## **Susan Treves**

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	RYR1-Related Rhabdomyolysis: A Spectrum of Hypermetabolic States Due to Ryanodine Receptor Dysfunction. Current Pharmaceutical Design, 2022, 28, 2-14.	1.9	11
2	Molecular basis of impaired muscle function in a mouse model of congenital myopathy due to compound heterozygous RYR1 mutations. Journal of General Physiology, 2022, 154, .	1.9	0
3	Improvement of muscle strength in a mouse model for congenital myopathy treated with HDAC and DNA methyltransferase inhibitors. ELife, 2022, 11, .	6.0	7
4	Pre-operative exercise and pyrexia as modifying factors in malignant hyperthermia (MH). Neuromuscular Disorders, 2022, 32, 628-634.	0.6	12
5	Rapid subcellular calcium responses and dynamics by calcium sensor G-CatchER+. IScience, 2021, 24, 102129.	4.1	19
6	Functional Characterization of Endogenously Expressed Human RYR1 Variants. Journal of Visualized Experiments, 2021, , .	0.3	0
7	The neuromuscular and multisystem features of RYR1-related malignant hyperthermia and rhabdomyolysis. Medicine (United States), 2021, 100, e26999.	1.0	8
8	Comment on "Overlapping Mechanisms of Exertional Heat Stroke and Malignant Hyperthermia: Evidence vs. Conjecture― Sports Medicine, 2021, , 1.	6.5	2
9	Bi-allelic expression of the RyR1 p.A4329D mutation decreases muscle strength in slow-twitch muscles in mice. Journal of Biological Chemistry, 2020, 295, 10331-10339.	3.4	4
10	Clinical, morphological and genetic characterization of Brody disease: an international study of 40 patients. Brain, 2020, 143, 452-466.	7.6	22
11	Molecular basis of impaired extraocular muscle function in a mouse model of congenital myopathy due to compound heterozygous Ryr1 mutations. Human Molecular Genetics, 2020, 29, 1330-1339.	2.9	5
12	Quantitative reduction of RyR1 protein caused by a single-allele frameshift mutation in RYR1 ex36 impairs the strength of adult skeletal muscle fibres. Human Molecular Genetics, 2019, 28, 1872-1884.	2.9	8
13	Extraocular muscle function is impaired in <i>ryr3</i> â^'/â^' mice. Journal of General Physiology, 2019, 151, 929-943.	1.9	11
14	Quantitative RyR1 reduction and loss of calcium sensitivity of RyR1Q1970fsX16+A4329D cause cores and loss of muscle strength. Human Molecular Genetics, 2019, 28, 2987-2999.	2.9	21
15	Aberrant regulation of epigenetic modifiers contributes to the pathogenesis in patients with selenoprotein N <i>â€</i> related myopathies. Human Mutation, 2019, 40, 962-974.	2.5	13
16	Biochemical and Epigenetic Modifications Occur in Muscles of Patients with Selenoprotein N Related Congenital Myopathy. Biophysical Journal, 2019, 116, 153a.	0.5	0
17	Characterization of an Animal Model for Congenital Myopathies Linked to Recessive RyR1 Mutations. Biophysical Journal, 2019, 116, 522a.	0.5	0
18	STIM1 over-activation generates a multi-systemic phenotype affecting the skeletal muscle, spleen, eye, skin, bones and immune system in mice. Human Molecular Genetics, 2019, 28, 1579-1593.	2.9	34

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19	Congenital myopathies: disorders of excitation–contraction coupling and muscle contraction. Nature Reviews Neurology, 2018, 14, 151-167.	10.1	212
20	Over-expression of a retinol dehydrogenase (SRP35/DHRS7C) in skeletal muscle activates mTORC2, enhances glucose metabolism and muscle performance. Scientific Reports, 2018, 8, 636.	3.3	19
21	Atypical periodic paralysis and myalgia. Neurology, 2018, 90, e412-e418.	1.1	39
22	Altered Eye Muscle Function in RYR3KO Mice. Biophysical Journal, 2018, 114, 469a.	0.5	0
23	<i>STAC3</i> variants cause a congenital myopathy with distinctive dysmorphic features and malignant hyperthermia susceptibility. Human Mutation, 2018, 39, 1980-1994.	2.5	42
24	Role of Calcium in Neutrophil Activation. , 2018, , 199-218.		0
25	Novel Local RYR1 Junctional Calcium Responses and Dynamics by Improved Calcium Sensor Catcher. Biophysical Journal, 2017, 112, 331a.	0.5	1
26	Biochemical and Mechanical Properties of Murine Extraocular Muscles. Biophysical Journal, 2017, 112, 101a-102a.	0.5	1
27	Dihydropyridine receptor (DHPR, CACNA1S) congenital myopathy. Acta Neuropathologica, 2017, 133, 517-533.	7.7	97
28	Current and future therapeutic approaches to the congenital myopathies. Seminars in Cell and Developmental Biology, 2017, 64, 191-200.	5.0	29
29	Ca2+ handling abnormalities in early-onset muscle diseases: Novel concepts and perspectives. Seminars in Cell and Developmental Biology, 2017, 64, 201-212.	5.0	19
30	Cellular, biochemical and molecular changes in muscles from patients with X-linked myotubular myopathy due toMTM1mutations. Human Molecular Genetics, 2016, 26, ddw388.	2.9	20
31	Functional characterization of orbicularis oculi and extraocular muscles. Journal of General Physiology, 2016, 147, 395-406.	1.9	9
32	Exertional rhabdomyolysis: physiological response or manifestation of an underlying myopathy?. BMJ Open Sport and Exercise Medicine, 2016, 2, e000151.	2.9	73
33	An <i>RYR1</i> mutation associated with malignant hyperthermia is also associated with bleeding abnormalities. Science Signaling, 2016, 9, ra68.	3.6	34
34	Role of the JP45-Calsequestrin Complex on Calcium Entry in Slow Twitch Skeletal Muscles. Journal of Biological Chemistry, 2016, 291, 14555-14565.	3.4	12
35	Functional characterization of the RYR1 mutation p.Arg4737Trp associated with susceptibility to malignant hyperthermia. Neuromuscular Disorders, 2016, 26, 21-25.	0.6	8
36	The metabolomic profile of gamma-irradiated human hepatoma and muscle cells reveals metabolic changes consistent with the Warburg effect. PeerJ, 2016, 4, e1624.	2.0	18

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37	Functional characterization of orbicularis oculi and extraocular muscles. Journal of Cell Biology, 2016, 213, 21330IA96.	5.2	0
38	Excitation-Contraction Coupling in Human Extraocular Muscles:There is more than Meets the Eye. Biophysical Journal, 2015, 108, 420a.	0.5	0
39	Differential Role of Calsequestrin Isoforms on Calcium Entry in Skeletal Muscle FDB Fibres. Biophysical Journal, 2015, 108, 269a.	0.5	Ο
40	Epigenetic changes as a common trigger of muscle weakness in congenital myopathies. Human Molecular Genetics, 2015, 24, 4636-4647.	2.9	44
41	Raptor ablation in skeletal muscle decreases Cav1.1 expression and affects the function of the excitation–contraction coupling supramolecular complex. Biochemical Journal, 2015, 466, 123-135.	3.7	10
42	Characterization of excitation–contraction coupling components in human extraocular muscles. Biochemical Journal, 2015, 466, 29-36.	3.7	10
43	Role of sarcoplasmic reticulum junctional proteins in skeletal muscle strength. BMC Anesthesiology, 2014, 14, .	1.8	Ο
44	Biochemical, Cellular and Electrophysiological Characterization of HMCL-7304 a Human Skeletal Muscle-Derived Cell Line. Biophysical Journal, 2014, 106, 446a.	0.5	0
45	Raptor Ablation in Skeletal Muscle Affects the Structure and Function of the Excitation-Contraction Coupling Macromolecular Complex. Biophysical Journal, 2014, 106, 123a.	0.5	Ο
46	RyR1 Deficiency in Congenital Myopathies Disrupts Excitation-Contraction Coupling. Human Mutation, 2013, 34, 986-996.	2.5	40
47	3D Structural Illumination Microscopy of the Skeletal Muscle Excitation-Contraction Coupling Macromolecular Complex. Biophysical Journal, 2013, 104, 105a.	0.5	2
48	Establishment of a human skeletal muscle-derived cell line: biochemical, cellular and electrophysiological characterization. Biochemical Journal, 2013, 455, 169-177.	3.7	19
49	Ectosomes of polymorphonuclear neutrophils activate multiple signaling pathways in macrophages. Immunobiology, 2013, 218, 382-392.	1.9	74
50	JP-45/ <i>JSRP1</i> Variants Affect Skeletal Muscle Excitation-Contraction Coupling by Decreasing the Sensitivity of the Dihydropyridine Receptor. Human Mutation, 2013, 34, 184-190.	2.5	12
51	Physiological Role(S) of RyR1 in Smooth Muscle Cells. Biophysical Journal, 2013, 104, 443a-444a.	0.5	0
52	Enhanced dihydropyridine receptor calcium channel activity restores muscle strength in JP45/CASQ1 double knockout mice. Nature Communications, 2013, 4, 1541.	12.8	35
53	Gain of function of the immune system caused by a ryanodine receptor 1 mutation. Journal of Cell Science, 2013, 126, 3485-92.	2.0	14
54	Variable Myopathic Presentation in a Single Family with Novel Skeletal RYR1 Mutation. PLoS ONE, 2013, 8. e69296.	2.5	12

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55	Remodeling of calcium handling in skeletal muscle through PGC-1α: impact on force, fatigability, and fiber type. American Journal of Physiology - Cell Physiology, 2012, 302, C88-C99.	4.6	51
56	Agrin regulates CLASP2-mediated capture of microtubules at the neuromuscular junction synaptic membrane. Journal of Cell Biology, 2012, 198, 421-437.	5.2	57
57	SRP-35, a newly identified protein of the skeletal muscle sarcoplasmic reticulum, is a retinol dehydrogenase. Biochemical Journal, 2012, 441, 731-741.	3.7	12
58	Downstream Effects of a RyR1 Mutation Linked to Malignant Hyperthermia on the Phenotypic and Functional Characteristic of Dendritic Cells. Biophysical Journal, 2012, 102, 307a.	0.5	0
59	Upregulation of Calcium Influx via Cav1.1 in Skeletal Muscle Fibers from JP-45 Calsequestrin 1 Double Knock Out Mice. Biophysical Journal, 2012, 102, 364a.	0.5	0
60	Mapping domains and mutations on the skeletal muscle ryanodine receptor channel. Trends in Molecular Medicine, 2012, 18, 644-657.	6.7	47
61	Endogenously determined restriction of food intake overcomes excitation–contraction uncoupling in JP45KO mice with aging. Experimental Gerontology, 2012, 47, 304-316.	2.8	6
62	Screening of the Ryanodine 1 Gene for Malignant Hyperthermia Causative Mutations by High Resolution Melt Curve Analysis. Anesthesia and Analgesia, 2011, 113, 1120-1128.	2.2	14
63	Alterations of excitation-contraction coupling and excitation coupled Ca2+ entry in human myotubes carrying CAV3 mutations linked to rippling muscle. Human Mutation, 2011, 32, 309-317.	2.5	15
64	Enhanced excitation-coupled Ca2+ entry induces nuclear translocation of NFAT and contributes to IL-6 release from myotubes from patients with central core disease. Human Molecular Genetics, 2011, 20, 589-600.	2.9	22
65	Functional Properties of RYR1 Mutations Identified in Swedish Patients with Malignant Hyperthermia and Central Core Disease. Anesthesia and Analgesia, 2010, 111, 185-190.	2.2	33
66	Frequent Calcium Oscillations Lead to NFAT Activation in Human Immature Dendritic Cells. Journal of Biological Chemistry, 2010, 285, 16003-16011.	3.4	29
67	Agonist-activated Ca2+ influx occurs at stable plasma membrane and endoplasmic reticulum junctions. Journal of Cell Science, 2010, 123, 4170-4181.	2.0	47
68	Ectosomes Released by Polymorphonuclear Neutrophils Induce a MerTK-dependent Anti-inflammatory Pathway in Macrophages. Journal of Biological Chemistry, 2010, 285, 39914-39921.	3.4	124
69	Multi-minicore disease and atypical periodic paralysis associated with novel mutations in the skeletal muscle ryanodine receptor (RYR1) gene. Neuromuscular Disorders, 2010, 20, 166-173.	0.6	78
70	Altering Skeletal Muscle EC Coupling by Ablating the Sarcoplasmic Reticulum Protein JP45 Affects Both Metabolism and Muscle Performance in Old Mice. Biophysical Journal, 2010, 98, 547a.	0.5	0
71	Novel Insights into the Role of Junctate in Calcium Homeostasis: Identification of Binding Domain on the InsP3R and Cellular Localization as Determined by TIRF Microscopy. Biophysical Journal, 2010, 98, 513a.	0.5	0
72	Calcium Influx Analysis by TIRF Microscopy on Cultured Primary Myotubes from Patients with RyR1 Mutations. Biophysical Journal, 2010, 98, 509a.	0.5	0

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73	A recessive ryanodine receptor 1 mutation in a CCD patient increases channel activity. Cell Calcium, 2009, 45, 192-197.	2.4	16
74	Increasing the number of diagnostic mutations in malignant hyperthermia. Human Mutation, 2009, 30, 590-598.	2.5	66
75	TIRF. Imaging & Microscopy, 2009, 11, 52-53.	0.1	0
76	Minor sarcoplasmic reticulum membrane components that modulate excitation–contraction coupling in striated muscles. Journal of Physiology, 2009, 587, 3071-3079.	2.9	64
77	Ryanodine receptor 1 signaling in dendritic cells. Biophysical Journal, 2009, 96, 122a.	0.5	0
78	SRP-35 A Putative NAD(P)H Binding Protein Of Skeletal Muscle Sarcoplasmic Reticulum Membrane. Biophysical Journal, 2009, 96, 235a.	0.5	0
79	Upstream stimulatory factors are involved in the P1 promoter directed transcription of the AbetaH-J-J locus. BMC Molecular Biology, 2008, 9, 110.	3.0	16
80	Congenital muscle disorders with cores: the ryanodine receptor calcium channel paradigm. Current Opinion in Pharmacology, 2008, 8, 319-326.	3.5	149
81	Ryanodine Receptor Activation by Cav1.2 Is Involved in Dendritic Cell Major Histocompatibility Complex Class II Surface Expression. Journal of Biological Chemistry, 2008, 283, 34913-34922.	3.4	29
82	SRP-27 is a novel component of the supramolecular signalling complex involved in skeletal muscle excitation–contraction coupling. Biochemical Journal, 2008, 411, 343-349.	3.7	20
83	Molecular mechanisms and phenotypic variation in RYR1-related congenital myopathies. Brain, 2007, 130, 2024-2036.	7.6	161
84	Loss of skeletal muscle strength by ablation of the sarcoplasmic reticulum protein JP45. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 20108-20113.	7.1	35
85	Ca2+ signaling through ryanodine receptor 1 enhances maturation and activation of human dendritic cells. Journal of Cell Science, 2007, 120, 2232-2240.	2.0	19
86	Centronuclear myopathy due to a de novo dominant mutation in the skeletal muscle ryanodine receptor (RYR1) gene. Neuromuscular Disorders, 2007, 17, 338-345.	0.6	105
87	Two central core disease (CCD) deletions in the C-terminal region of RYR1 alter muscle excitation-contraction (EC) coupling by distinct mechanisms. Human Mutation, 2007, 28, 61-68.	2.5	26
88	Increased Ca2+ storage capacity of the skeletal muscle sarcoplasmic reticulum of transgenic mice over-expressing membrane bound calcium binding protein junctate. Journal of Cellular Physiology, 2007, 213, 464-474.	4.1	23
89	Transcriptional activity and Sp 1/3 transcription factor binding to the P1 promoter sequences of the human AβHâ€Jâ€J locus. FEBS Journal, 2007, 274, 4476-4490.	4.7	14
90	Functional effects of mutations identified in patients with Multiminicore disease. IUBMB Life, 2007, 59, 14-20.	3.4	30

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91	Functional properties of ryanodine receptors carrying three amino acid substitutions identified in patients affected by multi-minicore disease and central core disease, expressed in immortalized lymphocytes. Biochemical Journal, 2006, 395, 259-266.	3.7	59
92	Multiple Levels of Control of the Expression of the Human AβH-J-J Locus Encoding Aspartyl-β-hydroxylase, Junctin, and Junctate. Annals of the New York Academy of Sciences, 2006, 1091, 184-190.	3.8	14
93	The junctional SR protein JP-45 affects the functional expression of the voltage-dependent Ca2+ channel Cav1.1. Journal of Cell Science, 2006, 119, 2145-2155.	2.0	30
94	Characterization of recessive RYR1 mutations in core myopathies. Human Molecular Genetics, 2006, 15, 2791-2803.	2.9	103
95	Myocyte Enhancer Factor 2 Activates Promoter Sequences of the Human AβH-J-J Locus, Encoding Aspartyl-β-Hydroxylase, Junctin, and Junctate. Molecular and Cellular Biology, 2005, 25, 3261-3275.	2.3	12
96	Junctate, an inositol 1,4,5-triphosphate receptor associated protein, is present in rodent sperm and binds TRPC2 and TRPC5 but not TRPC1 channels. Developmental Biology, 2005, 286, 326-337.	2.0	62
97	Ryanodine receptor 1 mutations, dysregulation of calcium homeostasis and neuromuscular disorders. Neuromuscular Disorders, 2005, 15, 577-587.	0.6	126
98	Junctate is a key element in calcium entry induced by activation of InsP3 receptors and/or calcium store depletion. Journal of Cell Biology, 2004, 166, 537-548.	5.2	116
99	Effect of Ryanodine Receptor Mutations on Interleukin-6 Release and Intracellular Calcium Homeostasis in Human Myotubes from Malignant Hyperthermia-susceptible Individuals and Patients Affected by Central Core Disease. Journal of Biological Chemistry, 2004, 279, 43838-43846.	3.4	96
100	Molecular Genetic Testing for Malignant Hyperthermia Susceptibility. Anesthesiology, 2004, 100, 1076-1080.	2.5	95
101	Clinical and functional effects of a deletion in a COOH-terminal lumenal loop of the skeletal muscle ryanodine receptor. Human Molecular Genetics, 2003, 12, 379-388.	2.9	35
102	The Novel Skeletal Muscle Sarcoplasmic Reticulum JP-45 Protein. Journal of Biological Chemistry, 2003, 278, 39987-39992.	3.4	36
103	B-lymphocytes from Malignant Hyperthermia-susceptible Patients Have an Increased Sensitivity to Skeletal Muscle Ryanodine Receptor Activators. Journal of Biological Chemistry, 2001, 276, 48077-48082.	3.4	74
104	Genotype-phenotype comparison of the Swiss malignant hyperthermia population. Human Mutation, 2001, 18, 357-358.	2.5	61
105	Identification of a novel 45ÂkDa protein (JP-45) from rabbit sarcoplasmic-reticulum junctional-face membrane. Biochemical Journal, 2000, 351, 537.	3.7	9
106	Identification of a novel 45 kDa protein (JP-45) from rabbit sarcoplasmic-reticulum junctional-face membrane. Biochemical Journal, 2000, 351, 537-543.	3.7	30
107	Methyl p-hydroxybenzoate (E-218) a preservative for drugs and food is an activator of the ryanodine receptor Ca2+ release channel. British Journal of Pharmacology, 2000, 131, 335-341.	5.4	8
108	Molecular Cloning, Expression, Functional Characterization, Chromosomal Localization, and Gene Structure of Junctate, a Novel Integral Calcium Binding Protein of Sarco(endo)plasmic Reticulum Membrane. Journal of Biological Chemistry, 2000, 275, 39555-39568.	3.4	87

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109	Nerve growth factor (NGF) influences differentiation and proliferation of myogenic cells in vitro via TrKA. International Journal of Developmental Neuroscience, 2000, 18, 869-885.	1.6	73
110	Interaction of S100A1 with the Ca2+Release Channel (Ryanodine Receptor) of Skeletal Muscleâ€. Biochemistry, 1997, 36, 11496-11503.	2.5	113
111	Malignant Hyperthermia Domain in the Regulation of Ca2+ Release Channel (Ryanodine Receptor). Trends in Cardiovascular Medicine, 1997, 7, 312-316.	4.9	3
112	Chlorocresol, an Additive to Commercial Succinylcholine, Induces Contracture of Human Malignant Hyperthermia-susceptible Muscles Via Activation of the Ryanodine Receptor Calcium sup 2+ Channel. Anesthesiology, 1996, 84, 1380-1385.	2.5	54
113	Role of Malignant Hyperthermia Domain in the Regulation of Ca2+ Release Channel (Ryanodine) Tj ETQq1 1 0.784 22759-22763.	314 rgBT 3.4	/Overlock 34
114	Calmodulin Binding Sites of the Skeletal, Cardiac, and Brain Ryanodine Receptor Ca2+ Channels: Modulation by the Catalytic Subunit of cAMP-Dependent Protein Kinase?. Biochemistry, 1995, 34, 5120-5129.	2.5	41
115	Identification and Characterization of Three Calmodulin Binding Sites of the Skeletal Muscle Rvanodine Receptor, Biochemistry, 1994, 33, 9078-9084.	2.5	75