The clinical and genetic characteristics in children with

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Citation Report

#	Article	IF	Citations
1	eRAM: encyclopedia of rare disease annotations for precision medicine. Nucleic Acids Research, 2018, 46, D937-D943.	6.5	56
2	Compound heterozygous missense and deep intronic variants in NDUFAF6 unraveled by exome sequencing and mRNA analysis. Journal of Human Genetics, 2018, 63, 563-568.	1.1	15
3	Clinical, Neuroimaging, and Pathological Analyses of 13 Chinese Leigh Syndrome Patients with Mitochondrial DNA Mutations. Chinese Medical Journal, 2018, 131, 2705-2712.	0.9	10
4	PedAM: a database for Pediatric Disease Annotation and Medicine. Nucleic Acids Research, 2018, 46, D977-D983.	6.5	27
5	Molecular Genetics Analysis of 70 Chinese Families With Muscular Dystrophy Using Multiplex Ligation-Dependent Probe Amplification and Next-Generation Sequencing. Frontiers in Pharmacology, 2019, 10, 814.	1.6	12
6	Next-Generation Sequencing Analysis Reveals Novel Pathogenic Variants in Four Chinese Siblings With Late-Infantile Neuronal Ceroid Lipofuscinosis. Frontiers in Genetics, 2019, 10, 370.	1.1	10
7	Transition from Leigh syndrome to MELAS syndrome in a patient with heteroplasmic MT-ND3 m.10158T>C. Brain and Development, 2019, 41, 803-807.	0.6	11
8	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
9	Clinical Assessments and EEG Analyses of Encephalopathies Associated With Dynamin-1 Mutation. Frontiers in Pharmacology, 2019, 10, 1454.	1.6	11
10	Mutations in the mitochondrial complex I assembly factor NDUFAF6 cause isolated bilateral striatal necrosis and progressive dystonia in childhood. Molecular Genetics and Metabolism, 2019, 126, 250-258.	0.5	20
11	Phenotype-Driven Virtual Panel Is an Effective Method to Analyze WES Data of Neurological Disease. Frontiers in Pharmacology, 2018, 9, 1529.	1.6	15
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14	Clinical and molecular characterization of pediatric mitochondrial disorders in south of China. European Journal of Medical Genetics, 2020, 63, 103898.	0.7	26
15	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
16	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
17	Mitochondrial Protein Translation: Emerging Roles and Clinical Significance in Disease. Frontiers in Cell and Developmental Biology, 2021, 9, 675465.	1.8	39
18	Phenotypes and genotypes of mitochondrial diseases with mtDNA variations in Chinese children: A multi-center study. Mitochondrion, 2022, 62, 139-150.	1.6	6

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19	NDUFV1 mutations in complex I deficiency: Case reports and review of symptoms. Genetics and Molecular Biology, 2021, 44, e20210149.	0.6	4
20	Evaluation of the Genetically Diagnosed Mitochondrial Disease Cases with Neuromuscular Involvement. Journal of Dr Behcet Uz Children S Hospital, 2022, 12, 27-36.	0.1	0
27	Phenotype-Genotype Analysis Based on Molecular Classification in 135 Children With Mitochondrial Disease. Pediatric Neurology, 2022, 132, 11-18.	1.0	4
28	NDUFAF6-Related Leigh Syndrome Caused by Rare Pathogenic Variants: A Case Report and the Focused Review of Literature. Frontiers in Pediatrics, 2022, 10, .	0.9	2
29	The genetic and phenotypic spectra of adult genetic leukoencephalopathies in a cohort of 309 patients. Brain, 2023, 146, 2364-2376.	3.7	10
30	Use of dual genomic sequencing to screen mitochondrial diseases in pediatrics: a retrospective analysis. Scientific Reports, 2023, 13, .	1.6	0
31	Phenotyping <scp>mtDNA</scp> â€related diseases in childhood: a cohort study of 150 patients. European Journal of Neurology, 0, , .	1.7	1