

# Susan Treves

## List of Publications by Year in descending order

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115  
papers

3,920  
citations

101543

36  
h-index

138484

58  
g-index

121  
all docs

121  
docs citations

121  
times ranked

3716  
citing authors

#	ARTICLE	IF	CITATIONS
1	Congenital myopathies: disorders of excitation-contraction coupling and muscle contraction. <i>Nature Reviews Neurology</i> , 2018, 14, 151-167.	10.1	212
2	Molecular mechanisms and phenotypic variation in RYR1-related congenital myopathies. <i>Brain</i> , 2007, 130, 2024-2036.	7.6	161
3	Congenital muscle disorders with cores: the ryanodine receptor calcium channel paradigm. <i>Current Opinion in Pharmacology</i> , 2008, 8, 319-326.	3.5	149
4	Ryanodine receptor 1 mutations, dysregulation of calcium homeostasis and neuromuscular disorders. <i>Neuromuscular Disorders</i> , 2005, 15, 577-587.	0.6	126
5	Ectosomes Released by Polymorphonuclear Neutrophils Induce a MerTK-dependent Anti-inflammatory Pathway in Macrophages. <i>Journal of Biological Chemistry</i> , 2010, 285, 39914-39921.	3.4	124
6	Junctate is a key element in calcium entry induced by activation of InsP3 receptors and/or calcium store depletion. <i>Journal of Cell Biology</i> , 2004, 166, 537-548.	5.2	116
7	Interaction of S100A1 with the Ca <sup>2+</sup> -Release Channel (Ryanodine Receptor) of Skeletal Muscle. <i>Biochemistry</i> , 1997, 36, 11496-11503.	2.5	113
8	Centronuclear myopathy due to a de novo dominant mutation in the skeletal muscle ryanodine receptor (RYR1) gene. <i>Neuromuscular Disorders</i> , 2007, 17, 338-345.	0.6	105
9	Characterization of recessive RYR1 mutations in core myopathies. <i>Human Molecular Genetics</i> , 2006, 15, 2791-2803.	2.9	103
10	Dihydropyridine receptor (DHPR, CACNA1S) congenital myopathy. <i>Acta Neuropathologica</i> , 2017, 133, 517-533.	7.7	97
11	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. <i>Journal of Biological Chemistry</i> , 2004, 279, 43838-43846.	3.4	96
12	Molecular Genetic Testing for Malignant Hyperthermia Susceptibility. <i>Anesthesiology</i> , 2004, 100, 1076-1080.	2.5	95
13	Molecular Cloning, Expression, Functional Characterization, Chromosomal Localization, and Gene Structure of Junctate, a Novel Integral Calcium Binding Protein of Sarco(endo)plasmic Reticulum Membrane. <i>Journal of Biological Chemistry</i> , 2000, 275, 39555-39568.	3.4	87
14	Multi-minicore disease and atypical periodic paralysis associated with novel mutations in the skeletal muscle ryanodine receptor (RYR1) gene. <i>Neuromuscular Disorders</i> , 2010, 20, 166-173.	0.6	78
15	Identification and Characterization of Three Calmodulin Binding Sites of the Skeletal Muscle Ryanodine Receptor. <i>Biochemistry</i> , 1994, 33, 9078-9084.	2.5	75
16	B-lymphocytes from Malignant Hyperthermia-susceptible Patients Have an Increased Sensitivity to Skeletal Muscle Ryanodine Receptor Activators. <i>Journal of Biological Chemistry</i> , 2001, 276, 48077-48082.	3.4	74
17	Ectosomes of polymorphonuclear neutrophils activate multiple signaling pathways in macrophages. <i>Immunobiology</i> , 2013, 218, 382-392.	1.9	74
18	Nerve growth factor (NGF) influences differentiation and proliferation of myogenic cells in vitro via TrKA. <i>International Journal of Developmental Neuroscience</i> , 2000, 18, 869-885.	1.6	73

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19	Exertional rhabdomyolysis: physiological response or manifestation of an underlying myopathy?. <i>BMJ Open Sport and Exercise Medicine</i> , 2016, 2, e000151.	2.9	73
20	Increasing the number of diagnostic mutations in malignant hyperthermia. <i>Human Mutation</i> , 2009, 30, 590-598.	2.5	66
21	Minor sarcoplasmic reticulum membrane components that modulate excitation-contraction coupling in striated muscles. <i>Journal of Physiology</i> , 2009, 587, 3071-3079.	2.9	64
22	Junctate, an inositol 1,4,5-triphosphate receptor associated protein, is present in rodent sperm and binds TRPC2 and TRPC5 but not TRPC1 channels. <i>Developmental Biology</i> , 2005, 286, 326-337.	2.0	62
23	Genotype-phenotype comparison of the Swiss malignant hyperthermia population. <i>Human Mutation</i> , 2001, 18, 357-358.	2.5	61
24	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. <i>Biochemical Journal</i> , 2006, 395, 259-266.	3.7	59
25	Agrin regulates CLASP2-mediated capture of microtubules at the neuromuscular junction synaptic membrane. <i>Journal of Cell Biology</i> , 2012, 198, 421-437.	5.2	57
26	Chlorocresol, an Additive to Commercial Succinylcholine, Induces Contracture of Human Malignant Hyperthermia-susceptible Muscles Via Activation of the Ryanodine Receptor Calcium $\text{sup } 2+$ Channel. <i>Anesthesiology</i> , 1996, 84, 1380-1385.	2.5	54
27	Remodeling of calcium handling in skeletal muscle through PGC-1 $\beta$ : impact on force, fatigability, and fiber type. <i>American Journal of Physiology - Cell Physiology</i> , 2012, 302, C88-C99.	4.6	51
28	Agonist-activated $\text{Ca}^{2+}$ influx occurs at stable plasma membrane and endoplasmic reticulum junctions. <i>Journal of Cell Science</i> , 2010, 123, 4170-4181.	2.0	47
29	Mapping domains and mutations on the skeletal muscle ryanodine receptor channel. <i>Trends in Molecular Medicine</i> , 2012, 18, 644-657.	6.7	47
30	Epigenetic changes as a common trigger of muscle weakness in congenital myopathies. <i>Human Molecular Genetics</i> , 2015, 24, 4636-4647.	2.9	44
31	<i>STAC3</i> variants cause a congenital myopathy with distinctive dysmorphic features and malignant hyperthermia susceptibility. <i>Human Mutation</i> , 2018, 39, 1980-1994.	2.5	42
32	Calmodulin Binding Sites of the Skeletal, Cardiac, and Brain Ryanodine Receptor $\text{Ca}^{2+}$ Channels: Modulation by the Catalytic Subunit of cAMP-Dependent Protein Kinase?. <i>Biochemistry</i> , 1995, 34, 5120-5129.	2.5	41
33	RyR1 Deficiency in Congenital Myopathies Disrupts Excitation-Contraction Coupling. <i>Human Mutation</i> , 2013, 34, 986-996.	2.5	40
34	Atypical periodic paralysis and myalgia. <i>Neurology</i> , 2018, 90, e412-e418.	1.1	39
35	The Novel Skeletal Muscle Sarcoplasmic Reticulum JP-45 Protein. <i>Journal of Biological Chemistry</i> , 2003, 278, 39987-39992.	3.4	36
36	Clinical and functional effects of a deletion in a COOH-terminal luminal loop of the skeletal muscle ryanodine receptor. <i>Human Molecular Genetics</i> , 2003, 12, 379-388.	2.9	35

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37	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
38	Enhanced dihydropyridine receptor calcium channel activity restores muscle strength in JP45/CASQ1 double knockout mice. Nature Communications, 2013, 4, 1541.	12.8	35
39	Role of Malignant Hyperthermia Domain in the Regulation of Ca <sup>2+</sup> Release Channel (Ryanodine) Tj ETQq1 1 0.784314 rgBT /Overlock 22759-22763.	3.4	34
40	An <i>RYR1</i> mutation associated with malignant hyperthermia is also associated with bleeding abnormalities. Science Signaling, 2016, 9, ra68.	3.6	34
41	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
42	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
43	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
44	The junctional SR protein JP-45 affects the functional expression of the voltage-dependent Ca <sup>2+</sup> channel Cav1.1. Journal of Cell Science, 2006, 119, 2145-2155.	2.0	30
45	Functional effects of mutations identified in patients with Multiminicore disease. IUBMB Life, 2007, 59, 14-20.	3.4	30
46	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
47	Frequent Calcium Oscillations Lead to NFAT Activation in Human Immature Dendritic Cells. Journal of Biological Chemistry, 2010, 285, 16003-16011.	3.4	29
48	Current and future therapeutic approaches to the congenital myopathies. Seminars in Cell and Developmental Biology, 2017, 64, 191-200.	5.0	29
49	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
50	Increased Ca <sup>2+</sup> 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
51	Enhanced excitation-coupled Ca <sup>2+</sup> 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
52	Clinical, morphological and genetic characterization of Brody disease: an international study of 40 patients. Brain, 2020, 143, 452-466.	7.6	22
53	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
54	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

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55	Cellular, biochemical and molecular changes in muscles from patients with X-linked myotubular myopathy due to MTM1 mutations. <i>Human Molecular Genetics</i> , 2016, 26, ddw388.	2.9	20
56	Ca <sup>2+</sup> signaling through ryanodine receptor 1 enhances maturation and activation of human dendritic cells. <i>Journal of Cell Science</i> , 2007, 120, 2232-2240.	2.0	19
57	Establishment of a human skeletal muscle-derived cell line: biochemical, cellular and electrophysiological characterization. <i>Biochemical Journal</i> , 2013, 455, 169-177.	3.7	19
58	Ca <sup>2+</sup> handling abnormalities in early-onset muscle diseases: Novel concepts and perspectives. <i>Seminars in Cell and Developmental Biology</i> , 2017, 64, 201-212.	5.0	19
59	Over-expression of a retinol dehydrogenase (SRP35/DHRS7C) in skeletal muscle activates mTORC2, enhances glucose metabolism and muscle performance. <i>Scientific Reports</i> , 2018, 8, 636.	3.3	19
60	Rapid subcellular calcium responses and dynamics by calcium sensor G-CatchER+. <i>IScience</i> , 2021, 24, 102129.	4.1	19
61	The metabolomic profile of gamma-irradiated human hepatoma and muscle cells reveals metabolic changes consistent with the Warburg effect. <i>PeerJ</i> , 2016, 4, e1624.	2.0	18
62	Upstream stimulatory factors are involved in the P1 promoter directed transcription of the AbetaH-JJ locus. <i>BMC Molecular Biology</i> , 2008, 9, 110.	3.0	16
63	A recessive ryanodine receptor 1 mutation in a CCD patient increases channel activity. <i>Cell Calcium</i> , 2009, 45, 192-197.	2.4	16
64	Alterations of excitation-contraction coupling and excitation coupled Ca <sup>2+</sup> entry in human myotubes carrying CAV3 mutations linked to rippling muscle. <i>Human Mutation</i> , 2011, 32, 309-317.	2.5	15
65	Multiple Levels of Control of the Expression of the Human $\text{A}^{\beta}\text{H-JJ}$ Locus Encoding Aspartyl- $\text{I}^2$ -hydroxylase, Junctin, and Junctate. <i>Annals of the New York Academy of Sciences</i> , 2006, 1091, 184-190.	3.8	14
66	Transcriptional activity and Sp 1/3 transcription factor binding to the P1 promoter sequences of the human $\text{A}^{\beta}\text{H-JJ}$ locus. <i>FEBS Journal</i> , 2007, 274, 4476-4490.	4.7	14
67	Screening of the Ryanodine 1 Gene for Malignant Hyperthermia Causative Mutations by High Resolution Melt Curve Analysis. <i>Anesthesia and Analgesia</i> , 2011, 113, 1120-1128.	2.2	14
68	Gain of function of the immune system caused by a ryanodine receptor 1 mutation. <i>Journal of Cell Science</i> , 2013, 126, 3485-92.	2.0	14
69	Aberrant regulation of epigenetic modifiers contributes to the pathogenesis in patients with selenoprotein N related myopathies. <i>Human Mutation</i> , 2019, 40, 962-974.	2.5	13
70	Myocyte Enhancer Factor 2 Activates Promoter Sequences of the Human $\text{A}^{\beta}\text{H-JJ}$ Locus, Encoding Aspartyl- $\text{I}^2$ -Hydroxylase, Junctin, and Junctate. <i>Molecular and Cellular Biology</i> , 2005, 25, 3261-3275.	2.3	12
71	SRP-35, a newly identified protein of the skeletal muscle sarcoplasmic reticulum, is a retinol dehydrogenase. <i>Biochemical Journal</i> , 2012, 441, 731-741.	3.7	12
72	JP-45/SRP Variants Affect Skeletal Muscle Excitation-Contraction Coupling by Decreasing the Sensitivity of the Dihydropyridine Receptor. <i>Human Mutation</i> , 2013, 34, 184-190.	2.5	12

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73	Role of the JP45-Calsequestrin Complex on Calcium Entry in Slow Twitch Skeletal Muscles. <i>Journal of Biological Chemistry</i> , 2016, 291, 14555-14565.	3.4	12
74	Variable Myopathic Presentation in a Single Family with Novel Skeletal RYR1 Mutation. <i>PLoS ONE</i> , 2013, 8, e69296.	2.5	12
75	Pre-operative exercise and pyrexia as modifying factors in malignant hyperthermia (MH). <i>Neuromuscular Disorders</i> , 2022, 32, 628-634.	0.6	12
76	Extraocular muscle function is impaired in <i>ryr3</i> mice. <i>Journal of General Physiology</i> , 2019, 151, 929-943.	1.9	11
77	RYR1-Related Rhabdomyolysis: A Spectrum of Hypermetabolic States Due to Ryanodine Receptor Dysfunction. <i>Current Pharmaceutical Design</i> , 2022, 28, 2-14.	1.9	11
78	Raptor ablation in skeletal muscle decreases Cav1.1 expression and affects the function of the excitation-contraction coupling supramolecular complex. <i>Biochemical Journal</i> , 2015, 466, 123-135.	3.7	10
79	Characterization of excitation-contraction coupling components in human extraocular muscles. <i>Biochemical Journal</i> , 2015, 466, 29-36.	3.7	10
80	Identification of a novel 45 kDa protein (JP-45) from rabbit sarcoplasmic-reticulum junctional-face membrane. <i>Biochemical Journal</i> , 2000, 351, 537.	3.7	9
81	Functional characterization of orbicularis oculi and extraocular muscles. <i>Journal of General Physiology</i> , 2016, 147, 395-406.	1.9	9
82	Methyl p-hydroxybenzoate (E-218) a preservative for drugs and food is an activator of the ryanodine receptor Ca <sup>2+</sup> release channel. <i>British Journal of Pharmacology</i> , 2000, 131, 335-341.	5.4	8
83	Functional characterization of the RYR1 mutation p.Arg4737Trp associated with susceptibility to malignant hyperthermia. <i>Neuromuscular Disorders</i> , 2016, 26, 21-25.	0.6	8
84	Quantitative reduction of RyR1 protein caused by a single-allele frameshift mutation in RYR1 ex36 impairs the strength of adult skeletal muscle fibres. <i>Human Molecular Genetics</i> , 2019, 28, 1872-1884.	2.9	8
85	The neuromuscular and multisystem features of RYR1-related malignant hyperthermia and rhabdomyolysis. <i>Medicine (United States)</i> , 2021, 100, e26999.	1.0	8
86	Improvement of muscle strength in a mouse model for congenital myopathy treated with HDAC and DNA methyltransferase inhibitors. <i>ELife</i> , 2022, 11, .	6.0	7
87	Endogenously determined restriction of food intake overcomes excitation-contraction uncoupling in JP45KO mice with aging. <i>Experimental Gerontology</i> , 2012, 47, 304-316.	2.8	6
88	Molecular basis of impaired extraocular muscle function in a mouse model of congenital myopathy due to compound heterozygous Ryr1 mutations. <i>Human Molecular Genetics</i> , 2020, 29, 1330-1339.	2.9	5
89	Bi-allelic expression of the RyR1 p.A4329D mutation decreases muscle strength in slow-twitch muscles in mice. <i>Journal of Biological Chemistry</i> , 2020, 295, 10331-10339.	3.4	4
90	Malignant Hyperthermia Domain in the Regulation of Ca <sup>2+</sup> Release Channel (Ryanodine Receptor). <i>Trends in Cardiovascular Medicine</i> , 1997, 7, 312-316.	4.9	3

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91	3D Structural Illumination Microscopy of the Skeletal Muscle Excitation-Contraction Coupling Macromolecular Complex. <i>Biophysical Journal</i> , 2013, 104, 105a.	0.5	2
92	Comment on "Overlapping Mechanisms of Exertional Heat Stroke and Malignant Hyperthermia: Evidence vs. Conjecture". <i>Sports Medicine</i> , 2021, , 1.	6.5	2
93	Novel Local RYR1 Junctional Calcium Responses and Dynamics by Improved Calcium Sensor Catcher. <i>Biophysical Journal</i> , 2017, 112, 331a.	0.5	1
94	Biochemical and Mechanical Properties of Murine Extraocular Muscles. <i>Biophysical Journal</i> , 2017, 112, 101a-102a.	0.5	1
95	TIRF. <i>Imaging &amp; Microscopy</i> , 2009, 11, 52-53.	0.1	0
96	Ryanodine receptor 1 signaling in dendritic cells. <i>Biophysical Journal</i> , 2009, 96, 122a.	0.5	0
97	SRP-35 A Putative NAD(P)H Binding Protein Of Skeletal Muscle Sarcoplasmic Reticulum Membrane. <i>Biophysical Journal</i> , 2009, 96, 235a.	0.5	0
98	Altering Skeletal Muscle EC Coupling by Ablating the Sarcoplasmic Reticulum Protein JP45 Affects Both Metabolism and Muscle Performance in Old Mice. <i>Biophysical Journal</i> , 2010, 98, 547a.	0.5	0
99	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. <i>Biophysical Journal</i> , 2010, 98, 513a.	0.5	0
100	Calcium Influx Analysis by TIRF Microscopy on Cultured Primary Myotubes from Patients with RyR1 Mutations. <i>Biophysical Journal</i> , 2010, 98, 509a.	0.5	0
101	Downstream Effects of a RyR1 Mutation Linked to Malignant Hyperthermia on the Phenotypic and Functional Characteristic of Dendritic Cells. <i>Biophysical Journal</i> , 2012, 102, 307a.	0.5	0
102	Upregulation of Calcium Influx via Cav1.1 in Skeletal Muscle Fibers from JP-45 Calsequestrin 1 Double Knock Out Mice. <i>Biophysical Journal</i> , 2012, 102, 364a.	0.5	0
103	Physiological Role(S) of RyR1 in Smooth Muscle Cells. <i>Biophysical Journal</i> , 2013, 104, 443a-444a.	0.5	0
104	Role of sarcoplasmic reticulum junctional proteins in skeletal muscle strength. <i>BMC Anesthesiology</i> , 2014, 14, .	1.8	0
105	Biochemical, Cellular and Electrophysiological Characterization of HMCL-7304 a Human Skeletal Muscle-Derived Cell Line. <i>Biophysical Journal</i> , 2014, 106, 446a.	0.5	0
106	Raptor Ablation in Skeletal Muscle Affects the Structure and Function of the Excitation-Contraction Coupling Macromolecular Complex. <i>Biophysical Journal</i> , 2014, 106, 123a.	0.5	0
107	Excitation-Contraction Coupling in Human Extraocular Muscles: There is more than Meets the Eye. <i>Biophysical Journal</i> , 2015, 108, 420a.	0.5	0
108	Differential Role of Calsequestrin Isoforms on Calcium Entry in Skeletal Muscle FDB Fibres. <i>Biophysical Journal</i> , 2015, 108, 269a.	0.5	0

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109	Altered Eye Muscle Function in RYR3KO Mice. Biophysical Journal, 2018, 114, 469a.	0.5	0
110	Biochemical and Epigenetic Modifications Occur in Muscles of Patients with Selenoprotein N Related Congenital Myopathy. Biophysical Journal, 2019, 116, 153a.	0.5	0
111	Characterization of an Animal Model for Congenital Myopathies Linked to Recessive RyR1 Mutations. Biophysical Journal, 2019, 116, 522a.	0.5	0
112	Functional Characterization of Endogenously Expressed Human RYR1 Variants. Journal of Visualized Experiments, 2021, , .	0.3	0
113	Functional characterization of orbicularis oculi and extraocular muscles. Journal of Cell Biology, 2016, 213, 21330IA96.	5.2	0
114	Role of Calcium in Neutrophil Activation. , 2018, , 199-218.		0
115	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