Kevin M Flanigan

List of Publications by Year
in descending order

156	11,370	24978	30848
papers	citations	57 h-index	102 g-index

169	169	169	10530
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Automated immunofluorescence analysis for sensitive and precise dystrophin quantification in muscle biopsies. Neuropathology and Applied Neurobiology, 2022, 48, .	1.8	9
2	Intron mutations and early transcription termination in Duchenne and Becker muscular dystrophy. Human Mutation, 2022, 43, 511-528.	1.1	16
3	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
4	Phenotypic Spectrum of Dystrophinopathy Due to Duchenne Muscular Dystrophy Exon 2 Duplications. Neurology, 2022, 98, .	1.5	6
5	Letter by Duan et al Regarding Article, â€œTherapeutic Exon Skipping Through a CRISPR-Guided Cytidine Deaminase Rescues Dystrophic Cardiomyopathy In Vivoâ€• Circulation, 2022, 145, e872-e873.	1.6	0
6	Gene editing and modulation for Duchenne muscular dystrophy. Progress in Molecular Biology and Translational Science, 2021, 182, 225-255.	0.9	7
7	Update in the Mucopolysaccharidoses. Seminars in Pediatric Neurology, 2021, 37, 100874.	1.0	27
8	Direct Reprogramming of Human Fibroblasts into Myoblasts to Investigate Therapies for Neuromuscular Disorders. Journal of Visualized Experiments, 2021, , .	0.2	0
9	Pre-clinical dose-escalation studies establish a therapeutic range for U7snRNA-mediated DMD exon 2 skipping. Molecular Therapy - Methods and Clinical Development, 2021, 21, 325-340.	1.8	21
10	Longitudinal MRI brain volume changes over one year in children with mucopolysaccharidosis types IIIA and IIIB. Molecular Genetics and Metabolism, 2021, 133, 193-200.	0.5	2
11	Health related quality of life in young, steroid-naÃ¯ve boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2021, 31, 1161-1168.	0.3	4
12	Absence of Significant Off-Target Splicing Variation with a U7snRNA Vector Targeting DMD Exon 2 Duplications. Human Gene Therapy, 2021, 32, 1346-1359.	1.4	8
13	Validity and Reliability of the Neuromuscular Gross Motor Outcome. Pediatric Neurology, 2021, 122, 21-26.	1.0	5
14	Evaluating longitudinal therapy effects via the North Star Ambulatory Assessment. Muscle and Nerve, 2021, 64, 614-619.	1.0	6
15	Lack of Toxicity in Nonhuman Primates Receiving Clinically Relevant Doses of an AAV9.U7snRNA Vector Designed to Induce DMD Exon 2 Skipping. Human Gene Therapy, 2021, 32, 882-894.	1.4	29
16	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
17	Truncating variants in UBAP1 associated with childhoodâ€onset nonsyndromic hereditary spastic paraplegia. Human Mutation, 2020, 41, 632-640.	1.1	15
18	Homozygous variants in AMPD2 and COL11A1 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.	0.7	5

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19	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.	1.0	14
20	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
21	X-linked muscular dystrophy in a Labrador Retriever strain: phenotypic and molecular characterisation. Skeletal Muscle, 2020, 10, 23.	1.9	12
22	Clinical Phenotypes of DMD Exon 51 Skip Equivalent Deletions: A Systematic Review. Journal of Neuromuscular Diseases, 2020, 7, 217-229.	1.1	18
23	The Genotypic and Phenotypic Spectrum of BICD2 Variants in Spinal Muscular Atrophy. Annals of Neurology, 2020, 87, 487-496.	2.8	18
24	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.	1.3	4
25	Diagnostic Utility of Whole Exome Sequencing in the Neuromuscular Clinic. Neuropediatrics, 2019, 50, 096-102.	0.3	28
26	Twiceâ€weekly glucocorticosteroids in infants and young boys with Duchenne muscular dystrophy. Muscle and Nerve, 2019, 59, 650-657.	1.0	32
27	Variants in MED12L, encoding a subunit of the mediator kinase module, are responsible for intellectual disability associated with transcriptional defect. Genetics in Medicine, 2019, 21, 2713-2722.	1.1	28
28	Evaluation of biomarkers for Sanfilippo syndrome. Molecular Genetics and Metabolism, 2019, 128, 68-74.	0.5	13
29	Update in Duchenne and Becker muscular dystrophy. Current Opinion in Neurology, 2019, 32, 722-727.	1.8	71
30	Alternate Translational Initiation of Dystrophin: A Novel Therapeutic Approach. , 2019, , 371-382.		0
31	Low-level dystrophin expression attenuating the dystrophinopathy phenotype. Neuromuscular Disorders, 2018, 28, 116-121.	0.3	27
32	Lowâ€level expression of EPG5 leads to an attenuated Vici syndrome phenotype. American Journal of Medical Genetics, Part A, 2018, 176, 1207-1211.	0.7	9
33	In-frame de novo mutation in BICD2 in two patients with muscular atrophy and arthrogryposis. Journal of Physical Education and Sports Management, 2018, 4, a003160.	0.5	14
34	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
35	Longâ€range genomic regulators of THBS1 and LTBP4 modify disease severity in duchenne muscular dystrophy. Annals of Neurology, 2018, 84, 234-245.	2.8	53
36	Natural history of echocardiographic abnormalities in mucopolysaccharidosis III. Molecular Genetics and Metabolism, 2018, 124, 131-134.	0.5	11

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37	An Isolated Limb Infusion Method Allows for Broad Distribution of rAAVrh74.MCK.GALGT2 to Leg Skeletal Muscles in the Rhesus Macaque. Molecular Therapy - Methods and Clinical Development, 2018, 10, 89-104.	1.8	14
38	Placeboâ€controlled Phase 2 Trial of Drisapersen for Duchenne Muscular Dystrophy. Annals of Clinical and Translational Neurology, 2018, 5, 913-926.	1.7	28
39	Comparison of Serum rAAV Serotype-Specific Antibodies in Patients with Duchenne Muscular Dystrophy, Becker Muscular Dystrophy, Inclusion Body Myositis, or GNE Myopathy. Human Gene Therapy, 2017, 28, 737-746.	1.4	27
40	ClinicopathologicÂConference: A Newborn With Hypotonia, Cleft Palate, Micrognathia, and Bilateral Club Feet. Pediatric Neurology, 2017, 74, 11-14.	1.0	6
41	The 100-meter timed test: Normative data in healthy males and comparative pilot outcome data for use in Duchenne muscular dystrophy clinical trials. Neuromuscular Disorders, 2017, 27, 452-457.	0.3	16
42	Follistatin Gene Therapy for Sporadic Inclusion Body Myositis Improves Functional Outcomes. Molecular Therapy, 2017, 25, 870-879.	3.7	84
43	General anesthesia with a native airway for patients with mucopolysaccharidosis type <scp>III</scp>. Paediatric Anaesthesia, 2017, 27, 370-376.	0.6	9
44	Developing standardized corticosteroid treatment for Duchenne muscular dystrophy. Contemporary Clinical Trials, 2017, 58, 34-39.	0.8	56
45	Efficient Skipping of Single Exon Duplications in DMD Patient-Derived Cell Lines Using an Antisense Oligonucleotide Approach. Journal of Neuromuscular Diseases, 2017, 4, 199-207.	1.1	27
46	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, 28, 187-196.	3.2	31
47	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
48	Clinical Outcomes in Duchenne Muscular Dystrophy: A Study of 5345 Patients from the TREAT-NMD DMD Global Database. Journal of Neuromuscular Diseases, 2017, 4, 293-306.	1.1	125
49	Modeling functional decline over time in sporadic inclusion body myositis. Muscle and Nerve, 2017, 55, 526-531.	1.0	12
50	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, , .	3.2	0
51	Impaired regeneration in calpain-3 null muscle is associated with perturbations in mTORC1 signaling and defective mitochondrial biogenesis. Skeletal Muscle, 2017, 7, 27.	1.9	29
52	Duchenne and Becker Muscular Dystrophies. , 2017, , 1106-1111.		0
53	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
54	N-terminal Î± Dystroglycan (Î±DG-N): AÂPotential Serum Biomarker for Duchenne Muscular Dystrophy. Journal of Neuromuscular Diseases, 2016, 3, 247-260.	1.1	10

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55	Reassessing carrier status for dystrophinopathies. Neurology: Genetics, 2016, 2, e108.	0.9	2
56	Association Study of Exon Variants in the NF-Î²B and TGFÎ² Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy. American Journal of Human Genetics, 2016, 99, 1163-1171.	2.6	71
57	Duchenne muscular dystrophy: meeting the therapeutic challenge. Lancet Neurology, The, 2016, 15, 785-787.	4.9	1
58	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.	1.0	4
59	EPG5-related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. Brain, 2016, 139, 765-781.	3.7	99
60	Chest Pain in a Boy With Duchenne Muscular Dystrophy and Cardiomyopathy (Visual Diagnosis). , 2016, , 53-58.		0
61	How a patient advocacy group developed the first proposed draft guidance document for industry for submission to the U.S. Food and Drug Administration. Orphanet Journal of Rare Diseases, 2015, 10, 82.	1.2	39
62	Dp412e: a novel human embryonic dystrophin isoform induced by BMP4 in early differentiated cells. Skeletal Muscle, 2015, 5, 40.	1.9	28
63	A GLP-Compliant Toxicology and Biodistribution Study: Systemic Delivery of an rAAV9 Vector for the Treatment of Mucopolysaccharidosis IIIB. Human Gene Therapy Clinical Development, 2015, 26, 228-242.	3.2	19
64	Outcome reliability in nonâ€Ambulatory Boys/Men with duchenne muscular dystrophy. Muscle and Nerve, 2015, 51, 522-532.	1.0	60
65	The TREAT-NMD advisory committee for therapeutics (TACT): an innovative de-risking model to foster orphan drug development. Orphanet Journal of Rare Diseases, 2015, 10, 49.	1.2	21
66	The first exon duplication mouse model of Duchenne muscular dystrophy: A tool for therapeutic development. Neuromuscular Disorders, 2015, 25, 827-834.	0.3	27
67	Reliability and validity of activeâ€seated: An outcome in dystrophinopathy. Muscle and Nerve, 2015, 52, 356-362.	1.0	15
68	Clinical phenotypes as predictors of the outcome of skipping around <scp>DMD</scp> exon 45. Annals of Neurology, 2015, 77, 668-674.	2.8	38
69	The TREAT-NMD DMD Global Database: Analysis of More than 7,000 Duchenne Muscular Dystrophy Mutations. Human Mutation, 2015, 36, 395-402.	1.1	507
70	204th ENMC International Workshop on Biomarkers in Duchenne Muscular Dystrophy 24â€“26 January 2014, Naarden, The Netherlands. Neuromuscular Disorders, 2015, 25, 184-198.	0.3	30
71	Early-progressive dilated cardiomyopathy in a family with Becker muscular dystrophy related to a novel frameshift mutation in the dystrophin gene exon 27. Journal of Human Genetics, 2015, 60, 151-155.	1.1	17
72	Genetics and Emerging Treatments for Duchenne and Becker Muscular Dystrophy. Pediatric Clinics of North America, 2015, 62, 723-742.	0.9	71

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73	A Phase 1/2a Follistatin Gene Therapy Trial for Becker Muscular Dystrophy. Molecular Therapy, 2015, 23, 192-201.	3.7	193
74	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. PLoS ONE, 2014, 9, e88226.	1.1	19
75	Targeted Exon Skipping to Correct Exon Duplications in the Dystrophin Gene. Molecular Therapy - Nucleic Acids, 2014, 3, e155.	2.3	37
76	Dystrophin quantification. Neurology, 2014, 83, 2062-2069.	1.5	73
77	Dexmedetomidine and ketamine sedation for muscle biopsies in patients with <scp>D</scp>uchenne muscular dystrophy. Paediatric Anaesthesia, 2014, 24, 851-856.	0.6	88
78	Ataluren treatment of patients with nonsense mutation dystrophinopathy. Muscle and Nerve, 2014, 50, 477-487.	1.0	357
79	Visual Diagnosis: Chest Pain in a Boy With Duchenne Muscular Dystrophy and Cardiomyopathy. Pediatrics in Review, 2014, 35, e64-e67.	0.2	5
80	Feasibility and Safety of Systemic rAAV9-hNAGLU Delivery for Treating Mucopolysaccharidosis IIIB: Toxicology, Biodistribution, and Immunological Assessments in Primates. Human Gene Therapy Clinical Development, 2014, 25, 72-84.	3.2	79
81	Dystrophin as a therapeutic biomarker: Are we ignoring data from the past?. Neuromuscular Disorders, 2014, 24, 463-466.	0.3	15
82	Pharmacokinetics and safety of single doses of drisapersen in non-ambulant subjects with Duchenne muscular dystrophy: Results of a double-blind randomized clinical trial. Neuromuscular Disorders, 2014, 24, 16-24.	0.3	62
83	Cryptic MHC class I-binding peptides are revealed by aminoglycoside-induced stop codon read-through into the 3â€² UTR. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 5670-5675.	3.3	43
84	Exome sequencing identifies a DNAJB6 mutation in a family with dominantly-inherited limb-girdle muscular dystrophy. Neuromuscular Disorders, 2014, 24, 431-435.	0.3	35
85	The ZZ Domain of Dystrophin in DMD: Making Sense of Missense Mutations. Human Mutation, 2014, 35, 257-264.	1.1	23
86	Recurrent central nervous system white matter changes in charcotâ€“Marieâ€“Tooth type X disease. Muscle and Nerve, 2014, 49, 451-454.	1.0	19
87	Duchenne and Becker Muscular Dystrophies. Neurologic Clinics, 2014, 32, 671-688.	0.8	227
88	Translation from a DMD exon 5 IRES results in a functional dystrophin isoform that attenuates dystrophinopathy in humans and mice. Nature Medicine, 2014, 20, 992-1000.	15.2	113
89	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
90	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.	1.2	0

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91	Visual Diagnosis: Chest Pain in a Boy With Duchenne Muscular Dystrophy and Cardiomyopathy. Pediatrics in Review, 2014, 35, e64-e67.	0.2	0
92	Anti-Dystrophin T Cell Responses in Duchenne Muscular Dystrophy: Prevalence and a Glucocorticoid Treatment Effect. Human Gene Therapy, 2013, 24, 797-806.	1.4	97
93	Camptocormia as a late presentation in a manifesting carrier of duchenne muscular dystrophy. Muscle and Nerve, 2013, 47, 124-127.	1.0	8
94	Becker muscular dystrophy with widespread muscle hypertrophy and a non-sense mutation of exon 2. Neuromuscular Disorders, 2013, 23, 192.	0.3	1
95	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
96	<scp>LTBP4</scp> genotype predicts age of ambulatory loss in duchenne muscular dystrophy. Annals of Neurology, 2013, 73, 481-488.	2.8	202
97	Diabetic Myonecrosis in a Cystic Fibrosis Patient. Respiratory Care, 2013, 58, e123-e125.	0.8	0
98	The TREAT-NMD Duchenne Muscular Dystrophy Registries: Conception, Design, and Utilization by Industry and Academia. Human Mutation, 2013, 34, 1449-1457.	1.1	94
99	Position of Glycine Substitutions in the Triple Helix ofCOL6A1,COL6A2, andCOL6A3is Correlated with Severity and Mode of Inheritance in Collagen VI Myopathies. Human Mutation, 2013, 34, 1558-1567.	1.1	79
100	Eteplirsen for the treatment of Duchenne muscular dystrophy. Annals of Neurology, 2013, 74, 637-647.	2.8	630
101	Correlation of knee strength to functional outcomes in becker muscular dystrophy. Muscle and Nerve, 2013, 47, 550-554.	1.0	16
102	Precordial R Wave Height Does Not Correlate with Echocardiographic Findings in Boys with Duchenne Muscular Dystrophy. Congenital Heart Disease, 2013, 8, 561-567.	0.0	6
103	Identification of New Dystroglycan Complexes in Skeletal Muscle. PLoS ONE, 2013, 8, e73224.	1.1	34
104	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
105	Proof of Concept of the Ability of the Kinect to Quantify Upper Extremity Function in Dystrophinopathy. PLOS Currents, 2013, 5, .	1.4	25
106	Phase 2a Study of Ataluren-Mediated Dystrophin Production in Patients with Nonsense Mutation Duchenne Muscular Dystrophy. PLoS ONE, 2013, 8, e81302.	1.1	201
107	The Muscular Dystrophies. Seminars in Neurology, 2012, 32, 255-263.	0.5	71
108	A Practical Approach to Molecular Diagnostic Testing in Neuromuscular Diseases. Physical Medicine and Rehabilitation Clinics of North America, 2012, 23, 589-608.	0.7	19

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109	Cardiac Management in Neuromuscular Diseases. Physical Medicine and Rehabilitation Clinics of North America, 2012, 23, 855-868.	0.7	12
110	RNA Interference Inhibits DUX4-induced Muscle Toxicity In Vivo: Implications for a Targeted FSHD Therapy. Molecular Therapy, 2012, 20, 1417-1423.	3.7	101
111	Evidenceâ€based path to newborn screening for duchenne muscular dystrophy. Annals of Neurology, 2012, 71, 304-313.	2.8	633
112	Effects of Angiotensin-Converting Enzyme Inhibitors and/or Beta Blockers on the Cardiomyopathy in Duchenne Muscular Dystrophy. American Journal of Cardiology, 2012, 110, 98-102.	0.7	137
113	Knee extensor strength exhibits potential to predict function in sporadic inclusionâ€body myositis. Muscle and Nerve, 2012, 45, 163-168.	1.0	25
114	Canine models of Duchenne muscular dystrophy and their use in therapeutic strategies. Mammalian Genome, 2012, 23, 85-108.	1.0	140
115	Becker muscular dystrophy due to an inversion of exons 23 and 24 of the DMD gene. Muscle and Nerve, 2011, 44, 822-825.	1.0	11
116	Nonsense mutation-associated Becker muscular dystrophy: interplay between exon definition and splicing regulatory elements within the DMD gene. Human Mutation, 2011, 32, 299-308.	1.1	103
117	Gentamicinâ€induced readthrough of stop codons in duchenne muscular dystrophy. Annals of Neurology, 2010, 67, 771-780.	2.8	238
118	Sustained alphaâ€sarcoglycan gene expression after gene transfer in limbâ€girdle muscular dystrophy, type 2D. Annals of Neurology, 2010, 68, 629-638.	2.8	214
119	Response to Letter by Yilmaz et al Regarding Article, â€œAnalysis of Dystrophin Deletion Mutations Predicts Age of Cardiomyopathy Onset in Becker Muscular Dystrophyâ€. Circulation: Cardiovascular Genetics, 2010, 3, .	5.1	0
120	Expanding the clinical, pathological and MRI phenotype of DNM2-related centronuclear myopathy. Neuromuscular Disorders, 2010, 20, 229-237.	0.3	100
121	Clinical and genetic characterization of manifesting carriers of DMD mutations. Neuromuscular Disorders, 2010, 20, 499-504.	0.3	136
122	Clinical, histological and genetic characterization of reducing body myopathy caused by mutations in FHL1. Brain, 2009, 132, 452-464.	3.7	88
123	Analysis of Dystrophin Deletion Mutations Predicts Age of Cardiomyopathy Onset in Becker Muscular Dystrophy. Circulation: Cardiovascular Genetics, 2009, 2, 544-551.	5.1	136
124	A mutation in theSEPN1selenocysteine redefinition element (SRE) reduces selenocysteine incorporation and leads toSEPN1-related myopathy. Human Mutation, 2009, 30, 411-416.	1.1	62
125	DMDexon 1 truncating point mutations: Amelioration of phenotype by alternative translation initiation in exon 6. Human Mutation, 2009, 30, 633-640.	1.1	66
126	Mutational spectrum of DMD mutations in dystrophinopathy patients: application of modern diagnostic techniques to a large cohort. Human Mutation, 2009, 30, 1657-1666.	1.1	279

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127	Utility of cystatin C to monitor renal function in duchenne muscular dystrophy. Muscle and Nerve, 2009, 40, 438-442.	1.0	84
128	Spinocerebellar Ataxia Type 31 Is Associated with â€œInsertedâ€•Penta-Nucleotide Repeats Containing (TGGAA)n. American Journal of Human Genetics, 2009, 85, 544-557.	2.6	260
129	A novel form of juvenile recessive ALS maps to loci on 6p25 and 21q22. Neuromuscular Disorders, 2009, 19, 279-287.	0.3	14
130	DMD Trp3X nonsense mutation associated with a founder effect in North American families with mild Becker muscular dystrophy. Neuromuscular Disorders, 2009, 19, 743-748.	0.3	43
131	DMD pseudoexon mutations: splicing efficiency, phenotype, and potential therapy. Annals of Neurology, 2008, 63, 81-89.	2.8	95
132	A phase I/IItrial of MYOâ€029 in adult subjects with muscular dystrophy. Annals of Neurology, 2008, 63, 561-571.	2.8	407
133	Mitochondrial DNA depletion syndrome due to mutations in the RRM2B gene. Neuromuscular Disorders, 2008, 18, 453-459.	0.3	87
134	Mitochondrial fusion and function in Charcotâ€“Marieâ€“Tooth type 2A patient fibroblasts with mitofusin 2 mutations. Experimental Neurology, 2008, 211, 115-127.	2.0	88
135	Selenoprotein N is required for ryanodine receptor calcium release channel activity in human and zebrafish muscle. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 12485-12490.	3.3	166
136	Proteomic identification of FHL1 as the protein mutated in human reducing body myopathy. Journal of Clinical Investigation, 2008, 118, 904-12.	3.9	126
137	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.	3.3	458
138	Abnormal expression of mu-crystallin in facioscapulohumeral muscular dystrophy. Experimental Neurology, 2007, 205, 583-586.	2.0	34
139	An unusual pathologic feature associated with dermatomyositis. Neuromuscular Disorders, 2006, 16, 391-393.	0.3	1
140	Sarcolemmal reorganization in facioscapulohumeral muscular dystrophy. Annals of Neurology, 2006, 59, 289-297.	2.8	25
141	Recoding elements located adjacent to a subset of eukaryal selenocysteine-specifying UGA codons. EMBO Journal, 2005, 24, 1596-1607.	3.5	84
142	Clinical and electrophysiologic features of CMT2A with mutations in the mitofusin 2 gene. Neurology, 2005, 65, 197-204.	1.5	160
143	Muscle MRI in Ullrich congenital muscular dystrophy and Bethlem myopathy. Neuromuscular Disorders, 2005, 15, 303-310.	0.3	154
144	Prenatal diagnosis of Ullrich congenital muscular dystrophy using haplotype analysis and collagen VI immunocytochemistry. Prenatal Diagnosis, 2004, 24, 440-444.	1.1	29

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145	Readthrough of dystrophin stop codon mutations induced by aminoglycosides. Annals of Neurology, 2004, 55, 422-426.	2.8	103
146	Cerebral proton magnetic resonance spectroscopy of a patient with giant axonal neuropathy. Brain and Development, 2003, 25, 45-50.	0.6	15
147	McArdle's disease presenting as recurrent cryptogenic renal failure due to occult seizures. Muscle and Nerve, 2003, 28, 640-643.	1.0	5
148	Rapid Direct Sequence Analysis of the Dystrophin Gene. American Journal of Human Genetics, 2003, 72, 931-939.	2.6	178
149	Expression profiling of FSHD muscle supports a defect in specific stages of myogenic differentiation. Human Molecular Genetics, 2003, 12, 2895-2907.	1.4	191
150	Genetic characterization of a large, historically significant Utah kindred with facioscapulohumeral dystrophy. Neuromuscular Disorders, 2001, 11, 525-529.	0.3	91
151	Congenital muscular dystrophy with rigid spine syndrome: A clinical, pathological, radiological, and genetic study. Annals of Neurology, 2000, 47, 152-161.	2.8	111
152	Sequence specificity of aminoglycoside-induced stop codon readthrough: Potential implications for treatment of Duchenne muscular dystrophy. Annals of Neurology, 2000, 48, 164-169.	2.8	233
153	Sequence specificity of aminoglycoside-induced stop codon readthrough: Potential implications for treatment of Duchenne muscular dystrophy. , 2000, 48, 164.		4
154	Localization of the giant axonal neuropathy gene to chromosome 16q24. Annals of Neurology, 1998, 43, 143-148.	2.8	37
155	AGE-RELATED BIOLOGY AND DISEASES OF MUSCLE AND NERVE. Neurologic Clinics, 1998, 16, 659-669.	0.8	22
156	Aminoglycosides and other nonsense suppression therapies for the treatment of dystrophinopathy. The Cochrane Library, 0, , .	1.5	2