Panlai Shi

List of Publications by Year
in descending order

9	77	1684188 5	1588992 8
papers	citations	h-index	g-index

12	12	12	26
all docs	docs citations	times ranked	citing authors

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