

Omar Hikmat

List of Publications by Year in descending order

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papers

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1307594

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#	ARTICLE	IF	CITATIONS
1	Phenotypic spectrum and clinical course of single large-scale mitochondrial DNA deletion disease in the paediatric population: a multicentre study. <i>Journal of Medical Genetics</i> , 2023, 60, 65-73.	3.2	2
2	Renal Phenotype in Mitochondrial Diseases: A Multicenter Study. <i>Kidney Diseases (Basel, Switzerland)</i> , 2022, 8, 148-159.	2.5	3
3	Diagnostic value of serum biomarkers <scp>FGF21</scp> and <scp>GDF15</scp> compared to muscle sample in mitochondrial disease. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 469-480.	3.6	34
4	Serum biomarkers in primary mitochondrial disorders. <i>Brain Communications</i> , 2021, 3, fcaa222.	3.3	14
5	A characteristic occipital epileptiform EEG pattern in ADCK3-related mitochondrial disease. <i>Epileptic Disorders</i> , 2021, 23, 281-290.	1.3	1
6	Expanding the phenotypic spectrum of <i>BCS1L</i>-related mitochondrial disease. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 2155-2165.	3.7	11
7	Simplifying the clinical classification of polymerase gamma (POLG) disease based on age of onset; studies using a cohort of 155 cases. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 726-736.	3.6	33
8	Mental health and health related quality of life in mitochondrial POLG disease. <i>Mitochondrion</i> , 2020, 55, 95-99.	3.4	4
9	The impact of gender, puberty, and pregnancy in patients with POLG disease. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 2019-2025.	3.7	7
10	Diseases of DNA Polymerase Gamma. , 2019, , 113-124.		0
11	Elevated cerebrospinal fluid protein in <i>POLG</i>-related epilepsy: Diagnostic and prognostic implications. <i>Epilepsia</i> , 2018, 59, 1595-1602.	5.1	6
12	The clinical spectrum and natural history of early-onset diseases due to DNA polymerase gamma mutations. <i>Genetics in Medicine</i> , 2017, 19, 1217-1225.	2.4	45
13	The presence of anaemia negatively influences survival in patients with POLG disease. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 861-866.	3.6	8
14	Progressive deafness-dystonia due to <i>SERAC1</i> mutations: A study of 67 cases. <i>Annals of Neurology</i> , 2017, 82, 1004-1015.	5.3	63
15	Understanding the Epilepsy in POLG Related Disease. <i>International Journal of Molecular Sciences</i> , 2017, 18, 1845.	4.1	30