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	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
2	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
3	Copy number variants implicate cardiac function and development pathways in earthquake-induced stress cardiomyopathy. Scientific Reports, 2018, 8, 7548.	1.6	8
4	Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. American Journal of Human Genetics, 2016, 99, 674-682.	2.6	48
5	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
6	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
7	Mutations in the planar cell polarity genesCELSR1andSCRIBare associated with the severe neural tube defect craniorachischisis. Human Mutation, 2012, 33, 440-447.	1.1	166
8	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
9	Abnormal folate metabolism in foetuses affected by neural tube defects. Brain, 2006, 130, 1043-1049.	3.7	48
10	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
11	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
12	Disruption of scribble (Scrb1) causes severe neural tube defects in the circletail mouse. Human Molecular Genetics, 2003, 12, 87-98.	1.4	266
13	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
14	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
15	Comparative Physical and Transcript Maps of â¼1 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
16	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
17	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
18	The Friedreich's ataxia gene encodes a novel phosphatidylinositolâ€“4â€“phosphate 5â€“kinase. Nature Genetics, 1996, 14, 157-162.	9.4	79

#	Article	IF	Citations
19	Friedreich's ataxia: a defect in signal transduction?. Human Molecular Genetics, 1995, 4, 1411-1419.	1.4	25