Yu Zheng

List of Publications by Year
in descending order

23	384	933447 10	794594 19
papers	citations	h-index	g-index

25	25	25	792
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Case series of congenital pseudarthrosis of the tibia unfulfilling neurofibromatosis type 1 diagnosis: 21% with somatic NF1 haploinsufficiency in the periosteum. Human Genetics, 2022, 141, 1371-1383.	3.8	4
2	A genotype and phenotype analysis of SMAD6 mutant patients with radioulnar synostosis. Molecular Genetics & Genomic Medicine, 2022, 10, e1850.	1.2	6
3	MECOM-related disorder: Radioulnar synostosis without hematological aberration due to unique variants. Genetics in Medicine, 2022, 24, 1139-1147.	2.4	9
4	Insulin alleviates LPS-induced ARDS via inhibiting CUL4B-mediated proteasomal degradation and restoring expression level of Na,K-ATPase Î±1 subunit through elevating HCF-1. Biochemical and Biophysical Research Communications, 2022, 611, 60-67.	2.1	1
5	Mutant B3GALT6 in a Multiplex Family: A Dominant Variant Co-Segregated With Moderate Malformations. Frontiers in Genetics, 2022, 13, .	2.3	2
6	Disorder of Sexual Development Males With XYY in Blood Have Exactly X/XY/XYY Mosaicism in Gonad Tissues. Frontiers in Genetics, 2021, 12, 616693.	2.3	1
7	Expanding the genotypes and phenotypes for 19 rare diseases by exome sequencing performed in pediatric intensive care unit. Human Mutation, 2021, 42, 1443-1460.	2.5	4
8	Combined surgery with 3-in-1 osteosynthesis in congenital pseudarthrosis of the tibia with intact fibula. Orphanet Journal of Rare Diseases, 2020, 15, 62.	2.7	14
9	Genetic and Clinical Analyses of 13 Chinese Families With Cystine Urolithiasis and Identification of 15 Novel Pathogenic Variants in SLC3A1 and SLC7A9. Frontiers in Genetics, 2020, 11, 74.	2.3	4
10	Capillary Malformationâ€“Arteriovenous Malformation Combined Alagille Syndrome in a Patient With Double Gene Variations of RASA1 and NOTCH2. Frontiers in Genetics, 2019, 10, 1088.	2.3	3
11	Identification and characterization of NF1 and non-NF1 congenital pseudarthrosis of the tibia based on germline NF1 variants: genetic and clinical analysis of 75 patients. Orphanet Journal of Rare Diseases, 2019, 14, 221.	2.7	23
12	SMAD6 is frequently mutated in nonsyndromic radioulnar synostosis. Genetics in Medicine, 2019, 21, 2577-2585.	2.4	22
13	CLCN7 and TCIRG1 mutations in a single family: Evidence for digenic inheritance of osteopetrosis. Molecular Medicine Reports, 2019, 19, 595-600.	2.4	3
14	Identification of ANKDD1B variants in an ankylosing spondylitis pedigree and a sporadic patient. BMC Medical Genetics, 2018, 19, 111.	2.1	7
15	Novel GATAD2B loss-of-function mutations cause intellectual disability in two unrelated cases. Journal of Human Genetics, 2017, 62, 513-516.	2.3	12
16	A novel NHS mutation causes Nance-Horan Syndrome in a Chinese family. BMC Medical Genetics, 2017, 18, 2.	2.1	12
17	WDR73 missense mutation causes infantile onset intellectual disability and cerebellar hypoplasia in a consanguineous family. Clinica Chimica Acta, 2017, 464, 24-29.	1.1	10
18	AKAP2identified as a novel gene mutated in a Chinese family with adolescent idiopathic scoliosis. Journal of Medical Genetics, 2016, 53, 488-493.	3.2	43

#	Article	IF	Citations
19	Novel SIL1 nonstop mutation in a Chinese consanguineous family with Marinesco-SjÃ¶gren syndrome and Dandy-Walker syndrome. Clinica Chimica Acta, 2016, 458, 1-4.	1.1	11
20	A novel de novo POGZ mutation in a patient with intellectual disability. Journal of Human Genetics, 2016, 61, 357-359.	2.3	31
21	GLA variation p.E66Q identified as the genetic etiology of Fabry disease using exome sequencing. Gene, 2016, 575, 363-367.	2.2	12
22	Mutations of P4HA2 encoding prolyl 4-hydroxylase 2 are associated with nonsyndromic high myopia. Genetics in Medicine, 2015, 17, 300-306.	2.4	63
23	Noninvasive Prenatal Testing for Wilson Disease by Use of Circulating Single-Molecule Amplification and Resequencing Technology (cSMART). Clinical Chemistry, 2015, 61, 172-181.	3.2	85