

Kalliopi Sofou

List of Publications by Year in descending order

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Version: 2024-02-01

23
papers

823
citations

687363

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642732

23
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24
all docs

24
docs citations

24
times ranked

1527
citing authors

#	ARTICLE	IF	CITATIONS
1	A multicenter study on Leigh syndrome: disease course and predictors of survival. Orphanet Journal of Rare Diseases, 2014, 9, 52.	2.7	182
2	Ketogenic diet in pyruvate dehydrogenase complex deficiency: short- and long-term outcomes. Journal of Inherited Metabolic Disease, 2017, 40, 237-245.	3.6	138
3	Whole exome sequencing reveals mutations in <i>NARS2</i> and <i>PARS2</i> , encoding the mitochondrial asparaginyl-tRNA synthetase and prolyl-tRNA synthetase, in patients with Alpers syndrome. Molecular Genetics & Genomic Medicine, 2015, 3, 59-68.	1.2	87
4	KIF1A variants are a frequent cause of autosomal dominant hereditary spastic paraplegia. European Journal of Human Genetics, 2020, 28, 40-49.	2.8	65
5	Phenotype-genotype correlations in Leigh syndrome: new insights from a multicentre study of 96 patients. Journal of Medical Genetics, 2018, 55, 21-27.	3.2	54
6	Management of Prolonged Seizures and Status Epilepticus in Childhood: A Systematic Review. Journal of Child Neurology, 2009, 24, 918-926.	1.4	53
7	MRI of the brain in childhood-onset mitochondrial disorders with central nervous system involvement. Mitochondrion, 2013, 13, 364-371.	3.4	40
8	Diagnostic value of serum biomarkers <i>FGF21</i> and <i>GDF15</i> compared to muscle sample in mitochondrial disease. Journal of Inherited Metabolic Disease, 2021, 44, 469-480.	3.6	34
9	Broad phenotypic variability in patients with complex I deficiency due to mutations in <i>NDUFS1</i> and <i>NDUFV1</i> . Mitochondrion, 2015, 21, 33-40.	3.4	30
10	Phenotypic and genotypic variability in Alpers syndrome. European Journal of Paediatric Neurology, 2012, 16, 379-389.	1.6	22
11	Mitochondrial complex IV deficiency caused by a novel frameshift variant in <i>MT-CO2</i> associated with myopathy and perturbed acylcarnitine profile. European Journal of Human Genetics, 2019, 27, 331-335.	2.8	17
12	One in five patients with Duchenne muscular dystrophy dies from other causes than cardiac or respiratory failure. European Journal of Epidemiology, 2022, 37, 147-156.	5.7	16
13	The phenotypic variability and natural history of <i>NARS2</i> associated disease. European Journal of Paediatric Neurology, 2021, 31, 31-37.	1.6	14
14	Ophthalmological characteristics in children with Leigh syndrome – A long-term follow-up. Acta Ophthalmologica, 2016, 94, 609-617.	1.1	13
15	Mitochondrial Disease. Journal of Child Neurology, 2013, 28, 663-667.	1.4	12
16	Cerebrospinal fluid neurofilament light is associated with survival in mitochondrial disease patients. Mitochondrion, 2019, 46, 228-235.	3.4	10
17	Novel imaging findings in pyruvate dehydrogenase complex (<i>PDHc</i>) deficiency – Results from a nationwide population-based study. Journal of Inherited Metabolic Disease, 2022, 45, 248-263.	3.6	9
18	Long term treatment with ataluren – the Swedish experience. BMC Musculoskeletal Disorders, 2021, 22, 837.	1.9	7

#	ARTICLE	IF	CITATIONS
19	Parental experiences of having a child with CLN3 disease (juvenile Batten disease) and how these experiences relate to family resilience. <i>Child: Care, Health and Development</i> , 2022, 48, 842-851.	1.7	6
20	SITUPS : a new tool for oral case presentation in the paediatric emergency department. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2019, 108, 1467-1474.	1.5	3
21	Renal Phenotype in Mitochondrial Diseases: A Multicenter Study. <i>Kidney Diseases (Basel, Switzerland)</i> , 2022, 8, 148-159.	2.5	3
22	Ophthalmologic involvement in Leigh syndrome. <i>Acta Ophthalmologica</i> , 2017, 95, e76.	1.1	1
23	Responding to feedback on the <scp>SITUPS</scp> mnemonic for medical students. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2020, 109, 210-210.	1.5	0