

Xiaomei Luo

List of Publications by Year in descending order

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Version: 2024-02-01

13
papers

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1163117

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330
citing authors

#	ARTICLE	IF	CITATIONS
1	Genetic Diagnostic Evaluation of Trio-Based Whole Exome Sequencing Among Children With Diagnosed or Suspected Autism Spectrum Disorder. <i>Frontiers in Genetics</i> , 2018, 9, 594.	2.3	25
2	A 29 Mainland Chinese cohort of patients with Phelanâ€“McDermid syndrome: genotypeâ€“phenotype correlations and the role of SHANK3 haploinsufficiency in the important phenotypes. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 335.	2.7	25
3	A pilot study of expanded newborn screening for 573 genes related to severe inherited disorders in China: results from 1,127 newborns. <i>Annals of Translational Medicine</i> , 2020, 8, 1058-1058.	1.7	21
4	Next-generation sequencing as a second-tier diagnostic test for newborn screening. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2018, 31, 927-931.	0.9	20
5	High Detection Rate of Copy Number Variations Using Capture Sequencing Data: A Retrospective Study. <i>Clinical Chemistry</i> , 2020, 66, 455-462.	3.2	16
6	Novel PYGL mutations in Chinese children leading to glycogen storage disease type VI: two case reports. <i>BMC Medical Genetics</i> , 2020, 21, 74.	2.1	12
7	Clinical, genetic profile and therapy evaluation of 55 children and 5 adults with sitosterolemia. <i>Journal of Clinical Lipidology</i> , 2022, 16, 40-51.	1.5	10
8	Breast tissue ablation with irreversible electroporation in rabbits: A safety and feasibility study. <i>PLoS ONE</i> , 2017, 12, e0181555.	2.5	9
9	The Effects of Irreversible Electroporation on the Colon in a Porcine Model. <i>PLoS ONE</i> , 2016, 11, e0167275.	2.5	9
10	Identification of RUNX2 variants associated with cleidocranial dysplasia. <i>Hereditas</i> , 2019, 156, 31.	1.4	8
11	Chromosomal microarray analysis in fetuses with high-risk prenatal indications: A retrospective study in China. <i>Taiwanese Journal of Obstetrics and Gynecology</i> , 2021, 60, 299-304.	1.3	7
12	The Safety of Irreversible Electroporation on Nerves Adjacent to Treated Tumors. <i>World Neurosurgery</i> , 2017, 108, 642-649.	1.3	4
13	Diagnosis and followâ€“up of glycogen storage disease (GSD) type VI from the largest GSD center in China. <i>Human Mutation</i> , 2022, 43, 557-567.	2.5	2