Mark Nellist

List of Publications by Year
in descending order

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Version: 2024-02-01

49	6,225	186265 28	189892 50
papers	citations	h-index	g-index

52	52	52	5137
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Subependymal giant cell astrocytomas are characterized by mTORC1 hyperactivation, a very low somatic mutation rate, and a unique gene expression profile. Modern Pathology, 2021, 34, 264-279.	5.5	16
2	G3BPs tether the TSC complex to lysosomes and suppress mTORC1 signaling. Cell, 2021, 184, 655-674.e27.	28.9	65
3	RHEB/mTOR hyperactivity causes cortical malformations and epileptic seizures through increased axonal connectivity. PLoS Biology, 2021, 19, e3001279.	5.6	27
4	Functional and structural analyses of novel Smith-Kingsmore Syndrome-Associated MTOR variants reveal potential new mechanisms and predictors of pathogenicity. PLoS Genetics, 2021, 17, e1009651.	3.5	9
5	TSC1 binding to lysosomal PIPs is required for TSC complex translocation and mTORC1 regulation. Molecular Cell, 2021, 81, 2705-2721.e8.	9.7	25
6	Comparison of the functional and structural characteristics of rare TSC2 variants with clinical and genetic findings. Human Mutation, 2020, 41, 759-773.	2.5	22
7	Structure of the TSC2 GAP Domain: Mechanistic Insight into Catalysis and Pathogenic Mutations. Structure, 2020, 28, 933-942.e4.	3.3	20
8	Mutational analysis of TSC1 and TSC2 in Danish patients with tuberous sclerosis complex. Scientific Reports, 2020, 10, 9909.	3.3	13
9	Examination of the genetic factors underlying the cognitive variability associated with neurofibromatosis type 1. Genetics in Medicine, 2020, 22, 889-897.	2.4	21
10	TSC2 c.1864C>T variant associated with mild cases of tuberous sclerosis complex. American Journal of Medical Genetics, Part A, 2017, 173, 771-775.	1.2	15
11	Subependymal giant cell astrocytomas in Tuberous Sclerosis Complex have consistent TSC1/TSC2 biallelic inactivation, and no BRAF mutations. Oncotarget, 2017, 8, 95516-95529.	1.8	49
12	Severe bleeding complications and multiple kidney transplants in a patient with tuberous sclerosis complex caused by a novel TSC2 missense variant. Croatian Medical Journal, 2017, 58, 416-423.	0.7	3
13	Variants WithinTSC2Exons 25 and 31 Are Very Unlikely to Cause Clinically Diagnosable Tuberous Sclerosis. Human Mutation, 2016, 37, 364-370.	2.5	16
14	TSC2 N-terminal lysine acetylation status affects to its stability modulating mTORC1 signaling and autophagy. Biochimica Et Biophysica Acta - Molecular Cell Research, 2016, 1863, 2658-2667.	4.1	31
15	Genotype and brain pathology phenotype in children with tuberous sclerosis complex. European Journal of Human Genetics, 2016, 24, 1688-1695.	2.8	35
16	Structure of the TBC1D7â€“TSC1 complex reveals that TBC1D7 stabilizes dimerization of the TSC1 C-terminal coiled coil region. Journal of Molecular Cell Biology, 2016, 8, 411-425.	3.3	37
17	Structural Basis of the Interaction between Tuberous Sclerosis Complex 1 (TSC1) and Tre2-Bub2-Cdc16 Domain Family Member 7 (TBC1D7). Journal of Biological Chemistry, 2016, 291, 8591-8601.	3.4	31
18	PAK2 is an effector of TSC1/2 signaling independent of mTOR and a potential therapeutic target for Tuberous Sclerosis Complex. Scientific Reports, 2015, 5, 14534.	3.3	40

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19	Preliminary Functional Assessment and Classification ofDEPDC5Variants Associated with Focal Epilepsy. Human Mutation, 2015, 36, 200-209.	2.5	28
20	Targeted Next Generation Sequencing reveals previously unidentified TSC1 and TSC2 mutations. BMC Medical Genetics, 2015, 16, 10.	2.1	62
21	Germline activating AKT3 mutation associated with megalencephaly, polymicrogyria, epilepsy and hypoglycemia. Molecular Genetics and Metabolism, 2015, 114, 467-473.	1.1	42
22	Identification of Regions Critical for the Integrity of the TSC1-TSC2-TBC1D7 Complex. PLoS ONE, 2014, 9, e93940.	2.5	32
23	Clinical significance of immunohistochemistry for detection of BAP1 mutations in uveal melanoma. Modern Pathology, 2014, 27, 1321-1330.	5.5	174
24	Tuberous Sclerosis Complex Diagnostic Criteria Update: Recommendations of the 2012 International Tuberous Sclerosis Complex Consensus Conference. Pediatric Neurology, 2013, 49, 243-254.	2.1	1,185
25	Tuberous Sclerosis Complex Surveillance and Management: Recommendations of the 2012 International Tuberous Sclerosis Complex Consensus Conference. Pediatric Neurology, 2013, 49, 255-265.	2.1	693
26	Functional Assessment ofTSC2 Variants Identified in Individuals with Tuberous Sclerosis Complex. Human Mutation, 2013, 34, 167-175.	2.5	60
27	Central TSC2 missense mutations are associated with a reduced risk of infantile spasms. Epilepsy Research, 2013, 103, 83-87.	1.6	26
28	TORC1â€dependent epilepsy caused by acute biallelic Tsc1 deletion in adult mice. Annals of Neurology, 2013, 74, 569-579.	5.3	68
29	The TSC1-TSC2 complex consists of multiple TSC1 and TSC2 subunits. BMC Biochemistry, 2012, 13, 18.	4.4	20
30	Functional assessment of TSC1 missense variants identified in individuals with tuberous sclerosis complex. Human Mutation, 2012, 33, 476-479.	2.5	45
31	Functional assessment of variants in the TSC1 and TSC2 genes identified in individuals with Tuberous Sclerosis Complex. Human Mutation, 2011, 32, 424-435.	2.5	73
32	Analysis of TSC1 truncations defines regions involved in TSC1 stability, aggregation and interaction. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2010, 1802, 774-781.	3.8	23
33	Identification of a region required for TSC1 stability by functional analysis of TSC1missense mutations found in individuals with tuberous sclerosis complex. BMC Medical Genetics, 2009, 10, 88.	2.1	29
34	Missense mutations to the TSC1 gene cause tuberous sclerosis complex. European Journal of Human Genetics, 2009, 17, 319-328.	2.8	25
35	A reliable cell-based assay for testing unclassified TSC2 gene variants. European Journal of Human Genetics, 2009, 17, 301-310.	2.8	26
36	Hamartin Variants That Are Frequent in Focal Dysplasias and Cortical Tubers Have Reduced Tuberin Binding and Aberrant Subcellular Distribution In Vitro. Journal of Neuropathology and Experimental Neurology, 2009, 68, 1136-1146.	1.7	12

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37	Functional characterisation of the TSC1â€“TSC2 complex to assess multiple TSC2 variants identified in single families affected by tuberous sclerosis complex. BMC Medical Genetics, 2008, 9, 10.	2.1	17
38	Clinicopathological and immunohistochemical findings in an autopsy case of tuberous sclerosis complex. Neuropathology, 2008, 28, 577-590.	1.2	96
39	Unusually mild tuberous sclerosis phenotype is associated withTSC2R905Q mutation. Annals of Neurology, 2006, 60, 528-539.	5.3	82
40	Distinct effects of single amino-acid changes to tuberin on the function of the tuberinâ€“hamartin complex. European Journal of Human Genetics, 2005, 13, 59-68.	2.8	73
41	Mutational analysis of the TSC1 and TSC2 genes in a diagnostic setting: genotype â€“ phenotype correlations and comparison of diagnostic DNA techniques in Tuberous Sclerosis Complex. European Journal of Human Genetics, 2005, 13, 731-741.	2.8	405
42	Phosphorylation and binding partner analysis of the TSC1â€“TSC2 complex. Biochemical and Biophysical Research Communications, 2005, 333, 818-826.	2.1	33
43	Identification and Characterization of the Interaction between Tuberin and 14-3-3Î¶. Journal of Biological Chemistry, 2002, 277, 39417-39424.	3.4	64
44	Analysis of TSC2 stop codon variants found in tuberous sclerosis patients. European Journal of Human Genetics, 2001, 9, 823-828.	2.8	5
45	Characterization of the Cytosolic Tuberin-Hamartin Complex. Journal of Biological Chemistry, 1999, 274, 35647-35652.	3.4	164
46	Identification of the Tuberous Sclerosis Gene TSC1 on Chromosome 9q34. Science, 1997, 277, 805-808.	12.6	1,550
47	Comparative Analysis and Genomic Structure of the Tuberous Sclerosis 2 (TSC2) Gene in Human and Pufferfish. Human Molecular Genetics, 1996, 5, 131-137.	2.9	66
48	Alternative Splicing of the Tuberous Sclerosis 2 (TSC2) Gene in Human and Mouse Tissues. Genomics, 1995, 27, 475-480.	2.9	64
49	Deletion of the TSC2 and PKD1 genes associated with severe infantile polycystic kidney disease â€” a contiguous gene syndrome. Nature Genetics, 1994, 8, 328-332.	21.4	466