Stephanie Schorge

List of Publications by Year in Descending Order

72 papers	2,840 citations	29 h-index	52 g-index
82 ext. papers	3,367 ext. citations	8.5 avg, IF	4.83 L-index

#	Paper	IF	Citations
72	Gene Therapy for Neurological Disease: State of the Art and Opportunities for Next-generation Approaches.. *Neuroscience*, **2022**,	3.9	2
71	BICS01 Mediates Reversible Anti-seizure Effects in Brain Slice Models of Epilepsy.. *Frontiers in Neurology*, **2021**, 12, 791608	4.1	
70	Gene therapy restores dopamine transporter expression and ameliorates pathology in iPSC and mouse models of infantile parkinsonism. *Science Translational Medicine*, **2021**, 13,	17.5	7
69	Recent advances in gene therapy for neurodevelopmental disorders with epilepsy. *Journal of Neurochemistry*, **2021**, 157, 229-262	6	12
68	Progressive myoclonus epilepsy KCNC1 variant causes a developmental dendritopathy. *Epilepsia*, **2021**, 62, 1256-1267	6.4	4
67	Translating genetic and functional data into clinical practice: a series of 223 families with myotonia. *Brain*, **2021**,	11.2	1
66	In vivo CRISPRa decreases seizures and rescues cognitive deficits in a rodent model of epilepsy. *Brain*, **2020**, 143, 891-905	11.2	40
65	A systems approach delivers a functional microRNA catalog and expanded targets for seizure suppression in temporal lobe epilepsy. *Proceedings of the National Academy of Sciences of the United States of America*, **2020**, 117, 15977-15988	11.5	19
64	Biological concepts in human sodium channel epilepsies and their relevance in clinical practice. *Epilepsia*, **2020**, 61, 387-399	6.4	35
63	Altered Hippocampal-Prefrontal Neural Dynamics in Mouse Models of Down Syndrome. *Cell Reports*, **2020**, 30, 1152-1163.e4	10.6	8
62	SCN1A variants from bench to bedside-improved clinical prediction from functional characterization. *Human Mutation*, **2020**, 41, 363-374	4.7	14
61	dCas9-Based Scn1a Gene Activation Restores Inhibitory Interneuron Excitability and Attenuates Seizures in Dravet Syndrome Mice. *Molecular Therapy*, **2020**, 28, 235-253	11.7	74
60	Antagonizing Increased Levels at the Chronic Stage of Experimental TLE Reduces Spontaneous Recurrent Seizures. *Journal of Neuroscience*, **2019**, 39, 5064-5079	6.6	18
59	Olanzapine: A potent agonist at the hM4D(Gi) DREADD amenable to clinical translation of chemogenetics. *Science Advances*, **2019**, 5, eaaw1567	14.3	29
58	Epilepsy Gene Therapy Using an Engineered Potassium Channel. *Journal of Neuroscience*, **2019**, 39, 3159-3169	6.6	41
57	Myotonia in a patient with a mutation in an S4 arginine residue associated with hypokalaemic periodic paralysis and a concomitant synonymous CLCN1 mutation. *Scientific Reports*, **2019**, 9, 17560	4.9	9
56	Spider toxin inhibits gating pore currents underlying periodic paralysis. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, 4495-4500	11.5	13

55	Biochemical autoregulatory gene therapy for focal epilepsy. *Nature Medicine*, **2018**, 24, 1324-1329	50.5	34
54	Spared CA1 pyramidal neuron function and hippocampal performance following antisense knockdown of microRNA-134. *Epilepsia*, **2018**, 59, 1518-1526	6.4	12
53	Rare coding variants in genes encoding GABA receptors in genetic generalised epilepsies: an exome-based case-control study. *Lancet Neurology, The*, **2018**, 17, 699-708	24.1	44
52	Semiology, clustering, periodicity and natural history of seizures in an experimental occipital cortical epilepsy model. *DMM Disease Models and Mechanisms*, **2018**, 11,	4.1	8
51	Conservation of alternative splicing in sodium channels reveals evolutionary focus on release from inactivation and structural insights into gating. *Journal of Physiology*, **2017**, 595, 5671-5685	3.9	6
50	Activity Clamp Provides Insights into Paradoxical Effects of the Anti-Seizure Drug Carbamazepine. *Journal of Neuroscience*, **2017**, 37, 5484-5495	6.6	6
49	Potent Anti-seizure Effects of Locked Nucleic Acid Antagomirs Targeting miR-134 in Multiple Mouse and Rat Models of Epilepsy. *Molecular Therapy - Nucleic Acids*, **2017**, 6, 45-56	10.7	43
48	Personalized translational epilepsy research - Novel approaches and future perspectives: Part II: Experimental and translational approaches. *Epilepsy and Behavior*, **2017**, 76, 7-12	3.2	11
47	Personalized translational epilepsy research - Novel approaches and future perspectives: Part I: Clinical and network analysis approaches. *Epilepsy and Behavior*, **2017**, 76, 13-18	3.2	18
46	MicroRNAs in epilepsy: pathophysiology and clinical utility. *Lancet Neurology, The*, **2016**, 15, 1368-1376	24.1	132
45	Lentiviral expression of GAD67 and CCK promoter-driven opsins to target interneurons in vitro and in vivo. *Journal of Gene Medicine*, **2016**, 18, 27-37	3.5	1
44	Changing channels in pain and epilepsy: Exploiting ion channel gene therapy for disorders of neuronal hyperexcitability. *FEBS Letters*, **2015**, 589, 1620-34	3.8	16
43	Mutations in SLC12A5 in epilepsy of infancy with migrating focal seizures. *Nature Communications*, **2015**, 6, 8038	17.4	104
42	Tracks through the genome to physiological events. *Experimental Physiology*, **2015**, 100, 1429-40	2.4	2
41	Chemical-genetic attenuation of focal neocortical seizures. *Nature Communications*, **2014**, 5, 3847	17.4	98
40	A common SCN1A splice-site polymorphism modifies the effect of carbamazepine on cortical excitability--a pharmacogenetic transcranial magnetic stimulation study. *Epilepsia*, **2014**, 55, 362-9	6.4	47
39	Gene therapy in epilepsy-is it time for clinical trials?. *Nature Reviews Neurology*, **2014**, 10, 300-4	15	52
38	From treatment to cure: stopping seizures, preventing seizures, and reducing brain propensity to seize. *International Review of Neurobiology*, **2014**, 114, 279-99	4.4	2

37	Insights from cerebellar transcriptomic analysis into the pathogenesis of ataxia. *JAMA Neurology*, **2014**, 71, 831-9	17.2	47
36	Ataxia and CACNA1A: Episodic or Progressive? **2014**, 27-45		
35	Clinical, genetic, neurophysiological and functional study of new mutations in episodic ataxia type 1. *Journal of Neurology, Neurosurgery and Psychiatry*, **2013**, 84, 1107-12	5.5	36
34	Nongenomic actions of progesterone and 17β-estradiol on the chloride conductance of skeletal muscle. *Muscle and Nerve*, **2013**, 48, 589-91	3.4	5
33	Gene therapy in status epilepticus. *Epilepsia*, **2013**, 54 Suppl 6, 43-5	6.4	9
32	Tackling obstacles for gene therapy targeting neurons: disrupting perineural nets with hyaluronidase improves transduction. *PLoS ONE*, **2013**, 8, e53269	3.7	7
31	Optogenetic and potassium channel gene therapy in a rodent model of focal neocortical epilepsy. *Science Translational Medicine*, **2012**, 4, 161ra152	17.5	179
30	Splice variants of Na(V)1.7 sodium channels have distinct β subunit-dependent biophysical properties. *PLoS ONE*, **2012**, 7, e41750	3.7	15
29	The P/Q channel in human disease: untangling the genetics and physiology. *Environmental Sciences Europe*, **2012**, 1, 311-320	5	6
28	New immunohistochemical method for improved myotonia and chloride channel mutation diagnostics. *Neurology*, **2012**, 79, 2194-200	6.5	8
27	A new explanation for recessive myotonia congenita: exon deletions and duplications in CLCN1. *Neurology*, **2012**, 78, 1953-8	6.5	26
26	Alternative splicing modulates inactivation of type 1 voltage-gated sodium channels by toggling an amino acid in the first S3-S4 linker. *Journal of Biological Chemistry*, **2011**, 286, 36700-8	5.4	32
25	Genetic and functional characterisation of the P/Q calcium channel in episodic ataxia with epilepsy. *Journal of Physiology*, **2010**, 588, 1905-13	3.9	70
24	Voltage sensor charge loss accounts for most cases of hypokalemic periodic paralysis. *Neurology*, **2010**, 74, 269; author reply 169-70	6.5	6
23	Nongenetic factors influence severity of episodic ataxia type 1 in monozygotic twins. *Neurology*, **2010**, 75, 367-72	6.5	32
22	PAW31 Clinical and genetic spectrum of the episodic ataxias: the UK perspective. *Journal of Neurology, Neurosurgery and Psychiatry*, **2010**, 81, e32-e32	5.5	
21	Human ataxias: a genetic dissection of inositol triphosphate receptor (ITPR1)-dependent signaling. *Trends in Neurosciences*, **2010**, 33, 211-9	13.3	76
20	Sodium channel mutations and epilepsy: association and causation. *Experimental Neurology*, **2010**, 226, 8-10	5.7	2

19	Dysfunction of the Ca(V)2.1 calcium channel in cerebellar ataxias. *F1000 Biology Reports*, **2010**, 2,		6
18	Large scale calcium channel gene rearrangements in episodic ataxia and hemiplegic migraine: implications for diagnostic testing. *Journal of Medical Genetics*, **2009**, 46, 786-91	5.8	48
17	Sodium channelopathy of peripheral nerve: tightening the genotype-phenotype relationship. *Brain*, **2009**, 132, 1690-2	11.2	2
16	Voltage sensor charge loss accounts for most cases of hypokalemic periodic paralysis. *Neurology*, **2009**, 72, 1544-7	6.5	133
15	A patient with episodic ataxia and paramyotonia congenita due to mutations in KCNA1 and SCN4A. *Neurology*, **2009**, 73, 993-5	6.5	8
14	A self-activating intrinsic brake on bursting in CA3 neurons. *Journal of Physiology*, **2009**, 587, 1143-4	3.9	2
13	Episodic ataxia type 1 mutations differentially affect neuronal excitability and transmitter release. *DMM Disease Models and Mechanisms*, **2009**, 2, 612-9	4.1	27
12	Non-genomic effects of sex hormones on CLC-1 may contribute to gender differences in myotonia congenita. *Neuromuscular Disorders*, **2008**, 18, 869-72	2.9	25
11	Episodic ataxia type 1: a neuronal potassium channelopathy. *Neurotherapeutics*, **2007**, 4, 258-66	6.4	84
10	Chloride channel myotonia: exon 8 hot-spot for dominant-negative interactions. *Brain*, **2007**, 130, 3265-74	11.2	93
9	Epileptogenesis is associated with enhanced glutamatergic transmission in the perforant path. *Journal of Neurophysiology*, **2006**, 95, 1213-20	3.2	46
8	Antagonism of ATP responses at P2X receptor subtypes by the pH indicator dye, Phenol red. *British Journal of Pharmacology*, **2005**, 145, 313-22	8.6	22
7	Maximum likelihood fitting of single channel NMDA activity with a mechanism composed of independent dimers of subunits. *Journal of Physiology*, **2005**, 569, 395-418	3.9	79
6	Genetic predictors of the maximum doses patients receive during clinical use of the anti-epileptic drugs carbamazepine and phenytoin. *Proceedings of the National Academy of Sciences of the United States of America*, **2005**, 102, 5507-12	11.5	278
5	Studies of NMDA receptor function and stoichiometry with truncated and tandem subunits. *Journal of Neuroscience*, **2003**, 23, 1151-8	6.6	194
4	Alternative splicing of a short cassette exon in alpha1B generates functionally distinct N-type calcium channels in central and peripheral neurons. *Journal of Neuroscience*, **1999**, 19, 5322-31	6.6	79
3	Calcium channel activation stabilizes a neuronal calcium channel mRNA. *Nature Neuroscience*, **1999**, 2, 785-90	25.5	25
2	Identification of functionally distinct isoforms of the N-type Ca2+ channel in rat sympathetic ganglia and brain. *Neuron*, **1997**, 18, 153-66	13.9	163

1 Olanzapine: a full and potent agonist at the hM4D(Gi) DREADD amenable to clinical translation of chemogenetics 1