StÃ©phanie Baulac

List of Publications by Year
in descending order

95 papers	8,662 citations	50244 46 h-index	46771 89 g-index
104 all docs	104 docs citations	104 times ranked	8956 citing authors

#	Article	IF	Citations
1	Mutations of SCN1A, encoding a neuronal sodium channel, in two families with GEFS+2. Nature Genetics, 2000, 24, 343-345.	9.4	910
2	First genetic evidence of GABAA receptor dysfunction in epilepsy: a mutation in the Î³2-subunit gene. Nature Genetics, 2001, 28, 46-48.	9.4	701
3	De Novo Mutations in Synaptic Transmission Genes Including DNM1 Cause Epileptic Encephalopathies. American Journal of Human Genetics, 2014, 95, 360-370.	2.6	388
4	Spectrum of SCN1A gene mutations associated with Dravet syndrome: analysis of 333 patients. Journal of Medical Genetics, 2008, 46, 183-191.	1.5	302
5	The phenotypic spectrum of SCN8A encephalopathy. Neurology, 2015, 84, 480-489.	1.5	246
6	Title is missing!. Nature Genetics, 2001, 28, 46-48.	9.4	241
7	Familial focal epilepsy with focal cortical dysplasia due to <scp>DEPDC</scp>5 mutations. Annals of Neurology, 2015, 77, 675-683.	2.8	231
8	De novo loss- or gain-of-function mutations in KCNA2 cause epileptic encephalopathy. Nature Genetics, 2015, 47, 393-399.	9.4	224
9	Mutations of DEPDC5 cause autosomal dominant focal epilepsies. Nature Genetics, 2013, 45, 552-555.	9.4	215
10	Dissecting the genetic basis of focal cortical dysplasia: a large cohort study. Acta Neuropathologica, 2019, 138, 885-900.	3.9	205
11	De novo mutations in HCN1 cause early infantile epileptic encephalopathy. Nature Genetics, 2014, 46, 640-645.	9.4	192
12	De Novo Loss-of-Function Mutations in CHD2 Cause a Fever-Sensitive Myoclonic Epileptic Encephalopathy Sharing Features with Dravet Syndrome. American Journal of Human Genetics, 2013, 93, 967-975.	2.6	188
13	A Second Locus for Familial Generalized Epilepsy with Febrile Seizures Plus Maps to Chromosome 2q21-q33. American Journal of Human Genetics, 1999, 65, 1078-1085.	2.6	182
14	Fever, genes, and epilepsy. Lancet Neurology, The, 2004, 3, 421-430.	4.9	179
15	Mutations in STX1B, encoding a presynaptic protein, cause fever-associated epilepsy syndromes. Nature Genetics, 2014, 46, 1327-1332.	9.4	178
16	Second-hit mosaic mutation in mTORC1 repressor DEPDC5 causes focal cortical dysplasiaâ€“associated epilepsy. Journal of Clinical Investigation, 2018, 128, 2452-2458.	3.9	171
17	Review: Mechanistic target of rapamycin (mTOR) pathway, focal cortical dysplasia and epilepsy. Neuropathology and Applied Neurobiology, 2018, 44, 6-17.	1.8	145
18	The landscape of epilepsy-related GATOR1 variants. Genetics in Medicine, 2019, 21, 398-408.	1.1	137

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19	Genome-wide association analysis of genetic generalized epilepsies implicates susceptibility loci at 1q43, 2p16.1, 2q22.3 and 17q21.32. Human Molecular Genetics, 2012, 21, 5359-5372.	1.4	134
20	Involvement of <scp>GATOR</scp> complex genes in familial focal epilepsies and focal cortical dysplasia. Epilepsia, 2016, 57, 994-1003.	2.6	133
21	Mechanisms for variable expressivity of inherited SCN1A mutations causing Dravet syndrome. Journal of Medical Genetics, 2010, 47, 404-410.	1.5	130
22	DEPDC5 mutations in families presenting as autosomal dominant nocturnal frontal lobe epilepsy. Neurology, 2014, 82, 2101-2106.	1.5	126
23	Germline and somatic mutations in the MTOR gene in focal cortical dysplasia and epilepsy. Neurology: Genetics, 2016, 2, e118.	0.9	125
24	Zebrafish lacking Alzheimer presenilin enhancerâ€ƒ2 (Pen-2) demonstrate excessive p53-dependent apoptosis and neuronal loss. Journal of Neurochemistry, 2006, 96, 1423-1440.	2.1	120
25	Electroclinical characterization of epileptic seizures in leucine-rich, glioma-inactivated 1-deficient mice. Brain, 2010, 133, 2749-2762.	3.7	118
26	Mutations and deletions in PCDH19 account for various familial or isolated epilepsies in females. Human Mutation, 2011, 32, E1959-E1975.	1.1	109
27	Functional Î³-secretase complex assembly in Golgi/trans-Golgi network: interactions among presenilin, nicastrin, Aph1, Pen-2, and Î³-secretase substrates. Neurobiology of Disease, 2003, 14, 194-204.	2.1	99
28	Role of the phosphoinositide phosphatase FIG4 gene in familial epilepsy with polymicrogyria. Neurology, 2014, 82, 1068-1075.	1.5	97
29	Parental mosaicism can cause recurrent transmission ofSCN1A mutations associated with severe myoclonic epilepsy of infancy. Human Mutation, 2006, 27, 389-389.	1.1	93
30	Evidence for digenic inheritance in a family with both febrile convulsions and temporal lobe epilepsy implicating chromosomes 18qter and 1q25-q31. Annals of Neurology, 2001, 49, 786-792.	2.8	92
31	The <scp>ILAE</scp> consensus classification of focal cortical dysplasia: An update proposed by an ad hoc task force of the <scp>ILAE</scp> diagnostic methods commission. Epilepsia, 2022, 63, 1899-1919.	2.6	88
32	GLUT1 mutations are a rare cause of familial idiopathic generalized epilepsy. Neurology, 2012, 78, 557-562.	1.5	86
33	Monogenic idiopathic epilepsies. Lancet Neurology, The, 2004, 3, 209-218.	4.9	78
34	Depdc5 knockout rat: A novel model of mTORopathy. Neurobiology of Disease, 2016, 89, 180-189.	2.1	78
35	A Recurrent Missense Variant in AP2M1 Impairs Clathrin-Mediated Endocytosis and Causes Developmental and Epileptic Encephalopathy. American Journal of Human Genetics, 2019, 104, 1060-1072.	2.6	78
36	Dominant partial epilepsies: A clinical, electrophysiological and genetic study of 19 European families. Brain, 2000, 123, 1247-1262.	3.7	76

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37	GABA_AReceptor Î²2 Subunit Mutations Linked to Human Epileptic Syndromes Differentially Affect Phasic and Tonic Inhibition. Journal of Neuroscience, 2007, 27, 14108-14116.	1.7	76
38	LGI1 tunes intrinsic excitability by regulating the density of axonal Kv1 channels. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, 7719-7724.	3.3	74
39	Distinctive binding properties of human monoclonal LGI1 autoantibodies determine pathogenic mechanisms. Brain, 2020, 143, 1731-1745.	3.7	74
40	Rare coding variants in genes encoding GABAA receptors in genetic generalised epilepsies: an exome-based case-control study. Lancet Neurology, The, 2018, 17, 699-708.	4.9	67
41	mTOR signaling pathway genes in focal epilepsies. Progress in Brain Research, 2016, 226, 61-79.	0.9	65
42	Frequent SLC35A2 brain mosaicism in mild malformation of cortical development with oligodendroglial hyperplasia in epilepsy (MOGHE). Acta Neuropathologica Communications, 2021, 9, 3.	2.4	62
43	Treatment Responsiveness in KCNT1-Related Epilepsy. Neurotherapeutics, 2019, 16, 848-857.	2.1	60
44	Increased DJ-1 expression under oxidative stress and in Alzheimer's disease brains. Molecular Neurodegeneration, 2009, 4, 12.	4.4	59
45	Dimerization of Parkinson's disease-causing DJ-1 and formation of high molecular weight complexes in human brain. Molecular and Cellular Neurosciences, 2004, 27, 236-246.	1.0	58
46	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. American Journal of Human Genetics, 2017, 100, 676-688.	2.6	54
47	Toward a better definition of focal cortical dysplasia: An iterative histopathological and genetic agreement trial. Epilepsia, 2021, 62, 1416-1428.	2.6	54
48	Recessive loss-of-function mutations in AP4S1 cause mild fever-sensitive seizures, developmental delay and spastic paraplegia through loss of AP-4 complex assembly. Human Molecular Genetics, 2015, 24, 2218-2227.	1.4	53
49	Neocortical development and epilepsy: insights from focal cortical dysplasia and brain tumours. Lancet Neurology, The, 2021, 20, 943-955.	4.9	47
50	Two Novel Epilepsy-Linked Mutations Leading to a Loss of Function of LGI1. Archives of Neurology, 2007, 64, 217.	4.9	44
51	Glutamatergic neuron-targeted loss of LGI1 epilepsy gene results in seizures. Brain, 2014, 137, 2984-2996.	3.7	43
52	Biallelic VARS variants cause developmental encephalopathy with microcephaly that is recapitulated in vars knockout zebrafish. Nature Communications, 2019, 10, 708.	5.8	40
53	New locus for febrile seizures with absence epilepsy on 3p and a possible modifier gene on 18p. Neurology, 2007, 68, 1374-1381.	1.5	39
54	LGI1 acts presynaptically to regulate excitatory synaptic transmission during early postnatal development. Scientific Reports, 2016, 6, 21769.	1.6	38

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55	Detection of Brain Somatic Mutations in <scp>Cerebrospinal Fluid</scp> from Refractory Epilepsy Patients. Annals of Neurology, 2021, 89, 1248-1252.	2.8	37
56	A Novel Locus for Generalized Epilepsy With Febrile Seizures Plus in French Families. Archives of Neurology, 2008, 65, 943-51.	4.9	36
57	A rat model for LGI1-related epilepsies. Human Molecular Genetics, 2012, 21, 3546-3557.	1.4	36
58	KCNT1-related epilepsies and epileptic encephalopathies: phenotypic and mutational spectrum. Brain, 2021, 144, 3635-3650.	3.7	34
59	New Analysis Workflow for MALDI Imaging Mass Spectrometry: Application to the Discovery and Identification of Potential Markers of Childhood Absence Epilepsy. Journal of Proteome Research, 2012, 11, 5453-5463.	1.8	32
60	Genomeâ€wide linkage metaâ€analysis identifies susceptibility loci at 2q34 and 13q31.3 for genetic generalized epilepsies. Epilepsia, 2012, 53, 308-318.	2.6	32
61	Depdc5 knockdown causes <scp>mTOR</scp>â€dependent motor hyperactivity in zebrafish. Annals of Clinical and Translational Neurology, 2018, 5, 510-523.	1.7	32
62	Kcna1-mutant rats dominantly display myokymia, neuromyotonia and spontaneous epileptic seizures. Brain Research, 2012, 1435, 154-166.	1.1	31
63	Novel GABRG2 mutations cause familial febrile seizures. Neurology: Genetics, 2015, 1, e35.	0.9	29
64	Genetics advances in autosomal dominant focal epilepsies. Progress in Brain Research, 2014, 213, 123-139.	0.9	28
65	Mild malformations of cortical development in sleepâ€related hypermotor epilepsy due to KCNT1 mutations. Annals of Clinical and Translational Neurology, 2019, 6, 386-391.	1.7	25
66	Absence of mutations in the LGI1 receptor ADAM22 gene in autosomal dominant lateral temporal epilepsy. Epilepsy Research, 2007, 76, 41-48.	0.8	24
67	Gradient of brain mosaic RHEB variants causes a continuum of cortical dysplasia. Annals of Clinical and Translational Neurology, 2021, 8, 485-490.	1.7	24
68	Refinement of the 2p11.1-q12.2 locus responsible for cortical tremor associated with epilepsy and exclusion of candidate genes. Neurogenetics, 2008, 9, 69-71.	0.7	22
69	Epilepsy GeneLGI1Regulates Postnatal Developmental Remodeling of Retinogeniculate Synapses. Journal of Neuroscience, 2012, 32, 903-910.	1.7	22
70	Genetic models of focal epilepsies. Journal of Neuroscience Methods, 2016, 260, 132-143.	1.3	22
71	Acute knockdown of Depdc5 leads to synaptic defects in mTOR-related epileptogenesis. Neurobiology of Disease, 2020, 139, 104822.	2.1	22
72	Lafora progressive myoclonus epilepsy: NHLRC1 mutations affect glycogen metabolism. Journal of Molecular Medicine, 2011, 89, 915-925.	1.7	20

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73	A novel three base-pair LGI1 deletion leading to loss of function in a family with autosomal dominant lateral temporal epilepsy and migraine-like episodes. Epilepsy Research, 2009, 85, 118-122.	0.8	19
74	Advances on the Genetics of Mendelian Idiopathic Epilepsies. Clinics in Laboratory Medicine, 2010, 30, 911-929.	0.7	18
75	Bi-allelic GAD1 variants cause a neonatal onset syndromic developmental and epileptic encephalopathy. Brain, 2020, 143, 1447-1461.	3.7	18
76	Advances on the Genetics of Mendelian Idiopathic Epilepsies. Neurologic Clinics, 2009, 27, 1041-1061.	0.8	15
77	Seizure semiology in autosomal dominant epilepsy with auditory features, due to novel LGI1 mutations. Epilepsy Research, 2013, 107, 311-317.	0.8	15
78	Cortical Dysplasia and the mTOR Pathway: How the Study of Human Brain Tissue Has Led to Insights into Epileptogenesis. International Journal of Molecular Sciences, 2022, 23, 1344.	1.8	14
79	Molecular diagnostics in drugâ€resistant focal epilepsy define new disease entities. Brain Pathology, 2021, 31, e12963.	2.1	13
80	The Nogo Receptor Ligand LGI1 Regulates Synapse Number and Synaptic Activity in Hippocampal and Cortical Neurons. ENeuro, 2018, 5, ENEURO.0185-18.2018.	0.9	13
81	Hyperactive behavior in a family with autosomal dominant lateral temporal lobe epilepsy caused by a mutation in the LGI1/epitempin gene. Epilepsy and Behavior, 2013, 28, 41-46.	0.9	12
82	Cardiac Investigations in Sudden Unexpected Death in <scp>DEPDC5</scp>â€Related Epilepsy. Annals of Neurology, 2022, 91, 101-116.	2.8	11
83	A locus for bilateral occipital polymicrogyria maps to chromosome 6q16â€“q22. Neurogenetics, 2009, 10, 35-42.	0.7	10
84	Clinical and genetic study of Tunisian families with genetic generalized epilepsy: contribution of CACNA1H and MAST4 genes. Neurogenetics, 2018, 19, 165-178.	0.7	10
85	Defective lipid signalling caused by mutations inPIK3C2Bunderlies focal epilepsy. Brain, 2022, 145, 2313-2331.	3.7	10
86	mTOR pathway in familial focal epilepsies. Oncotarget, 2017, 8, 5674-5675.	0.8	8
87	Association of ultraâ€rare coding variants with genetic generalized epilepsy: A caseâ€“control whole exome sequencing study. Epilepsia, 2022, 63, 723-735.	2.6	8
88	Involvement of ADGRV1 Gene in Familial Forms of Genetic Generalized Epilepsy. Frontiers in Neurology, 2021, 12, 738272.	1.1	7
89	Genes in infantile epileptic encephalopathies. Epilepsia, 2010, 51, 69-69.	2.6	5
90	Genetics of inherited human epilepsies. Dialogues in Clinical Neuroscience, 2001, 3, 47-57.	1.8	3

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91	Juvenile myoclonic epilepsy phenotype in a family with Unverrichtâ€Lundborg disease. Epileptic Disorders, 2019, 21, 359-365.	0.7	3
92	Reply to â€œImproving Specificity of <scp>CSF</scp> Liquid Biopsy for Genetic Testingâ€• Annals of Neurology, 2021, 90, 694-695.	2.8	2
93	Ã‰pilepsies, convulsions fÃ©briles et canaux ioniques : le dÃ©but d'une longue histoire.. Medecine/Sciences, 2001, 17, 999.	0.0	2
94	Choking Fits During Sleep Related to Epilepsy. American Journal of Medicine, 2016, 129, e137-e138.	0.6	1
95	Canalopathies Ã©pileptiques. Epilepsies, 2010, 22, 226-229.	0.0	0