Zhimei Liu

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/7369164/publications.pdf

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17 papers	215 citations	7 h-index	1058333 14 g-index
20	20	20	181
all docs	docs citations	times ranked	citing authors

#	Article	IF	CITATIONS
1	eRAM: encyclopedia of rare disease annotations for precision medicine. Nucleic Acids Research, 2018, 46, D937-D943.	6.5	56
2	The clinical and genetic characteristics in children with mitochondrial disease in China. Science China Life Sciences, 2017, 60, 746-757.	2.3	32
3	PedAM: a database for Pediatric Disease Annotation and Medicine. Nucleic Acids Research, 2018, 46, D977-D983.	6.5	27
4	Whole exome sequencing identifies a novel homozygous MECR mutation in a Chinese patient with childhood-onset dystonia and basal ganglia abnormalities, without optic atrophy. Mitochondrion, 2021, 57, 222-229.	1.6	13
5	Clinical Assessments and EEG Analyses of Encephalopathies Associated With Dynamin-1 Mutation. Frontiers in Pharmacology, 2019, 10, 1454.	1.6	11
6	Novel ECHS1 mutations in Leigh syndrome identified by whole-exome sequencing in five Chinese families: case report. BMC Medical Genetics, 2020, 21, 149.	2.1	10
7	Report of the Largest Chinese Cohort With SLC19A3 Gene Defect and Literature Review. Frontiers in Genetics, 2021, 12, 683255.	1.1	10
8	Leigh Syndrome: A Study of 209 Patients at the Beijing Children's Hospital. Annals of Neurology, 2022, 91, 466-482.	2.8	10
9	Compound Heterozygous CHAT Gene Mutations of a Large Deletion and a Missense Variant in a Chinese Patient With Severe Congenital Myasthenic Syndrome With Episodic Apnea. Frontiers in Pharmacology, 2019, 10, 259.	1.6	9
10	Biallelic COA7-Variants Leading to Developmental Regression With Progressive Spasticity and Brain Atrophy in a Chinese Patient. Frontiers in Genetics, 2021, 12, 685035.	1.1	7
11	Identification and characterization of novel <scp><i>MPC1</i></scp> gene variants causing mitochondrial pyruvate carrier deficiency. Journal of Inherited Metabolic Disease, 2022, 45, 264-277.	1.7	7
12	Phenotypes and genotypes of mitochondrial diseases with mtDNA variations in Chinese children: A multi-center study. Mitochondrion, 2022, 62, 139-150.	1.6	6
13	Whole genome and exome sequencing identify <i>NDUFV2</i> mutations as a new cause of progressive cavitating leukoencephalopathy. Journal of Medical Genetics, 2022, 59, 351-357.	1.5	5
14	Cinical, Metabolic, and Genetic Analysis and Follow-Up of Eight Patients With HIBCH Mutations Presenting With Leigh/Leigh-Like Syndrome. Frontiers in Pharmacology, 2021, 12, 605803.	1.6	4
15	Identification of a Novel m.3955G>A Variant in MT-ND1 Associated with Leigh Syndrome. Mitochondrion, 2021, 62, 13-23.	1.6	3
16	Identification of a Novel Variant in MT-CO3 Causing MELAS. Frontiers in Genetics, 2021, 12, 638749.	1.1	2
17	Generation of an iPSC line from a patient with early-onset epileptic encephalopathy carrying CARS2 (p.G476R) mutation. Stem Cell Research, 2022, 59, 102633.	0.3	O