## Atsuko Imai-Okazaki

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/5992668/publications.pdf Version: 2024-02-01



ATSUKO IMALOKAZAKI

#	Article	IF	CITATIONS
1	Cardiomyopathy in children with mitochondrial disease: Prognosis and genetic background. International Journal of Cardiology, 2019, 279, 115-121.	1.7	35
2	Fatal Perinatal Mitochondrial Cardiac Failure Caused by Recurrent De Novo Duplications in the ATAD3 Locus. Med, 2021, 2, 49-73.e10.	4.4	33
3	Mortality of Japanese patients with Leigh syndrome: Effects of age at onset and genetic diagnosis. Journal of Inherited Metabolic Disease, 2020, 43, 819-826.	3.6	32
4	Barth Syndrome: Different Approaches to Diagnosis. Journal of Pediatrics, 2018, 193, 256-260.	1.8	14
5	Long-term prognosis and genetic background of cardiomyopathy in 223 pediatric mitochondrial disease patients. International Journal of Cardiology, 2021, 341, 48-55.	1.7	14
6	A novel homozygous variant in <i>MICOS13</i> / <i>QIL1</i> causes hepatoâ€encephalopathy with mitochondrial DNA depletion syndrome. Molecular Genetics & Genomic Medicine, 2020, 8, e1427.	1.2	12
7	Neonatal-onset mitochondrial disease: clinical features, molecular diagnosis and prognosis. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2022, 107, 329-334.	2.8	9
8	NAD(P)HX dehydratase protein-truncating mutations are associated with neurodevelopmental disorder exacerbated by acute illness. Brain, 2020, 143, e54-e54.	7.6	7
9	HDR-del: A tool based on Hamming distance for prioritizing pathogenic chromosomal deletions in exome sequencing. Human Mutation, 2017, 38, 1796-1800.	2.5	6
10	Heterozygosity mapping for human dominant trait variants. Human Mutation, 2019, 40, 996-1004.	2.5	4
11	Development of Leigh syndrome with a high probability of cardiac manifestations in infantile-onset patients with m.14453GÂ>ÂA. Mitochondrion, 2022, 63, 1-8.	3.4	2
12	Clinical heterogeneity in patients with m.4412G > A MT-TM mutation and different heteroplasmy levels. Mitochondrion, 2021, 59, 214-215.	3.4	0