

Kevin M Flanigan

List of Publications by Year in descending order

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156
papers

11,370
citations

28736

57
h-index

35168

102
g-index

169
all docs

169
docs citations

169
times ranked

11337
citing authors

#	ARTICLE	IF	CITATIONS
1	Automated immunofluorescence analysis for sensitive and precise dystrophin quantification in muscle biopsies. <i>Neuropathology and Applied Neurobiology</i> , 2022, 48, .	1.8	9
2	Intron mutations and early transcription termination in Duchenne and Becker muscular dystrophy. <i>Human Mutation</i> , 2022, 43, 511-528.	1.1	16
3	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.	3.8	43
4	Phenotypic Spectrum of Dystrophinopathy Due to Duchenne Muscular Dystrophy Exon 2 Duplications. <i>Neurology</i> , 2022, 98, .	1.5	6
5	Letter by Duan et al Regarding Article, "Therapeutic Exon Skipping Through a CRISPR-Guided Cytidine Deaminase Rescues Dystrophic Cardiomyopathy In Vivo" <i>Circulation</i> , 2022, 145, e872-e873.	1.6	0
6	Gene editing and modulation for Duchenne muscular dystrophy. <i>Progress in Molecular Biology and Translational Science</i> , 2021, 182, 225-255.	0.9	7
7	Update in the Mucopolysaccharidoses. <i>Seminars in Pediatric Neurology</i> , 2021, 37, 100874.	1.0	27
8	Direct Reprogramming of Human Fibroblasts into Myoblasts to Investigate Therapies for Neuromuscular Disorders. <i>Journal of Visualized Experiments</i> , 2021, , .	0.2	0
9	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.	1.8	21
10	Longitudinal MRI brain volume changes over one year in children with mucopolysaccharidosis types IIIA and IIIB. <i>Molecular Genetics and Metabolism</i> , 2021, 133, 193-200.	0.5	2
11	Health related quality of life in young, steroid-naïve boys with Duchenne muscular dystrophy. <i>Neuromuscular Disorders</i> , 2021, 31, 1161-1168.	0.3	4
12	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.	1.4	8
13	Validity and Reliability of the Neuromuscular Gross Motor Outcome. <i>Pediatric Neurology</i> , 2021, 122, 21-26.	1.0	5
14	Evaluating longitudinal therapy effects via the North Star Ambulatory Assessment. <i>Muscle and Nerve</i> , 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 <i>DMD</i> Exon 2 Skipping. <i>Human Gene Therapy</i> , 2021, 32, 882-894.	1.4	29
16	Meta-analyses of deflazacort versus prednisone/prednisolone in patients with nonsense mutation Duchenne muscular dystrophy. <i>Journal of Comparative Effectiveness Research</i> , 2021, 10, 1337-1347.	0.6	6
17	Truncating variants in <i>UBAP1</i> associated with childhood-onset nonsyndromic hereditary spastic paraplegia. <i>Human Mutation</i> , 2020, 41, 632-640.	1.1	15
18	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. <i>American Journal of Medical Genetics, Part A</i> , 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. <i>Pediatric Neurology</i> , 2020, 113, 15-20.	1.0	14
20	Meta-analyses of ataluren randomized controlled trials in nonsense mutation Duchenne muscular dystrophy. <i>Journal of Comparative Effectiveness Research</i> , 2020, 9, 973-984.	0.6	41
21	X-linked muscular dystrophy in a Labrador Retriever strain: phenotypic and molecular characterisation. <i>Skeletal Muscle</i> , 2020, 10, 23.	1.9	12
22	Clinical Phenotypes of DMD Exon 51 Skip Equivalent Deletions: A Systematic Review. <i>Journal of Neuromuscular Diseases</i> , 2020, 7, 217-229.	1.1	18
23	The Genotypic and Phenotypic Spectrum of <i>BICD2</i> Variants in Spinal Muscular Atrophy. <i>Annals of Neurology</i> , 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. <i>Circulation: Cardiovascular Imaging</i> , 2019, 12, e008919.	1.3	4
25	Diagnostic Utility of Whole Exome Sequencing in the Neuromuscular Clinic. <i>Neuropediatrics</i> , 2019, 50, 096-102.	0.3	28
26	Twice-weekly glucocorticosteroids in infants and young boys with Duchenne muscular dystrophy. <i>Muscle and Nerve</i> , 2019, 59, 650-657.	1.0	32
27	Variants in <i>MED12L</i> , 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.	1.1	28
28	Evaluation of biomarkers for Sanfilippo syndrome. <i>Molecular Genetics and Metabolism</i> , 2019, 128, 68-74.	0.5	13
29	Update in Duchenne and Becker muscular dystrophy. <i>Current Opinion in Neurology</i> , 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. <i>Neuromuscular Disorders</i> , 2018, 28, 116-121.	0.3	27
32	Low-level expression of <i>EPG5</i> leads to an attenuated Vici syndrome phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1207-1211.	0.7	9
33	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.	0.5	14
34	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.	0.7	26
35	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.	2.8	53
36	Natural history of echocardiographic abnormalities in mucopolysaccharidosis III. <i>Molecular Genetics and Metabolism</i> , 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. <i>Molecular Therapy - Methods and Clinical Development</i> , 2018, 10, 89-104.	1.8	14
38	Placebo-controlled Phase 2 Trial of Drisapersen for Duchenne Muscular Dystrophy. <i>Annals of Clinical and Translational Neurology</i> , 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. <i>Human Gene Therapy</i> , 2017, 28, 737-746.	1.4	27
40	Clinicopathologic Conference: A Newborn With Hypotonia, Cleft Palate, Micrognathia, and Bilateral Club Feet. <i>Pediatric Neurology</i> , 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. <i>Neuromuscular Disorders</i> , 2017, 27, 452-457.	0.3	16
42	Follistatin Gene Therapy for Sporadic Inclusion Body Myositis Improves Functional Outcomes. <i>Molecular Therapy</i> , 2017, 25, 870-879.	3.7	84
43	General anesthesia with a native airway for patients with mucopolysaccharidosis type <scp>III</scp>. <i>Paediatric Anaesthesia</i> , 2017, 27, 370-376.	0.6	9
44	Developing standardized corticosteroid treatment for Duchenne muscular dystrophy. <i>Contemporary Clinical Trials</i> , 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. <i>Journal of Neuromuscular Diseases</i> , 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. <i>Human Gene Therapy Clinical Development</i> , 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. <i>Lancet, The</i> , 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. <i>Journal of Neuromuscular Diseases</i> , 2017, 4, 293-306.	1.1	125
49	Modeling functional decline over time in sporadic inclusion body myositis. <i>Muscle and Nerve</i> , 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. <i>Human Gene Therapy Clinical Development</i> , 2017, , .	3.2	0
51	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.	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. <i>Muscle and Nerve</i> , 2016, 54, 681-689.	1.0	29
54	N-terminal β -Dystroglycan (β DG-N): A Potential Serum Biomarker for Duchenne Muscular Dystrophy. <i>Journal of Neuromuscular Diseases</i> , 2016, 3, 247-260.	1.1	10

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55	Reassessing carrier status for dystrophinopathies. <i>Neurology: Genetics</i> , 2016, 2, e108.	0.9	2
56	Association Study of Exon Variants in the NF- κ B and TGF β 2 Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy. <i>American Journal of Human Genetics</i> , 2016, 99, 1163-1171.	2.6	71
57	Duchenne muscular dystrophy: meeting the therapeutic challenge. <i>Lancet Neurology</i> , 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. <i>Pediatric Neurology</i> , 2016, 63, 76-79.	1.0	4
59	<i>EPG5</i>-related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. <i>Brain</i> , 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. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 82.	1.2	39
62	Dp412e: a novel human embryonic dystrophin isoform induced by BMP4 in early differentiated cells. <i>Skeletal Muscle</i> , 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. <i>Human Gene Therapy Clinical Development</i> , 2015, 26, 228-242.	3.2	19
64	Outcome reliability in non-ambulatory Boys/Men with duchenne muscular dystrophy. <i>Muscle and Nerve</i> , 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. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 49.	1.2	21
66	The first exon duplication mouse model of Duchenne muscular dystrophy: A tool for therapeutic development. <i>Neuromuscular Disorders</i> , 2015, 25, 827-834.	0.3	27
67	Reliability and validity of active-seated: An outcome in dystrophinopathy. <i>Muscle and Nerve</i> , 2015, 52, 356-362.	1.0	15
68	Clinical phenotypes as predictors of the outcome of skipping around <sc><i>DMD</i></sc> exon 45. <i>Annals of Neurology</i> , 2015, 77, 668-674.	2.8	38
69	The TREAT-NMD DMD Global Database: Analysis of More than 7,000 Duchenne Muscular Dystrophy Mutations. <i>Human Mutation</i> , 2015, 36, 395-402.	1.1	507
70	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.	0.3	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. <i>Journal of Human Genetics</i> , 2015, 60, 151-155.	1.1	17
72	Genetics and Emerging Treatments for Duchenne and Becker Muscular Dystrophy. <i>Pediatric Clinics of North America</i> , 2015, 62, 723-742.	0.9	71

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73	A Phase 1/2a Follistatin Gene Therapy Trial for Becker Muscular Dystrophy. <i>Molecular Therapy</i> , 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. <i>PLoS ONE</i> , 2014, 9, e88226.	1.1	19
75	Targeted Exon Skipping to Correct Exon Duplications in the Dystrophin Gene. <i>Molecular Therapy - Nucleic Acids</i> , 2014, 3, e155.	2.3	37
76	Dystrophin quantification. <i>Neurology</i> , 2014, 83, 2062-2069.	1.5	73
77	Dexmedetomidine and ketamine sedation for muscle biopsies in patients with Duchenne muscular dystrophy. <i>Paediatric Anaesthesia</i> , 2014, 24, 851-856.	0.6	88
78	Ataluren treatment of patients with nonsense mutation dystrophinopathy. <i>Muscle and Nerve</i> , 2014, 50, 477-487.	1.0	357
79	Visual Diagnosis: Chest Pain in a Boy With Duchenne Muscular Dystrophy and Cardiomyopathy. <i>Pediatrics in Review</i> , 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. <i>Human Gene Therapy Clinical Development</i> , 2014, 25, 72-84.	3.2	79
81	Dystrophin as a therapeutic biomarker: Are we ignoring data from the past?. <i>Neuromuscular Disorders</i> , 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. <i>Neuromuscular Disorders</i> , 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 3' UTR. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, 5670-5675.	3.3	43
84	Exome sequencing identifies a DNAJB6 mutation in a family with dominantly-inherited limb-girdle muscular dystrophy. <i>Neuromuscular Disorders</i> , 2014, 24, 431-435.	0.3	35
85	The ZZ Domain of Dystrophin in DMD: Making Sense of Missense Mutations. <i>Human Mutation</i> , 2014, 35, 257-264.	1.1	23
86	Recurrent central nervous system white matter changes in charcot-Marie-Tooth type X disease. <i>Muscle and Nerve</i> , 2014, 49, 451-454.	1.0	19
87	Duchenne and Becker Muscular Dystrophies. <i>Neurologic Clinics</i> , 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. <i>Nature Medicine</i> , 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. <i>Pediatric Neurology</i> , 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. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, P7.	1.2	0

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91	Visual Diagnosis: Chest Pain in a Boy With Duchenne Muscular Dystrophy and Cardiomyopathy. <i>Pediatrics in Review</i> , 2014, 35, e64-e67.	0.2	0
92	Anti-Dystrophin T Cell Responses in Duchenne Muscular Dystrophy: Prevalence and a Glucocorticoid Treatment Effect. <i>Human Gene Therapy</i> , 2013, 24, 797-806.	1.4	97
93	Camptocormia as a late presentation in a manifesting carrier of duchenne muscular dystrophy. <i>Muscle and Nerve</i> , 2013, 47, 124-127.	1.0	8
94	Becker muscular dystrophy with widespread muscle hypertrophy and a non-sense mutation of exon 2. <i>Neuromuscular Disorders</i> , 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. <i>Neuromuscular Disorders</i> , 2013, 23, 529-539.	0.3	79
96	<i>LTBP4</i> genotype predicts age of ambulatory loss in duchenne muscular dystrophy. <i>Annals of Neurology</i> , 2013, 73, 481-488.	2.8	202
97	Diabetic Myonecrosis in a Cystic Fibrosis Patient. <i>Respiratory Care</i> , 2013, 58, e123-e125.	0.8	0
98	The TREAT-NMD Duchenne Muscular Dystrophy Registries: Conception, Design, and Utilization by Industry and Academia. <i>Human Mutation</i> , 2013, 34, 1449-1457.	1.1	94
99	Position of Glycine Substitutions in the Triple Helix of<i>COL6A1</i>,<i>COL6A2</i>, and<i>COL6A3</i>is Correlated with Severity and Mode of Inheritance in Collagen VI Myopathies. <i>Human Mutation</i> , 2013, 34, 1558-1567.	1.1	79
100	Eteplirsen for the treatment of Duchenne muscular dystrophy. <i>Annals of Neurology</i> , 2013, 74, 637-647.	2.8	630
101	Correlation of knee strength to functional outcomes in becker muscular dystrophy. <i>Muscle and Nerve</i> , 2013, 47, 550-554.	1.0	16
102	Precordial R Wave Height Does Not Correlate with Echocardiographic Findings in Boys with Duchenne Muscular Dystrophy. <i>Congenital Heart Disease</i> , 2013, 8, 561-567.	0.0	6
103	Identification of New Dystroglycan Complexes in Skeletal Muscle. <i>PLoS ONE</i> , 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. <i>PLOS Currents</i> , 2013, 5, .	1.4	42
105	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
106	Phase 2a Study of Ataluren-Mediated Dystrophin Production in Patients with Nonsense Mutation Duchenne Muscular Dystrophy. <i>PLoS ONE</i> , 2013, 8, e81302.	1.1	201
107	The Muscular Dystrophies. <i>Seminars in Neurology</i> , 2012, 32, 255-263.	0.5	71
108	A Practical Approach to Molecular Diagnostic Testing in Neuromuscular Diseases. <i>Physical Medicine and Rehabilitation Clinics of North America</i> , 2012, 23, 589-608.	0.7	19

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109	Cardiac Management in Neuromuscular Diseases. <i>Physical Medicine and Rehabilitation Clinics of North America</i> , 2012, 23, 855-868.	0.7	12
110	RNA Interference Inhibits DUX4-induced Muscle Toxicity In Vivo: Implications for a Targeted FSHD Therapy. <i>Molecular Therapy</i> , 2012, 20, 1417-1423.	3.7	101
111	Evidence-based path to newborn screening for duchenne muscular dystrophy. <i>Annals of Neurology</i> , 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. <i>American Journal of Cardiology</i> , 2012, 110, 98-102.	0.7	137
113	Knee extensor strength exhibits potential to predict function in sporadic inclusion-body myositis. <i>Muscle and Nerve</i> , 2012, 45, 163-168.	1.0	25
114	Canine models of Duchenne muscular dystrophy and their use in therapeutic strategies. <i>Mammalian Genome</i> , 2012, 23, 85-108.	1.0	140
115	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.	1.0	11
116	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.	1.1	103
117	Gentamicin-induced readthrough of stop codons in duchenne muscular dystrophy. <i>Annals of Neurology</i> , 2010, 67, 771-780.	2.8	238
118	Sustained alpha-sarcoglycan gene expression after gene transfer in limb-girdle muscular dystrophy, type 2D. <i>Annals of Neurology</i> , 2010, 68, 629-638.	2.8	214
119	Response to Letter by Yilmaz et al Regarding Article, "Analysis of Dystrophin Deletion Mutations Predicts Age of Cardiomyopathy Onset in Becker Muscular Dystrophy". <i>Circulation: Cardiovascular Genetics</i> , 2010, 3, .	5.1	0
120	Expanding the clinical, pathological and MRI phenotype of DNM2-related centronuclear myopathy. <i>Neuromuscular Disorders</i> , 2010, 20, 229-237.	0.3	100
121	Clinical and genetic characterization of manifesting carriers of DMD mutations. <i>Neuromuscular Disorders</i> , 2010, 20, 499-504.	0.3	136
122	Clinical, histological and genetic characterization of reducing body myopathy caused by mutations in FHL1. <i>Brain</i> , 2009, 132, 452-464.	3.7	88
123	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
124	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.	1.1	62
125	<i>DMD</i> exon 1 truncating point mutations: Amelioration of phenotype by alternative translation initiation in exon 6. <i>Human Mutation</i> , 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. <i>Human Mutation</i> , 2009, 30, 1657-1666.	1.1	279

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127	Utility of cystatin C to monitor renal function in duchenne muscular dystrophy. <i>Muscle and Nerve</i> , 2009, 40, 438-442.	1.0	84
128	Spinocerebellar Ataxia Type 31 Is Associated with α -Penta-Nucleotide Repeats Containing (TGGAA) _n . <i>American Journal of Human Genetics</i> , 2009, 85, 544-557.	2.6	260
129	A novel form of juvenile recessive ALS maps to loci on 6p25 and 21q22. <i>Neuromuscular Disorders</i> , 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. <i>Neuromuscular Disorders</i> , 2009, 19, 743-748.	0.3	43
131	DMD pseudoexon mutations: splicing efficiency, phenotype, and potential therapy. <i>Annals of Neurology</i> , 2008, 63, 81-89.	2.8	95
132	A phase I/II trial of MYO29 in adult subjects with muscular dystrophy. <i>Annals of Neurology</i> , 2008, 63, 561-571.	2.8	407
133	Mitochondrial DNA depletion syndrome due to mutations in the RRM2B gene. <i>Neuromuscular Disorders</i> , 2008, 18, 453-459.	0.3	87
134	Mitochondrial fusion and function in Charcot-Marie-Tooth type 2A patient fibroblasts with mitofusin 2 mutations. <i>Experimental Neurology</i> , 2008, 211, 115-127.	2.0	88
135	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.	3.3	166
136	Proteomic identification of FHL1 as the protein mutated in human reducing body myopathy. <i>Journal of Clinical Investigation</i> , 2008, 118, 904-12.	3.9	126
137	Distinctive patterns of microRNA expression in primary muscular disorders. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2007, 104, 17016-17021.	3.3	458
138	Abnormal expression of mu-crystallin in facioscapulohumeral muscular dystrophy. <i>Experimental Neurology</i> , 2007, 205, 583-586.	2.0	34
139	An unusual pathologic feature associated with dermatomyositis. <i>Neuromuscular Disorders</i> , 2006, 16, 391-393.	0.3	1
140	Sarcolemmal reorganization in facioscapulohumeral muscular dystrophy. <i>Annals of Neurology</i> , 2006, 59, 289-297.	2.8	25
141	Recoding elements located adjacent to a subset of eukaryal selenocysteine-specifying UGA codons. <i>EMBO Journal</i> , 2005, 24, 1596-1607.	3.5	84
142	Clinical and electrophysiologic features of CMT2A with mutations in the mitofusin 2 gene. <i>Neurology</i> , 2005, 65, 197-204.	1.5	160
143	Muscle MRI in Ullrich congenital muscular dystrophy and Bethlem myopathy. <i>Neuromuscular Disorders</i> , 2005, 15, 303-310.	0.3	154
144	Prenatal diagnosis of Ullrich congenital muscular dystrophy using haplotype analysis and collagen VI immunocytochemistry. <i>Prenatal Diagnosis</i> , 2004, 24, 440-444.	1.1	29

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