

Ningning Hu

List of Publications by Year in descending order

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16
papers

568
citations

759233

12
h-index

996975

15
g-index

16
all docs

16
docs citations

16
times ranked

862
citing authors

#	ARTICLE	IF	CITATIONS
1	<i>De novo GABRG2</i> mutations associated with epileptic encephalopathies. <i>Brain</i> , 2017, 140, 49-67.	7.6	80
2	Epileptic encephalopathy de novo <i>GABRB</i> mutations impair γ -aminobutyric acid type A receptor function. <i>Annals of Neurology</i> , 2016, 79, 806-825.	5.3	71
3	<i>SCN8A</i> mutations in Chinese children with early onset epilepsy and intellectual disability. <i>Epilepsia</i> , 2015, 56, 431-438.	5.1	61
4	Synaptic clustering differences due to different <i>GABRB3</i> mutations cause variable epilepsy syndromes. <i>Brain</i> , 2019, 142, 3028-3044.	7.6	57
5	<i>GABRB3</i> Mutation, G32R, Associated with Childhood Absence Epilepsy Alters γ -Aminobutyric Acid Type A (GABA) Receptor Expression and Channel Gating. <i>Journal of Biological Chemistry</i> , 2012, 287, 12083-12097.	3.4	52
6	The <i>GABRG2</i> nonsense mutation, Q40X, associated with Dravet syndrome activated NMD and generated a truncated subunit that was partially rescued by aminoglycoside-induced stop codon read-through. <i>Neurobiology of Disease</i> , 2012, 48, 115-123.	4.4	43
7	The <i>GABRA6</i> mutation, <i>R46W</i> , associated with childhood absence epilepsy, alters γ and β GABA _A receptor channel gating and expression. <i>Journal of Physiology</i> , 2011, 589, 5857-5878.	2.9	36
8	Three epilepsy-associated <i>GABRG2</i> missense mutations at the β interface disrupt GABA _A receptor assembly and trafficking by similar mechanisms but to different extents. <i>Neurobiology of Disease</i> , 2014, 68, 167-179.	4.4	36
9	Altered inhibitory synapses in de novo <i>GABRA5</i> and <i>GABRA1</i> mutations associated with early onset epileptic encephalopathies. <i>Brain</i> , 2019, 142, 1938-1954.	7.6	32
10	Deleterious Rare Variants Reveal Risk for Loss of GABA _A Receptor Function in Patients with Genetic Epilepsy and in the General Population. <i>PLoS ONE</i> , 2016, 11, e0162883.	2.5	27
11	Altered Channel Conductance States and Gating of GABA _A Receptors by a Pore Mutation Linked to Dravet Syndrome. <i>ENeuro</i> , 2017, 4, ENEURO.0251-16.2017.	1.9	26
12	Dravet syndrome-associated mutations in <i>GABRA1</i> , <i>GABRB2</i> and <i>GABRG2</i> define the genetic landscape of defects of GABA _A receptors. <i>Brain Communications</i> , 2021, 3, fcab033.	3.3	21
13	GABA beyond the synapse: defining the subtype-specific pharmacodynamics of non-synaptic GABA _A receptors. <i>Journal of Physiology</i> , 2018, 596, 4475-4495.	2.9	17
14	The K328M substitution in the human GABA _A receptor gamma2 subunit causes GEFS+ and premature sudden death in knock-in mice. <i>Neurobiology of Disease</i> , 2021, 152, 105296.	4.4	7
15	Whole exome sequencing reveals novel NOV and DCAF13 variants in a Chinese pedigree with familial cortical myoclonic tremor with epilepsy. <i>Neuroscience Letters</i> , 2018, 684, 115-120.	2.1	2
16	Reply. <i>Annals of Neurology</i> , 2016, 80, 312-313.	5.3	0