Judith Conroy

List of Publications by Year in descending order

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759233 996975 4,108 16 12 15 h-index citations g-index papers 16 16 16 7780 docs citations times ranked citing authors all docs

#	Article	IF	CITATIONS
1	Functional impact of global rare copy number variation in autism spectrum disorders. Nature, 2010, 466, 368-372.	27.8	1,803
2	Convergence of Genes and Cellular Pathways Dysregulated in Autism Spectrum Disorders. American Journal of Human Genetics, 2014, 94, 677-694.	6.2	819
3	A genome-wide scan for common alleles affecting risk for autism. Human Molecular Genetics, 2010, 19, 4072-4082.	2.9	538
4	Individual common variants exert weak effects on the risk for autism spectrum disorders. Human Molecular Genetics, 2012, 21, 4781-4792.	2.9	334
5	Unexplained early onset epileptic encephalopathy: Exome screening and phenotype expansion. Epilepsia, 2016, 57, e12-7.	5.1	164
6	TDP2 protects transcription from abortive topoisomerase activity and is required for normal neural function. Nature Genetics, 2014, 46, 516-521.	21,4	122
7	Clinical spectrum and genotype–phenotype associations of KCNA2-related encephalopathies. Brain, 2017, 140, 2337-2354.	7.6	117
8	The variable phenotypes of <scp>KCNQ</scp> â€related epilepsy. Epilepsia, 2014, 55, e99-105.	5.1	109
9	Towards the identification of a genetic basis for <scp>L</scp> andauâ€ <scp>K</scp> leffner <scp>s</scp> yndrome. Epilepsia, 2014, 55, 858-865.	5.1	44
10	Novel European SLC1A4 variant: infantile spasms and population ancestry analysis. Journal of Human Genetics, 2016, 61, 761-764.	2.3	18
11	Chromosomal microarray in unexplained severe early onset epilepsy – A single centre cohort. European Journal of Paediatric Neurology, 2015, 19, 390-394.	1.6	14
12	Novel <i><scp>SMC</scp>1A</i> variant and epilepsy of infancy with migrating focal seizures: Expansion of the phenotype. Epilepsia, 2017, 58, 1301-1302.	5.1	12
13	Atypical benign partial epilepsy of childhood with acquired neurocognitive, lexical semantic, and autistic spectrum disorder. Epilepsy & Behavior Case Reports, 2016, 6, 42-48.	1.5	10
14	Cost of exome sequencing in epileptic encephalopathy: is it â€~worth it'?. Archives of Disease in Childhood, 2018, 103, 304-304.	1.9	3
15	Symmetrical thalamic calcification: A trio whole exome sequencing negative series. Brain and Development, 2017, 39, 426-430.	1.1	1
16	OC48â€Re-interrogation of whole exome sequencing data in developmental epileptic encephalopathies. , 2019, , .		0