrophin as a therapeutic biomarker: Are we ignoring data from the past?. <i>Neuromuscular Disorders</i> , 2014, 24, 463-466.	0.6	15



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109	Reliability and validity of activeâ€seated: An outcome in dystrophinopathy. <i>Muscle and Nerve</i> , 2015, 52, 356-362.	2.2	15
110	Truncating variants in <i>UBAP1</i> associated with childhoodâ€onset nonsyndromic hereditary spastic paraplegia. <i>Human Mutation</i> , 2020, 41, 632-640.	2.5	15
111	A novel form of juvenile recessive ALS maps to loci on 6p25 and 21q22. <i>Neuromuscular Disorders</i> , 2009, 19, 279-287.	0.6	14
112	In-frame de novo mutation in <i>BICD2</i> in two patients with muscular atrophy and arthrogryposis. <i>Journal of Physical Education and Sports Management</i> , 2018, 4, a003160.	1.2	14
113	An Isolated Limb Infusion Method Allows for Broad Distribution of rAAVrh74.MCK.GALGT2 to Leg Skeletal Muscles in the Rhesus Macaque. <i>Molecular Therapy - Methods and Clinical Development</i> , 2018, 10, 89-104.	4.1	14
114	Natural History of Steroid-Treated Young Boys With Duchenne Muscular Dystrophy Using the NSAA, 100m, and Timed Functional Tests. <i>Pediatric Neurology</i> , 2020, 113, 15-20.	2.1	14
115	Evaluation of biomarkers for Sanfilippo syndrome. <i>Molecular Genetics and Metabolism</i> , 2019, 128, 68-74.	1.1	13
116	Cardiac Management in Neuromuscular Diseases. <i>Physical Medicine and Rehabilitation Clinics of North America</i> , 2012, 23, 855-868.	1.3	12
117	Modeling functional decline over time in sporadic inclusion body myositis. <i>Muscle and Nerve</i> , 2017, 55, 526-531.	2.2	12
118	X-linked muscular dystrophy in a Labrador Retriever strain: phenotypic and molecular characterisation. <i>Skeletal Muscle</i> , 2020, 10, 23.	4.2	12
119	Becker muscular dystrophy due to an inversion of exons 23 and 24 of the <i>DMD</i> gene. <i>Muscle and Nerve</i> , 2011, 44, 822-825.	2.2	11
120	Natural history of echocardiographic abnormalities in mucopolysaccharidosis III. <i>Molecular Genetics and Metabolism</i> , 2018, 124, 131-134.	1.1	11
121	N-terminal Î± Dystroglycan (Î±DG-N): AÂPotential Serum Biomarker for Duchenne Muscular Dystrophy. <i>Journal of Neuromuscular Diseases</i> , 2016, 3, 247-260.	2.6	10
122	General anesthesia with a native airway for patients with mucopolysaccharidosis type <scp>III</scp>. <i>Paediatric Anaesthesia</i> , 2017, 27, 370-376.	1.1	9
123	Lowâ€level expression of EPG5 leads to an attenuated Vici syndrome phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1207-1211.	1.2	9
124	Automated immunofluorescence analysis for sensitive and precise dystrophin quantification in muscle biopsies. <i>Neuropathology and Applied Neurobiology</i> , 2022, 48, .	3.2	9
125	Campptocormia as a late presentation in a manifesting carrier of duchenne muscular dystrophy. <i>Muscle and Nerve</i> , 2013, 47, 124-127.	2.2	8
126	Absence of Significant Off-Target Splicing Variation with a U7snRNA Vector Targeting <i>DMD</i> Exon 2 Duplications. <i>Human Gene Therapy</i> , 2021, 32, 1346-1359.	2.7	8



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127	Gene editing and modulation for Duchenne muscular dystrophy. Progress in Molecular Biology and Translational Science, 2021, 182, 225-255.	1.7	7
128	Precordial R Wave Height Does Not Correlate with Echocardiographic Findings in Boys with Duchenne Muscular Dystrophy. Congenital Heart Disease, 2013, 8, 561-567.	0.2	6
129	Clinicopathologic Conference: A Newborn With Hypotonia, Cleft Palate, Micrognathia, and Bilateral Club Feet. Pediatric Neurology, 2017, 74, 11-14.	2.1	6
130	Evaluating longitudinal therapy effects via the North Star Ambulatory Assessment. Muscle and Nerve, 2021, 64, 614-619.	2.2	6
131	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
132	Phenotypic Spectrum of Dystrophinopathy Due to Duchenne Muscular Dystrophy Exon 2 Duplications. Neurology, 2022, 98, .	1.1	6
133	McArdle's disease presenting as recurrent cryptogenic renal failure due to occult seizures. Muscle and Nerve, 2003, 28, 640-643.	2.2	5
134	Visual Diagnosis: Chest Pain in a Boy With Duchenne Muscular Dystrophy and Cardiomyopathy. Pediatrics in Review, 2014, 35, e64-e67.	0.4	5
135	Homozygous variants in <i>AMPD2</i> and <i>COL11A1</i> lead to a complex phenotype of pontocerebellar hypoplasia type 9 and Stickler syndrome type 2. American Journal of Medical Genetics, Part A, 2020, 182, 557-560.	1.2	5
136	Validity and Reliability of the Neuromuscular Gross Motor Outcome. Pediatric Neurology, 2021, 122, 21-26.	2.1	5
137	Recurrent Fat Embolic Strokes in a Patient With Duchenne Muscular Dystrophy With Long Bone Fractures and a Patent Foramen Ovale. Pediatric Neurology, 2016, 63, 76-79.	2.1	4
138	Young Becker Muscular Dystrophy Patients Demonstrate Fibrosis Associated With Abnormal Left Ventricular Ejection Fraction on Cardiac Magnetic Resonance Imaging. Circulation: Cardiovascular Imaging, 2019, 12, e008919.	2.6	4
139	Health related quality of life in young, steroid-naïve boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2021, 31, 1161-1168.	0.6	4
140	Sequence specificity of aminoglycoside-induced stop codon readthrough: Potential implications for treatment of Duchenne muscular dystrophy. , 2000, 48, 164.		4
141	Aminoglycosides and other nonsense suppression therapies for the treatment of dystrophinopathy. The Cochrane Library, 0, , .	2.8	2
142	Reassessing carrier status for dystrophinopathies. Neurology: Genetics, 2016, 2, e108.	1.9	2
143	Longitudinal MRI brain volume changes over one year in children with mucopolysaccharidosis types IIIA and IIIB. Molecular Genetics and Metabolism, 2021, 133, 193-200.	1.1	2
144	An unusual pathologic feature associated with dermatomyositis. Neuromuscular Disorders, 2006, 16, 391-393.	0.6	1

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145	Becker muscular dystrophy with widespread muscle hypertrophy and a non-sense mutation of exon 2. Neuromuscular Disorders, 2013, 23, 192.	0.6	1
146	Duchenne muscular dystrophy: meeting the therapeutic challenge. Lancet Neurology, The, 2016, 15, 785-787.	10.2	1
147	Response to Letter by Yilmaz et al Regarding Article, "Analysis of Dystrophin Deletion Mutations Predicts Age of Cardiomyopathy Onset in Becker Muscular Dystrophy". Circulation: Cardiovascular Genetics, 2010, 3, .	5.1	0
148	Diabetic Myonecrosis in a Cystic Fibrosis Patient. Respiratory Care, 2013, 58, e123-e125.	1.6	0
149	Sanfilippo syndrome registry project and natural history studies: an example of patients, parents and researchers collaborating for a cure. Orphanet Journal of Rare Diseases, 2014, 9, P7.	2.7	0
150	Differential prevalence of antibodies against adeno-associated virus in healthy children and patients with mucopolysaccharidosis III: perspective for AAV-mediated gene therapy. Human Gene Therapy Clinical Development, 2017, , .	3.1	0
151	Duchenne and Becker Muscular Dystrophies. , 2017, , 1106-1111.		0
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