List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	The Tuberous Sclerosis Complex. New England Journal of Medicine, 2006, 355, 1345-1356.	13.9	1,570
2	Tuberous Sclerosis Complex Diagnostic Criteria Update: Recommendations of the 2012 International Tuberous Sclerosis Complex Consensus Conference. Pediatric Neurology, 2013, 49, 243-254.	1.0	1,185
3	Tuberous Sclerosis Complex Surveillance and Management: Recommendations of the 2012 International Tuberous Sclerosis Complex Consensus Conference. Pediatric Neurology, 2013, 49, 255-265.	1.0	693
4	The tuberous sclerosis complex. Annals of the New York Academy of Sciences, 2010, 1184, 87-105.	1.8	344
5	The mTOR signalling cascade: paving new roads to cure neurological disease. Nature Reviews Neurology, 2016, 12, 379-392.	4.9	283
6	mTOR cascade activation distinguishes tubers from focal cortical dysplasia. Annals of Neurology, 2004, 56, 478-487.	2.8	238
7	mTOR: A pathogenic signaling pathway in developmental brain malformations. Trends in Molecular Medicine, 2011, 17, 734-742.	3.5	224
8	Expression of ICAM-1, TNF-α, NFκB, and MAP kinase in tubers of the tuberous sclerosis complex. Neurobiology of Disease, 2003, 14, 279-290.	2.1	134
9	Biallelic <i>TSC</i> gene inactivation in tuberous sclerosis complex. Neurology, 2010, 74, 1716-1723.	1.5	134
10	Gene Expression Analysis of Tuberous Sclerosis Complex Cortical Tubers Reveals Increased Expression of Adhesion and Inflammatory Factors. Brain Pathology, 2010, 20, 704-719.	2.1	132
11	Increased Expression of the Neuronal Glutamate Transporter (EAAT3/EAAC1) in Hippocampal and Neocortical Epilepsy. Epilepsia, 2002, 43, 211-218.	2.6	131
12	mTOR Signaling in Epilepsy: Insights from Malformations of Cortical Development. Cold Spring Harbor Perspectives in Medicine, 2015, 5, a022442-a022442.	2.9	127
13	Evolving neurobiology of tuberous sclerosis complex. Acta Neuropathologica, 2013, 125, 317-332.	3.9	125
14	Fetal Brain Lesions in Tuberous Sclerosis Complex: <scp>TORC1</scp> Activation and Inflammation. Brain Pathology, 2013, 23, 45-59.	2.1	123
15	Familial cortical dysplasia caused by mutation in the mammalian target of rapamycin regulator <i>NPRL3</i> . Annals of Neurology, 2016, 79, 132-137.	2.8	116
16	Neurodevelopmental Disorders as a Cause of Seizures: Neuropathologic, Genetic, and Mechanistic Considerations. Brain Pathology, 2002, 12, 212-233.	2.1	105
17	Polyhydramnios, megalencephaly and symptomatic epilepsy caused by a homozygous 7-kilobase deletion in LYK5. Brain, 2007, 130, 1929-1941.	3.7	105
18	Rapamycin Prevents Seizures After Depletion of STRADA in a Rare Neurodevelopmental Disorder. Science Translational Medicine, 2013, 5, 182ra53.	5.8	99

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19	Focal Cortical Dysplasia: Gene Mutations, Cell Signaling, and Therapeutic Implications. Annual Review of Pathology: Mechanisms of Disease, 2017, 12, 547-571.	9.6	98
20	STRADα deficiency results in aberrant mTORC1 signaling during corticogenesis in humans and mice. Journal of Clinical Investigation, 2010, 120, 1591-1602.	3.9	96
21	Familial cortical dysplasia type <scp>IIA</scp> caused by a germline mutation in <i><scp>DEPDC</scp>5</i> . Annals of Clinical and Translational Neurology, 2015, 2, 575-580.	1.7	95
22	Somatic <i>SLC35A2</i> variants in the brain are associated with intractable neocortical epilepsy. Annals of Neurology, 2018, 83, 1133-1146.	2.8	95
23	Genetics of tuberous sclerosis complex: implications for clinical practice. The Application of Clinical Genetics, 2017, Volume 10, 1-8.	1.4	94
24	Fetal Brain mTOR Signaling Activation in Tuberous Sclerosis Complex. Cerebral Cortex, 2014, 24, 315-327.	1.6	92
25	Molecular Pathogenesis of Tuber Formation in Tuberous Sclerosis Complex. Journal of Child Neurology, 2004, 19, 716-725.	0.7	86
26	Internexin, MAP1B, and nestin in cortical dysplasia as markers of developmental maturity. Acta Neuropathologica, 1997, 93, 619-627.	3.9	83
27	Focal brain malformations: Seizures, signaling, sequencing. Epilepsia, 2009, 50, 3-8.	2.6	74
28	Molecular Pathogenesis of Focal Cortical Dysplasia and Hemimegalencephaly. Journal of Child Neurology, 2005, 20, 330-336.	0.7	69
29	Novel Histopathological Patterns in Cortical Tubers of Epilepsy Surgery Patients with Tuberous Sclerosis Complex. PLoS ONE, 2016, 11, e0157396.	1.1	69
30	mTOR pathway inhibition prevents neuroinflammation and neuronal death in a mouse model of cerebral palsy. Neurobiology of Disease, 2016, 85, 144-154.	2.1	63
31	Recessive nephrocerebellar syndrome on the Galloway-Mowat syndrome spectrum is caused by homozygous protein-truncating mutations of <i>WDR73</i> . Brain, 2015, 138, 2173-2190.	3.7	60
32	Focal Brain Malformations: A Spectrum of Disorders along the mTOR Cascade. Novartis Foundation Symposium, 2007, 288, 260-275.	1.2	58
33	Focal Cortical Dysplasia. Seminars in Neurology, 2015, 35, 201-208.	0.5	57
34	Effects of rapamycin on gene expression, morphology, and electrophysiological properties of rat hippocampal neurons. Epilepsy Research, 2007, 77, 85-92.	0.8	55
35	Co-expression of cyclin D1 and phosphorylated ribosomal S6 proteins in hemimegalencephaly. Acta Neuropathologica, 2007, 114, 287-293.	3.9	54
36	Mutations in TBCK, Encoding TBC1-Domain-Containing Kinase, Lead to a Recognizable Syndrome of Intellectual Disability and Hypotonia. American Journal of Human Genetics, 2016, 98, 782-788.	2.6	50

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37	Impaired Reelin-Dab1 Signaling Contributes to Neuronal Migration Deficits of Tuberous Sclerosis Complex. Cell Reports, 2015, 12, 965-978.	2.9	49
38	Coding and small non-coding transcriptional landscape of tuberous sclerosis complex cortical tubers: implications for pathophysiology and treatment. Scientific Reports, 2017, 7, 8089.	1.6	47
39	<scp>GATOR</scp> opathies: The role of amino acid regulatory gene mutations in epilepsy and cortical malformations. Epilepsia, 2019, 60, 2163-2173.	2.6	45
40	Magnetic resonsance imaging of the cauda equina in chronic inflammatory demyelination polyneuropathy. Annals of Neurology, 2004, 33, 311-313.	2.8	43
41	Tuberous sclerosis complex. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 148, 813-822.	1.0	41
42	Somatic variants in diverse genes leads to a spectrum of focal cortical malformations. Brain, 2022, 145, 2704-2720.	3.7	33
43	DEPDC5 and NPRL3 modulate cell size, filopodial outgrowth, and localization of mTOR in neural progenitor cells and neurons. Neurobiology of Disease, 2018, 114, 184-193.	2.1	32
44	Homozygous boricua <i>TBCK</i> mutation causes neurodegeneration and aberrant autophagy. Annals of Neurology, 2018, 83, 153-165.	2.8	32
45	Transcription of intermediate filament genes is enhanced in focal cortical dysplasia. Acta Neuropathologica, 2001, 102, 141-148.	3.9	29
46	Targeted Gene Expression Analysis in Hemimegalencephaly: Activation of β atenin Signaling. Brain Pathology, 2005, 15, 179-186.	2.1	29
47	Gene Expression, Genetics, and Genomics in Epilepsy: Some Answers, More Questions. Epilepsia, 2007, 48, 42-50.	2.6	28
48	The pathophysiology of tuberous sclerosis complex. Epilepsia, 2010, 51, 27-29.	2.6	23
49	Mechanistic target of rapamycin (mTOR) signaling in status epilepticus. Epilepsy and Behavior, 2019, 101, 106550.	0.9	23
50	Epilepsy and cortical dysplasias. Current Treatment Options in Neurology, 2000, 2, 543-551.	0.7	21
51	Malformations of Cortical Development: Molecular Pathogenesis and Experimental Strategies. Advances in Experimental Medicine and Biology, 2004, 548, 175-191.	0.8	18
52	BK channel properties correlate with neurobehavioral severity in three KCNMA1-linked channelopathy mouse models. ELife, 0, 11, .	2.8	17
53	<i>STRADA</i> â€mutant human cortical organoids model megalencephaly and exhibit delayed neuronal differentiation. Developmental Neurobiology, 2021, 81, 696-709.	1.5	16
54	Dysregulation of PINCH signaling in mesial temporal epilepsy. Journal of Clinical Neuroscience, 2017, 36, 43-52.	0.8	15

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55	The role of somatic mutational events in the pathogenesis of epilepsy. Current Opinion in Neurology, 2019, 32, 191-197.	1.8	15
56	NPRL3 loss alters neuronal morphology, mTOR localization, cortical lamination and seizure threshold. Brain, 2022, 145, 3872-3885.	3.7	15
57	Analysis of mRNA Populations from Single Live and Fixed Cells of the Central Nervous System. Current Protocols in Neuroscience, 1997, 00, 5.3.1-5.3.15.	2.6	14
58	mTORopathies: A Road Well-Traveled. Epilepsy Currents, 2020, 20, 64S-66S.	0.4	14
59	Association of Epileptic and Nonepileptic Seizures and Changes in Circulating Plasma Proteins Linked to Neuroinflammation. Neurology, 2021, 96, e1443-e1452.	1.5	13
60	Gene Profiling in Temporal Lobe Epilepsy Tissue and Dysplastic Lesions. Epilepsia, 2006, 47, 1608-1616.	2.6	12
61	Do We Have a Cure for Tuberous Sclerosis Complex?. Epilepsy Currents, 2008, 8, 159-162.	0.4	10
62	Multimodal Analysis of STRADA Function in Brain Development. Frontiers in Cellular Neuroscience, 2020, 14, 122.	1.8	10
63	Somatic uniparental disomy of Chromosome 16p in hemimegalencephaly. Journal of Physical Education and Sports Management, 2017, 3, a001735.	0.5	9
64	Viral expression of constitutively active AKT3 induces CST axonal sprouting and regeneration, but also promotes seizures. Experimental Neurology, 2022, 349, 113961.	2.0	9
65	mTOR and epileptogenesis in developmental brain malformations. Epilepsia, 2010, 51, 72-72.	2.6	7
66	The enlarging spectrum of focal cortical dysplasias: Figure 1. Brain, 2015, 138, 1446-1448.	3.7	6
67	Gene expression analysis as a strategy to understand the molecular pathogenesis of infantile spasms. International Review of Neurobiology, 2002, 49, 367-389.	0.9	5
68	Sending Mixed Signals: The Expanding Role of Molecular Cascade Mutations in Malformations of Cortical Development and Epilepsy. Epilepsy Currents, 2016, 16, 158-163.	0.4	5
69	Polymicrogyria and GRIN1 mutations: altered connections, altered excitability. Brain, 2018, 141, 622-623.	3.7	5
70	MicroRNAâ€34a activation in tuberous sclerosis complex during early brain development may lead to impaired corticogenesis. Neuropathology and Applied Neurobiology, 2021, 47, 796-811.	1.8	5
71	Down-regulation of the brain-specific cell-adhesion molecule contactin-3 in tuberous sclerosis complex during the early postnatal period. Journal of Neurodevelopmental Disorders, 2022, 14, 8.	1.5	4
72	Knockout of a Tuberous Sclerosis Gene Highlights Role of Glia in Epileptogenesis. Epilepsy Currents, 2003, 3, 139-141.	0.4	3

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73	A Patient Perspective on Seizure Detection and Forecasting. Frontiers in Neurology, 2022, 13, 779551.	1.1	3
74	Dynamic analysis of 4E-BP1 phosphorylation in neurons with Tsc2 or Depdc5 knockout. Experimental Neurology, 2020, 334, 113432.	2.0	2
75	Abnormal activation of Yap/Taz contributes to the pathogenesis of tuberous sclerosis complex. Human Molecular Genetics, 2022, , .	1.4	1