Valerio Conti

List of Publications by Year
in descending order

48	2,234	304743	276875
papers	citations	22 h-index	41 g-index

53	53	53	4228
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Profiling PI3K-AKT-MTOR variants in focal brain malformations reveals new insights for diagnostic care. Brain, 2022, 145, 925-938.	7.6	25
2	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. Journal of Personalized Medicine, 2022, 12, 527.	2.5	7
3	Phenotypic and genetic spectrum of ATP6V1A encephalopathy: a disorder of lysosomal homeostasis. Brain, 2022, 145, 2687-2703.	7.6	11
4	An Enhanced Distributed Computational Platform for Developmental and Epileptic Encephalopathies. , 2022, , .		0
5	Somatic Focal Copy Number Gains of Noncoding Regions of Receptor Tyrosine Kinase Genes in Treatment-Resistant Epilepsy. Journal of Neuropathology and Experimental Neurology, 2021, 80, 160-168.	1.7	7
6	Is Focal Cortical Dysplasia/Epilepsy Caused by Somatic MTOR Mutations Always a Unilateral Disorder?. Neurology: Genetics, 2021, 7, e540.	1.9	26
7	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
8	Angiocentric glioma-associated seizures: The possible role of EATT2, pyruvate carboxylase and glutamine synthetase. Seizure: the Journal of the British Epilepsy Association, 2021, 86, 152-154.	2.0	8
9	Large-scale, cell-resolution volumetric mapping allows layer-specific investigation of human brain cytoarchitecture. Biomedical Optics Express, 2021, 12, 3684.	2.9	18
10	Lesional and non-lesional epilepsies: A blurring genetic boundary. European Journal of Paediatric Neurology, 2020, 24, 24-29.	1.6	8
11	International consensus recommendations on the diagnostic work-up for malformations of cortical development. Nature Reviews Neurology, 2020, 16, 618-635.	10.1	53
12	The landscape of epilepsy-related GATOR1 variants. Genetics in Medicine, 2019, 21, 398-408.	2.4	137
13	Somatic double-hit in MTOR and RPS6 in hemimegalencephaly with intractable epilepsy. Human Molecular Genetics, 2019, 28, 3755-3765.	2.9	42
14	Biallelic DMXL2 mutations impair autophagy and cause Ohtahara syndrome with progressive course. Brain, 2019, 142, 3876-3891.	7.6	23
15	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
16	Genomic <scp>DNA</scp> methylation distinguishes subtypes of human focal cortical dysplasia. Epilepsia, 2019, 60, 1091-1103.	5.1	61
17	Analysis of 17 genes detects mutations in 81% of 811 patients with lissencephaly. Genetics in Medicine, 2018, 20, 1354-1364.	2.4	92
18	De novo mutations of the ATP6V1A gene cause developmental encephalopathy with epilepsy. Brain, 2018, 141, 1703-1718.	7.6	69

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19	Multimodal fiberâ€probe spectroscopy allows detecting epileptogenic focal cortical dysplasia in children. Journal of Biophotonics, 2017, 10, 896-904.	2.3	11
20	Fiber-probe optical spectroscopy discriminates normal brain from focal cortical dysplasia in pediatric subjects. , 2017, , .		0
21	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. Brain, 2017, 140, 2610-2622.	7.6	102
22	Multimodal fiber-probe spectroscopy as a clinical tool for diagnosing and classifying biological tissues. , 2017, , .		0
23	Human Mutations Associated With Brain Malformations Resulting in Hyperexcitability in Rodents. , 2017, , 827-844.		1
24	A Novel Strategy Combining Array-CGH, Whole-exome Sequencing and In Utero Electroporation in Rodents to Identify Causative Genes for Brain Malformations. Journal of Visualized Experiments, 2017, , .	0.3	0
25	Association ofMTORMutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. JAMA Neurology, 2016, 73, 836.	9.0	234
26	Towards automated neuron tracing via global and local 3D image analysis. , 2016, , .		1
27	Genetic Basis of Brain Malformations. Molecular Syndromology, 2016, 7, 220-233.	0.8	156
28	Germline and somatic mutations in the MTOR gene in focal cortical dysplasia and epilepsy. Neurology: Genetics, 2016, 2, e118.	1.9	125
29	Probing focal cortical dysplasia in formalin fixed samples using tissue optical spectroscopy. , 2016, , .		0
30	PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. JCI Insight, 2016, 1, .	5.0	134
31	Computer-based automatic identification of neurons in gigavoxel-sized 3D human brain images. , 2015, 2015, 7724-7.		2
32	A versatile clearing agent for multi-modal brain imaging. Scientific Reports, 2015, 5, 9808.	3.3	228
33	Nocturnal frontal lobe epilepsy with paroxysmal arousals due to CHRNA2 loss of function. Neurology, 2015, 84, 1520-1528.	1.1	32
34	Characterisation of mutations of the phosphoinositide-3-kinase regulatory subunit, PIK3R2, in perisylvian polymicrogyria: a next-generation sequencing study. Lancet Neurology, The, 2015, 14, 1182-1195.	10.2	74
35	Focal dysplasia of the cerebral cortex and infantile spasms associated with somatic 1q21.1â€q44 duplication including the <scp>AKT3</scp> gene. Clinical Genetics, 2015, 88, 241-247.	2.0	60
36	Autism-epilepsy phenotype with macrocephaly suggests PTEN, but not GLIALCAM, genetic screening. BMC Medical Genetics, 2014, 15, 26.	2.1	55

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37	Periventricular heterotopia in 6q terminal deletion syndrome: role of the C6orf70 gene. Brain, 2013, 136, 3378-3394.	7.6	85
38	PRRT2 mutations in familial infantile seizures, paroxysmal dyskinesia, and hemiplegic migraine. Neurology, 2012, 79, 2109-2114.	1.1	106
39	Albuminuria and Glomerular Damage in Mice Lacking the Metabotropic Glutamate Receptor 1. American Journal of Pathology, 2011, 178, 1257-1269.	3.8	31
40	Contractions in the second polyA tract of ARX are rare, nonâ€pathogenic polymorphisms. American Journal of Medical Genetics, Part A, 2011, 155, 164-167.	1.2	2
41	Corpus callosum agenesis, severe mental retardation, epilepsy, and dyskinetic quadriparesis due to a novel mutation in the homeodomain of ARX. , 2011, 155, 892-897.		11
42	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. Journal of Theoretical Biology, 2011, 270, 134-142.	1.7	1
43	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
44	Presynaptic mGlu1 and mGlu5 autoreceptors facilitate glutamate exocytosis from mouse cortical nerve endings. Neuropharmacology, 2008, 55, 474-482.	4.1	49
45	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
46	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
47	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
48	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