Gemma L Carvill

List of Publications by Year
in descending order

14	160	1478505 6	1281871 11
papers	citations	h-index	g-index

14	14	14	365
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A 2020 View on the Genetics of Developmental and Epileptic Encephalopathies. Epilepsy Currents, 2020, 20, 90-96.	0.8	39
2	Damaging de novo missense variants inEEF1A2lead to a developmental and degenerative epilepticâ€dyskinetic encephalopathy. Human Mutation, 2020, 41, 1263-1279.	2.5	24
3	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. Annals of Neurology, 2018, 83, 926-934.	5.3	20
4	CACNA1H variants are not a cause of monogenic epilepsy. Human Mutation, 2020, 41, 1138-1144.	2.5	19
5	Dravet syndrome in South African infants: Tools for an early diagnosis. Seizure: the Journal of the British Epilepsy Association, 2018, 62, 99-105.	2.0	12
6	Phenotypic Spectrum of Seizure Disorders in MBD5-Associated Neurodevelopmental Disorder. Neurology: Genetics, 2021, 7, e579.	1.9	8
7	Unravelling the genetic architecture of autosomal recessive epilepsy in the genomic era. Journal of Neurogenetics, 2018, 32, 295-312.	1.4	7
8	Reanalysis and optimisation of bioinformatic pipelines is critical for mutation detection. Human Mutation, 2019, 40, 374-379.	2.5	7
9	Pathogenic <scp>MAST3</scp> Variants in the <scp>STK</scp> Domain Are Associated with Epilepsy. Annals of Neurology, 2021, 90, 274-284.	5.3	7
10	Pathogenic mechanisms underlying SLC6A1 variant-mediated neurodevelopmental disorders. Brain, 2021, 144, 2237-2239.	7.6	5
11	Genetic convergence of developmental and epileptic encephalopathies and intellectual disability. Developmental Medicine and Child Neurology, 2021, 63, 1441-1447.	2.1	4
12	Wholeâ€exome sequencing and adrenocorticotropic hormone therapy in individuals with infantile spasms. Developmental Medicine and Child Neurology, 2022, 64, 633-640.	2.1	4
13	Epilepsy Genetics: What Once Was Rare, Is Now Common. Epilepsy Currents, 2020, 20, 221-223.	0.8	3
14	Free as a BRD4: Bromodomain Inhibition Ameliorates Disease Phenotypes in a Model of MECP2 Deficiency and Is a Potential Therapy for Rett Syndrome. Epilepsy Currents, 2020, 20, 390-392.	0.8	1