

Peter B Kang

List of Publications by Year in descending order

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Version: 2024-02-01

129
papers

4,973
citations

117453

34
h-index

102304

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. <i>Science Translational Medicine</i> , 2017, 9, .	5.8	516
2	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
3	Observational study of spinal muscular atrophy type I and implications for clinical trials. <i>Neurology</i> , 2014, 83, 810-817.	1.5	367
4	Prospective cohort study of spinal muscular atrophy types 2 and 3. <i>Neurology</i> , 2012, 79, 1889-1897.	1.5	207
5	Observational Study of Spinal Muscular Atrophy Type 2 and 3. <i>Archives of Neurology</i> , 2011, 68, 779.	4.9	142
6	MicroRNA-199a is induced in dystrophic muscle and affects WNT signaling, cell proliferation, and myogenic differentiation. <i>Cell Death and Differentiation</i> , 2013, 20, 1194-1208.	5.0	140
7	MicroRNA-486-dependent modulation of DOCK3/PTEN/AKT signaling pathways improves muscular dystrophy-associated symptoms. <i>Journal of Clinical Investigation</i> , 2014, 124, 2651-2667.	3.9	128
8	Mutation spectrum in the large GTPase dynamin 2, and genotype-phenotype correlation in autosomal dominant centronuclear myopathy. <i>Human Mutation</i> , 2012, 33, 949-959.	1.1	115
9	A novel syndrome caused by the E410K amino acid substitution in the neuronal β -tubulin isotype 3. <i>Brain</i> , 2013, 136, 522-535.	3.7	112
10	Juvenile myasthenia gravis. <i>Muscle and Nerve</i> , 2009, 39, 423-431.	1.0	110
11	Mutation Update and Genotype-Phenotype Correlations of Novel and Previously Described Mutations in <i>TPM2</i> and <i>TPM3</i> Causing Congenital Myopathies. <i>Human Mutation</i> , 2014, 35, 779-790.	1.1	92
12	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
13	Evidence-based guideline summary: Evaluation, diagnosis, and management of congenital muscular dystrophy. <i>Neurology</i> , 2015, 84, 1369-1378.	1.5	88
14	Emery-Dreifuss muscular dystrophy. <i>Muscle and Nerve</i> , 2020, 61, 436-448.	1.0	84
15	Regulation of DMD pathology by an ankyrin-encoded miRNA. <i>Skeletal Muscle</i> , 2011, 1, 27.	1.9	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. <i>Neuromuscular Disorders</i> , 2013, 23, 529-539.	0.3	79
17	The sensitivity of exome sequencing in identifying pathogenic mutations for LGMD in the United States. <i>Journal of Human Genetics</i> , 2017, 62, 243-252.	1.1	73
18	POMK mutations disrupt muscle development leading to a spectrum of neuromuscular presentations. <i>Human Molecular Genetics</i> , 2014, 23, 5781-5792.	1.4	72

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19	Mutations in the satellite cell gene MEGF10 cause a recessive congenital myopathy with minicores. <i>Neurogenetics</i> , 2012, 13, 115-124.	0.7	68
20	Variants in EXOSC9 Disrupt the RNA Exosome and Result in Cerebellar Atrophy with Spinal Motor Neuronopathy. <i>American Journal of Human Genetics</i> , 2018, 102, 858-873.	2.6	65
21	Comparison of Plasmapheresis and Intravenous Immunoglobulin as Maintenance Therapies for Juvenile Myasthenia Gravis. <i>JAMA Neurology</i> , 2014, 71, 575.	4.5	64
22	Serum Transaminase Levels in Boys With Duchenne and Becker Muscular Dystrophy. <i>Pediatrics</i> , 2011, 127, e132-e136.	1.0	63
23	Update on the Genetics of Limb Girdle Muscular Dystrophy. <i>Seminars in Pediatric Neurology</i> , 2012, 19, 211-218.	1.0	62
24	Childhood chronic inflammatory demyelinating polyradiculoneuropathy: Combined analysis of a large cohort and eleven published series. <i>Neuromuscular Disorders</i> , 2013, 23, 103-111.	0.3	62
25	Identifying diagnostic DNA methylation profiles for facioscapulohumeral muscular dystrophy in blood and saliva using bisulfite sequencing. <i>Clinical Epigenetics</i> , 2014, 6, 23.	1.8	61
26	Outcome reliability in non-ambulatory Boys/Men with duchenne muscular dystrophy. <i>Muscle and Nerve</i> , 2015, 51, 522-532.	1.0	60
27	Deficiency of base excision repair enzyme NEIL3 drives increased predisposition to autoimmunity. <i>Journal of Clinical Investigation</i> , 2016, 126, 4219-4236.	3.9	56
28	The child neurology clinical workforce in 2015. <i>Neurology</i> , 2016, 87, 1384-1392.	1.5	49
29	Update on juvenile myasthenia gravis. <i>Current Opinion in Pediatrics</i> , 2013, 25, 694-700.	1.0	43
30	AAV-Mediated TAZ Gene Replacement Restores Mitochondrial and Cardioskeletal Function in Barth Syndrome. <i>Human Gene Therapy</i> , 2019, 30, 139-154.	1.4	40
31	Transcriptional profile of postmortem skeletal muscle. <i>Physiological Genomics</i> , 2004, 16, 222-228.	1.0	38
32	Congenital Myasthenic Syndrome With Episodic Apnea. <i>Pediatric Neurology</i> , 2009, 41, 42-45.	1.0	38
33	Multifocal slowing of nerve conduction in metachromatic leukodystrophy. <i>Muscle and Nerve</i> , 2004, 29, 531-536.	1.0	37
34	Glycogen storage disease type IV: novel mutations and molecular characterization of a heterogeneous disorder. <i>Journal of Inherited Metabolic Disease</i> , 2010, 33, 83-90.	1.7	37
35	PLIN2 inhibits insulin-induced glucose uptake in myoblasts through the activation of the NLRP3 inflammasome. <i>International Journal of Molecular Medicine</i> , 2015, 36, 839-844.	1.8	36
36	The influence of muscle type and dystrophin deficiency on murine expression profiles. <i>Mammalian Genome</i> , 2005, 16, 739-748.	1.0	35

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37	The motor neuron response to SMN1 deficiency in spinal muscular atrophy. <i>Muscle and Nerve</i> , 2014, 49, 636-644.	1.0	34
38	Clinical trials in spinal muscular atrophy. <i>Current Opinion in Pediatrics</i> , 2007, 19, 675-679.	1.0	32
39	Exome sequencing identifies a novel SMCHD1 mutation in facioscapulohumeral muscular dystrophy 2. <i>Neuromuscular Disorders</i> , 2013, 23, 975-980.	0.3	32
40	Diagnostic value of electromyography and muscle biopsy in arthrogryposis multiplex congenita. <i>Annals of Neurology</i> , 2003, 54, 790-795.	2.8	31
41	Involvement of superficial peroneal sensory nerve in common peroneal neuropathy. <i>Muscle and Nerve</i> , 2005, 31, 725-729.	1.0	30
42	Consequences of MEGF10 deficiency on myoblast function and Notch1 interactions. <i>Human Molecular Genetics</i> , 2017, 26, 2984-3000.	1.4	30
43	Lactic Acid Elevation in Extramitochondrial Childhood Neurodegenerative Diseases. <i>Journal of Child Neurology</i> , 2001, 16, 657-660.	0.7	29
44	LGMD2I in a North American population. <i>BMC Musculoskeletal Disorders</i> , 2007, 8, 115.	0.8	29
45	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
46	Variations in gene expression among different types of human skeletal muscle. <i>Muscle and Nerve</i> , 2005, 32, 483-491.	1.0	28
47	Transcriptome-scale similarities between mouse and human skeletal muscles with normal and myopathic phenotypes. <i>BMC Musculoskeletal Disorders</i> , 2006, 7, 23.	0.8	28
48	The role of thymectomy in the treatment of juvenile myasthenia gravis: a systematic review. <i>Pediatric Surgery International</i> , 2017, 33, 683-694.	0.6	26
49	Autoimmune Neuromuscular Disorders in Childhood. <i>Current Treatment Options in Neurology</i> , 2011, 13, 590-607.	0.7	24
50	Recent developments in the treatment of Duchenne muscular dystrophy and spinal muscular atrophy. <i>Therapeutic Advances in Neurological Disorders</i> , 2013, 6, 147-160.	1.5	24
51	Silencing of drpr Leads to Muscle and Brain Degeneration in Adult Drosophila. <i>American Journal of Pathology</i> , 2014, 184, 2653-2661.	1.9	23
52	Advances in Muscular Dystrophies. <i>JAMA Neurology</i> , 2015, 72, 741.	4.5	23
53	Child Neurology: Chronic inflammatory demyelinating polyradiculoneuropathy in children. <i>Neurology</i> , 2008, 71, e74-8.	1.5	22
54	Clinical correlates of charcotâ€œmarieâ€œtooth disease in patients with pes cavus deformities. <i>Muscle and Nerve</i> , 2013, 47, 488-492.	1.0	22

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55	Inefficient dystrophin expression after cord blood transplantation in Duchenne muscular dystrophy. <i>Muscle and Nerve</i> , 2010, 41, 746-750.	1.0	21
56	Expanding the Phenotypic Spectrum and Variability of Endocrine Abnormalities Associated With TUBB3 E410K Syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015, 100, E473-E477.	1.8	21
57	POLRMT mutations impair mitochondrial transcription causing neurological disease. <i>Nature Communications</i> , 2021, 12, 1135.	5.8	21
58	Ambulatory foot temperature measurement: A new technique in polyneuropathy evaluation. <i>Muscle and Nerve</i> , 2003, 27, 737-742.	1.0	20
59	A Case of Congenital Glycogen Storage Disease Type IV With a Novel GBE1 Mutation. <i>Journal of Child Neurology</i> , 2008, 23, 349-352.	0.7	19
60	Response of Motor Complications in Cockayne Syndrome to Carbidopa-Levodopa. <i>Archives of Neurology</i> , 2008, 65, 1117-21.	4.9	17
61	Rapid reversal of uremic neuropathy following renal transplantation in an adolescent. <i>Pediatric Transplantation</i> , 2012, 16, E296-300.	0.5	17
62	Referral and diagnostic trends in pediatric electromyography in the molecular era. <i>Muscle and Nerve</i> , 2014, 50, 244-249.	1.0	17
63	AAV9-TAZ Gene Replacement Ameliorates Cardiac TMT Proteomic Profiles in a Mouse Model of Barth Syndrome. <i>Molecular Therapy - Methods and Clinical Development</i> , 2019, 13, 167-179.	1.8	17
64	The ties that bind: functional clusters in limb-girdle muscular dystrophy. <i>Skeletal Muscle</i> , 2020, 10, 22.	1.9	17
65	Cardiac electrophysiological characteristics of the mdx 5cv mouse model of Duchenne muscular dystrophy. <i>Journal of Interventional Cardiac Electrophysiology</i> , 2007, 20, 1-7.	0.6	16
66	Cysteine mutations cause defective tyrosine phosphorylation in MEGF10 myopathy. <i>FEBS Letters</i> , 2013, 587, 2952-2957.	1.3	16
67	Infantile Leukoencephalopathy Owing to Mitochondrial Enzyme Dysfunction. <i>Journal of Child Neurology</i> , 2002, 17, 421-428.	0.7	15
68	Efficient identification of novel mutations in patients with limb girdle muscular dystrophy. <i>Neurogenetics</i> , 2010, 11, 449-455.	0.7	15
69	Child Neurology Recruitment and Training: Views of Residents and Child Neurologists From the 2015 AAP/CNS Workforce Survey. <i>Pediatric Neurology</i> , 2017, 66, 89-95.	1.0	15
70	Impact of PYROXD1 deficiency on cellular respiration and correlations with genetic analyses of limb-girdle muscular dystrophy in Saudi Arabia and Sudan. <i>Physiological Genomics</i> , 2018, 50, 929-939.	1.0	15
71	Child neurology in the 21st century. <i>Neurology</i> , 2020, 94, 75-82.	1.5	15
72	A form of muscular dystrophy associated with pathogenic variants in JAG2. <i>American Journal of Human Genetics</i> , 2021, 108, 840-856.	2.6	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.	0.3	14
75	Utility and practice of electrodiagnostic testing in the pediatric population: An AANEM consensus statement. Muscle and Nerve, 2020, 61, 143-155.	1.0	14
76	Atypical presentations of spinal muscular atrophy type III (Kugelbergâ€“Welander disease). Neuromuscular Disorders, 2006, 16, 492-494.	0.3	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.	1.0	13
79	Homozygous nonsense mutation in <i>SGCA</i> is a common cause of limb-girdle muscular dystrophy in Assiut, Egypt. Muscle and Nerve, 2016, 54, 690-695.	1.0	12
80	Longitudinal Patterns of Thalidomide Neuropathy in Children and Adolescents. Journal of Pediatrics, 2016, 178, 227-232.	0.9	12
81	Medial medullary injury during adenoidectomy. Journal of Pediatrics, 2001, 138, 772-774.	0.9	11
82	Electrophysiologic features of fibular neuropathy in childhood and adolescence. Muscle and Nerve, 2017, 55, 693-697.	1.0	11
83	A slowly progressive form of limbâ€“girdle muscular dystrophy type 2C associated with founder mutation in the <i>SGCG</i> gene in Puerto Rican Hispanics. Molecular Genetics & Genomic Medicine, 2015, 3, 92-98.	0.6	10
84	Novel biallelic variants expand the SLC5A6-related phenotypic spectrum. European Journal of Human Genetics, 2022, 30, 439-449.	1.4	10
85	Diagnostic capabilities of nanopore longâ€“read sequencing in muscular dystrophy. Annals of Clinical and Translational Neurology, 2022, 9, 1302-1309.	1.7	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.	1.8	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.	0.6	9
88	Intravenous Immunoglobulin as a Therapeutic Option for Mycoplasma pneumoniae Encephalitis. Journal of Child Neurology, 2019, 34, 687-691.	0.7	8
89	Megf10 deficiency impairs skeletal muscle stem cell migration and muscle regeneration. FEBS Open Bio, 2021, 11, 114-123.	1.0	8
90	Selective serotonin reuptake inhibitors ameliorate MEGF10 myopathy. Human Molecular Genetics, 2019, 28, 2365-2377.	1.4	7

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91	Ethical decision-making for children with neuromuscular disorders in the COVID-19 crisis. <i>Neurology</i> , 2020, 95, 260-265.	1.5	7
92	Rituximab as Adjunct Maintenance Therapy for Refractory Juvenile Myasthenia Gravis. <i>Pediatric Neurology</i> , 2020, 111, 40-43.	1.0	7
93	<scp>hnRNP L</scp> is essential for myogenic differentiation and modulates myotonic dystrophy pathologies. <i>Muscle and Nerve</i> , 2021, 63, 928-940.	1.0	7
94	An Opportune Time for Newborn Screening in Duchenne Muscular Dystrophy. <i>JAMA Neurology</i> , 2021, 78, 901.	4.5	7
95	Phenotypic implications of pathogenic variant types in Pompe disease. <i>Journal of Human Genetics</i> , 2021, 66, 1089-1099.	1.1	6
96	Pediatric monomelic amyotrophy: Evidence for poliomyelitis in vulnerable populations. <i>Muscle and Nerve</i> , 2009, 40, 860-863.	1.0	5
97	Ethical issues in neurogenetic disorders. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2013, 118, 265-276.	1.0	5
98	Electrophysiologic Features of Radial Neuropathy in Childhood and Adolescence. <i>Pediatric Neurology</i> , 2018, 81, 14-18.	1.0	5
99	Dollars and antisense for Duchenne muscular dystrophy. <i>Neurology</i> , 2018, 90, 1091-1092.	1.5	5
100	The impact of Megf10/Drpr gain-of-function on muscle development in <i>Drosophila</i> . <i>FEBS Letters</i> , 2019, 593, 680-696.	1.3	5
101	Clinical, electrophysiological, and imaging findings in childhood brachial plexus injury. <i>Developmental Medicine and Child Neurology</i> , 2022, , .	1.1	5
102	Ethical issues in the evaluation of adults with suspected genetic neuromuscular disorders. <i>Muscle and Nerve</i> , 2016, 54, 997-1006.	1.0	4
103	Electrophysiologic features of ulnar neuropathy in childhood and adolescence. <i>Clinical Neurophysiology</i> , 2017, 128, 751-755.	0.7	4
104	Neurodevelopmental outcomes at 9-14 months gestational age after treatment of neonatal seizures due to brain injury. <i>Child's Nervous System</i> , 2019, 35, 1571-1578.	0.6	4
105	Growth charts in Cockayne syndrome type 1 and type 2. <i>European Journal of Medical Genetics</i> , 2021, 64, 104105.	0.7	4
106	Beyond the Gowers sign: measuring outcomes in Duchenne muscular dystrophy. <i>Muscle and Nerve</i> , 2013, 48, 315-317.	1.0	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. <i>Neurology</i> , 2015, 85, 1432-1433.	1.5	3
108	Neuropathic and Myopathic Pain. <i>Seminars in Pediatric Neurology</i> , 2016, 23, 242-247.	1.0	3

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109	Patient reported quality of life in limb girdle muscular dystrophy. <i>Neuromuscular Disorders</i> , 2022, 32, 57-64.	0.3	3
110	Lumbosacral ventral spinal nerve root atrophy identified on MRI in a case of spinal muscular atrophy type II. <i>Clinical Imaging</i> , 2019, 53, 134-137.	0.8	2
111	Presymptomatic and Early Symptomatic Genetic Testing. <i>CONTINUUM Lifelong Learning in Neurology</i> , 2011, 17, 343-346.	0.4	1
112	The struggle to model muscular dystrophy. <i>Muscle and Nerve</i> , 2011, 44, 157-159.	1.0	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. <i>Pediatric Neurology</i> , 2020, 102, 1-2.	1.0	1
116	Hunting for the perfect test: Neuromuscular diagnosis in the age of genomic bounty. <i>Muscle and Nerve</i> , 2021, 63, 282-284.	1.0	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. <i>Current Opinion in Pediatrics</i> , 1997, 9, 558-564.	1.0	0
121	The New Frontier of Genetically Targeted Therapies for Muscle Disease. <i>CONTINUUM Lifelong Learning in Neurology</i> , 2013, 19, 1698-1702.	0.4	0
122	Reply. <i>Muscle and Nerve</i> , 2014, 50, 458-459.	1.0	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. <i>FASEB Journal</i> , 2007, 21, A399.	0.2	0
126	Silencing of drpr leads to muscle and brain degeneration in adult <i>Drosophila</i> . <i>FASEB Journal</i> , 2013, 27, 873.14.	0.2	0

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127	Modeling Human MEGF10 Myopathy in <i>Drosophila melanogaster</i> . <i>FASEB Journal</i> , 2015, 29, 613.9.	0.2	0
128	Motor Unit Number Estimation. , 2017, , 151-155.		0
129	Muscle Analysis. , 2017, , 115-121.		0