Kristen J Nowak

List of Publications by Year
in descending order

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70 papers	3,552 citations	159585 30 h-index	138484 58 g-index
72 all docs	72 docs citations	72 times ranked	3742 citing authors

#	Article	IF	Citations
1	Mutations in the skeletal muscle Î±-actin gene in patients with actin myopathy and nemaline myopathy. Nature Genetics, 1999, 23, 208-212.	21.4	389
2	Molecular mechanisms of muscular dystrophies: old and new players. Nature Reviews Molecular Cell Biology, 2006, 7, 762-773.	37.0	300
3	Duchenne muscular dystrophy and dystrophin: pathogenesis and opportunities for treatment. EMBO Reports, 2004, 5, 872-876.	4.5	276
4	Mutations and polymorphisms of the skeletal muscle ÃŽÂ±-actin gene (ACTA1). Human Mutation, 2009, 30, 1267-1277.	2.5	198
5	Muscle disease caused by mutations in the skeletal muscle alpha-actin gene (ACTA1). Neuromuscular Disorders, 2003, 13, 519-531.	0.6	192
6	Dominant Mutations in KBTBD13, a Member of the BTB/Kelch Family, Cause Nemaline Myopathy with Cores. American Journal of Human Genetics, 2010, 87, 842-847.	6.2	143
7	Nemaline Myopathies. Seminars in Pediatric Neurology, 2011, 18, 230-238.	2.0	139
8	Genotype?phenotype correlations in nemaline myopathy caused by mutations in the genes for nebulin and skeletal muscle ?-actin. Neuromuscular Disorders, 2004, 14, 461-470.	0.6	107
9	Skeletal muscle Î±-actin diseases (actinopathies): pathology and mechanisms. Acta Neuropathologica, 2013, 125, 19-32.	7.7	98
10	Nemaline myopathy caused by absence of Î±-skeletal muscle actin. Annals of Neurology, 2007, 61, 175-184.	5.3	91
11	Evidence for a dominant-negative effect in ACTA1 nemaline myopathy caused by abnormal folding, aggregation and altered polymerization of mutant actin isoforms. Human Molecular Genetics, 2004, 13, 1727-1743.	2.9	89
12	Myopathy mutations in Î±-skeletal-muscle actin cause a range of molecular defects. Journal of Cell Science, 2004, 117, 3367-3377.	2.0	84
13	Fatal hypertrophic cardiomyopathy and nemaline myopathy associated with ACTA1 K336E mutation. Neuromuscular Disorders, 2006, 16, 548-552.	0.6	83
14	When contractile proteins go bad: the sarcomere and skeletal muscle disease. BioEssays, 2005, 27, 809-822.	2.5	82
15	Familial dilated cardiomyopathy mutations uncouple troponin I phosphorylation from changes in myofibrillar Ca2+ sensitivity. Cardiovascular Research, 2013, 99, 65-73.	3.8	68
16	Rescue of skeletal muscle Î±-actinâ€“null mice by cardiac (fetal) Î±-actin. Journal of Cell Biology, 2009, 185, 903-915.	5.2	67
17	Mouse models of dominant ACTA1 disease recapitulate human disease and provide insight into therapies. Brain, 2011, 134, 1101-1115.	7.6	60
18	Characterization of human muscle type cofilin (CFL2) in normal and regenerating muscle. FEBS Journal, 2001, 268, 3473-3482.	0.2	58

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19	Optimizing Precision Medicine for Public Health. Frontiers in Public Health, 2019, 7, 42.	2.7	58
20	Integrated Guidance for Enhancing the Care of Familial Hypercholesterolaemia in Australia. Heart Lung and Circulation, 2021, 30, 324-349.	0.4	51
21	The Evolution of Public Health Genomics: Exploring Its Past, Present, and Future. Frontiers in Public Health, 2018, 6, 247.	2.7	49
22	Zebrafish models for nemaline myopathy reveal a spectrum of nemaline bodies contributing to reduced muscle function. Acta Neuropathologica, 2015, 130, 389-406.	7.7	47
23	Variants in the Oxidoreductase PYROXD1 Cause Early-Onset Myopathy with Internalized Nuclei and Myofibrillar Disorganization. American Journal of Human Genetics, 2016, 99, 1086-1105.	6.2	45
24	K7del is a common TPM2 gene mutation associated with nemaline myopathy and raised myofibre calcium sensitivity. Brain, 2013, 136, 494-507.	7.6	42
25	Nebulin interactions with actin and tropomyosin are altered by disease-causing mutations. Skeletal Muscle, 2014, 4, 15.	4.2	39
26	Muscle weakness inTPM3-myopathy is due to reduced Ca²⁺-sensitivity and impaired acto-myosin cross-bridge cycling in slow fibres. Human Molecular Genetics, 2015, 24, 6278-6292.	2.9	38
27	Genomic Testing for Human Health and Disease Across the Life Cycle: Applications and Ethical, Legal, and Social Challenges. Frontiers in Public Health, 2019, 7, 40.	2.7	37
28	Dissociated flexor digitorum brevis myofiber culture systemâ€"A more mature muscle culture system. Cytoskeleton, 2007, 64, 727-738.	4.4	36
29	<scp>TPM</scp>3 deletions cause a hypercontractile congenital muscle stiffness phenotype. Annals of Neurology, 2015, 78, 982-994.	5.3	36
30	Severe nemaline myopathy caused by mutations of the stop codon of the skeletal muscle alpha actin gene (ACTA1). Neuromuscular Disorders, 2006, 16, 541-547.	0.6	35
31	Actin Nemaline Myopathy Mouse Reproduces Disease, Suggests Other Actin Disease Phenotypes and Provides Cautionary Note on Muscle Transgene Expression. PLoS ONE, 2011, 6, e28699.	2.5	30
32	Recent advances in understanding congenital myopathies. F1000Research, 2018, 7, 1921.	1.6	28
33	Targeted gene panel use in 2249 neuromuscular patients: the Australasian referral center experience. Annals of Clinical and Translational Neurology, 2020, 7, 353-362.	3.7	28
34	Disease Severity and Thin Filament Regulation in M9RTPM3Nemaline Myopathy. Journal of Neuropathology and Experimental Neurology, 2008, 67, 867-877.	1.7	27
35	Nemaline Myopathy-Related Skeletal Muscle Î±-Actin (ACTA1) Mutation, Asp286Gly, Prevents Proper Strong Myosin Binding and Triggers Muscle Weakness. PLoS ONE, 2012, 7, e45923.	2.5	27
36	Myostatin inhibition using mRK35 produces skeletal muscle growth and tubular aggregate formation in wild type and TgACTA1D286G nemaline myopathy mice. Human Molecular Genetics, 2018, 27, 638-648.	2.9	27

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37	Investigation of changes in skeletal muscle Î±-actin expression in normal and pathological human and mouse hearts. Journal of Muscle Research and Cell Motility, 2010, 31, 207-214.	2.0	25
38	Variable cardiac Î±-actin (Actc1) expression in early adult skeletal muscle correlates with promoter methylation. Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms, 2017, 1860, 1025-1036.	1.9	23
39	A novel ACTA1 mutation resulting in a severe congenital myopathy with nemaline bodies, intranuclear rods and type I fibre predominance. Neuromuscular Disorders, 2011, 21, 31-36.	0.6	22
40	Cardiac Â-actin over-expression therapy in dominant ACTA1 disease. Human Molecular Genetics, 2013, 22, 3987-3997.	2.9	22
41	Developmental and muscle-type-specific expression of mouse nebulin exons 127 and 128. Genomics, 2006, 88, 489-495.	2.9	20
42	Variable outcomes of human heart attack recapitulated in genetically diverse mice. Npj Regenerative Medicine, 2019, 4, 5.	5.2	20
43	Expression of cardiac Î±-actin spares extraocular muscles in skeletal muscle Î±-actin diseases â€“ Quantification of striated Î±-actins by MRM-mass spectrometry. Neuromuscular Disorders, 2008, 18, 953-958.	0.6	18
44	L-tyrosine supplementation does not ameliorate skeletal muscle dysfunction in zebrafish and mouse models of dominant skeletal muscle Î±-actin nemaline myopathy. Scientific Reports, 2018, 8, 11490.	3.3	18
45	Healthcare System Priorities for Successful Integration of Genomics: An Australian Focus. Frontiers in Public Health, 2019, 7, 41.	2.7	18
46	Production of human skeletal Î±-actin proteins by the baculovirus expression system. Biochemical and Biophysical Research Communications, 2003, 307, 74-79.	2.1	16
47	Multimodal MRI and 31P-MRS Investigations of the ACTA1(Asp286Gly) Mouse Model of Nemaline Myopathy Provide Evidence of Impaired In Vivo Muscle Function, Altered Muscle Structure and Disturbed Energy Metabolism. PLoS ONE, 2013, 8, e72294.	2.5	15
48	Skeletal and cardiac Î±-actin isoforms differently modulate myosin cross-bridge formation and myofibre force production. Human Molecular Genetics, 2013, 22, 4398-4404.	2.9	13
49	Systemic AAV8-mediated delivery of a functional copy of muscle glycogen phosphorylase (Pygm) ameliorates disease in a murine model of McArdle disease. Human Molecular Genetics, 2020, 29, 20-30.	2.9	12
50	Clinical utility gene card for: Nemaline myopathy â€“ update 2015. European Journal of Human Genetics, 2015, 23, 4-5.	2.8	11
51	Novel application of flow cytometry: Determination of muscle fiber types and protein levels in whole murine skeletal muscles and heart. Cytoskeleton, 2007, 64, 914-925.	4.4	10
52	X-ray recordings reveal how a human disease-linked skeletal muscle Î±-actin mutation leads to contractile dysfunction. Journal of Structural Biology, 2015, 192, 331-335.	2.8	10
53	Direct visualisation and kinetic analysis of normal and nemaline myopathy actin polymerisation using total internal reflection microscopy. Journal of Muscle Research and Cell Motility, 2009, 30, 85-92.	2.0	9
54	Extramuscular manifestations in children with severe congenital myopathy due to ACTA1 gene mutations. Neuromuscular Disorders, 2011, 21, 489-493.	0.6	9

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55	Clinical utility gene card for: Nemaline myopathy. European Journal of Human Genetics, 2012, 20, 713-713.	2.8	7
56	Incidental inequity. European Journal of Human Genetics, 2018, 26, 616-617.	2.8	7
57	Nebulin nemaline myopathy recapitulated in a compound heterozygous mouse model with both a missense and a nonsense mutation in Neb. Acta Neuropathologica Communications, 2020, 8, 18.	5.2	7
58	Molecular Consequences of the Myopathy-Related D286G Mutation on Actin Function. Frontiers in Physiology, 2018, 9, 1756.	2.8	6
59	Clinical utility gene card for McArdle disease. European Journal of Human Genetics, 2018, 26, 758-764.	2.8	4
60	Ovine congenital progressive muscular dystrophy (OCPMD) is a model of TNNT1 congenital myopathy. Acta Neuropathologica Communications, 2020, 8, 142.	5.2	4
61	Essentials of a new clinical practice guidance on familial hypercholesterolaemia for physicians. Internal Medicine Journal, 2021, 51, 769-779.	0.8	4
62	Generation of two isogenic induced pluripotent stem cell lines from a 4-month-old severe nemaline myopathy patient with a heterozygous dominant c.553CÂ>ÂA (p.Arg183Ser) variant in the ACTA1 gene. Stem Cell Research, 2021, 53, 102273.	0.7	4
63	CUGC for Duchenne muscular dystrophy (DMD). European Journal of Human Genetics, 2018, 26, 749-757.	2.8	3
64	Clinical Utility Gene Card for: Becker muscular dystrophy. European Journal of Human Genetics, 2018, 26, 1065-1071.	2.8	2
65	Descriptive epidemiological study of rare, less common and common cancers in Western Australia. BMC Cancer, 2021, 21, 779.	2.6	2
66	Therapeutic Approaches for the Sarcomeric Protein Diseases. Advances in Experimental Medicine and Biology, 2008, 642, 207-223.	1.6	2
67	Clinical Utility Gene Card for: autosomal dominant myotonia congenita (Thomsen Disease). European Journal of Human Genetics, 2018, 26, 1072-1077.	2.8	1
68	Generation of two isogenic induced pluripotent stem cell lines from a 10-year-old typical nemaline myopathy patient with a heterozygous dominant c.541G>A (p.Asp179Asn) pathogenic variant in the ACTA1 gene. Stem Cell Research, 2021, 55, 102482.	0.7	1
69	Generation of two isogenic induced pluripotent stem cell lines from a 1-month-old nemaline myopathy patient harbouring a homozygous recessive c.121C>T (p.Arg39Ter) variant in the ACTA1 gene. Stem Cell Research, 2022, , 102830.	0.7	1
70	Generation of an induced pluripotent stem cell line from a 3-month-old nemaline myopathy patient with a heterozygous dominant c.515C>A (p.Ala172Glu) variant in the ACTA1 gene. Stem Cell Research, 2022, , 102829.	0.7	0