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	Electromyography guides toward subgroups of mutations in muscle channelopathies. Annals of Neurology, 2004, 56, 650-661.	2.8	265
2	The non-dystrophic myotonias: molecular pathogenesis, diagnosis and treatment. Brain, 2010, 133, 9-22.	3.7	244
3	Ageing muscle: clonal expansions of mitochondrial DNA point mutations and deletions cause focal impairment of mitochondrial function. Neuromuscular Disorders, 2002, 12, 484-493.	0.3	180
4	Hypokalaemic periodic paralysis type 2 caused by mutations at codon 672 in the muscle sodium channel gene SCN4A. Brain, 2001, 124, 1091-1099.	3.7	179
5	Cold extends electromyography distinction between ion channel mutations causing myotonia. Annals of Neurology, 2006, 60, 356-365.	2.8	136
6	Mutations in CNTNAP1 and ADCY6 are responsible for severe arthrogryposis multiplex congenita with axoglial defects. Human Molecular Genetics, 2014, 23, 2279-2289.	1.4	98
7	Agrin mutations lead to a congenital myasthenic syndrome with distal muscle weakness and atrophy. Brain, 2014, 137, 2429-2443.	3.7	86
8	Human skeletal muscle sodium channelopathies. Neurological Sciences, 2005, 26, 194-202.	0.9	81
9	Evaluation of Parental Mitochondrial Inheritance in Neonates Born after Intracytoplasmic Sperm Injection. American Journal of Human Genetics, 1999, 65, 463-473.	2.6	75
10	Impaired Presynaptic High-Affinity Choline Transporter Causes a Congenital Myasthenic Syndrome with Episodic Apnea. American Journal of Human Genetics, 2016, 99, 753-761.	2.6	68
11	The neuronal endopeptidase ECEL1 is associated with a distinct form of recessive distal arthrogryposis. Human Molecular Genetics, 2013, 22, 1483-1492.	1.4	66
12	Mitochondrial Damage Associated With Long-Term Antiretroviral Treatment: Associated Alteration or Causal Disorder?. Journal of Acquired Immune Deficiency Syndromes (1999), 2002, 31, 299-308.	0.9	58
13	Cystic fibrosis transmembrane conductance regulator (CFTR) gene defects in patients with primary sclerosing cholangitis. Journal of Hepatology, 2002, 37, 192-197.	1.8	58
14	Severe neonatal non-dystrophic myotonia secondary to a novel mutation of the voltage-gated sodium channel (SCN4A) gene. American Journal of Medical Genetics, Part A, 2008, 146A, 380-383.	0.7	57
15	Two large French pedigrees with non syndromic sensorineural deafness and the mitochondrial DNA T7511C mutation: evidence for a modulatory factor. European Journal of Human Genetics, 2002, 10, 851-856.	1.4	50
16	Truncating Mutations of MAGEL2, a Gene within the Prader-Willi Locus, Are Responsible for Severe Arthrogryposis. American Journal of Human Genetics, 2015, 97, 616-620.	2.6	49
17	A recessive Na _v 1.4 mutation underlies congenital myasthenic syndrome with periodic paralysis. Neurology, 2016, 86, 161-169.	1.5	49
18	Episodic weakness due to mitochondrial DNA MT-ATP6/8 mutations. Neurology, 2013, 81, 1810-1818.	1.5	48

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19	Functional characterization and cold sensitivity of T1313A, a new mutation of the skeletal muscle sodium channel causing paramyotonia congenita in humans. Journal of Physiology, 2004, 554, 635-647.	1.3	46
20	Comparative efficacy of repetitive nerve stimulation, exercise, and cold in differentiating myotonic disorders. Muscle and Nerve, 2007, 36, 643-650.	1.0	45
21	Clinical, electrophysiologic, and genetic study of non-dystrophic myotonia in French-Canadians. Neuromuscular Disorders, 2009, 19, 330-334.	0.3	44
22	Physiological and Pathophysiological Insights of Nav1.4 and Nav1.5 Comparison. Frontiers in Pharmacology, 2015, 6, 314.	1.6	40
23	In vivoandin vitrofunctional characterization of Andersen's syndrome mutations. Journal of Physiology, 2005, 565, 731-741.	1.3	34
24	Diagnosis and Outcome of SCN4A-Related Severe Neonatal Episodic Laryngospasm (SNEL): 2 New Cases. Pediatrics, 2013, 132, e784-e787.	1.0	34
25	Diseaseâ€causing mutations C277R and C277Y modify gating of human ClCâ€1 chloride channels in myotonia congenita. Journal of Physiology, 2012, 590, 3449-3464.	1.3	32
26	Gating defects of a novel Na+ channel mutant causing hypokalemic periodic paralysis. Biochemical and Biophysical Research Communications, 2006, 348, 653-661.	1.0	31
27	Hypokalemic periodic paralysis: A model for a clinical and research approach to a rare disorder. Neurotherapeutics, 2007, 4, 225-232.	2.1	29
28	Heterozygous CLCN1 mutations can modulate phenotype in sodium channel myotonia. Neuromuscular Disorders, 2014, 24, 953-959.	0.3	27
29	A National French consensus on gene lists for the diagnosis of myopathies using next-generation sequencing. European Journal of Human Genetics, 2019, 27, 349-352.	1.4	27
30	Mutations in GFPT1-related congenital myasthenic syndromes are associated with synaptic morphological defects and underlie a tubular aggregate myopathy with synaptopathy. Journal of Neurology, 2017, 264, 1791-1803.	1.8	26
31	Pharmacogenetics of myotonic hNav1.4 sodium channel variants situated near the fast inactivation gate. Pharmacological Research, 2019, 141, 224-235.	3.1	25
32	Phenotypic spectrum and genomics of undiagnosed arthrogryposis multiplex congenita. Journal of Medical Genetics, 2022, 59, 559-567.	1.5	25
33	Coldâ€induced disruption of Na⁺ channel slow inactivation underlies paralysis in highly thermosensitive paramyotonia. Journal of Physiology, 2009, 587, 1705-1714.	1.3	24
34	Homozygosity for dominant mutations increases severity of muscle channelopathies. Muscle and Nerve, 2010, 41, 470-477.	1.0	24
35	Glucocorticoids may trigger attacks in several types of periodic paralysis. Neuromuscular Disorders, 2009, 19, 217-219.	0.3	22
36	Clinical, morphological and genetic characterization of Brody disease: an international study of 40 patients. Brain, 2020, 143, 452-466.	3.7	22

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37	Dosage Effect of a Dominant CLCN1 Mutation: A Novel Syndrome. Journal of Child Neurology, 2008, 23, 163-166.	0.7	21
38	Impaired surface membrane insertion of homo- and heterodimeric human muscle chloride channels carrying amino-terminal myotonia-causing mutations. Scientific Reports, 2015, 5, 15382.	1.6	21
39	A large german kindred with cold-aggravated myotonia and a heterozygous A1481D mutation in theSCN4A gene. Muscle and Nerve, 2007, 35, 599-606.	1.0	20
40	Functional Mitochondrial Heterogeneity in Heteroplasmic Cells Carrying the Mitochondrial DNA Mutation Associated with the MELAS Syndrome (Mitochondrial Encephalopathy, Lactic Acidosis, and) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	1.8	18
41	A1152D mutation of the Na+channel causes paramyotonia congenita and emphasizes the role of DIII/S4-S5 linker in fast inactivation. Journal of Physiology, 2005, 565, 415-427.	1.3	18
42	Mechanisms underlying a lifeâ€threatening skeletal muscle Na⁺ channel disorder. Journal of Physiology, 2011, 589, 3115-3124.	1.3	16
43	How chromosomal deletions can unmask recessive mutations? Deletions in 10q11.2 associated with CHAT or SLC18A3 mutations lead to congenital myasthenic syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 151-155.	0.7	16
44	Substitutions of the S4DIV R2 residue (R1451) in NaV1.4 lead to complex forms of paramyotonia congenita and periodic paralyses. Scientific Reports, 2018, 8, 2041.	1.6	14
45	A Novel Mitochondrial DNA tRNAIle (m.4322dupC) Mutation Associated With Idiopathic Dilated Cardiomyopathy. Diagnostic Molecular Pathology, 2007, 16, 238-242.	2.1	12
46	Hypokalaemic periodic paralysis with a charge-retaining substitution in the voltage sensor. Brain Communications, 2020, 2, fcaa103.	1.5	11
47	Hypokalaemic periodic paralysis due to the CACNA1S R1239H mutation in a large African family. Neuromuscular Disorders, 2007, 17, 419-422.	0.3	9
48	Prolonged attacks of weakness with hypokalemia in SCN4Aâ€related paramyotonia congenita. Muscle and Nerve, 2018, 58, E27-E28.	1.0	6
49	Non-dystrophic myotonia Chilean cohort with predominance of the SCN4A Gly1306Glu variant. Neuromuscular Disorders, 2020, 30, 554-561.	0.3	6
50	Focal and abnormally persistent paralysis associated with congenital paramyotonia. BMJ Case Reports, 2014, 2014, bcr2014204430-bcr2014204430.	0.2	6
51	New recessive mutations in SYT2 causing severe presynaptic congenital myasthenic syndromes. Neurology: Genetics, 2020, 6, e534.	0.9	6
52	A204E mutation in Nav1.4 DIS3 exerts gain- and loss-of-function effects that lead to periodic paralysis combining hyper- with hypo-kalaemic signs. Scientific Reports, 2018, 8, 16681.	1.6	5
53	Atypical nuclear abnormalities in a patient with Brody disease. Neuromuscular Disorders, 2015, 25, 773-779.	0.3	4
54	A TOR1AIP1 variant segregating with an early onset limb girdle myastheniaâ€”Support for the role of LAP1 in NMJ function and disease. Neuropathology and Applied Neurobiology, 2022, 48, .	1.8	4

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55	Brody myopathy demonstrates a pseudoâ€increment on repetitive nerve stimulation. Muscle and Nerve, 2020, 61, 491-495.	1.0	2
56	Relapses and remissions of hypokalaemic periodic paralysis and multiple sclerosis in the same patient. Clinical Neurology and Neurosurgery, 2011, 113, 683-685.	0.6	1
57	Syndromes myasthÃ©niques congÃ©nitaux â€" Lâ€™expÃ©rience franÃ§aise. Bulletin De L'Academie Nationale De Medecine, 2014, 198, 257-271.	0.0	1
58	Phenotypical variability and atypical presentations in a French cohort of Andersenâ€“Tawil syndrome. European Journal of Neurology, 2022, 29, 2398-2411.	1.7	1
59	Hypokalaemia and dysmorphia, is there a link?. CKJ: Clinical Kidney Journal, 2009, 2, 222-224.	1.4	0