Kit Doudney

List of Publications by Year
in descending order

19 papers	1,220 citations	686830 13 h-index	794141 19 g-index
20 all docs	20 docs citations	20 times ranked	1526 citing authors

#	Article	IF	Citations
1	Disruption of scribble (Scrb1) causes severe neural tube defects in the circletail mouse. Human Molecular Genetics, 2003, 12, 87-98.	1.4	266
2	The T-box transcription factor gene TBX22 is mutated in X-linked cleft palate and ankyloglossia. Nature Genetics, 2001, 29, 179-183.	9.4	245
3	Mutations in the planar cell polarity genesCELSR1andSCRIBare associated with the severe neural tube defect craniorachischisis. Human Mutation, 2012, 33, 440-447.	1.1	166
4	Craniofacial expression of human and murine TBX22 correlates with the cleft palate and ankyloglossia phenotype observed in CPX patients. Human Molecular Genetics, 2002, 11, 2793-2804.	1.4	87
5	TBX22 Missense Mutations Found in Patients with X-Linked Cleft Palate Affect DNA Binding, Sumoylation, and Transcriptional Repression. American Journal of Human Genetics, 2007, 81, 700-712.	2.6	84
6	The Friedreich's ataxia gene encodes a novel phosphatidylinositolâ€“4â€“phosphate 5â€“kinase. Nature Genetics, 1996, 14, 157-162.	9.4	79
7	Abnormal folate metabolism in foetuses affected by neural tube defects. Brain, 2006, 130, 1043-1049.	3.7	48
8	Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. American Journal of Human Genetics, 2016, 99, 674-682.	2.6	48
9	Epithelial cell polarity genes are required for neural tube closure. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2005, 135C, 42-47.	0.7	42
10	G-Quadruplex Structures and CpG Methylation Cause Drop-Out of the Maternal Allele in Polymerase Chain Reaction Amplification of the Imprinted MEST Gene Promoter. PLoS ONE, 2014, 9, e113955.	1.1	30
11	Cloning and Characterization of Igsf9 in Mouse and Human: A New Member of the Immunoglobulin Superfamily Expressed in the Developing Nervous System. Genomics, 2002, 79, 663-670.	1.3	27
12	Friedreich's ataxia: a defect in signal transduction?. Human Molecular Genetics, 1995, 4, 1411-1419.	1.4	25
13	Valproic acid exposure leads to upregulation and increased promoter histone acetylation of sepiapterin reductase in a serotonergic cell line. Neuropharmacology, 2015, 99, 79-88.	2.0	21
14	Comparative Physical and Transcript Maps of âˆ¼1 Mb around loop-tail, a Gene for Severe Neural Tube Defects on Distal Mouse Chromosome 1 and Human Chromosome 1q22â€“q23. Genomics, 2001, 72, 180-192.	1.3	13
15	Genomic organization and embryonic expression of igsf8, an immunoglobulin superfamily member implicated in development of the nervous system and organ epithelia. Molecular and Cellular Neurosciences, 2003, 22, 62-74.	1.0	10
16	Comparing the variants of takotsubo syndrome: an observational study of the ECG and structural changes from a New Zealand tertiary hospital. BMJ Open, 2019, 9, e025253.	0.8	10
17	Copy number variants implicate cardiac function and development pathways in earthquake-induced stress cardiomyopathy. Scientific Reports, 2018, 8, 7548.	1.6	8
18	PPA2-associated sudden cardiac death: extending the clinical and allelic spectrum in 20 new families. Genetics in Medicine, 2021, 23, 2415-2425.	1.1	8

#	Article	IF	Citations
19	Exonâ€“Intron Structure of a 2.7-kb Transcript of the STM7 Gene with Phosphatidylinositol-4-phosphate 5-Kinase Activity. Genomics, 1997, 42, 170-172.	1.3	2