Kalliopi Sofou

List of Publications by Year
in descending order

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23 papers	823 citations	687363 13 h-index	642732 23 g-index
24 all docs	24 docs citations	24 times ranked	1527 citing authors

#	Article	IF	Citations
1	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
2	Renal Phenotype in Mitochondrial Diseases: A Multicenter Study. Kidney Diseases (Basel, Switzerland), 2022, 8, 148-159.	2.5	3
3	Parental experiences of having a child with CLN3 disease (juvenile Batten disease) and how these experiences relate to family resilience. Child: Care, Health and Development, 2022, 48, 842-851.	1.7	6
4	Novel imaging findings in pyruvate dehydrogenase complex (<scp>PDHc</scp>) deficiencyâ€"Results from a nationwide populationâ€based study. Journal of Inherited Metabolic Disease, 2022, 45, 248-263.	3.6	9
5	Diagnostic value of serum biomarkers <scp>FGF21</scp> and <scp>GDF15</scp> compared to muscle sample in mitochondrial disease. Journal of Inherited Metabolic Disease, 2021, 44, 469-480.	3.6	34
6	The phenotypic variability and natural history of NARS2 associated disease. European Journal of Paediatric Neurology, 2021, 31, 31-37.	1.6	14
7	Long term treatment with atalurenâ€"the Swedish experience. BMC Musculoskeletal Disorders, 2021, 22, 837.	1.9	7
8	Responding to feedback on the <scp>SITUPS</scp> mnemonic for medical students. Acta Paediatrica, International Journal of Paediatrics, 2020, 109, 210-210.	1.5	0
9	KIF1A variants are a frequent cause of autosomal dominant hereditary spastic paraplegia. European Journal of Human Genetics, 2020, 28, 40-49.	2.8	65
10	Cerebrospinal fluid neurofilament light is associated with survival in mitochondrial disease patients. Mitochondrion, 2019, 46, 228-235.	3.4	10
11	Mitochondrial complex IV deficiency caused by a novel frameshift variant in MT-CO2 associated with myopathy and perturbed acylcarnitine profile. European Journal of Human Genetics, 2019, 27, 331-335.	2.8	17
12	SITUPS : a new tool for oral case presentation in the paediatric emergency department. Acta Paediatrica, International Journal of Paediatrics, 2019, 108, 1467-1474.	1.5	3
13	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
14	Ophthalmologic involvement in Leigh syndrome. Acta Ophthalmologica, 2017, 95, e76.	1.1	1
15	Ketogenic diet in pyruvate dehydrogenase complex deficiency: shortâ€and longâ€term outcomes. Journal of Inherited Metabolic Disease, 2017, 40, 237-245.	3.6	138
16	Ophthalmological characteristics in children with Leigh syndrome â€“ A longâ€term followâ€up. Acta Ophthalmologica, 2016, 94, 609-617.	1.1	13
17	Broad phenotypic variability in patients with complex I deficiency due to mutations in NDUFS1 and NDUFV1. Mitochondrion, 2015, 21, 33-40.	3.4	30
18	Whole exome sequencing reveals mutations inNARS2andPARS2, 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

#	Article	IF	Citations
19	A multicenter study on Leigh syndrome: disease course and predictors of survival. Orphanet Journal of Rare Diseases, 2014, 9, 52.	2.7	182
20	MRI of the brain in childhood-onset mitochondrial disorders with central nervous system involvement. Mitochondrion, 2013, 13, 364-371.	3.4	40
21	Mitochondrial Disease. Journal of Child Neurology, 2013, 28, 663-667.	1.4	12
22	Phenotypic and genotypic variability in Alpers syndrome. European Journal of Paediatric Neurology, 2012, 16, 379-389.	1.6	22
23	Management of Prolonged Seizures and Status Epilepticus in Childhood: A Systematic Review. Journal of Child Neurology, 2009, 24, 918-926.	1.4	53