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	Cardiac Investigations in Sudden Unexpected Death in <scp>DEPDC5</scp>â€Related Epilepsy. Annals of Neurology, 2022, 91, 101-116.	2.8	11
2	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
3	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
4	Defective lipid signalling caused by mutations inPIK3C2Bunderlies focal epilepsy. Brain, 2022, 145, 2313-2331.	3.7	10
5	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
6	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
7	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
8	Detection of Brain Somatic Mutations in <scp>Cerebrospinal Fluid</scp> from Refractory Epilepsy Patients. Annals of Neurology, 2021, 89, 1248-1252.	2.8	37
9	Toward a better definition of focal cortical dysplasia: An iterative histopathological and genetic agreement trial. Epilepsia, 2021, 62, 1416-1428.	2.6	54
10	KCNT1-related epilepsies and epileptic encephalopathies: phenotypic and mutational spectrum. Brain, 2021, 144, 3635-3650.	3.7	34
11	Molecular diagnostics in drugâ€resistant focal epilepsy define new disease entities. Brain Pathology, 2021, 31, e12963.	2.1	13
12	Reply to â€œImproving Specificity of <scp>CSF</scp> Liquid Biopsy for Genetic Testingâ€• Annals of Neurology, 2021, 90, 694-695.	2.8	2
13	Involvement of ADGRV1 Gene in Familial Forms of Genetic Generalized Epilepsy. Frontiers in Neurology, 2021, 12, 738272.	1.1	7
14	Neocortical development and epilepsy: insights from focal cortical dysplasia and brain tumours. Lancet Neurology, The, 2021, 20, 943-955.	4.9	47
15	Distinctive binding properties of human monoclonal LGI1 autoantibodies determine pathogenic mechanisms. Brain, 2020, 143, 1731-1745.	3.7	74
16	Acute knockdown of Depdc5 leads to synaptic defects in mTOR-related epileptogenesis. Neurobiology of Disease, 2020, 139, 104822.	2.1	22
17	Bi-allelic GAD1 variants cause a neonatal onset syndromic developmental and epileptic encephalopathy. Brain, 2020, 143, 1447-1461.	3.7	18
18	The landscape of epilepsy-related GATOR1 variants. Genetics in Medicine, 2019, 21, 398-408.	1.1	137

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19	Dissecting the genetic basis of focal cortical dysplasia: a large cohort study. Acta Neuropathologica, 2019, 138, 885-900.	3.9	205
20	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
21	Treatment Responsiveness in KCNT1-Related Epilepsy. Neurotherapeutics, 2019, 16, 848-857.	2.1	60
22	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
23	Biallelic VARS variants cause developmental encephalopathy with microcephaly that is recapitulated in vars knockout zebrafish. Nature Communications, 2019, 10, 708.	5.8	40
24	Juvenile myoclonic epilepsy phenotype in a family with Unverrichtâ€Lundborg disease. Epileptic Disorders, 2019, 21, 359-365.	0.7	3
25	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
26	Review: Mechanistic target of rapamycin (mTOR) pathway, focal cortical dysplasia and epilepsy. Neuropathology and Applied Neurobiology, 2018, 44, 6-17.	1.8	145
27	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
28	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
29	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
30	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
31	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
32	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
33	mTOR pathway in familial focal epilepsies. Oncotarget, 2017, 8, 5674-5675.	0.8	8
34	Involvement of <scp>GATOR</scp> complex genes in familial focal epilepsies and focal cortical dysplasia. Epilepsia, 2016, 57, 994-1003.	2.6	133
35	LGI1 acts presynaptically to regulate excitatory synaptic transmission during early postnatal development. Scientific Reports, 2016, 6, 21769.	1.6	38
36	Choking Fits During Sleep Related to Epilepsy. American Journal of Medicine, 2016, 129, e137-e138.	0.6	1

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37	mTOR signaling pathway genes in focal epilepsies. Progress in Brain Research, 2016, 226, 61-79.	0.9	65
38	Germline and somatic mutations in the MTOR gene in focal cortical dysplasia and epilepsy. Neurology: Genetics, 2016, 2, e118.	0.9	125
39	Depdc5 knockout rat: A novel model of mTORopathy. Neurobiology of Disease, 2016, 89, 180-189.	2.1	78
40	Genetic models of focal epilepsies. Journal of Neuroscience Methods, 2016, 260, 132-143.	1.3	22
41	Novel GABRG2 mutations cause familial febrile seizures. Neurology: Genetics, 2015, 1, e35.	0.9	29
42	The phenotypic spectrum of SCN8A encephalopathy. Neurology, 2015, 84, 480-489.	1.5	246
43	Familial focal epilepsy with focal cortical dysplasia due to <scp>DEPDC</scp>5 mutations. Annals of Neurology, 2015, 77, 675-683.	2.8	231
44	De novo loss- or gain-of-function mutations in KCNA2 cause epileptic encephalopathy. Nature Genetics, 2015, 47, 393-399.	9.4	224
45	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
46	Glutamatergic neuron-targeted loss of LGI1 epilepsy gene results in seizures. Brain, 2014, 137, 2984-2996.	3.7	43
47	DEPDC5 mutations in families presenting as autosomal dominant nocturnal frontal lobe epilepsy. Neurology, 2014, 82, 2101-2106.	1.5	126
48	Role of the phosphoinositide phosphatase FIG4 gene in familial epilepsy with polymicrogyria. Neurology, 2014, 82, 1068-1075.	1.5	97
49	De novo mutations in HCN1 cause early infantile epileptic encephalopathy. Nature Genetics, 2014, 46, 640-645.	9.4	192
50	Mutations in STX1B, encoding a presynaptic protein, cause fever-associated epilepsy syndromes. Nature Genetics, 2014, 46, 1327-1332.	9.4	178
51	Genetics advances in autosomal dominant focal epilepsies. Progress in Brain Research, 2014, 213, 123-139.	0.9	28
52	De Novo Mutations in Synaptic Transmission Genes Including DNM1 Cause Epileptic Encephalopathies. American Journal of Human Genetics, 2014, 95, 360-370.	2.6	388
53	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
54	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

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55	Seizure semiology in autosomal dominant epilepsy with auditory features, due to novel LGI1 mutations. Epilepsy Research, 2013, 107, 311-317.	0.8	15
56	Mutations of DEPDC5 cause autosomal dominant focal epilepsies. Nature Genetics, 2013, 45, 552-555.	9.4	215
57	Epilepsy GeneLGI1Regulates Postnatal Developmental Remodeling of Retinogeniculate Synapses. Journal of Neuroscience, 2012, 32, 903-910.	1.7	22
58	A rat model for LGI1-related epilepsies. Human Molecular Genetics, 2012, 21, 3546-3557.	1.4	36
59	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
60	GLUT1 mutations are a rare cause of familial idiopathic generalized epilepsy. Neurology, 2012, 78, 557-562.	1.5	86
61	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
62	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
63	Kcna1-mutant rats dominantly display myokymia, neuromyotonia and spontaneous epileptic seizures. Brain Research, 2012, 1435, 154-166.	1.1	31
64	Lafora progressive myoclonus epilepsy: NHLRC1 mutations affect glycogen metabolism. Journal of Molecular Medicine, 2011, 89, 915-925.	1.7	20
65	Mutations and deletions in PCDH19 account for various familial or isolated epilepsies in females. Human Mutation, 2011, 32, E1959-E1975.	1.1	109
66	Genes in infantile epileptic encephalopathies. Epilepsia, 2010, 51, 69-69.	2.6	5
67	Canalopathies Ã©pileptiques. Epilepsies, 2010, 22, 226-229.	0.0	0
68	Electroclinical characterization of epileptic seizures in leucine-rich, glioma-inactivated 1-deficient mice. Brain, 2010, 133, 2749-2762.	3.7	118
69	Advances on the Genetics of Mendelian Idiopathic Epilepsies. Clinics in Laboratory Medicine, 2010, 30, 911-929.	0.7	18
70	Mechanisms for variable expressivity of inherited SCN1A mutations causing Dravet syndrome. Journal of Medical Genetics, 2010, 47, 404-410.	1.5	130
71	A locus for bilateral occipital polymicrogyria maps to chromosome 6q16â€“q22. Neurogenetics, 2009, 10, 35-42.	0.7	10
72	Increased DJ-1 expression under oxidative stress and in Alzheimer's disease brains. Molecular Neurodegeneration, 2009, 4, 12.	4.4	59

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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. Neurologic Clinics, 2009, 27, 1041-1061.	0.8	15
75	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
76	Spectrum of SCN1A gene mutations associated with Dravet syndrome: analysis of 333 patients. Journal of Medical Genetics, 2008, 46, 183-191.	1.5	302
77	A Novel Locus for Generalized Epilepsy With Febrile Seizures Plus in French Families. Archives of Neurology, 2008, 65, 943-51.	4.9	36
78	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
79	Two Novel Epilepsy-Linked Mutations Leading to a Loss of Function of LGI1. Archives of Neurology, 2007, 64, 217.	4.9	44
80	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
81	Absence of mutations in the LGI1 receptor ADAM22 gene in autosomal dominant lateral temporal epilepsy. Epilepsy Research, 2007, 76, 41-48.	0.8	24
82	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
83	Parental mosaicism can cause recurrent transmission ofSCN1A mutations associated with severe myoclonic epilepsy of infancy. Human Mutation, 2006, 27, 389-389.	1.1	93
84	Monogenic idiopathic epilepsies. Lancet Neurology, The, 2004, 3, 209-218.	4.9	78
85	Fever, genes, and epilepsy. Lancet Neurology, The, 2004, 3, 421-430.	4.9	179
86	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
87	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
88	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
89	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
90	Title is missing!. Nature Genetics, 2001, 28, 46-48.	9.4	241

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91	Genetics of inherited human epilepsies. Dialogues in Clinical Neuroscience, 2001, 3, 47-57.	1.8	3
92	Ã‰pilepsies, convulsions fÃ©briles et canaux ioniques : le dÃ©but d'une longue histoire.. Medecine/Sciences, 2001, 17, 999.	0.0	2
93	Mutations of SCN1A, encoding a neuronal sodium channel, in two families with GEFS+2. Nature Genetics, 2000, 24, 343-345.	9.4	910
94	Dominant partial epilepsies: A clinical, electrophysiological and genetic study of 19 European families. Brain, 2000, 123, 1247-1262.	3.7	76
95	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