Gianina Ravenscroft

List of Publications by Year
in descending order

92 papers	3,444 citations	136950 32 h-index	161849 54 g-index
112 all docs	112 docs citations	112 times ranked	4545 citing authors

#	Article	IF	Citations
1	A TOR1AIP1 variant segregating with an early onset limb girdle myastheniaâ€"Support for the role of LAP1 in NMJ function and disease. Neuropathology and Applied Neurobiology, 2022, 48, .	3.2	4
2	Comprehensive genetic diagnosis of tandem repeat expansion disorders with programmable targeted nanopore sequencing. Science Advances, 2022, 8, eabm5386.	10.3	68
3	Bi-allelic MYH3 loss-of-function variants cause a lethal form of contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B. Neuromuscular Disorders, 2022, 32, 445-449.	0.6	2
4	FXR1-related congenital myopathy: expansion of the clinical and genetic spectrum. Journal of Medical Genetics, 2022, 59, 1069-1074.	3.2	1
5	Bi-allelic loss-of-function OBSCN variants predispose individuals to severe recurrent rhabdomyolysis. Brain, 2022, 145, 3985-3998.	7.6	6
6	Partial lossâ€ofâ€function variant in neuregulin 1 identified in family with heritable peripheral neuropathy. Human Mutation, 2022, 43, 1216-1223.	2.5	4
7	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
8	Neurogenetic fetal akinesia and arthrogryposis: genetics, expanding genotype-phenotypes and functional genomics. Journal of Medical Genetics, 2021, 58, 609-618.	3.2	46
9	Heterozygous CAPN3 missense variants causing autosomalâ€dominant calpainopathy in seven unrelated families. Neuropathology and Applied Neurobiology, 2021, 47, 283-296.	3.2	23
10	Biallelic hypomorphic variants in ALDH1A2 cause a novel lethal human multiple congenital anomaly syndrome encompassing diaphragmatic, pulmonary, and cardiovascular defects. Human Mutation, 2021, 42, 506-519.	2.5	12
11	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
12	Genetic neuromuscular disorders: what is the best that we can do?. Neuromuscular Disorders, 2021, 31, 1081-1089.	0.6	4
13	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
14	Improved CRISPR/Cas9 gene editing in primary human myoblasts using low confluency cultures on Matrigel. Skeletal Muscle, 2021, 11, 23.	4.2	4
15	Another step towards defining the genetic landscape of rhabdomyolysis. Brain, 2021, 144, 2560-2561.	7.6	1
16	Pseudoperoxidase activity, conformational stability and aggregation propensity of the His98Tyr myoglobin variant: Implications for the onset of myoglobinopathy. FEBS Journal, 2021, , .	4.7	1
17	Victor(iou)â€™s myologists: snapshots of a legacy. Neuromuscular Disorders, 2021, 31, 1096-1099.	0.6	0
18	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

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19	Cylindrical spirals in two families: Clinical and genetic investigations. Neuromuscular Disorders, 2020, 30, 151-158.	0.6	7
20	The Impact of Next-Generation Sequencing on the Diagnosis, Treatment, and Prevention of Hereditary Neuromuscular Disorders. Molecular Diagnosis and Therapy, 2020, 24, 641-652.	3.8	7
21	A novel RFC1 repeat motif (ACAGG) in two Asia-Pacific CANVAS families. Brain, 2020, 143, 2904-2910.	7.6	53
22	A MÄori specific RFC1 pathogenic repeat configuration in CANVAS, likely due to a founder allele. Brain, 2020, 143, 2673-2680.	7.6	45
23	Ovine congenital progressive muscular dystrophy (OCPMD) is a model of TNNT1 congenital myopathy. Acta Neuropathologica Communications, 2020, 8, 142.	5.2	4
24	NEMF mutations that impair ribosome-associated quality control are associated with neuromuscular disease. Nature Communications, 2020, 11, 4625.	12.8	47
25	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
26	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
27	Cerebellar ataxia, neuropathy, vestibular areflexia syndrome due to RFC1 repeat expansion. Brain, 2020, 143, 480-490.	7.6	140
28	A homozygous UBA5 pathogenic variant causes a fatal congenital neuropathy. Journal of Medical Genetics, 2020, 57, 835-842.	3.2	16
29	O.16Diagnosis of fetal akinesia and arthrogryposis by panel sequencing and functional genomics. Neuromuscular Disorders, 2019, 29, S119.	0.6	0
30	Study protocol of a multicentre cohort pilot study implementing an expanded preconception carrier-screening programme in metropolitan and regional Western Australia. BMJ Open, 2019, 9, e028209.	1.9	3
31	Altered myogenesis and premature senescence underlie human TRIM32-related myopathy. Acta Neuropathologica Communications, 2019, 7, 30.	5.2	24
32	Myoglobinopathy is an adult-onset autosomal dominant myopathy with characteristic sarcoplasmic inclusions. Nature Communications, 2019, 10, 1396.	12.8	11
33	Recessive MYH7-related myopathy in two families. Neuromuscular Disorders, 2019, 29, 456-467.	0.6	14
34	Ryanodine receptor type 3 (<scp>RYR</scp>3) as a novel gene associated with a myopathy with nemaline bodies. European Journal of Neurology, 2018, 25, 841-847.	3.3	31
35	Mutations in ATP1A1 Cause Dominant Charcot-Marie-Tooth Type 2. American Journal of Human Genetics, 2018, 102, 505-514.	6.2	59
36	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	5.3	93

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37	Recent advances in understanding congenital myopathies. F1000Research, 2018, 7, 1921.	1.6	28
38	Biallelic mutations in nucleoporin NUP88 cause lethal fetal akinesia deformation sequence. PLoS Genetics, 2018, 14, e1007845.	3.5	26
39	Bi-allelic mutations in MYL1 cause a severe congenital myopathy. Human Molecular Genetics, 2018, 27, 4263-4272.	2.9	31
40	Variants in <scp>ACTG</scp>2 underlie a substantial number of Australasian patients with primary chronic intestinal pseudoâ€obstruction. Neurogastroenterology and Motility, 2018, 30, e13371.	3.0	23
41	Genetics of neuromuscular fetal akinesia in the genomics era. Journal of Medical Genetics, 2018, 55, 505-514.	3.2	35
42	Measuring the impact of genetic knowledge on intentions and attitudes of the community towards expanded preconception carrier screening. Journal of Medical Genetics, 2018, 55, 744-752.	3.2	34
43	Lethal multiple pterygium syndrome: A severe phenotype associated with a novel mutation in the nebulin gene. Neuromuscular Disorders, 2017, 27, 537-541.	0.6	18
44	Expanding the phenotypic spectrum associated with mutations of DYNC1H1. Neuromuscular Disorders, 2017, 27, 607-615.	0.6	29
45	TOR1A variants cause a severe arthrogryposis with developmental delay, strabismus and tremor. Neuromuscular Disorders, 2017, 27, S243.	0.6	0
46	New era in genetics of early-onset muscle disease: Breakthroughs and challenges. Seminars in Cell and Developmental Biology, 2017, 64, 160-170.	5.0	24
47	TOR1A variants cause a severe arthrogryposis with developmental delay, strabismus and tremor. Brain, 2017, 140, 2851-2859.	7.6	42
48	CAV3 mutations causing exercise intolerance, myalgia and rhabdomyolysis: Expanding the phenotypic spectrum of caveolinopathies. Neuromuscular Disorders, 2016, 26, 504-510.	0.6	38
49	Ryanodine receptor type 3 (RYR3) as a novel gene associated with nemaline myopathy and fibre type disproportion. Neuromuscular Disorders, 2016, 26, S137.	0.6	3
50	Biallelic PPA2 Mutations Cause Sudden Unexpected Cardiac Arrest in Infancy. American Journal of Human Genetics, 2016, 99, 666-673.	6.2	39
51	Recurrent de novo BICD2 mutation associated with arthrogryposis multiplex congenita and bilateral perisylvian polymicrogyria. Neuromuscular Disorders, 2016, 26, 744-748.	0.6	44
52	Loss-of-function mutations inSCN4Acause severe foetal hypokinesia or â€˜classicalâ€™ congenital myopathy. Brain, 2016, 139, 674-691.	7.6	100
53	Next generation sequencing in a large cohort of patients presenting with neuromuscular disease before or at birth. Orphanet Journal of Rare Diseases, 2015, 10, 148.	2.7	94
54	Mutations of GPR126 Are Responsible for Severe Arthrogryposis Multiplex Congenita. American Journal of Human Genetics, 2015, 96, 955-961.	6.2	92

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55	Pathophysiological concepts in the congenital myopathies: blurring the boundaries, sharpening the focus. Brain, 2015, 138, 246-268.	7.6	70
56	Expanding the phenotype of GMPPB mutations. Brain, 2015, 138, 836-844.	7.6	54
57	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
58	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
59	Pathology provides clarity in the next-generation sequencing era. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 479-480.	1.9	2
60	Distal arthrogryposis type 5D with novel clinical features and compound heterozygous mutations in ECEL1. American Journal of Medical Genetics, Part A, 2014, 164, 1846-1849.	1.2	14
61	SPEG Interacts with Myotubularin, and Its Deficiency Causes Centronuclear Myopathy with Dilated Cardiomyopathy. American Journal of Human Genetics, 2014, 95, 218-226.	6.2	143
62	Novel cofilin-2 (CFL2) four base pair deletion causing nemaline myopathy. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 1058-1060.	1.9	24
63	G.P.18. Neuromuscular Disorders, 2014, 24, 799-800.	0.6	3
64	KLHL40 deficiency destabilizes thin filament proteins and promotes nemaline myopathy. Journal of Clinical Investigation, 2014, 124, 3529-3539.	8.2	103
65	Leiomodin-3 dysfunction results in thin filament disorganization and nemaline myopathy. Journal of Clinical Investigation, 2014, 124, 4693-4708.	8.2	153
66	Identification of KLHL41 Mutations Implicates BTB-Kelch-Mediated Ubiquitination as an Alternate Pathway to Myofibrillar Disruption in Nemaline Myopathy. American Journal of Human Genetics, 2013, 93, 1108-1117.	6.2	147
67	Mutations in KLHL40 Are a Frequent Cause of Severe Autosomal-Recessive Nemaline Myopathy. American Journal of Human Genetics, 2013, 93, 6-18.	6.2	186
68	Familial dilated cardiomyopathy mutations uncouple troponin I phosphorylation from changes in myofibrillar Ca2+ sensitivity. Cardiovascular Research, 2013, 99, 65-73.	3.8	68
69	Skeletal muscle Î±-actin diseases (actinopathies): pathology and mechanisms. Acta Neuropathologica, 2013, 125, 19-32.	7.7	98
70	Whole exome sequencing in foetal akinesia expands the genotypeâ€“phenotype spectrum of GBE1 glycogen storage disease mutations. Neuromuscular Disorders, 2013, 23, 165-169.	0.6	39
71	K7del is a common TPM2 gene mutation associated with nemaline myopathy and raised myofibre calcium sensitivity. Brain, 2013, 136, 494-507.	7.6	42
72	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

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73	Cardiac Â-actin over-expression therapy in dominant ACTA1 disease. Human Molecular Genetics, 2013, 22, 3987-3997.	2.9	22
74	Nemaline myopathy with stiffness and hypertonia associated with an ACTA1 mutation. Neurology, 2012, 78, 1100-1103.	1.1	43
75	Skeletal Muscle Myopathy Mutations at the Actin-Tropomyosin Interface that Cause Gain Or Loss of Function. Biophysical Journal, 2012, 102, 231a.	0.5	0
76	D.P.6 Whole exome sequencing applied to foetal akinesia. Neuromuscular Disorders, 2012, 22, 809.	0.6	0
77	C.O.3 Efficacy of cardiac actin over-expression therapy for ACTA1 disease seems mutation specific. Neuromuscular Disorders, 2012, 22, 839.	0.6	0
78	C.P.18 Skeletal muscle myopathy mutations at the actin tropomyosin interface that cause gain or loss of function. Neuromuscular Disorders, 2012, 22, 846.	0.6	0
79	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
80	Fetal akinesia: review of the genetics of the neuromuscular causes. Journal of Medical Genetics, 2011, 48, 793-801.	3.2	65
81	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
82	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
83	Dominant Mutations in KBTBD13, a Member of the BTB/Kelch Family, Cause Nemaline Myopathy with Cores. American Journal of Human Genetics, 2011, 88, 122.	6.2	0
84	Mutations in the N-terminal Actin-Binding Domain of Filamin C Cause a Distal Myopathy. American Journal of Human Genetics, 2011, 88, 729-740.	6.2	124
85	Mouse models of dominant ACTA1 disease recapitulate human disease and provide insight into therapies. Brain, 2011, 134, 1101-1115.	7.6	60
86	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
87	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
88	Evidence of Altered Guinea Pig Ventricular Cardiomyocyte Protein Expression and Growth in Response to a 5 min in vitro Exposure to H₂O₂. Journal of Proteome Research, 2010, 9, 1985-1994.	3.7	26
89	Rescue of skeletal muscle Î±-actinâ€“null mice by cardiac (fetal) Î±-actin. Journal of Cell Biology, 2009, 185, 903-915.	5.2	67
90	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

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91	Dissociated flexor digitorum brevis myofiber culture systemâ€"A more mature muscle culture system. Cytoskeleton, 2007, 64, 727-738.	4.4	36
92	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