Marc Bartoli

List of Publications by Year
in descending order

108 papers	3,194 citations	136885 32 h-index	168321 53 g-index
118 all docs	118 docs citations	118 times ranked	3840 citing authors

#	Article	IF	Citations
1	Constitutive Activation of the Calcium Sensor STIM1 Causes Tubular-Aggregate Myopathy. American Journal of Human Genetics, 2013, 92, 271-278.	2.6	169
2	Calpains in muscle wasting. International Journal of Biochemistry and Cell Biology, 2005, 37, 2115-2133.	1.2	155
3	Calpain 3 Is Activated through Autolysis within the Active Site and Lyses Sarcomeric and Sarcolemmal Components. Molecular and Cellular Biology, 2003, 23, 9127-9135.	1.1	136
4	VarAFT: a variant annotation and filtration system for human next generation sequencing data. Nucleic Acids Research, 2018, 46, W545-W553.	6.5	136
5	Efficient recovery of dysferlin deficiency by dual adeno-associated vector-mediated gene transfer. Human Molecular Genetics, 2010, 19, 1897-1907.	1.4	122
6	Imaging Calpain Protease Activity by Multiphoton FRET in Living Mice. Journal of Molecular Biology, 2005, 346, 215-222.	2.0	115
7	Calpain 3: a key regulator of the sarcomere?. FEBS Journal, 2006, 273, 3427-3436.	2.2	115
8	A novel calmodulin-binding protein, belonging to the WD-repeat family, is localized in dendrites of a subset of CNS neurons.. Journal of Cell Biology, 1996, 134, 1051-1062.	2.3	114
9	Interactions of the Rapsyn RING-H2 Domain with Dystroglycan. Journal of Biological Chemistry, 2001, 276, 24911-24917.	1.6	89
10	Differential DNA methylation of the D4Z4 repeat in patients with FSHD and asymptomatic carriers. Neurology, 2014, 83, 733-742.	1.5	82
11	Efficient bypass of mutations in dysferlin deficient patient cells by antisense-induced exon skipping. Human Mutation, 2010, 31, 136-142.	1.1	80
12	AAV-mediated delivery of a mutated myostatin propeptide ameliorates calpain 3 but not Î±-sarcoglycan deficiency. Gene Therapy, 2007, 14, 733-740.	2.3	74
13	Deregulation of the Protocadherin Gene FAT1 Alters Muscle Shapes: Implications for the Pathogenesis of Facioscapulohumeral Dystrophy. PLoS Genetics, 2013, 9, e1003550.	1.5	73
14	A Naturally Occurring Human Minidysferlin Protein Repairs Sarcolemmal Lesions in a Mouse Model of Dysferlinopathy. Science Translational Medicine, 2010, 2, 50ra69.	5.8	69
15	Phenotypic Correction of Î±-Sarcoglycan Deficiency by Intra-arterial Injection of a Muscle-specific Serotype 1 rAAV Vector. Molecular Therapy, 2007, 15, 53-61.	3.7	63
16	Safety and Efficacy of AAV-Mediated Calpain 3 Gene Transfer in a Mouse Model of Limb-Girdle Muscular Dystrophy Type 2A. Molecular Therapy, 2006, 13, 250-259.	3.7	62
17	Calcium-dependent plasma membrane repair requires m- or Î¼-calpain, but not calpain-3, the proteasome, or caspases. Biochimica Et Biophysica Acta - Molecular Cell Research, 2009, 1793, 1886-1893.	1.9	61
18	Lack of Correlation between Outcomes of Membrane Repair Assay and Correction of Dystrophic Changes in Experimental Therapeutic Strategy in Dysferlinopathy. PLoS ONE, 2012, 7, e38036.	1.1	61

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19	Striatin, a calmodulin-dependent scaffolding protein, directly binds caveolin-1. FEBS Letters, 2001, 508, 49-52.	1.3	56
20	Cardiac ankyrin repeat protein is a marker of skeletal muscle pathological remodelling. FEBS Journal, 2009, 276, 669-684.	2.2	56
21	NFâ€NF<scp>â€Î°</scp>BBâ€dependent expression of the antiapoptotic factor câ€FLIP is regulated by calpain 3, the protein involved in limbâ€girdle muscular dystrophy type 2A. FASEB Journal, 2008, 22, 1521-1529.	0.2	55
22	Mannosidase I inhibition rescues the human Î±-sarcoglycan R77C recurrent mutation. Human Molecular Genetics, 2008, 17, 1214-1221.	1.4	54
23	Interaction of Calmodulin with Striatin, a WD-repeat Protein Present in Neuronal Dendritic Spines. Journal of Biological Chemistry, 1998, 273, 22248-22253.	1.6	50
24	A third of LGMD2A biopsies have normal calpain 3 proteolytic activity as determined by an in vitro assay. Neuromuscular Disorders, 2007, 17, 148-156.	0.3	46
25	Dysregulation of 4q35- and muscle-specific genes in fetuses with a short D4Z4 array linked to facio-scapulo-humeral dystrophy. Human Molecular Genetics, 2013, 22, 4206-4214.	1.4	45
26	Down-regulation of striatin, a neuronal calmodulin-binding protein, impairs rat locomotor activity. , 1999, 40, 234-243.		43
27	Ins and outs of therapy in limb girdle muscular dystrophies. International Journal of Biochemistry and Cell Biology, 2007, 39, 1608-1624.	1.2	42
28	Restriction of Calpain3 Expression to the Skeletal Muscle Prevents Cardiac Toxicity and Corrects Pathology in a Murine Model of Limb-Girdle Muscular Dystrophy. Circulation, 2013, 128, 1094-1104.	1.6	40
29	Molecular combing reveals complex 4q35 rearrangements in Facioscapulohumeral dystrophy. Human Mutation, 2017, 38, 1432-1441.	1.1	39
30	Identification of Variants in the 4q35 GeneFAT1in Patients with a Facioscapulohumeral Dystrophy-Like Phenotype. Human Mutation, 2015, 36, 443-453.	1.1	38
31	A new pathway encompassing calpain 3 and its newly identified substrate cardiac ankyrin repeat protein is involved in the regulation of the nuclear factorâ€Î°B pathway in skeletal muscle. FEBS Journal, 2010, 277, 4322-4337.	2.2	37
32	Translational Research and Therapeutic Perspectives in Dysferlinopathies. Molecular Medicine, 2011, 17, 875-882.	1.9	36
33	Rescue of sarcoglycan mutations by inhibition of endoplasmic reticulum quality control is associated with minimal structural modifications. Human Mutation, 2012, 33, 429-439.	1.1	36
34	A human skeletal muscle interactome centered on proteins involved in muscular dystrophies: LGMD interactome. Skeletal Muscle, 2013, 3, 3.	1.9	36
35	UMD-DYSF, a novel locus specific database for the compilation and interactive analysis of mutations in the dysferlin gene. Human Mutation, 2012, 33, E2317-E2331.	1.1	35
36	A codon-optimized Mecp2 transgene corrects breathing deficits and improves survival in a mouse model of Rett syndrome. Neurobiology of Disease, 2017, 99, 1-11.	2.1	35

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37	Low penetrance in facioscapulohumeral muscular dystrophy type 1 with large pathological D4Z4 alleles: a cross-sectional multicenter study. Orphanet Journal of Rare Diseases, 2015, 10, 2.	1.2	32
38	Noninvasive monitoring of therapeutic gene transfer in animal models of muscular dystrophies. Gene Therapy, 2006, 13, 20-28.	2.3	30
39	Muscle Cells Fix Breaches by Orchestrating a Membrane Repair Ballet. Journal of Neuromuscular Diseases, 2018, 5, 21-28.	1.1	30
40	Exon 32 Skipping of Dysferlin Rescues Membrane Repair in Patientsâ€™ Cells. Journal of Neuromuscular Diseases, 2015, 2, 281-290.	1.1	29
41	Exome sequencing as a secondâ€tier diagnostic approach for clinically suspected dysferlinopathy patients. Muscle and Nerve, 2014, 50, 1007-1010.	1.0	26
42	Distribution of Striatin, a newly identified calmodulin-binding protein in the rat brain: An in situ hybridization and immunocytochemical study. , 1998, 397, 41-59.		25
43	Cloning of Human Striatin cDNA (STRN), Gene Mapping to 2p22â€“p21, and Preferential Expression in Brain. Genomics, 1998, 51, 136-139.	1.3	25
44	A Mouse Model for Monitoring Calpain Activity under Physiological and Pathological Conditions. Journal of Biological Chemistry, 2006, 281, 39672-39680.	1.6	24
45	Truncated prelamin A expression in HGPS-like patients: a transcriptional study. European Journal of Human Genetics, 2015, 23, 1051-1061.	1.4	24
46	Eosinophilic infiltration related to CAPN3 mutations: a pathophysiological component of primary calpainopathy?. Clinical Genetics, 2011, 80, 398-402.	1.0	22
47	Identification of Splicing Defects Caused by Mutations in the Dysferlin Gene. Human Mutation, 2014, 35, 1532-1541.	1.1	22
48	Relationships between striatin-containing neurons and cortical or thalamic afferent fibres in the rat striatum. An ultrastructural study by dual labelling. Neuroscience, 1998, 85, 111-122.	1.1	21
49	Down-regulation of striatin, a neuronal calmodulin-binding protein, impairs rat locomotor activity. Journal of Neurobiology, 1999, 40, 234-43.	3.7	21
50	Improving molecular diagnosis of distal myopathies by targeted next-generation sequencing: TableÂ1. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 340-342.	0.9	20
51	Correction of pseudoexon splicing caused by a novel intronic dysferlin mutation. Annals of Clinical and Translational Neurology, 2019, 6, 642-654.	1.7	20
52	Novel CAPN3 variant associated with an autosomal dominant calpainopathy. Neuropathology and Applied Neurobiology, 2020, 46, 564-578.	1.8	20
53	Comparing targeted exome and whole exome approaches for genetic diagnosis of neuromuscular disorders. Applied & Translational Genomics, 2015, 7, 26-31.	2.1	18
54	Loss of Cajal bodies in motor neurons from patients with novel mutations in VRK1. Human Molecular Genetics, 2019, 28, 2378-2394.	1.4	17

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55	Methylation hotspots evidenced by deep sequencing in patients with facioscapulohumeral dystrophy and mosaicism. Neurology: Genetics, 2019, 5, e372.	0.9	16
56	Segregation between SMCHD1 mutation, D4Z4 hypomethylation and Facio-Scapulo-Humeral Dystrophy: a case report. BMC Medical Genetics, 2016, 17, 66.	2.1	13
57	Distribution of striatin, a newly identified calmodulin-binding protein in the rat brain: an in situ hybridization and immunocytochemical study. Journal of Comparative Neurology, 1998, 397, 41-59.	0.9	13
58	Immunolabelling and flow cytometry as new tools to explore dysferlinopathies. Neuromuscular Disorders, 2010, 20, 57-60.	0.3	12
59	Clinical heterogeneity and phenotype/genotype findings in 5 families with GYG1 deficiency. Neurology: Genetics, 2017, 3, e208.	0.9	12
60	Novel Pathogenic Variants in a French Cohort Widen the Mutational Spectrum ofÂGNE Myopathy. Journal of Neuromuscular Diseases, 2015, 2, 131-136.	1.1	11
61	Clinical massively parallel sequencing for the diagnosis of myopathies. Revue Neurologique, 2015, 171, 558-571.	0.6	11
62	Retrospective analysis and reclassification of DYSF variants in a large French series of dysferlinopathy patients. Genetics in Medicine, 2021, 23, 1574-1577.	1.1	11
63	Rs488087 single nucleotide polymorphism as predictive risk factor for pancreatic cancers. Oncotarget, 2015, 6, 39855-39864.	0.8	11
64	G.O.5 Partial functionality of a Mini-dysferlin molecule identified in a patient affected with moderately severe primary dysferlinopathy. Neuromuscular Disorders, 2008, 18, 781.	0.3	10
65	Splicing impact of deep exonic missense variants in CAPN3 explored systematically by minigene functional assay. Human Mutation, 2020, 41, 1797-1810.	1.1	9
66	Exclusion of Mutations in the Dysferlin Alternative Exons 1 of DYSF-v1, 5a, and 40a in a Cohort of 26 Patients. Genetic Testing and Molecular Biomarkers, 2010, 14, 153-154.	0.3	8
67	Validation of comparative genomic hybridization arrays for the detection of genomic rearrangements of the calpainâ€3 and dysferlin genes. Clinical Genetics, 2012, 81, 99-101.	1.0	8
68	Therapeutic exon â€˜switchingâ€™ for dysferlinopathies?. European Journal of Human Genetics, 2010, 18, 969-970.	1.4	7
69	Autosomal dominant segregation of CAPN3 c.598_612del15 associated with a mild form of calpainopathy. Annals of Clinical and Translational Neurology, 2020, 7, 2538-2540.	1.7	7
70	Genetic Characterization of a French Cohort of GNEâ€mutation negative inclusion body myopathy patients with exome sequencing. Muscle and Nerve, 2017, 56, 993-997.	1.0	6
71	Entire CAPN3 gene deletion in a patient with limbâ€girdle muscular dystrophy type 2A. Muscle and Nerve, 2014, 50, 448-453.	1.0	5
72	Dysferlin Exon 32 Skipping in Patient Cells. Methods in Molecular Biology, 2018, 1828, 489-496.	0.4	4

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73	First characterization of congenital myasthenic syndrome type 5 in North Africa. Molecular Biology Reports, 2021, 48, 6999-7006.	1.0	4
74	Coverage analysis of lists of genes involved in heterogeneous genetic diseases following benchtop exome sequencing using the ion proton. Journal of Genetics, 2016, 95, 203-208.	0.4	3
75	A novel biâ€allelic lossâ€ofâ€function mutation in <scp>STIM1</scp> expands the phenotype of <scp>STIM1</scp>â€related diseases. Clinical Genetics, 2021, 100, 84-89.	1.0	3
76	Altered action potential waveform and shorter axonal initial segment in hiPSC-derived motor neurons with mutations in VRK1. Neurobiology of Disease, 2022, 164, 105609.	2.1	3
77	Genetic Profile of Patients with Limb-Girdle Muscle Weakness in the Chilean Population. Genes, 2022, 13, 1076.	1.0	3
78	Dystrophies musculaires des ceinturesÂ: stratÃ©gie diagnostique, bases molÃ©culaires. Revue Du Rhumatisme (Edition Francaise), 2008, 75, 142-150.	0.0	2
79	Further heterogeneity in myopathy with tubular aggregates?. Muscle and Nerve, 2012, 46, 983-984.	1.0	2
80	Eosinophils in Human Disease. , 2013, , 431-536.		2
81	Comment on: A novel dysferlinâ€mutant pseudoexon bypassed with antisense oligonucleotides. Annals of Clinical and Translational Neurology, 2015, 2, 783-784.	1.7	2
82	Extension of the phenotypic spectrum of GLE1â€related disorders to a mild congenital form resembling congenital myopathy. Molecular Genetics & Genomic Medicine, 2020, 8, e1277.	0.6	2
83	Identification of novel mutations by targeted NGS in Moroccan families clinically diagnosed with a neuromuscular disorder. Clinica Chimica Acta, 2022, 524, 51-58.	0.5	2
84	The Dysferlin Transcript Containing the Alternative Exon 40a is Essential for Myocyte Functions. Frontiers in Cell and Developmental Biology, 2021, 9, 754555.	1.8	2
85	G.P.4.10 Functional evaluation of a putative mini-dysferlin identified in a patient with moderate Miyoshi myopathy phenotype. Neuromuscular Disorders, 2007, 17, 790.	0.3	1
86	P1.15 DNA micro-arrays for revisiting molecular pathology in neuromuscular disorders. Neuromuscular Disorders, 2010, 20, 604.	0.3	1
87	Calpainopathy in Chile, first cases reported. Neuromuscular Disorders, 2016, 26, S91.	0.3	1
88	Integrated analysis of the large-scale sequencing project â€œMyocaptureâ€•to identify novel genes for myopathies. Neuromuscular Disorders, 2017, 27, S195.	0.3	1
89	A new tool CovReport generates easy-to-understand sequencing coverage summary for diagnostic reports. Scientific Reports, 2020, 10, 6247.	1.6	1
90	Refining NGS diagnosis of muscular disorders. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 223-225.	0.9	1

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91	Le sÃ©quenÃ§age de nouvelle gÃ©nÃ©ration (Next-Generation Sequencing, ou NGS) appliquÃ© au diagnostic de maladies monogÃ©niques hÃ©tÃ©rogÃ¨nes. Les Cahiers De Myologie, 2016, , 31-33.	0.0	1
92	Cloning and characterization of a new member of transducing proteins. Biology of the Cell, 1995, 84, 102-102.	0.7	0
93	T.P.1 07 Therapeutic benefit of AAV-mediated injection of a mutated propeptide of myostatin in calpain 3 deficient mice. Neuromuscular Disorders, 2006, 16, 686.	0.3	0
94	Nouvelles stratÃ©gies thÃ©rapeutiques des dystrophies musculaires. Revue Du Rhumatisme (Edition) Tj ETQq0 0 0 rgBT /Overlock 10 T	0.6	0
95	G.P.8.12 Mannosidase I inhibition rescues the human Î±-sarcoglycan R77C recurrent mutation. Neuromuscular Disorders, 2008, 18, 785.	0.3	0
96	G.P.6.06 Systematic screening for genomic deletions and duplications in the dysferlin gene using multiplex ligation-dependant probe amplification and CGH microarrays. Neuromuscular Disorders, 2009, 19, 585-586.	0.3	0
97	T.O.2 Efficient recovery of dysferlin deficiency by dual adeno associated vector mediated gene transfer. Neuromuscular Disorders, 2009, 19, 659.	0.3	0
98	O.17 Efficient bypass of mutations in dysferlin deficient patient cells by antisense-induced exon skipping. Neuromuscular Disorders, 2010, 20, 640.	0.3	0
99	T.P.27 Characterization of the modular domains of dysferlin for gene transfer. Neuromuscular Disorders, 2012, 22, 860-861.	0.3	0
100	G.O.3 A human skeletal muscle interactome centered on proteins involved in muscular dystrophies. Neuromuscular Disorders, 2012, 22, 873.	0.3	0
101	Mutations in the EF hands of STIM1 lead to different clinical severity. Neuromuscular Disorders, 2015, 25, S289.	0.3	0
102	Next generation sequencing technologies in the genetic diagnosis of congenital myasthenic syndrome. Neuromuscular Disorders, 2017, 27, S196-S197.	0.3	0
103	Genetic characterization of a French cohort of GNE -mutation negative inclusion body myopathy patients using exome sequencing. Neuromuscular Disorders, 2017, 27, S149.	0.3	0
104	Characterization of the eosinophilic myositis caused by CAPN3 mutations on a mouse model. Neuromuscular Disorders, 2017, 27, S143-S144.	0.3	0
105	P.252LGMD, exercise intolerance, ptosis, ophthalmoplegia and dermatologic features: the phenotypic pleiotropy of plectinopathies in 8 French families. Neuromuscular Disorders, 2019, 29, S140.	0.3	0
106	Tumor protein 53â€induced nuclear protein 1 deficiency alters mouse gastrocnemius muscle function and bioenergetics inÂvivo. Physiological Reports, 2019, 7, e14055.	0.7	0
107	FROM THE SPINAL CORD TO THE MUSCLE. Neuromuscular Disorders, 2020, 30, S149.	0.3	0
108	Commentary: Long-Term Exercise Reduces Formation of Tubular Aggregates and Promotes Maintenance of Ca2+ Entry Units in Aged Muscle. Frontiers in Physiology, 2021, 12, 663677.	1.3	0