Ying Peng

List of Publications by Year
in descending order

17 papers	259 citations	1307594 7 h-index	940533 16 g-index
18 all docs	18 docs citations	18 times ranked	514 citing authors

#	Article	IF	Citations
1	Clinical and genetic analysis of classical Ehlersâ€Ðanlos syndrome patient caused by synonymous mutation in COL5A2. Molecular Genetics & Genomic Medicine, 2021, 9, e1632.	1.2	4
2	Implementation of fragile X syndrome carrier screening during prenatal diagnosis: A pilot study at a single center. Molecular Genetics & Genomic Medicine, 2021, 9, e1711.	1.2	2
3	Prenatal case of Simpsonâ€“Golabiâ€“Behmel syndrome with a de novo 370Kbâ€sized microdeletion of Xq26.2 compassing partial GPC3 gene and review. Molecular Genetics & Genomic Medicine, 2021, 9, e1750.	1.2	4
4	Whole Exome Sequencing Analysis in Fetal Skeletal Dysplasia Detected by Ultrasonography: An Analysis of 38 Cases. Frontiers in Genetics, 2021, 12, 728544.	2.3	15
5	Whole genome sequencing reveals translocation breakpoints disrupting TP63 gene underlying split hand/foot malformation in a Chinese family. Molecular Genetics & Genomic Medicine, 2021, 9, e1604.	1.2	4
6	Identification of a novel gross deletion of TCOF1 in a Chinese prenatal case with Treacher Collins syndrome. Molecular Genetics & Genomic Medicine, 2020, 8, e1313.	1.2	9
7	Clinical and molecular characterization of 12 prenatal cases of Criâ€duâ€chat syndrome. Molecular Genetics & Genomic Medicine, 2020, 8, e1312.	1.2	10
8	De novo exonic deletion of KDM6A in a Chinese girl with Kabuki syndrome: A case report and brief literature review. American Journal of Medical Genetics, Part A, 2016, 170, 1613-1621.	1.2	19
9	Three novel mutations of STK11 gene in Chinese patients with Peutzâ€“Jeghers syndrome. BMC Medical Genetics, 2016, 17, 77.	2.1	8
10	De Novo ring chromosome 11 and non-reciprocal translocation of 11p15.3-pter to 21qter in a patient with congenital heart disease. Molecular Cytogenetics, 2015, 8, 88.	0.9	4
11	Rare intracranial cholesterol deposition and a homozygous mutation of LDLR in a familial hypercholesterolemia patient. Gene, 2015, 569, 313-317.	2.2	5
12	PMP22-Related neuropathies and other clinical manifestations in Chinese han patients with charcot-marie-tooth disease type 1. Muscle and Nerve, 2015, 52, 69-75.	2.2	5
13	Novel de novo nonsense mutation of the PHEX gene (p.Lys50Ter) in a Chinese patient with hypophosphatemic rickets. Gene, 2015, 565, 150-154.	2.2	4
14	Partial trisomy 2q33.3-q37.3 in a patient with an inverted duplicated neocentric marker chromosome. Molecular Cytogenetics, 2015, 8, 10.	0.9	4
15	DMD mutation spectrum analysis in 613 Chinese patients with dystrophinopathy. Journal of Human Genetics, 2015, 60, 435-442.	2.3	47
16	Pachygyria, seizures, hypotonia, and growth retardation in a patient with an atypical 1.33Mb inherited microduplication at 22q11.23. Gene, 2015, 569, 46-50.	2.2	9
17	Copy Number Variation Sequencing for Comprehensive Diagnosis of Chromosome Disease Syndromes. Journal of Molecular Diagnostics, 2014, 16, 519-526.	2.8	106