

Valerio Conti

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/6379997/publications.pdf>

Version: 2024-02-01

48
papers

2,234
citations

304743

22
h-index

276875

41
g-index

53
all docs

53
docs citations

53
times ranked

4228
citing authors

#	ARTICLE	IF	CITATIONS
1	Association of <i>MTOR</i> Mutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. <i>JAMA Neurology</i> , 2016, 73, 836.	9.0	234
2	A versatile clearing agent for multi-modal brain imaging. <i>Scientific Reports</i> , 2015, 5, 9808.	3.3	228
3	Genetic Basis of Brain Malformations. <i>Molecular Syndromology</i> , 2016, 7, 220-233.	0.8	156
4	The landscape of epilepsy-related GATOR1 variants. <i>Genetics in Medicine</i> , 2019, 21, 398-408.	2.4	137
5	PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. <i>JCI Insight</i> , 2016, 1, .	5.0	134
6	Germline and somatic mutations in the <i>MTOR</i> gene in focal cortical dysplasia and epilepsy. <i>Neurology: Genetics</i> , 2016, 2, e118.	1.9	125
7	<i>PRRT2</i> mutations in familial infantile seizures, paroxysmal dyskinesia, and hemiplegic migraine. <i>Neurology</i> , 2012, 79, 2109-2114.	1.1	106
8	Mutations of <i>AKT3</i> are associated with a wide spectrum of developmental disorders including extreme megalencephaly. <i>Brain</i> , 2017, 140, 2610-2622.	7.6	102
9	Analysis of 17 genes detects mutations in 81% of 811 patients with lissencephaly. <i>Genetics in Medicine</i> , 2018, 20, 1354-1364.	2.4	92
10	Periventricular heterotopia in 6q terminal deletion syndrome: role of the <i>C6orf70</i> gene. <i>Brain</i> , 2013, 136, 3378-3394.	7.6	85
11	Characterisation of mutations of the phosphoinositide-3-kinase regulatory subunit, <i>PIK3R2</i> , in perisylvian polymicrogyria: a next-generation sequencing study. <i>Lancet Neurology</i> , The, 2015, 14, 1182-1195.	10.2	74
12	De novo mutations of the <i>ATP6V1A</i> gene cause developmental encephalopathy with epilepsy. <i>Brain</i> , 2018, 141, 1703-1718.	7.6	69
13	Genomic DNA methylation distinguishes subtypes of human focal cortical dysplasia. <i>Epilepsia</i> , 2019, 60, 1091-1103.	5.1	61
14	Focal dysplasia of the cerebral cortex and infantile spasms associated with somatic 1q21.1q44 duplication including the <i>AKT3</i> gene. <i>Clinical Genetics</i> , 2015, 88, 241-247.	2.0	60
15	Autism-epilepsy phenotype with macrocephaly suggests <i>PTEN</i> , but not <i>GLIALCAM</i> , genetic screening. <i>BMC Medical Genetics</i> , 2014, 15, 26.	2.1	55
16	International consensus recommendations on the diagnostic work-up for malformations of cortical development. <i>Nature Reviews Neurology</i> , 2020, 16, 618-635.	10.1	53
17	Presynaptic mGlu1 and mGlu5 autoreceptors facilitate glutamate exocytosis from mouse cortical nerve endings. <i>Neuropharmacology</i> , 2008, 55, 474-482.	4.1	49
18	Somatic double-hit in <i>MTOR</i> and <i>RPS6</i> in hemimegalencephaly with intractable epilepsy. <i>Human Molecular Genetics</i> , 2019, 28, 3755-3765.	2.9	42

#	ARTICLE	IF	CITATIONS
19	crv4, a mouse model for human ataxia associated with kyphoscoliosis caused by an mRNA splicing mutation of the metabotropic glutamate receptor 1 (Grm1). International Journal of Molecular Medicine, 2006, 18, 593-600.	4.0	36
20	Expression of Tsga10 sperm tail protein in embryogenesis and neural development: From cilium to cell division. Biochemical and Biophysical Research Communications, 2006, 344, 1102-1110.	2.1	33
21	Nocturnal frontal lobe epilepsy with paroxysmal arousals due to CHRNA2 loss of function. Neurology, 2015, 84, 1520-1528.	1.1	32
22	Albuminuria and Glomerular Damage in Mice Lacking the Metabotropic Glutamate Receptor 1. American Journal of Pathology, 2011, 178, 1257-1269.	3.8	31
23	Is Focal Cortical Dysplasia/Epilepsy Caused by Somatic <i>MTOR</i> Mutations Always a Unilateral Disorder?. Neurology: Genetics, 2021, 7, e540.	1.9	26
24	Profiling PI3K-AKT-MTOR variants in focal brain malformations reveals new insights for diagnostic care. Brain, 2022, 145, 925-938.	7.6	25
25	Biallelic DMXL2 mutations impair autophagy and cause Ohtahara syndrome with progressive course. Brain, 2019, 142, 3876-3891.	7.6	23
26	Multiple genomic copy number variants associated with periventricular nodular heterotopia indicate extreme genetic heterogeneity. European Journal of Human Genetics, 2019, 27, 909-918.	2.8	21
27	Large-scale, cell-resolution volumetric mapping allows layer-specific investigation of human brain cytoarchitecture. Biomedical Optics Express, 2021, 12, 3684.	2.9	18
28	C620R mutation of the murineret proto-oncogene: Loss of function effect in homozygotes and possible gain of function effect in heterozygotes. International Journal of Cancer, 2007, 121, 292-300.	5.1	15
29	Circulating tumor cells and palbociclib treatment in patients with ER-positive, HER2-negative advanced breast cancer: results from a translational sub-study of the TREnd trial. Breast Cancer Research, 2021, 23, 38.	5.0	14
30	Low-copy repeats on chromosome 22q11.2 show replication timing switches, DNA flexibility peaks and stress inducible asynchrony, sharing instability features with fragile sites. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2010, 686, 74-83.	1.0	11
31	Corpus callosum agenesis, severe mental retardation, epilepsy, and dyskinetic quadriplegia due to a novel mutation in the homeodomain of ARX. , 2011, 155, 892-897.		11
32	Multimodal fiber-optic probe spectroscopy allows detecting epileptogenic focal cortical dysplasia in children. Journal of Biophotonics, 2017, 10, 896-904.	2.3	11
33	Phenotypic and genetic spectrum of ATP6V1A encephalopathy: a disorder of lysosomal homeostasis. Brain, 2022, 145, 2687-2703.	7.6	11
34	crv4, a mouse model for human ataxia associated with kyphoscoliosis caused by an mRNA splicing mutation of the metabotropic glutamate receptor 1 (Grm1). International Journal of Molecular Medicine, 2006, 18, 593.	4.0	9
35	Lesional and non-lesional epilepsies: A blurring genetic boundary. European Journal of Paediatric Neurology, 2020, 24, 24-29.	1.6	8
36	Angiocentric glioma-associated seizures: The possible role of EAT2, pyruvate carboxylase and glutamine synthetase. Seizure: the Journal of the British Epilepsy Association, 2021, 86, 152-154.	2.0	8

#	ARTICLE	IF	CITATIONS
37	Somatic Focal Copy Number Gains of Noncoding Regions of Receptor Tyrosine Kinase Genes in Treatment-Resistant Epilepsy. <i>Journal of Neuropathology and Experimental Neurology</i> , 2021, 80, 160-168.	1.7	7
38	Prospective Evaluation of Ghrelin and Des-Acyl Ghrelin Plasma Levels in Children with Newly Diagnosed Epilepsy: Evidence for Reduced Ghrelin-to-Des-Acyl Ghrelin Ratio in Generalized Epilepsies. <i>Journal of Personalized Medicine</i> , 2022, 12, 527.	2.5	7
39	Contractions in the second polyA tract of ARX are rare, nonâ€pathogenic polymorphisms. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 164-167.	1.2	2
40	Computer-based automatic identification of neurons in gigavoxel-sized 3D human brain images. , 2015, 7724-7.		2
41	A top-down linguistic approach to the analysis of genomic sequences: The metabotropic glutamate receptors 1 and 5 in human and in mouse as a case study. <i>Journal of Theoretical Biology</i> , 2011, 270, 134-142.	1.7	1
42	Towards automated neuron tracing via global and local 3D image analysis. , 2016, , .		1
43	Human Mutations Associated With Brain Malformations Resulting in Hyperexcitability in Rodents. , 2017, , 827-844.		1
44	Probing focal cortical dysplasia in formalin fixed samples using tissue optical spectroscopy. , 2016, , .		0
45	Fiber-probe optical spectroscopy discriminates normal brain from focal cortical dysplasia in pediatric subjects. , 2017, , .		0
46	Multimodal fiber-probe spectroscopy as a clinical tool for diagnosing and classifying biological tissues. , 2017, , .		0
47	A Novel Strategy Combining Array-CGH, Whole-exome Sequencing and <i>In Utero</i> Electroporation in Rodents to Identify Causative Genes for Brain Malformations. <i>Journal of Visualized Experiments</i> , 2017, , .	0.3	0
48	An Enhanced Distributed Computational Platform for Developmental and Epileptic Encephalopathies. , 2022, , .		0