Kit Sing Au

List of Publications by Year
in descending order

33 papers	1,208 citations	623574 14 h-index	477173 29 g-index
36 all docs	36 docs citations	36 times ranked	1564 citing authors

#	Article	IF	Citations
1	Genotype/phenotype correlation in 325 individuals referred for a diagnosis of tuberous sclerosis complex in the United States. Genetics in Medicine, 2007, 9, 88-100.	1.1	353
2	Epidemiologic and genetic aspects of spina bifida and other neural tube defects. Developmental Disabilities Research Reviews, 2010, 16, 6-15.	2.9	269
3	Presentation and Diagnosis of Tuberous Sclerosis Complex in Infants. Pediatrics, 2017, 140, .	1.0	90
4	Genetics, genomics, and genotypeâ€“phenotype correlations of TSC: Insights for clinical practice. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 281-290.	0.7	65
5	Genes in Glucose Metabolism and Association With Spina Bifida. Reproductive Sciences, 2008, 15, 51-58.	1.1	44
6	Genetic association study of putative functional single nucleotide polymorphisms of genes in folate metabolism and spina bifida. American Journal of Obstetrics and Gynecology, 2009, 201, 394.e1-394.e11.	0.7	42
7	Association of folate receptor (folr1, folr2, folr3) and reduced folate carrier (slc19a1) genes with meningomyelocele. Birth Defects Research Part A: Clinical and Molecular Teratology, 2010, 88, 689-694.	1.6	36
8	Finding the genetic mechanisms of folate deficiency and neural tube defectsâ€”Leaving no stone unturned. American Journal of Medical Genetics, Part A, 2017, 173, 3042-3057.	0.7	34
9	Copy number variation analysis implicates the cell polarity gene glypican 5 as a human spina bifida candidate gene. Human Molecular Genetics, 2013, 22, 1097-1111.	1.4	29
10	Characteristics of a spina bifida population including North American Caucasian and Hispanic individuals. Birth Defects Research Part A: Clinical and Molecular Teratology, 2008, 82, 692-700.	1.6	27
11	Tuberous Sclerosis Complex Genotypes and Developmental Phenotype. Pediatric Neurology, 2019, 96, 58-63.	1.0	21
12	Genetic association of the glycine cleavage system genes and myelomeningocele. Birth Defects Research Part A: Clinical and Molecular Teratology, 2016, 106, 847-853.	1.6	19
13	Association of copperâ€zinc superoxide dismutase (SOD1) and manganese superoxide dismutase (SOD2) genes with nonsyndromic myelomeningocele. Birth Defects Research Part A: Clinical and Molecular Teratology, 2012, 94, 762-769.	1.6	18
14	Mutations in folate transporter genes and risk for human myelomeningocele. American Journal of Medical Genetics, Part A, 2017, 173, 2973-2984.	0.7	17
15	Tuberous sclerosis complex: disease modifiers and treatments. Current Opinion in Pediatrics, 2008, 20, 628-633.	1.0	15
16	TSC2 c.1864C>T variant associated with mild cases of tuberous sclerosis complex. American Journal of Medical Genetics, Part A, 2017, 173, 771-775.	0.7	15
17	Folate Metabolism Gene 5,10-Methylenetetrahydrofolate Reductase (MTHFR) Is Associated with ADHD in Myelomeningocele Patients. PLoS ONE, 2012, 7, e51330.	1.1	14
18	Genetic studies of the cystathionine betaâ€synthase gene and myelomeningocele. Birth Defects Research Part A: Clinical and Molecular Teratology, 2012, 94, 52-56.	1.6	13

#	Article	IF	Citations
19	Deep sequencing study of the MTHFR gene to identify variants associated with myelomeningocele. Birth Defects Research Part A: Clinical and Molecular Teratology, 2012, 94, 84-90.	1.6	10
20	Association of facilitated glucose transporter 2 gene variants with the myelomeningocele phenotype. Birth Defects Research Part A: Clinical and Molecular Teratology, 2015, 103, 479-487.	1.6	10
21	Catenin delta-1 (CTNND1) phosphorylation controls the mesenchymal to epithelial transition in astrocytic tumors. Human Molecular Genetics, 2016, 25, 4201-4210.	1.4	10
22	Snx3 is important for mammalian neural tube closure via its role in canonical and non-canonical WNT signaling. Development (Cambridge), 2020, 147, .	1.2	10
23	Epilepsy Risk Prediction Model for Patients With Tuberous Sclerosis Complex. Pediatric Neurology, 2020, 113, 46-50.	1.0	9
24	Human myelomeningocele risk and ultra-rare deleterious variants in genes associated with cilium, WNT-signaling, ECM, cytoskeleton and cell migration. Scientific Reports, 2021, 11, 3639.	1.6	8
25	Genetic variations in the GLUT3 gene associated with myelomeningocele. American Journal of Obstetrics and Gynecology, 2014, 211, 305.e1-305.e8.	0.7	7
26	Burden of rare deleterious variants in WNT signaling genes among 511 myelomeningocele patients. PLoS ONE, 2020, 15, e0239083.	1.1	7
27	Identification of novel candidate risk genes for myelomeningocele within the glucose homeostasis/oxidative stress and folate/oneâ€carbon metabolism networks. Molecular Genetics & Genomic Medicine, 2020, 8, e1495.	0.6	5
28	Maternal geneâ€“micronutrient interactions related to oneâ€carbon metabolism and the risk of myelomeningocele among offspring. Birth Defects Research, 2017, 109, 99-105.	0.8	3
29	Whole exome sequencing identifies potential candidate genes for spina bifida derived from mouse models. American Journal of Medical Genetics, Part A, 2022, , .	0.7	2
30	Burden of rare deleterious variants in WNT signaling genes among 511 myelomeningocele patients. , 2020, 15, e0239083.		0
31	Burden of rare deleterious variants in WNT signaling genes among 511 myelomeningocele patients. , 2020, 15, e0239083.		0
32	Burden of rare deleterious variants in WNT signaling genes among 511 myelomeningocele patients. , 2020, 15, e0239083.		0
33	Burden of rare deleterious variants in WNT signaling genes among 511 myelomeningocele patients. , 2020, 15, e0239083.		0