

Isabelle Marty

List of Publications by Year in descending order

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Version: 2024-02-01

77
papers

3,656
citations

109264

35
h-index

133188

59
g-index

81
all docs

81
docs citations

81
times ranked

3874
citing authors

#	ARTICLE	IF	CITATIONS
1	Increased neuronal nitric oxide synthase-derived NO production in the failing human heart. <i>Lancet, The</i> , 2004, 363, 1365-1367.	6.3	234
2	Absence of triadin, a protein of the calcium release complex, is responsible for cardiac arrhythmia with sudden death in human. <i>Human Molecular Genetics</i> , 2012, 21, 2759-2767.	1.4	227
3	Biochemical evidence for a complex involving dihydropyridine receptor and ryanodine receptor in triad junctions of skeletal muscle.. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1994, 91, 2270-2274.	3.3	153
4	Recessive mutations in RYR1 are a common cause of congenital fiber type disproportion. <i>Human Mutation</i> , 2010, 31, E1544-E1550.	1.1	153
5	Role of Myocardial Neuronal Nitric Oxide Synthaseâ€‘Derived Nitric Oxide in Î²-Adrenergic Hyporesponsiveness After Myocardial Infarctionâ€‘Induced Heart Failure in Rat. <i>Circulation</i> , 2004, 110, 2368-2375.	1.6	135
6	A homozygous splicing mutation causing a depletion of skeletal muscle RYR1 is associated with multi-minicore disease congenital myopathy with ophthalmoplegia. <i>Human Molecular Genetics</i> , 2003, 12, 1171-1178.	1.4	129
7	Oxidative stress and successful antioxidant treatment in models of RYR1-related myopathy. <i>Brain</i> , 2012, 135, 1115-1127.	3.7	114
8	Recessive RYR1 mutations cause unusual congenital myopathy with prominent nuclear internalization and large areas of myofibrillar disorganization. <i>Neuropathology and Applied Neurobiology</i> , 2011, 37, 271-284.	1.8	97
9	Null mutations causing depletion of the type 1 ryanodine receptor (RYR1) are commonly associated with recessive structural congenital myopathies with cores. <i>Human Mutation</i> , 2008, 29, 670-678.	1.1	89
10	Single Delivery of an Adeno-Associated Viral Construct to Transfer the <i>CASQ2</i> Gene to Knock-In Mice Affected by Catecholaminergic Polymorphic Ventricular Tachycardia Is Able to Cure the Disease From Birth to Advanced Age. <i>Circulation</i> , 2014, 129, 2673-2681.	1.6	88
11	Different Compartments of Sarcoplasmic Reticulum Participate in the Excitation-Contraction Coupling Process in Human Atrial Myocytes. <i>Circulation Research</i> , 1997, 80, 345-353.	2.0	88
12	WASP is required for Amphiphysinâ€‘dependent nuclear positioning and triad organization in skeletal muscle and is involved in the pathophysiology of centronuclear myopathy. <i>EMBO Molecular Medicine</i> , 2014, 6, 1455-1475.	3.3	87
13	Intracellular Ca ²⁺ Handling in Vascular Smooth Muscle Cells Is Affected by Proliferation. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2000, 20, 1225-1235.	1.1	85
14	Defects in ryanodine receptor calcium release in skeletal muscle from postmyocardial infarcted rats. <i>FASEB Journal</i> , 2003, 17, 1-18.	0.2	78
15	Cardiomyocyte Overexpression of Neuronal Nitric Oxide Synthase Delays Transition Toward Heart Failure in Response to Pressure Overload by Preserving Calcium Cycling. <i>Circulation</i> , 2008, 117, 3187-3198.	1.6	73
16	Functional Interaction of the Cytoplasmic Domain of Triadin with the Skeletal Ryanodine Receptor. <i>Journal of Biological Chemistry</i> , 1999, 274, 12278-12283.	1.6	72
17	Triadin Deletion Induces Impaired Skeletal Muscle Function. <i>Journal of Biological Chemistry</i> , 2009, 284, 34918-34929.	1.6	71
18	Triadin Binding to the C-Terminal Luminal Loop of the Ryanodine Receptor is Important for Skeletal Muscle Excitationâ€‘Contraction Coupling. <i>Journal of General Physiology</i> , 2007, 130, 365-378.	0.9	70

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19	Topography of the membrane-bound ADP/ATP carrier assessed by enzymic proteolysis. <i>Biochemistry</i> , 1992, 31, 4058-4065.	1.2	69
20	Tubular aggregates are from whole sarcoplasmic reticulum origin: alterations in calcium binding protein expression in mouse skeletal muscle during aging. <i>Neuromuscular Disorders</i> , 2004, 14, 208-216.	0.3	68
21	The neuronal endopeptidase ECEL1 is associated with a distinct form of recessive distal arthrogyriposis. <i>Human Molecular Genetics</i> , 2013, 22, 1483-1492.	1.4	66
22	Molecular interaction of dihydropyridine receptors with type-1 ryanodine receptors in rat brain. <i>Biochemical Journal</i> , 2001, 354, 597.	1.7	65
23	Effects of sustained low-flow ischemia on myocardial function and calcium-regulating proteins in adult and senescent rat hearts. <i>Cardiovascular Research</i> , 1998, 38, 169-180.	1.8	63
24	Transduction of the Scorpion Toxin Maurocalcine into Cells. <i>Journal of Biological Chemistry</i> , 2005, 280, 12833-12839.	1.6	62
25	DHPR $\hat{1}$ S subunit controls skeletal muscle mass and morphogenesis. <i>EMBO Journal</i> , 2010, 29, 643-654.	3.5	59
26	Absence of $\hat{2}$ -tropomyosin is a new cause of Escobar syndrome associated with nemaline myopathy. <i>Neuromuscular Disorders</i> , 2009, 19, 118-123.	0.3	58
27	Developmental expression of the calcium release channels during early neurogenesis of the mouse cerebral cortex. <i>European Journal of Neuroscience</i> , 2001, 14, 1613-1622.	1.2	57
28	An Integrated Diagnosis Strategy for Congenital Myopathies. <i>PLoS ONE</i> , 2013, 8, e67527.	1.1	53
29	Multiple determinants in voltage-dependent P/Q calcium channels control their retention in the endoplasmic reticulum. <i>European Journal of Neuroscience</i> , 2002, 16, 883-895.	1.2	48
30	Cloning and Characterization of a New Isoform of Skeletal Muscle Triadin. <i>Journal of Biological Chemistry</i> , 2000, 275, 8206-8212.	1.6	47
31	Critical Amino Acid Residues Determine the Binding Affinity and the Ca ²⁺ Release Efficacy of Maurocalcine in Skeletal Muscle Cells. <i>Journal of Biological Chemistry</i> , 2003, 278, 37822-37831.	1.6	43
32	Cardiac Calcium Release Channel (Ryanodine Receptor) in Control and Cardiomyopathic Human Hearts: mRNA and Protein Contents are Differentially Regulated. <i>Journal of Molecular and Cellular Cardiology</i> , 1997, 29, 1237-1246.	0.9	40
33	Triadin and CLIMP-63 form a link between triads and microtubules in muscle cells. <i>Journal of Cell Science</i> , 2016, 129, 3744-3755.	1.2	37
34	Triadin: what possible function 20 years later?. <i>Journal of Physiology</i> , 2009, 587, 3117-3121.	1.3	36
35	Triadin regulation of the ryanodine receptor complex. <i>Journal of Physiology</i> , 2015, 593, 3261-3266.	1.3	36
36	Involvement of the Dihydropyridine Receptor and Internal Ca ²⁺ Stores in Myoblast Fusion. <i>Experimental Cell Research</i> , 1996, 223, 301-307.	1.2	35

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37	Localization of the N-terminal and C-terminal ends of triadin with respect to the sarcoplasmic reticulum membrane of rabbit skeletal muscle. <i>Biochemical Journal</i> , 1995, 307, 769-774.	1.7	33
38	Triadin (Trisk 95) Overexpression Blocks Excitation-Contraction Coupling in Rat Skeletal Myotubes. <i>Journal of Biological Chemistry</i> , 2005, 280, 39302-39308.	1.6	33
39	Triadins Are Not Triad-specific Proteins. <i>Journal of Biological Chemistry</i> , 2005, 280, 28601-28609.	1.6	33
40	Human skeletal muscle triadin: gene organization and cloning of the major isoform, Trisk 51. <i>Biochemical and Biophysical Research Communications</i> , 2003, 303, 669-675.	1.0	32
41	“Dusty core disease” (DuCD): expanding morphological spectrum of RYR1 recessive myopathies. <i>Acta Neuropathologica Communications</i> , 2019, 7, 3.	2.4	31
42	Transmembrane orientation of the N-terminal and C-terminal ends of the ryanodine receptor in the sarcoplasmic reticulum of rabbit skeletal muscle. <i>Biochemical Journal</i> , 1994, 298, 743-749.	1.7	30
43	Altered expression of triadin 95 causes parallel changes in localized Ca ²⁺ release events and global Ca ²⁺ signals in skeletal muscle cells in culture. <i>Journal of Physiology</i> , 2008, 586, 5803-5818.	1.3	29
44	Calcium-dependent translocation of synaptotagmin to the plasma membrane in the dendrites of developing neurones. <i>Molecular Brain Research</i> , 2001, 96, 1-13.	2.5	28
45	First genomic rearrangement of the RYR1 gene associated with an atypical presentation of lethal neonatal hypotonia. <i>Neuromuscular Disorders</i> , 2009, 19, 680-684.	0.3	27
46	Exon Skipping as a Therapeutic Strategy Applied to an RYR1 Mutation with Pseudo-Exon Inclusion Causing a Severe Core Myopathy. <i>Human Gene Therapy</i> , 2013, 24, 702-713.	1.4	27
47	Abnormal Distribution of Calcium-Handling Proteins. <i>Journal of Neuropathology and Experimental Neurology</i> , 2007, 66, 57-65.	0.9	25
48	Expression of the cardiac ryanodine receptor in the compensated phase of hypertrophy in rat heart. <i>Cardiovascular Research</i> , 1996, 32, 258-265.	1.8	23
49	Excitation-Contraction Coupling Alterations in Myopathies. <i>Journal of Neuromuscular Diseases</i> , 2016, 3, 443-453.	1.1	22
50	Quiescence of human muscle stem cells is favored by culture on natural biopolymeric films. <i>Stem Cell Research and Therapy</i> , 2017, 8, 104.	2.4	22
51	Deletion of the microtubule-associated protein 6 (MAP6) results in skeletal muscle dysfunction. <i>Skeletal Muscle</i> , 2018, 8, 30.	1.9	21
52	Role of Triadin in the Organization of Reticulum Membrane at the Muscle Triad. <i>Journal of Cell Science</i> , 2012, 125, 3443-53.	1.2	20
53	Caveolin 3 Is Associated with the Calcium Release Complex and Is Modified via in Vivo Triadin Modification. <i>Biochemistry</i> , 2010, 49, 6130-6135.	1.2	18
54	Interplay between Triadin and Calsequestrin in the Pathogenesis of CPVT in the Mouse. <i>Molecular Therapy</i> , 2020, 28, 171-179.	3.7	17

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55	A ryanodine-sensitive calcium store in ascidian eggs monitored by whole-cell patch-clamp recordings. <i>Cell Calcium</i> , 1997, 21, 93-101.	1.1	16
56	Functional Characterization of a Central Core Disease RyR1 Mutation (p.Y4864H) Associated with Quantitative Defect in RyR1 Protein. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, 421-432.	1.1	16
57	Triadin: a multi-protein family for which purpose?. <i>Cellular and Molecular Life Sciences</i> , 2004, 61, 1850-1853.	2.4	15
58	Functional Characterization and Rescue of a Deep Intronic Mutation in <i>OCRL</i> Gene Responsible for Lowe Syndrome. <i>Human Mutation</i> , 2017, 38, 152-159.	1.1	13
59	Functional analysis reveals splicing mutations of the <i>CASQ2</i> gene in patients with CPVT: implication for genetic counselling and clinical management. <i>Human Mutation</i> , 2011, 32, 995-999.	1.1	12
60	Clinical phenotype and loss of the slow skeletal muscle troponin T in three new patients with recessive <i>TNNT1</i> nemaline myopathy. <i>Journal of Medical Genetics</i> , 2021, 58, 602-608.	1.5	11
61	Retrograde regulation of store-operated calcium channels by the ryanodine receptor-associated protein triadin 95 in rat skeletal myotubes. <i>Cell Calcium</i> , 2007, 41, 179-185.	1.1	10
62	Alteration of Sarcoplasmic Reticulum Ca^{2+} Release in Skeletal Muscle from Calpain 3-Deficient Mice. <i>International Journal of Cell Biology</i> , 2009, 2009, 1-12.	1.1	10
63	Phenotype-guided whole genome analysis in a patient with genetically elusive long QT syndrome yields a novel <i>TRDN</i> -encoded triadin pathogenetic substrate for triadin knockout syndrome and reveals a novel primate-specific cardiac <i>TRDN</i> transcript. <i>Heart Rhythm</i> , 2020, 17, 1017-1024.	0.3	10
64	Kinetics of nucleotide transport in rat heart mitochondria studied by a rapid filtration technique. <i>Biochemistry</i> , 1990, 29, 9720-9727.	1.2	9
65	In vivo RyR1 reduction in muscle triggers a core-like myopathy. <i>Acta Neuropathologica Communications</i> , 2020, 8, 192.	2.4	9
66	Variations in the <i>TRPV1</i> gene are associated to exertional heat stroke. <i>Journal of Science and Medicine in Sport</i> , 2020, 23, 1021-1027.	0.6	7
67	<i>SPEG</i> binds with desmin and its deficiency causes defects in triad and focal adhesion proteins. <i>Human Molecular Genetics</i> , 2021, 29, 3882-3891.	1.4	6
68	Therapies for RYR1-Related Myopathies: Where We Stand and the Perspectives. <i>Current Pharmaceutical Design</i> , 2022, 28, 15-25.	0.9	5
69	Trisk 32 regulates IP3 receptors in rat skeletal myoblasts. <i>Pflugers Archiv European Journal of Physiology</i> , 2011, 462, 599-610.	1.3	4
70	Triadin Function In Sarcoplasmic Reticulum Structure?. <i>Biophysical Journal</i> , 2009, 96, 237a.	0.2	1
71	Familial deep cavitating state with a glutathione metabolism defect. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 2573-2578.	1.7	1
72	Dynamics of triadin, a muscle-specific triad protein, within sarcoplasmic reticulum subdomains. <i>Molecular Biology of the Cell</i> , 2020, 31, 261-272.	0.9	1

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73	Ryanodine Receptor 1 and Associated Pathologies. , 2014, , 167-187.		1
74	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.	2.3	1
75	Quantification of the calcium signaling deficit in muscles devoid of triadin. PLoS ONE, 2022, 17, e0264146.	1.1	1
76	Trisk 95 as a novel skin mirror for normal and diabetic systemic glucose level. Scientific Reports, 2020, 10, 12246.	1.6	0
77	Dynamique de lâ€™organisation des triades. Les Cahiers De Myologie, 2016, , 97-98.	0.0	0