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	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
2	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
3	Epilepsy Risk Prediction Model for Patients With Tuberous Sclerosis Complex. Pediatric Neurology, 2020, 113, 46-50.	1.0	9
4	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
5	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
6	Burden of rare deleterious variants in WNT signaling genes among 511 myelomeningocele patients. PLoS ONE, 2020, 15, e0239083.	1.1	7
7	Burden of rare deleterious variants in WNT signaling genes among 511 myelomeningocele patients. , 2020, 15, e0239083.		0
8	Burden of rare deleterious variants in WNT signaling genes among 511 myelomeningocele patients. , 2020, 15, e0239083.		0
9	Burden of rare deleterious variants in WNT signaling genes among 511 myelomeningocele patients. , 2020, 15, e0239083.		0
10	Burden of rare deleterious variants in WNT signaling genes among 511 myelomeningocele patients. , 2020, 15, e0239083.		0
11	Tuberous Sclerosis Complex Genotypes and Developmental Phenotype. Pediatric Neurology, 2019, 96, 58-63.	1.0	21
12	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
13	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
14	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
15	Mutations in folate transporter genes and risk for human myelomeningocele. American Journal of Medical Genetics, Part A, 2017, 173, 2973-2984.	0.7	17
16	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
17	Presentation and Diagnosis of Tuberous Sclerosis Complex in Infants. Pediatrics, 2017, 140, .	1.0	90
18	Catenin delta-1 (CTNND1) phosphorylation controls the mesenchymal to epithelial transition in astrocytic tumors. Human Molecular Genetics, 2016, 25, 4201-4210.	1.4	10

#	Article	IF	Citations
19	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
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	Genetic variations in the GLUT3 gene associated with myelomeningocele. American Journal of Obstetrics and Gynecology, 2014, 211, 305.e1-305.e8.	0.7	7
22	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
23	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
24	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
25	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
26	Folate Metabolism Gene 5,10-Methylenetetrahydrofolate Reductase (MTHFR) Is Associated with ADHD in Myelomeningocele Patients. PLoS ONE, 2012, 7, e51330.	1.1	14
27	Epidemiologic and genetic aspects of spina bifida and other neural tube defects. Developmental Disabilities Research Reviews, 2010, 16, 6-15.	2.9	269
28	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
29	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
30	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
31	Genes in Glucose Metabolism and Association With Spina Bifida. Reproductive Sciences, 2008, 15, 51-58.	1.1	44
32	Tuberous sclerosis complex: disease modifiers and treatments. Current Opinion in Pediatrics, 2008, 20, 628-633.	1.0	15
33	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