Peter B Kang

List of Publications by Year
in descending order

129 papers	4,973 citations	117625 34 h-index	102487 66 g-index
131 all docs	131 docs citations	131 times ranked	7337 citing authors

#	Article	IF	Citations
1	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. Science Translational Medicine, 2017, 9, .	12.4	516
2	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
3	Observational study of spinal muscular atrophy type I and implications for clinical trials. Neurology, 2014, 83, 810-817.	1.1	367
4	Prospective cohort study of spinal muscular atrophy types 2 and 3. Neurology, 2012, 79, 1889-1897.	1.1	207
5	Observational Study of Spinal Muscular Atrophy Type 2 and 3. Archives of Neurology, 2011, 68, 779.	4.5	142
6	MicroRNA-199a is induced in dystrophic muscle and affects WNT signaling, cell proliferation, and myogenic differentiation. Cell Death and Differentiation, 2013, 20, 1194-1208.	11.2	140
7	MicroRNA-486â€“dependent modulation of DOCK3/PTEN/AKT signaling pathways improves muscular dystrophyâ€“associated symptoms. Journal of Clinical Investigation, 2014, 124, 2651-2667.	8.2	128
8	Mutation spectrum in the large GTPase dynamin 2, and genotype-phenotype correlation in autosomal dominant centronuclear myopathy. Human Mutation, 2012, 33, 949-959.	2.5	115
9	A novel syndrome caused by the E410K amino acid substitution in the neuronal Î²-tubulin isotype 3. Brain, 2013, 136, 522-535.	7.6	112
10	Juvenile myasthenia gravis. Muscle and Nerve, 2009, 39, 423-431.	2.2	110
11	Mutation Update and Genotype-Phenotype Correlations of Novel and Previously Described Mutations inTPM2andTPM3Causing Congenital Myopathies. Human Mutation, 2014, 35, 779-790.	2.5	92
12	Mitochondrial fusion and function in Charcotâ€“Marieâ€“Tooth type 2A patient fibroblasts with mitofusin 2 mutations. Experimental Neurology, 2008, 211, 115-127.	4.1	88
13	Evidence-based guideline summary: Evaluation, diagnosis, and management of congenital muscular dystrophy. Neurology, 2015, 84, 1369-1378.	1.1	88
14	Emeryâ€Dreifuss muscular dystrophy. Muscle and Nerve, 2020, 61, 436-448.	2.2	84
15	Regulation of DMD pathology by an ankyrin-encoded miRNA. Skeletal Muscle, 2011, 1, 27.	4.2	81
16	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
17	The sensitivity of exome sequencing in identifying pathogenic mutations for LGMD in the United States. Journal of Human Genetics, 2017, 62, 243-252.	2.3	73
18	POMK mutations disrupt muscle development leading to a spectrum of neuromuscular presentations. Human Molecular Genetics, 2014, 23, 5781-5792.	2.9	72

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19	Mutations in the satellite cell gene MEGF10 cause a recessive congenital myopathy with minicores. Neurogenetics, 2012, 13, 115-124.	1.4	68
20	Variants in EXOSC9 Disrupt the RNA Exosome and Result in Cerebellar Atrophy with Spinal Motor Neuronopathy. American Journal of Human Genetics, 2018, 102, 858-873.	6.2	65
21	Comparison of Plasmapheresis and Intravenous Immunoglobulin as Maintenance Therapies for Juvenile Myasthenia Gravis. JAMA Neurology, 2014, 71, 575.	9.0	64
22	Serum Transaminase Levels in Boys With Duchenne and Becker Muscular Dystrophy. Pediatrics, 2011, 127, e132-e136.	2.1	63
23	Update on the Genetics of Limb Girdle Muscular Dystrophy. Seminars in Pediatric Neurology, 2012, 19, 211-218.	2.0	62
24	Childhood chronic inflammatory demyelinating polyradiculoneuropathy: Combined analysis of a large cohort and eleven published series. Neuromuscular Disorders, 2013, 23, 103-111.	0.6	62
25	Identifying diagnostic DNA methylation profiles for facioscapulohumeral muscular dystrophy in blood and saliva using bisulfite sequencing. Clinical Epigenetics, 2014, 6, 23.	4.1	61
26	Outcome reliability in nonâ€Ambulatory Boys/Men with duchenne muscular dystrophy. Muscle and Nerve, 2015, 51, 522-532.	2.2	60
27	Deficiency of base excision repair enzyme NEIL3 drives increased predisposition to autoimmunity. Journal of Clinical Investigation, 2016, 126, 4219-4236.	8.2	56
28	The child neurology clinical workforce in 2015. Neurology, 2016, 87, 1384-1392.	1.1	49
29	Update on juvenile myasthenia gravis. Current Opinion in Pediatrics, 2013, 25, 694-700.	2.0	43
30	AAV-MediatedTAZGene Replacement Restores Mitochondrial and Cardioskeletal Function in Barth Syndrome. Human Gene Therapy, 2019, 30, 139-154.	2.7	40
31	Transcriptional profile of postmortem skeletal muscle. Physiological Genomics, 2004, 16, 222-228.	2.3	38
32	Congenital Myasthenic Syndrome With Episodic Apnea. Pediatric Neurology, 2009, 41, 42-45.	2.1	38
33	Multifocal slowing of nerve conduction in metachromatic leukodystrophy. Muscle and Nerve, 2004, 29, 531-536.	2.2	37
34	Glycogen storage disease type IV: novel mutations and molecular characterization of a heterogeneous disorder. Journal of Inherited Metabolic Disease, 2010, 33, 83-90.	3.6	37
35	PLIN2 inhibits insulin-induced glucose uptake in myoblasts through the activation of the NLRP3 inflammasome. International Journal of Molecular Medicine, 2015, 36, 839-844.	4.0	36
36	The influence of muscle type and dystrophin deficiency on murine expression profiles. Mammalian Genome, 2005, 16, 739-748.	2.2	35

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37	The motor neuron response to SMN1 deficiency in spinal muscular atrophy. Muscle and Nerve, 2014, 49, 636-644.	2.2	34
38	Clinical trials in spinal muscular atrophy. Current Opinion in Pediatrics, 2007, 19, 675-679.	2.0	32
39	Exome sequencing identifies a novel SMCHD1 mutation in facioscapulohumeral muscular dystrophy 2. Neuromuscular Disorders, 2013, 23, 975-980.	0.6	32
40	Diagnostic value of electromyography and muscle biopsy in arthrogryposis multiplex congenita. Annals of Neurology, 2003, 54, 790-795.	5.3	31
41	Involvement of superficial peroneal sensory nerve in common peroneal neuropathy. Muscle and Nerve, 2005, 31, 725-729.	2.2	30
42	Consequences of MEGF10 deficiency on myoblast function and Notch1 interactions. Human Molecular Genetics, 2017, 26, 2984-3000.	2.9	30
43	Lactic Acid Elevation in Extramitochondrial Childhood Neurodegenerative Diseases. Journal of Child Neurology, 2001, 16, 657-660.	1.4	29
44	LGMD2I in a North American population. BMC Musculoskeletal Disorders, 2007, 8, 115.	1.9	29
45	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
46	Variations in gene expression among different types of human skeletal muscle. Muscle and Nerve, 2005, 32, 483-491.	2.2	28
47	Transcriptome-scale similarities between mouse and human skeletal muscles with normal and myopathic phenotypes. BMC Musculoskeletal Disorders, 2006, 7, 23.	1.9	28
48	The role of thymectomy in the treatment of juvenile myasthenia gravis: a systematic review. Pediatric Surgery International, 2017, 33, 683-694.	1.4	26
49	Autoimmune Neuromuscular Disorders in Childhood. Current Treatment Options in Neurology, 2011, 13, 590-607.	1.8	24
50	Recent developments in the treatment of Duchenne muscular dystrophy and spinal muscular atrophy. Therapeutic Advances in Neurological Disorders, 2013, 6, 147-160.	3.5	24
51	Silencing of drpr Leads to Muscle and Brain Degeneration in Adult Drosophila. American Journal of Pathology, 2014, 184, 2653-2661.	3.8	23
52	Advances in Muscular Dystrophies. JAMA Neurology, 2015, 72, 741.	9.0	23
53	Child Neurology: Chronic inflammatory demyelinating polyradiculoneuropathy in children. Neurology, 2008, 71, e74-8.	1.1	22
54	Clinical correlates of charcotâ€“marieâ€“tooth disease in patients with pes cavus deformities. Muscle and Nerve, 2013, 47, 488-492.	2.2	22

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55	Inefficient dystrophin expression after cord blood transplantation in Duchenne muscular dystrophy. Muscle and Nerve, 2010, 41, 746-750.	2.2	21
56	Expanding the Phenotypic Spectrum and Variability of Endocrine Abnormalities Associated With TUBB3 E410K Syndrome. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E473-E477.	3.6	21
57	POLRMT mutations impair mitochondrial transcription causing neurological disease. Nature Communications, 2021, 12, 1135.	12.8	21
58	Ambulatory foot temperature measurement: A new technique in polyneuropathy evaluation. Muscle and Nerve, 2003, 27, 737-742.	2.2	20
59	A Case of Congenital Glycogen Storage Disease Type IV With a Novel GBE1 Mutation. Journal of Child Neurology, 2008, 23, 349-352.	1.4	19
60	Response of Motor Complications in Cockayne Syndrome to Carbidopa-Levodopa. Archives of Neurology, 2008, 65, 1117-21.	4.5	17
61	Rapid reversal of uremic neuropathy following renal transplantation in an adolescent. Pediatric Transplantation, 2012, 16, E296-300.	1.0	17
62	Referral and diagnostic trends in pediatric electromyography in the molecular era. Muscle and Nerve, 2014, 50, 244-249.	2.2	17
63	AAV9-TAZ Gene Replacement Ameliorates Cardiac TMT Proteomic Profiles in a Mouse Model of Barth Syndrome. Molecular Therapy - Methods and Clinical Development, 2019, 13, 167-179.	4.1	17
64	The ties that bind: functional clusters in limb-girdle muscular dystrophy. Skeletal Muscle, 2020, 10, 22.	4.2	17
65	Cardiac electrophysiological characteristics of the mdx 5cv mouse model of Duchenne muscular dystrophy. Journal of Interventional Cardiac Electrophysiology, 2007, 20, 1-7.	1.3	16
66	Cysteine mutations cause defective tyrosine phosphorylation in MEGF10 myopathy. FEBS Letters, 2013, 587, 2952-2957.	2.8	16
67	Infantile Leukoencephalopathy Owing to Mitochondrial Enzyme Dysfunction. Journal of Child Neurology, 2002, 17, 421-428.	1.4	15
68	Efficient identification of novel mutations in patients with limb girdle muscular dystrophy. Neurogenetics, 2010, 11, 449-455.	1.4	15
69	Child Neurology Recruitment and Training: Views of Residents and Child Neurologists From the 2015 AAP/CNS Workforce Survey. Pediatric Neurology, 2017, 66, 89-95.	2.1	15
70	Impact of PYROXD1 deficiency on cellular respiration and correlations with genetic analyses of limb-girdle muscular dystrophy in Saudi Arabia and Sudan. Physiological Genomics, 2018, 50, 929-939.	2.3	15
71	Child neurology in the 21st century. Neurology, 2020, 94, 75-82.	1.1	15
72	A form of muscular dystrophy associated with pathogenic variants in JAG2. American Journal of Human Genetics, 2021, 108, 840-856.	6.2	15

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73	Pediatric Nerve Conduction Studies and EMG. , 2007, , 369-389.		15
74	Novel MPZ mutations and congenital hypomyelinating neuropathy. Neuromuscular Disorders, 2010, 20, 725-729.	0.6	14
75	Utility and practice of electrodiagnostic testing in the pediatric population: An AANEM consensus statement. Muscle and Nerve, 2020, 61, 143-155.	2.2	14
76	Atypical presentations of spinal muscular atrophy type III (Kugelbergâ€“Welander disease). Neuromuscular Disorders, 2006, 16, 492-494.	0.6	13
77	Molecular diagnosis of hereditary inclusion body myopathy by linkage analysis and identification of a novel splice site mutation in GNE. BMC Medical Genetics, 2011, 12, 87.	2.1	13
78	The Spectrum of Myotonic and Myopathic Disorders in a Pediatric Electromyography Laboratory Over 12 Years. Pediatric Neurology, 2012, 47, 97-100.	2.1	13
79	Homozygous nonsense mutation inSGCAis a common cause of limb-girdle muscular dystrophy in Assiut, Egypt. Muscle and Nerve, 2016, 54, 690-695.	2.2	12
80	Longitudinal Patterns of Thalidomide Neuropathy in Children and Adolescents. Journal of Pediatrics, 2016, 178, 227-232.	1.8	12
81	Medial medullary injury during adenoidectomy. Journal of Pediatrics, 2001, 138, 772-774.	1.8	11
82	Electrophysiologic features of fibular neuropathy in childhood and adolescence. Muscle and Nerve, 2017, 55, 693-697.	2.2	11
83	A slowly progressive form of limbâ€girdle muscular dystrophy type 2C associated with founder mutation in the SGCG gene in Puerto Rican Hispanics. Molecular Genetics & Genomic Medicine, 2015, 3, 92-98.	1.2	10
84	Novel biallelic variants expand the SLC5A6-related phenotypic spectrum. European Journal of Human Genetics, 2022, 30, 439-449.	2.8	10
85	Diagnostic capabilities of nanopore longâ€read sequencing in muscular dystrophy. Annals of Clinical and Translational Neurology, 2022, 9, 1302-1309.	3.7	10
86	Increased mtDNA Abundance and Improved Function in Human Barth Syndrome Patient Fibroblasts Following AAV-TAZ Gene Delivery. International Journal of Molecular Sciences, 2019, 20, 3416.	4.1	9
87	Identification of a pathogenic mutation in ATP2A1 via in silico analysis of exome data for cryptic aberrant splice sites. Molecular Genetics & Genomic Medicine, 2019, 7, e552.	1.2	9
88	Intravenous Immunoglobulin as a Therapeutic Option for Mycoplasma pneumoniae Encephalitis. Journal of Child Neurology, 2019, 34, 687-691.	1.4	8
89	Megf10 deficiency impairs skeletal muscle stem cell migration and muscle regeneration. FEBS Open Bio, 2021, 11, 114-123.	2.3	8
90	Selective serotonin reuptake inhibitors ameliorate MEGF10 myopathy. Human Molecular Genetics, 2019, 28, 2365-2377.	2.9	7

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91	Ethical decision-making for children with neuromuscular disorders in the COVID-19 crisis. Neurology, 2020, 95, 260-265.	1.1	7
92	Rituximab as Adjunct Maintenance Therapy for Refractory Juvenile Myasthenia Gravis. Pediatric Neurology, 2020, 111, 40-43.	2.1	7
93	<scp>hnRNP L</scp> is essential for myogenic differentiation and modulates myotonic dystrophy pathologies. Muscle and Nerve, 2021, 63, 928-940.	2.2	7
94	An Opportune Time for Newborn Screening in Duchenne Muscular Dystrophy. JAMA Neurology, 2021, 78, 901.	9.0	7
95	Phenotypic implications of pathogenic variant types in Pompe disease. Journal of Human Genetics, 2021, 66, 1089-1099.	2.3	6
96	Pediatric monomelic amyotrophy: Evidence for poliomyelitis in vulnerable populations. Muscle and Nerve, 2009, 40, 860-863.	2.2	5
97	Ethical issues in neurogenetic disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 118, 265-276.	1.8	5
98	Electrophysiologic Features of Radial Neuropathy in Childhood and Adolescence. Pediatric Neurology, 2018, 81, 14-18.	2.1	5
99	Dollars and antisense for Duchenne muscular dystrophy. Neurology, 2018, 90, 1091-1092.	1.1	5
100	The impact of Megf10/Drpr gainâ€ofâ€function on muscle development inÂDrosophila. FEBS Letters, 2019, 593, 680-696.	2.8	5
101	Clinical, electrophysiological, and imaging findings in childhood brachial plexus injury. Developmental Medicine and Child Neurology, 2022, , .	2.1	5
102	Ethical issues in the evaluation of adults with suspected genetic neuromuscular disorders. Muscle and Nerve, 2016, 54, 997-1006.	2.2	4
103	Electrophysiologic features of ulnar neuropathy in childhood and adolescence. Clinical Neurophysiology, 2017, 128, 751-755.	1.5	4
104	Neurodevelopmental outcomes at 9â€“14Âmonths gestational age after treatment of neonatal seizures due to brain injury. Child's Nervous System, 2019, 35, 1571-1578.	1.1	4
105	Growth charts in Cockayne syndrome type 1 and type 2. European Journal of Medical Genetics, 2021, 64, 104105.	1.3	4
106	Beyond the Gowers sign: measuring outcomes in Duchenne muscular dystrophy. Muscle and Nerve, 2013, 48, 315-317.	2.2	3
107	Evidence-based guideline summary: Evaluation, diagnosis, and management of congenital muscular dystrophy: Report of the Guideline Development Subcommittee of the American Academy of Neurology and the Practice Issues Review Panel of the American Association of Neuromuscular & Electrodiagnostic Medicine. Neurology, 2015, 85, 1432-1433.	1.1	3
108	Neuropathic and Myopathic Pain. Seminars in Pediatric Neurology, 2016, 23, 242-247.	2.0	3

#	Article	IF	Citations
109	Patient reported quality of life in limb girdle muscular dystrophy. Neuromuscular Disorders, 2022, 32, 57-64.	0.6	3
110	Lumbosacral ventral spinal nerve root atrophy identified on MRI in a case of spinal muscular atrophy type II. Clinical Imaging, 2019, 53, 134-137.	1.5	2
111	Presymptomatic and Early Symptomatic Genetic Testing. CONTINUUM Lifelong Learning in Neurology, 2011, 17, 343-346.	0.8	1
112	The struggle to model muscular dystrophy. Muscle and Nerve, 2011, 44, 157-159.	2.2	1
113	Chronic Inflammatory Demyelinating Polyradiculoneuropathy. , 2015, , 398-417.		1
114	Juvenile and Neonatal Myasthenia Gravis. , 2015, , 482-496.		1
115	The End of the Beginning: The Journey to Molecular Therapies for Spinal Muscular Atrophy. Pediatric Neurology, 2020, 102, 1-2.	2.1	1
116	Hunting for the perfect test: Neuromuscular diagnosis in the age of genomic bounty. Muscle and Nerve, 2021, 63, 282-284.	2.2	1
117	Approach to Electrodiagnostic Testing in Children. , 2017, , 23-27.		1
118	Neuromuscular Transmission Disorders. , 2020, , 1257-1279.		1
119	Hereditary and Acquired Myopathies. , 2020, , 1281-1349.		1
120	A two-year-old girl with acute onset of seizures and progressive encephalopathy. Current Opinion in Pediatrics, 1997, 9, 558-564.	2.0	0
121	The New Frontier of Genetically Targeted Therapies for Muscle Disease. CONTINUUM Lifelong Learning in Neurology, 2013, 19, 1698-1702.	0.8	0
122	Reply. Muscle and Nerve, 2014, 50, 458-459.	2.2	0
123	Electromyography in Pediatrics. , 2015, , 32-45.		0
124	Laboratory Assessment of the Child with Suspected Neuromuscular Disorders. , 2017, , 1038-1043.		0
125	Neuromuscular pathology in Vici syndrome. FASEB Journal, 2007, 21, A399.	0.5	0
126	Silencing of drpr leads to muscle and brain degeneration in adult Drosophila. FASEB Journal, 2013, 27, 873.14.	0.5	0

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127	Modeling Human MEGF10 Myopathy in Drosophila melanogaster. FASEB Journal, 2015, 29, 613.9.	0.5	0
128	Motor Unit Number Estimation. , 2017, , 151-155.		0
129	Muscle Analysis. , 2017, , 115-121.		0