# Isabelle Marty

List of Publications by Year
in descending order

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|---|---|---|---|
| 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 | Therapies for RYR1-Related Myopathies: Where We Stand and the Perspectives. Current Pharmaceutical Design, 2022, 28, 15-25. | 0.9 | 5 |
| 2 | Quantification of the calcium signaling deficit in muscles devoid of triadin. PLoS ONE, 2022, 17, e0264146. | 1.1 | 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. | 1.5 | 11 |
| 4 | SPEG binds with desmin and its deficiency causes defects in triad and focal adhesion proteins. Human Molecular Genetics, 2021, 29, 3882-3891. | 1.4 | 6 |
| 5 | Interplay between Triadin and Calsequestrin in the Pathogenesis of CPVT in the Mouse. Molecular Therapy, 2020, 28, 171-179. | 3.7 | 17 |
| 6 | In vivo RyR1 reduction in muscle triggers a core-like myopathy. Acta Neuropathologica Communications, 2020, 8, 192. | 2.4 | 9 |
| 7 | Trisk 95 as a novel skin mirror for normal and diabetic systemic glucose level. Scientific Reports, 2020, 10, 12246. | 1.6 | 0 |
| 8 | Variations in the TRPV1 gene are associated to exertional heat stroke. Journal of Science and Medicine in Sport, 2020, 23, 1021-1027. | 0.6 | 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. | 0.3 | 10 |
| 10 | Dynamics of triadin, a muscle-specific triad protein, within sarcoplasmic reticulum subdomains. Molecular Biology of the Cell, 2020, 31, 261-272. | 0.9 | 1 |
| 11 | Familial deep cavitating state with a glutathione metabolism defect. Annals of Clinical and Translational Neurology, 2019, 6, 2573-2578. | 1.7 | 1 |
| 12 | â€˜Dusty core diseaseâ€™ (DuCD): expanding morphological spectrum of RYR1 recessive myopathies. Acta Neuropathologica Communications, 2019, 7, 3. | 2.4 | 31 |
| 13 | Deletion of the microtubule-associated protein 6 (MAP6) results in skeletal muscle dysfunction. Skeletal Muscle, 2018, 8, 30. | 1.9 | 21 |
| 14 | Quiescence of human muscle stem cells is favored by culture on natural biopolymeric films. Stem Cell Research and Therapy, 2017, 8, 104. | 2.4 | 22 |
| 15 | Functional Characterization and Rescue of a Deep Intronic Mutation inOCRLGene Responsible for Lowe Syndrome. Human Mutation, 2017, 38, 152-159. | 1.1 | 13 |
| 16 | Excitation-Contraction Coupling Alterations in Myopathies. Journal of Neuromuscular Diseases, 2016, 3, 443-453. | 1.1 | 22 |
| 17 | Triadin and CLIMP-63 form a link between triads and microtubules in muscle cells. Journal of Cell Science, 2016, 129, 3744-3755. | 1.2 | 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. | 1.1 | 16 |
| 20 | Triadin regulation of the ryanodine receptor complex. Journal of Physiology, 2015, 593, 3261-3266. | 1.3 | 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. | 3.3 | 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. | 1.4 | 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. | 1.4 | 27 |
| 26 | An Integrated Diagnosis Strategy for Congenital Myopathies. PLoS ONE, 2013, 8, e67527. | 1.1 | 53 |
| 27 | Role of Triadin in the Organization of Reticulum Membrane at the Muscle Triad. Journal of Cell Science, 2012, 125, 3443-53. | 1.2 | 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. | 1.4 | 227 |
| 29 | Oxidative stress and successful antioxidant treatment in models of RYR1-related myopathy. Brain, 2012, 135, 1115-1127. | 3.7 | 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. | 1.8 | 97 |
| 31 | Trisk 32 regulates IP3 receptors in rat skeletal myoblasts. Pflugers Archiv European Journal of Physiology, 2011, 462, 599-610. | 1.3 | 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. | 1.1 | 12 |
| 33 | Recessive mutations in RYR1 are a common cause of congenital fiber type disproportion. Human Mutation, 2010, 31, E1544-E1550. | 1.1 | 153 |
| 34 | DHPR Î±1S subunit controls skeletal muscle mass and morphogenesis. EMBO Journal, 2010, 29, 643-654. | 3.5 | 59 |
| 35 | Caveolin 3 Is Associated with the Calcium Release Complex and Is Modified via in Vivo Triadin Modification. Biochemistry, 2010, 49, 6130-6135. | 1.2 | 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. | | 10 |

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|---|---|---|---|
| 37 | Triadin Deletion Induces Impaired Skeletal Muscle Function. Journal of Biological Chemistry, 2009, 284, 34918-34929. | 1.6 | 71 |
| 38 | Triadin: what possible function 20 years later?. Journal of Physiology, 2009, 587, 3117-3121. | 1.3 | 36 |
| 39 | Absence of Î²-tropomyosin is a new cause of Escobar syndrome associated with nemaline myopathy. Neuromuscular Disorders, 2009, 19, 118-123. | 0.3 | 58 |
| 40 | First genomic rearrangement of the RYR1 gene associated with an atypical presentation of lethal neonatal hypotonia. Neuromuscular Disorders, 2009, 19, 680-684. | 0.3 | 27 |
| 41 | Triadin Function In Sarcoplasmic Reticulum Structure?. Biophysical Journal, 2009, 96, 237a. | 0.2 | 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. | 1.1 | 89 |
| 43 | Altered expression of triadin 95 causes parallel changes in localized Ca<sup>2+</sup> release events and global Ca<sup>2+</sup> signals in skeletal muscle cells in culture. Journal of Physiology, 2008, 586, 5803-5818. | 1.3 | 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. | 0.9 | 70 |
| 46 | Abnormal Distribution of Calcium-Handling Proteins. Journal of Neuropathology and Experimental Neurology, 2007, 66, 57-65. | 0.9 | 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. | 1.1 | 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. | 2.3 | 1 |
| 49 | Triadin (Trisk 95) Overexpression Blocks Excitation-Contraction Coupling in Rat Skeletal Myotubes. Journal of Biological Chemistry, 2005, 280, 39302-39308. | 1.6 | 33 |
| 50 | Transduction of the Scorpion Toxin Maurocalcine into Cells. Journal of Biological Chemistry, 2005, 280, 12833-12839. | 1.6 | 62 |
| 51 | Triadins Are Not Triad-specific Proteins. Journal of Biological Chemistry, 2005, 280, 28601-28609. | 1.6 | 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. | 2.4 | 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. | 0.3 | 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. | 6.3 | 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. | 1.6 | 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. | 1.0 | 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. | 1.4 | 129 |
| 59 | Defects in ryanodine receptor calcium release in skeletal muscle from postmyocardial infarcted rats. FASEB Journal, 2003, 17, 1-18. | 0.2 | 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. | 1.2 | 48 |
| 61 | Calcium-dependent translocation of synaptotagmin to the plasma membrane in the dendrites of developing neurones. Molecular Brain Research, 2001, 96, 1-13. | 2.5 | 28 |
| 62 | Molecular interaction of dihydropyridine receptors with type-1 ryanodine receptors in rat brain. Biochemical Journal, 2001, 354, 597. | 1.7 | 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. | 1.2 | 57 |
| 64 | Cloning and Characterization of a New Isoform of Skeletal Muscle Triadin. Journal of Biological Chemistry, 2000, 275, 8206-8212. | 1.6 | 47 |
| 65 | Intracellular Ca 2+ Handling in Vascular Smooth Muscle Cells Is Affected by Proliferation. Arteriosclerosis, Thrombosis, and Vascular Biology, 2000, 20, 1225-1235. | 1.1 | 85 |
| 66 | Functional Interaction of the Cytoplasmic Domain of Triadin with the Skeletal Ryanodine Receptor. Journal of Biological Chemistry, 1999, 274, 12278-12283. | 1.6 | 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. | 1.8 | 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. | 0.9 | 40 |
| 69 | A ryanodine-sensitive calcium store in ascidian eggs monitored by whole-cell patch-clamp recordings. Cell Calcium, 1997, 21, 93-101. | 1.1 | 16 |
| 70 | Different Compartments of Sarcoplasmic Reticulum Participate in the Excitation-Contraction Coupling Process in Human Atrial Myocytes. Circulation Research, 1997, 80, 345-353. | 2.0 | 88 |
| 71 | Involvement of the Dihydropyridine Receptor and Internal Ca2+Stores in Myoblast Fusion. Experimental Cell Research, 1996, 223, 301-307. | 1.2 | 35 |
| 72 | Expression of the cardiac ryanodine receptor in the compensated phase of hypertrophy in rat heart. Cardiovascular Research, 1996, 32, 258-265. | 1.8 | 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. | 1.7 | 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. | 1.7 | 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. | 3.3 | 153 |
| 76 | Topography of the membrane-bound ADP/ATP carrier assessed by enzymic proteolysis. Biochemistry, 1992, 31, 4058-4065. | 1.2 | 69 |
| 77 | Kinetics of nucleotide transport in rat heart mitochondria studied by a rapid filtration technique. Biochemistry, 1990, 29, 9720-9727. | 1.2 | 9 |