

Panlai Shi

List of Publications by Year in descending order

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papers

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1684188

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times ranked

26
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutation analysis in the <i>F8</i> gene in 485 families with haemophilia A and prenatal diagnosis in China. <i>Haemophilia</i> , 2021, 27, e88-e92.	2.1	8
2	Usefulness of copy number variant detection following monogenic disease exclusion in prenatal diagnosis. <i>Journal of Obstetrics and Gynaecology Research</i> , 2021, 47, 1002-1008.	1.3	1
3	Prenatal and postnatal diagnoses and phenotype of 8p23.3p22 duplication in one family. <i>BMC Medical Genomics</i> , 2021, 14, 88.	1.5	1
4	Incorporation of exome-based CNV analysis makes trio-WES a more powerful tool for clinical diagnosis in neurodevelopmental disorders: A retrospective study. <i>Human Mutation</i> , 2021, 42, 990-1004.	2.5	25
5	Hermansky-Pudlak syndrome: Five Chinese patients with novel variants in HPS1 and HPS6. <i>European Journal of Medical Genetics</i> , 2021, 64, 104228.	1.3	2
6	The potential of expanded noninvasive prenatal screening for detection of microdeletion and microduplication syndromes. <i>Prenatal Diagnosis</i> , 2021, 41, 1332-1342.	2.3	19
7	Novel mutations of the CYP17A1 gene in four Chinese 46,XX cases with partial 17 α -hydroxylase/17,20-lyase deficiency. <i>Steroids</i> , 2021, 173, 108873.	1.8	8
8	Novel Partial Exon 51 Deletion in the Duchenne Muscular Dystrophy Gene Identified via Whole Exome Sequencing and Long-Read Whole-Genome Sequencing. <i>Frontiers in Genetics</i> , 2021, 12, 762987.	2.3	4
9	Influence of validating the parental origin on the clinical interpretation of fetal copy number variations in 141 core family cases. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e00944.	1.2	9