

Damien Sternberg

List of Publications by Year in descending order

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59
papers

2,622
citations

201385

27
h-index

189595

50
g-index

60
all docs

60
docs citations

60
times ranked

2814
citing authors

#	ARTICLE	IF	CITATIONS
1	Phenotypic spectrum and genomics of undiagnosed arthrogryposis multiplex congenita. <i>Journal of Medical Genetics</i> , 2022, 59, 559-567.	1.5	25
2	A <i>TOR1AIP1</i> variant segregating with an early onset limb girdle myasthenia—Support for the role of LAP1 in NMJ function and disease. <i>Neuropathology and Applied Neurobiology</i> , 2022, 48, .	1.8	4
3	Phenotypical variability and atypical presentations in a French cohort of Andersen—Tawil syndrome. <i>European Journal of Neurology</i> , 2022, 29, 2398-2411.	1.7	1
4	Clinical, morphological and genetic characterization of Brody disease: an international study of 40 patients. <i>Brain</i> , 2020, 143, 452-466.	3.7	22
5	Hypokalaemic periodic paralysis with a charge-retaining substitution in the voltage sensor. <i>Brain Communications</i> , 2020, 2, fcaa103.	1.5	11
6	Non-dystrophic myotonia Chilean cohort with predominance of the SCN4A Gly1306Glu variant. <i>Neuromuscular Disorders</i> , 2020, 30, 554-561.	0.3	6
7	Brody myopathy demonstrates a pseudo-increment on repetitive nerve stimulation. <i>Muscle and Nerve</i> , 2020, 61, 491-495.	1.0	2
8	New recessive mutations in <i>SYT2</i> causing severe presynaptic congenital myasthenic syndromes. <i>Neurology: Genetics</i> , 2020, 6, e534.	0.9	6
9	A National French consensus on gene lists for the diagnosis of myopathies using next-generation sequencing. <i>European Journal of Human Genetics</i> , 2019, 27, 349-352.	1.4	27
10	Pharmacogenetics of myotonic hNav1.4 sodium channel variants situated near the fast inactivation gate. <i>Pharmacological Research</i> , 2019, 141, 224-235.	3.1	25
11	Substitutions of the S4DIV R2 residue (R1451) in Nav1.4 lead to complex forms of paramyotonia congenita and periodic paralyses. <i>Scientific Reports</i> , 2018, 8, 2041.	1.6	14
12	How chromosomal deletions can unmask recessive mutations? Deletions in 10q11.2 associated with <i>CHAT</i> or <i>SLC18A3</i> mutations lead to congenital myasthenic syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 151-155.	0.7	16
13	A204E mutation in Nav1.4 DIS3 exerts gain- and loss-of-function effects that lead to periodic paralysis combining hyper- with hypo-kalaemic signs. <i>Scientific Reports</i> , 2018, 8, 16681.	1.6	5
14	Prolonged attacks of weakness with hypokalemia in <i>SCN4A</i> -related paramyotonia congenita. <i>Muscle and Nerve</i> , 2018, 58, E27-E28.	1.0	6
15	Mutations in GFPT1-related congenital myasthenic syndromes are associated with synaptic morphological defects and underlie a tubular aggregate myopathy with synaptopathy. <i>Journal of Neurology</i> , 2017, 264, 1791-1803.	1.8	26
16	Impaired Presynaptic High-Affinity Choline Transporter Causes a Congenital Myasthenic Syndrome with Episodic Apnea. <i>American Journal of Human Genetics</i> , 2016, 99, 753-761.	2.6	68
17	A recessive Na ^v 1.4 mutation underlies congenital myasthenic syndrome with periodic paralysis. <i>Neurology</i> , 2016, 86, 161-169.	1.5	49
18	Impaired surface membrane insertion of homo- and heterodimeric human muscle chloride channels carrying amino-terminal myotonia-causing mutations. <i>Scientific Reports</i> , 2015, 5, 15382.	1.6	21

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19	Atypical nuclear abnormalities in a patient with Brody disease. <i>Neuromuscular Disorders</i> , 2015, 25, 773-779.	0.3	4
20	Truncating Mutations of MAGEL2, a Gene within the Prader-Willi Locus, Are Responsible for Severe Arthrogyrosis. <i>American Journal of Human Genetics</i> , 2015, 97, 616-620.	2.6	49
21	Physiological and Pathophysiological Insights of Nav1.4 and Nav1.5 Comparison. <i>Frontiers in Pharmacology</i> , 2015, 6, 314.	1.6	40
22	Mutations in CNTNAP1 and ADCY6 are responsible for severe arthrogyrosis multiplex congenita with axoglial defects. <i>Human Molecular Genetics</i> , 2014, 23, 2279-2289.	1.4	98
23	Agrin mutations lead to a congenital myasthenic syndrome with distal muscle weakness and atrophy. <i>Brain</i> , 2014, 137, 2429-2443.	3.7	86
24	Heterozygous CLCN1 mutations can modulate phenotype in sodium channel myotonia. <i>Neuromuscular Disorders</i> , 2014, 24, 953-959.	0.3	27
25	Syndromes myasthéniques congénitaux "L'expérience française". <i>Bulletin De L'Academie Nationale De Medecine</i> , 2014, 198, 257-271.	0.0	1
26	Focal and abnormally persistent paralysis associated with congenital paramyotonia. <i>BMJ Case Reports</i> , 2014, 2014, bcr2014204430-bcr2014204430.	0.2	6
27	The neuronal endopeptidase ECEL1 is associated with a distinct form of recessive distal arthrogyrosis. <i>Human Molecular Genetics</i> , 2013, 22, 1483-1492.	1.4	66
28	Diagnosis and Outcome of <i>SCN4A</i> -Related Severe Neonatal Episodic Laryngospasm (SNEL): 2 New Cases. <i>Pediatrics</i> , 2013, 132, e784-e787.	1.0	34
29	Episodic weakness due to mitochondrial DNA <i>MT-ATP6/8</i> mutations. <i>Neurology</i> , 2013, 81, 1810-1818.	1.5	48
30	Disease-causing mutations C277R and C277Y modify gating of human <i>ClC-1</i> chloride channels in myotonia congenita. <i>Journal of Physiology</i> , 2012, 590, 3449-3464.	1.3	32
31	Relapses and remissions of hypokalaemic periodic paralysis and multiple sclerosis in the same patient. <i>Clinical Neurology and Neurosurgery</i> , 2011, 113, 683-685.	0.6	1
32	Mechanisms underlying a life-threatening skeletal muscle Na^{+} channel disorder. <i>Journal of Physiology</i> , 2011, 589, 3115-3124.	1.3	16
33	Homozygosity for dominant mutations increases severity of muscle channelopathies. <i>Muscle and Nerve</i> , 2010, 41, 470-477.	1.0	24
34	The non-dystrophic myotonias: molecular pathogenesis, diagnosis and treatment. <i>Brain</i> , 2010, 133, 9-22.	3.7	244
35	Hypokalaemia and dysmorphia, is there a link?. <i>CKJ: Clinical Kidney Journal</i> , 2009, 2, 222-224.	1.4	0
36	Cold-induced disruption of Na^{+} channel slow inactivation underlies paralysis in highly thermosensitive paramyotonia. <i>Journal of Physiology</i> , 2009, 587, 1705-1714.	1.3	24

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37	Clinical, electrophysiologic, and genetic study of non-dystrophic myotonia in French-Canadians. <i>Neuromuscular Disorders</i> , 2009, 19, 330-334.	0.3	44
38	Glucocorticoids may trigger attacks in several types of periodic paralysis. <i>Neuromuscular Disorders</i> , 2009, 19, 217-219.	0.3	22
39	Severe neonatal non-dystrophic myotonia secondary to a novel mutation of the voltage-gated sodium channel (SCN4A) gene. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 380-383.	0.7	57
40	Dosage Effect of a Dominant CLCN1 Mutation: A Novel Syndrome. <i>Journal of Child Neurology</i> , 2008, 23, 163-166.	0.7	21
41	A Novel Mitochondrial DNA tRNA ^{Leu} (m.4322dupC) Mutation Associated With Idiopathic Dilated Cardiomyopathy. <i>Diagnostic Molecular Pathology</i> , 2007, 16, 238-242.	2.1	12
42	Hypokalaemic periodic paralysis due to the CACNA1S R1239H mutation in a large African family. <i>Neuromuscular Disorders</i> , 2007, 17, 419-422.	0.3	9
43	A large german kindred with cold-aggravated myotonia and a heterozygous A1481D mutation in the SCN4A gene. <i>Muscle and Nerve</i> , 2007, 35, 599-606.	1.0	20
44	Comparative efficacy of repetitive nerve stimulation, exercise, and cold in differentiating myotonic disorders. <i>Muscle and Nerve</i> , 2007, 36, 643-650.	1.0	45
45	Hypokalemic periodic paralysis: A model for a clinical and research approach to a rare disorder. <i>Neurotherapeutics</i> , 2007, 4, 225-232.	2.1	29
46	Gating defects of a novel Na ⁺ channel mutant causing hypokalemic periodic paralysis. <i>Biochemical and Biophysical Research Communications</i> , 2006, 348, 653-661.	1.0	31
47	Cold extends electromyography distinction between ion channel mutations causing myotonia. <i>Annals of Neurology</i> , 2006, 60, 356-365.	2.8	136
48	A1152D mutation of the Na ⁺ channel causes paramyotonia congenita and emphasizes the role of DIII/S4-S5 linker in fast inactivation. <i>Journal of Physiology</i> , 2005, 565, 415-427.	1.3	18
49	In vivo and in vitro functional characterization of Andersen's syndrome mutations. <i>Journal of Physiology</i> , 2005, 565, 731-741.	1.3	34
50	Human skeletal muscle sodium channelopathies. <i>Neurological Sciences</i> , 2005, 26, 194-202.	0.9	81
51	Functional characterization and cold sensitivity of T1313A, a new mutation of the skeletal muscle sodium channel causing paramyotonia congenita in humans. <i>Journal of Physiology</i> , 2004, 554, 635-647.	1.3	46
52	Electromyography guides toward subgroups of mutations in muscle channelopathies. <i>Annals of Neurology</i> , 2004, 56, 650-661.	2.8	265
53	Mitochondrial Damage Associated With Long-Term Antiretroviral Treatment: Associated Alteration or Causal Disorder?. <i>Journal of Acquired Immune Deficiency Syndromes (1999)</i> , 2002, 31, 299-308.	0.9	58
54	Cystic fibrosis transmembrane conductance regulator (CFTR) gene defects in patients with primary sclerosing cholangitis. <i>Journal of Hepatology</i> , 2002, 37, 192-197.	1.8	58

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55	Ageing muscle: clonal expansions of mitochondrial DNA point mutations and deletions cause focal impairment of mitochondrial function. <i>Neuromuscular Disorders</i> , 2002, 12, 484-493.	0.3	180
56	Two large French pedigrees with non syndromic sensorineural deafness and the mitochondrial DNA T7511C mutation: evidence for a modulatory factor. <i>European Journal of Human Genetics</i> , 2002, 10, 851-856.	1.4	50
57	Hypokalaemic periodic paralysis type 2 caused by mutations at codon 672 in the muscle sodium channel gene SCN4A. <i>Brain</i> , 2001, 124, 1091-1099.	3.7	179
58	Functional Mitochondrial Heterogeneity in Heteroplasmic Cells Carrying the Mitochondrial DNA Mutation Associated with the MELAS Syndrome (Mitochondrial Encephalopathy, Lactic Acidosis, and) Tj ETQq0 0 0rgBT /Over 10 Tf		
59	Evaluation of Parental Mitochondrial Inheritance in Neonates Born after Intracytoplasmic Sperm Injection. <i>American Journal of Human Genetics</i> , 1999, 65, 463-473.	2.6	75