Sarah von Spiczak

List of Publications by Year
in descending order

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24 papers	3,254 citations	471509 17 h-index	610901 24 g-index
24 all docs	24 docs citations	24 times ranked	5466 citing authors

#	Article	IF	Citations
1	Iterative phenotyping of 15q11.2, 15q13.3 and 16p13.11 microdeletion carriers in pediatric epilepsies. Epilepsy Research, 2014, 108, 109-116.	1.6	21
2	Atypical Vitamin B₆Deficiency. Journal of Child Neurology, 2014, 29, 704-707.	1.4	16
3	GABRA1 and STXBP1 : Novel genetic causes of Dravet syndrome. Neurology, 2014, 82, 1245-1253.	1.1	229
4	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.	6.2	188
5	Mutations in GRIN2A cause idiopathic focal epilepsy with rolandic spikes. Nature Genetics, 2013, 45, 1067-1072.	21.4	391
6	The role of SLC2A1 in early onset and childhood absence epilepsies. Epilepsy Research, 2013, 105, 229-233.	1.6	13
7	Myoclonic astatic epilepsy (Doose syndrome) â€“ A lamotrigine responsive epilepsy?. European Journal of Paediatric Neurology, 2013, 17, 29-35.	1.6	18
8	Variability of <scp>EEG</scp>â€f<scp>MRI</scp> findings in patients with <scp>SCN1A</scp>â€positive <scp>D</scp>ravet syndrome. Epilepsia, 2013, 54, 918-926.	5.1	26
9	Reduction of seizure frequency after epilepsy surgery in a patient with <scp>STXBP1</scp> encephalopathy and clinical description of six novel mutation carriers. Epilepsia, 2013, 54, e74-80.	5.1	59
10	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.	2.9	134
11	A retrospective study of the relation between vaccination and occurrence of seizures in Dravet syndrome. Epilepsia, 2011, 52, 175-178.	5.1	43
12	A retrospective populationâ€based study on seizures related to childhood vaccination. Epilepsia, 2011, 52, 1506-1512.	5.1	17
13	Comprehensive analysis of candidate genes for photosensitivity using a complementary bioinformatic and experimental approach. Epilepsia, 2011, 52, e143-e147.	5.1	8
14	Absence seizures with intellectual disability as a phenotype of the 15q13.3 microdeletion syndrome. Epilepsia, 2011, 52, e194-8.	5.1	29
15	Association Study of TRPC4 as a Candidate Gene for Generalized Epilepsy with Photosensitivity. NeuroMolecular Medicine, 2010, 12, 292-299.	3.4	25
16	Role of GRM4 in idiopathic generalized epilepsies analysed by genetic association and sequence analysis. Epilepsy Research, 2010, 89, 319-326.	1.6	19
17	A duplication in 1q21.3 in a family with early onset and childhood absence epilepsy. Epilepsia, 2010, 51, 2453-2456.	5.1	12
18	Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. Nature Genetics, 2010, 42, 1021-1026.	21.4	431

#	Article	IF	Citations
19	Recurrent microdeletions at 15q11.2 and 16p13.11 predispose to idiopathic generalized epilepsies. Brain, 2010, 133, 23-32.	7.6	406
20	Genome-Wide Copy Number Variation in Epilepsy: Novel Susceptibility Loci in Idiopathic Generalized and Focal Epilepsies. PLoS Genetics, 2010, 6, e1000962.	3.5	414
21	15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. Nature Genetics, 2009, 41, 160-162.	21.4	511
22	Non-response to antiepileptic pharmacotherapy is associated with the ABCC2 â^'24C>T polymorphism in young and adult patients with epilepsy. Pharmacogenetics and Genomics, 2009, 19, 353-362.	1.5	87
23	The role of opioids in restless legs syndrome: an [11C]diprenorphine PET study. Brain, 2005, 128, 906-917.	7.6	140
24	Opioid binding in DYT1 primary torsion dystonia: An11C-diprenorphine PET study. Movement Disorders, 2004, 19, 1498-1503.	3.9	17