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	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
2	Mutations of P4HA2 encoding prolyl 4-hydroxylase 2 are associated with nonsyndromic high myopia. Genetics in Medicine, 2015, 17, 300-306.	2.4	63
3	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
4	A novel de novo POGZ mutation in a patient with intellectual disability. Journal of Human Genetics, 2016, 61, 357-359.	2.3	31
5	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
6	SMAD6 is frequently mutated in nonsyndromic radioulnar synostosis. Genetics in Medicine, 2019, 21, 2577-2585.	2.4	22
7	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
8	GLA variation p.E66Q identified as the genetic etiology of Fabry disease using exome sequencing. Gene, 2016, 575, 363-367.	2.2	12
9	Novel GATAD2B loss-of-function mutations cause intellectual disability in two unrelated cases. Journal of Human Genetics, 2017, 62, 513-516.	2.3	12
10	A novel NHS mutation causes Nance-Horan Syndrome in a Chinese family. BMC Medical Genetics, 2017, 18, 2.	2.1	12
11	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
12	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
13	MECOM-related disorder: Radioulnar synostosis without hematological aberration due to unique variants. Genetics in Medicine, 2022, 24, 1139-1147.	2.4	9
14	Identification of ANKDD1B variants in an ankylosing spondylitis pedigree and a sporadic patient. BMC Medical Genetics, 2018, 19, 111.	2.1	7
15	A genotype and phenotype analysis of SMAD6 mutant patients with radioulnar synostosis. Molecular Genetics & Genomic Medicine, 2022, 10, e1850.	1.2	6
16	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
17	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
18	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

#	Article	IF	Citations
19	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
20	CLCN7 and TCIRG1 mutations in a single family: Evidence for digenic inheritance of osteopetrosis. Molecular Medicine Reports, 2019, 19, 595-600.	2.4	3
21	Mutant B3GALT6 in a Multiplex Family: A Dominant Variant Co-Segregated With Moderate Malformations. Frontiers in Genetics, 2022, 13, .	2.3	2
22	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
23	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