Isabelle Marty

List of Publications by Year
in descending order

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77 papers	3,656 citations	109321 35 h-index	133252 59 g-index
81 all docs	81 docs citations	81 times ranked	3874 citing authors

#	Article	IF	Citations
1	Therapies for RYR1-Related Myopathies: Where We Stand and the Perspectives. Current Pharmaceutical Design, 2022, 28, 15-25.	1.9	5
2	Quantification of the calcium signaling deficit in muscles devoid of triadin. PLoS ONE, 2022, 17, e0264146.	2.5	1
3	Clinical phenotype and loss of the slow skeletal muscle troponin T in three new patients with recessive TNNT1 nemaline myopathy. Journal of Medical Genetics, 2021, 58, 602-608.	3.2	11
4	SPEG binds with desmin and its deficiency causes defects in triad and focal adhesion proteins. Human Molecular Genetics, 2021, 29, 3882-3891.	2.9	6
5	Interplay between Triadin and Calsequestrin in the Pathogenesis of CPVT in the Mouse. Molecular Therapy, 2020, 28, 171-179.	8.2	17
6	In vivo RyR1 reduction in muscle triggers a core-like myopathy. Acta Neuropathologica Communications, 2020, 8, 192.	5.2	9
7	Trisk 95 as a novel skin mirror for normal and diabetic systemic glucose level. Scientific Reports, 2020, 10, 12246.	3.3	0
8	Variations in the TRPV1 gene are associated to exertional heat stroke. Journal of Science and Medicine in Sport, 2020, 23, 1021-1027.	1.3	7
9	Phenotype-guided whole genome analysis in a patient with genetically elusive long QT syndrome yields a novel TRDN-encoded triadin pathogenetic substrate for triadin knockout syndrome and reveals a novel primate-specific cardiac TRDN transcript. Heart Rhythm, 2020, 17, 1017-1024.	0.7	10
10	Dynamics of triadin, a muscle-specific triad protein, within sarcoplasmic reticulum subdomains. Molecular Biology of the Cell, 2020, 31, 261-272.	2.1	1
11	Familial deep cavitating state with a glutathione metabolism defect. Annals of Clinical and Translational Neurology, 2019, 6, 2573-2578.	3.7	1
12	â€˜Dusty core diseaseâ€™ (DuCD): expanding morphological spectrum of RYR1 recessive myopathies. Acta Neuropathologica Communications, 2019, 7, 3.	5.2	31
13	Deletion of the microtubule-associated protein 6 (MAP6) results in skeletal muscle dysfunction. Skeletal Muscle, 2018, 8, 30.	4.2	21
14	Quiescence of human muscle stem cells is favored by culture on natural biopolymeric films. Stem Cell Research and Therapy, 2017, 8, 104.	5.5	22
15	Functional Characterization and Rescue of a Deep Intronic Mutation inOCRLGene Responsible for Lowe Syndrome. Human Mutation, 2017, 38, 152-159.	2.5	13
16	Excitation-Contraction Coupling Alterations in Myopathies. Journal of Neuromuscular Diseases, 2016, 3, 443-453.	2.6	22
17	Triadin and CLIMP-63 form a link between triads and microtubules in muscle cells. Journal of Cell Science, 2016, 129, 3744-3755.	2.0	37
18	Dynamique de lâ€™organisation des triades. Les Cahiers De Myologie, 2016, , 97-98.	0.0	0

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19	Functional Characterization of a Central Core Disease RyR1 Mutation (p.Y4864H) Associated with Quantitative Defect in RyR1 Protein. Journal of Neuromuscular Diseases, 2015, 2, 421-432.	2.6	16
20	Triadin regulation of the ryanodine receptor complex. Journal of Physiology, 2015, 593, 3261-3266.	2.9	36
21	Single Delivery of an Adeno-Associated Viral Construct to Transfer the CASQ2 Gene to Knock-In Mice Affected by Catecholaminergic Polymorphic Ventricular Tachycardia Is Able to Cure the Disease From Birth to Advanced Age. Circulation, 2014, 129, 2673-2681.	1.6	88
22	Nâ€<scp>WASP</scp> is required for Amphiphysinâ€2/<scp>BIN</scp>1â€dependent nuclear positioning and triad organization in skeletal muscle and is involved in the pathophysiology of centronuclear myopathy. EMBO Molecular Medicine, 2014, 6, 1455-1475.	6.9	87
23	Ryanodine Receptor 1 and Associated Pathologies. , 2014, , 167-187.		1
24	The neuronal endopeptidase ECEL1 is associated with a distinct form of recessive distal arthrogryposis. Human Molecular Genetics, 2013, 22, 1483-1492.	2.9	66
25	Exon Skipping as a Therapeutic Strategy Applied to anRYR1Mutation with Pseudo-Exon Inclusion Causing a Severe Core Myopathy. Human Gene Therapy, 2013, 24, 702-713.	2.7	27
26	An Integrated Diagnosis Strategy for Congenital Myopathies. PLoS ONE, 2013, 8, e67527.	2.5	53
27	Role of Triadin in the Organization of Reticulum Membrane at the Muscle Triad. Journal of Cell Science, 2012, 125, 3443-53.	2.0	20
28	Absence of triadin, a protein of the calcium release complex, is responsible for cardiac arrhythmia with sudden death in human. Human Molecular Genetics, 2012, 21, 2759-2767.	2.9	227
29	Oxidative stress and successful antioxidant treatment in models of RYR1-related myopathy. Brain, 2012, 135, 1115-1127.	7.6	114
30	Recessive RYR1 mutations cause unusual congenital myopathy with prominent nuclear internalization and large areas of myofibrillar disorganization. Neuropathology and Applied Neurobiology, 2011, 37, 271-284.	3.2	97
31	Trisk 32 regulates IP3 receptors in rat skeletal myoblasts. Pflugers Archiv European Journal of Physiology, 2011, 462, 599-610.	2.8	4
32	Functional analysis reveals splicing mutations of the CASQ2 gene in patients with CPVT: implication for genetic counselling and clinical management. Human Mutation, 2011, 32, 995-999.	2.5	12
33	Recessive mutations in RYR1 are a common cause of congenital fiber type disproportion. Human Mutation, 2010, 31, E1544-E1550.	2.5	153
34	DHPR Î±1S subunit controls skeletal muscle mass and morphogenesis. EMBO Journal, 2010, 29, 643-654.	7.8	59
35	Caveolin 3 Is Associated with the Calcium Release Complex and Is Modified via in Vivo Triadin Modification. Biochemistry, 2010, 49, 6130-6135.	2.5	18
36	Alteration of Sarcoplasmic Reticulum<mml:math xmlns:mml="http://www.w3.org/1998/Math/MathML"><mml:mrow><mml:msup><mml:mrow><mml:mtext>Ca</mml:mtext></mml:mrow> in Skeletal Muscle from Calpain 3-Deficient Mice. International Journal of Cell Biology, 2009, 2009, 1-12.	2.5	10

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37	Triadin Deletion Induces Impaired Skeletal Muscle Function. Journal of Biological Chemistry, 2009, 284, 34918-34929.	3.4	71
38	Triadin: what possible function 20 years later?. Journal of Physiology, 2009, 587, 3117-3121.	2.9	36
39	Absence of Î²-tropomyosin is a new cause of Escobar syndrome associated with nemaline myopathy. Neuromuscular Disorders, 2009, 19, 118-123.	0.6	58
40	First genomic rearrangement of the RYR1 gene associated with an atypical presentation of lethal neonatal hypotonia. Neuromuscular Disorders, 2009, 19, 680-684.	0.6	27
41	Triadin Function In Sarcoplasmic Reticulum Structure?. Biophysical Journal, 2009, 96, 237a.	0.5	1
42	Null mutations causing depletion of the type 1 ryanodine receptor (RYR1) are commonly associated with recessive structural congenital myopathies with cores. Human Mutation, 2008, 29, 670-678.	2.5	89
43	Altered expression of triadin 95 causes parallel changes in localized Ca²⁺ release events and global Ca²⁺ signals in skeletal muscle cells in culture. Journal of Physiology, 2008, 586, 5803-5818.	2.9	29
44	Cardiomyocyte Overexpression of Neuronal Nitric Oxide Synthase Delays Transition Toward Heart Failure in Response to Pressure Overload by Preserving Calcium Cycling. Circulation, 2008, 117, 3187-3198.	1.6	73
45	Triadin Binding to the C-Terminal Luminal Loop of the Ryanodine Receptor is Important for Skeletal Muscle Excitationâ€“Contraction Coupling. Journal of General Physiology, 2007, 130, 365-378.	1.9	70
46	Abnormal Distribution of Calcium-Handling Proteins. Journal of Neuropathology and Experimental Neurology, 2007, 66, 57-65.	1.7	25
47	Retrograde regulation of store-operated calcium channels by the ryanodine receptor-associated protein triadin 95 in rat skeletal myotubes. Cell Calcium, 2007, 41, 179-185.	2.4	10
48	Triadin Binding to the C-Terminal Luminal Loop of the Ryanodine Receptor is Important for Skeletal Muscle Excitationâ€“Contraction Coupling. Journal of Cell Biology, 2007, 179, i2-i2.	5.2	1
49	Triadin (Trisk 95) Overexpression Blocks Excitation-Contraction Coupling in Rat Skeletal Myotubes. Journal of Biological Chemistry, 2005, 280, 39302-39308.	3.4	33
50	Transduction of the Scorpion Toxin Maurocalcine into Cells. Journal of Biological Chemistry, 2005, 280, 12833-12839.	3.4	62
51	Triadins Are Not Triad-specific Proteins. Journal of Biological Chemistry, 2005, 280, 28601-28609.	3.4	33
52	Role of Myocardial Neuronal Nitric Oxide Synthaseâ€“Derived Nitric Oxide in Î²-Adrenergic Hyporesponsiveness After Myocardial Infarctionâ€“Induced Heart Failure in Rat. Circulation, 2004, 110, 2368-2375.	1.6	135
53	Triadin: a multi-protein family for which purpose?. Cellular and Molecular Life Sciences, 2004, 61, 1850-1853.	5.4	15
54	Tubular aggregates are from whole sarcoplasmic reticulum origin: alterations in calcium binding protein expression in mouse skeletal muscle during aging. Neuromuscular Disorders, 2004, 14, 208-216.	0.6	68

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55	Increased neuronal nitric oxide synthase-derived NO production in the failing human heart. Lancet, The, 2004, 363, 1365-1367.	13.7	234
56	Critical Amino Acid Residues Determine the Binding Affinity and the Ca2+ Release Efficacy of Maurocalcine in Skeletal Muscle Cells. Journal of Biological Chemistry, 2003, 278, 37822-37831.	3.4	43
57	Human skeletal muscle triadin: gene organization and cloning of the major isoform, Trisk 51. Biochemical and Biophysical Research Communications, 2003, 303, 669-675.	2.1	32
58	A homozygous splicing mutation causing a depletion of skeletal muscle RYR1 is associated with multi-minicore disease congenital myopathy with ophthalmoplegia. Human Molecular Genetics, 2003, 12, 1171-1178.	2.9	129
59	Defects in ryanodine receptor calcium release in skeletal muscle from postmyocardial infarcted rats. FASEB Journal, 2003, 17, 1-18.	0.5	78
60	Multiple determinants in voltage-dependent P/Q calcium channels control their retention in the endoplasmic reticulum. European Journal of Neuroscience, 2002, 16, 883-895.	2.6	48
61	Calcium-dependent translocation of synaptotagmin to the plasma membrane in the dendrites of developing neurones. Molecular Brain Research, 2001, 96, 1-13.	2.3	28
62	Molecular interaction of dihydropyridine receptors with type-1 ryanodine receptors in rat brain. Biochemical Journal, 2001, 354, 597.	3.7	65
63	Developmental expression of the calcium release channels during early neurogenesis of the mouse cerebral cortex. European Journal of Neuroscience, 2001, 14, 1613-1622.	2.6	57
64	Cloning and Characterization of a New Isoform of Skeletal Muscle Triadin. Journal of Biological Chemistry, 2000, 275, 8206-8212.	3.4	47
65	Intracellular Ca 2+ Handling in Vascular Smooth Muscle Cells Is Affected by Proliferation. Arteriosclerosis, Thrombosis, and Vascular Biology, 2000, 20, 1225-1235.	2.4	85
66	Functional Interaction of the Cytoplasmic Domain of Triadin with the Skeletal Ryanodine Receptor. Journal of Biological Chemistry, 1999, 274, 12278-12283.	3.4	72
67	Effects of sustained low-flow ischemia on myocardial function and calcium-regulating proteins in adult and senescent rat hearts. Cardiovascular Research, 1998, 38, 169-180.	3.8	63
68	Cardiac Calcium Release Channel (Ryanodine Receptor) in Control and Cardiomyopathic Human Hearts: mRNA and Protein Contents are Differentially Regulated. Journal of Molecular and Cellular Cardiology, 1997, 29, 1237-1246.	1.9	40
69	A ryanodine-sensitive calcium store in ascidian eggs monitored by whole-cell patch-clamp recordings. Cell Calcium, 1997, 21, 93-101.	2.4	16
70	Different Compartments of Sarcoplasmic Reticulum Participate in the Excitation-Contraction Coupling Process in Human Atrial Myocytes. Circulation Research, 1997, 80, 345-353.	4.5	88
71	Involvement of the Dihydropyridine Receptor and Internal Ca2+Stores in Myoblast Fusion. Experimental Cell Research, 1996, 223, 301-307.	2.6	35
72	Expression of the cardiac ryanodine receptor in the compensated phase of hypertrophy in rat heart. Cardiovascular Research, 1996, 32, 258-265.	3.8	23

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73	Localization of the N-terminal and C-terminal ends of triadin with respect to the sarcoplasmic reticulum membrane of rabbit skeletal muscle. Biochemical Journal, 1995, 307, 769-774.	3.7	33
74	Transmembrane orientation of the N-terminal and C-terminal ends of the ryanodine receptor in the sarcoplasmic reticulum of rabbit skeletal muscle. Biochemical Journal, 1994, 298, 743-749.	3.7	30
75	Biochemical evidence for a complex involving dihydropyridine receptor and ryanodine receptor in triad junctions of skeletal muscle.. Proceedings of the National Academy of Sciences of the United States of America, 1994, 91, 2270-2274.	7.1	153
76	Topography of the membrane-bound ADP/ATP carrier assessed by enzymic proteolysis. Biochemistry, 1992, 31, 4058-4065.	2.5	69
77	Kinetics of nucleotide transport in rat heart mitochondria studied by a rapid filtration technique. Biochemistry, 1990, 29, 9720-9727.	2.5	9