Gittan Kollberg

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/1545765/publications.pdf

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19	707	623734	713466
papers	citations	14 h-index	21 g-index

21	21	21	1248
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Phenotypic spectrum and clinical course of single large-scale mitochondrial DNA deletion disease in the paediatric population: a multicentre study. Journal of Medical Genetics, 2023, 60, 65-73.	3.2	2
2	The phenotypic variability and natural history of NARS2 associated disease. European Journal of Paediatric Neurology, 2021, 31, 31-37.	1.6	14
3	Functional analysis of a novel POLÎ³A mutation associated with a severe perinatal mitochondrial encephalomyopathy. Neuromuscular Disorders, 2021, 31, 348-358.	0.6	2
4	Expression pattern of mitochondrial respiratory chain enzymes in skeletal muscle of patients with mitochondrial myopathy associated with the homoplasmic m.14674T>C variant. Brain Pathology, 2021, , e13038.	4.1	4
5	TANGO2 deficiency as a cause of neurodevelopmental delay with indirect effects on mitochondrial energy metabolism. Journal of Inherited Metabolic Disease, 2019, 42, 898-908.	3.6	32
6	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
7	Identification of a large intronic transposal insertion in SLC17A5 causing sialic acid storage disease. Orphanet Journal of Rare Diseases, 2017, 12, 28.	2.7	14
8	Leukoencephalopathy due to Complex II Deficiency and Bi-Allelic SDHB Mutations: Further Cases and Implications for Genetic Counselling. JIMD Reports, 2016, 33, 69-77.	1.5	17
9	Broad phenotypic variability in patients with complex I deficiency due to mutations in NDUFS1 and NDUFV1. Mitochondrion, 2015, 21, 33-40.	3.4	30
10	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
11	Phenotypic and genotypic variability in Alpers syndrome. European Journal of Paediatric Neurology, 2012, 16, 379-389.	1.6	22
12	Transient restoration of succinate dehydrogenase activity after rhabdomyolysis in ironâ€“sulphur cluster deficiency myopathy. Neuromuscular Disorders, 2011, 21, 115-120.	0.6	20
13	Clinical manifestation and a new ISCU mutation in ironâ€“sulphur cluster deficiency myopathy. Brain, 2009, 132, 2170-2179.	7.6	91
14	Antisense oligonucleotide therapeutics for ironâ€“sulphur cluster deficiency myopathy. Neuromuscular Disorders, 2009, 19, 833-836.	0.6	17
15	Cardiomyopathy and Exercise Intolerance in Muscle Glycogen Storage Disease 0. New England Journal of Medicine, 2007, 357, 1507-1514.	27.0	123
16	POLG1Mutations Associated With Progressive Encephalopathy in Childhood. Journal of Neuropathology and Experimental Neurology, 2006, 65, 758-768.	1.7	86
17	Mitochondrial (Mt)Dna Changes in Tissue May Not be Reflected by Depletion of Mtdna in Peripheral Blood Mononuclear Cells in HIV-Infected Patients. Antiviral Therapy, 2006, 11, 601-608.	1.0	48
18	Low frequency of mtDNA point mutations in patients with PEO associated with POLG1 mutations. European Journal of Human Genetics, 2005, 13, 463-469.	2.8	32

#	Article	IF	Citations
19	Mitochondrial Myopathy and Rhabdomyolysis Associated with a Novel Nonsense Mutation in the Gene Encoding CytochromecOxidase Subunit I. Journal of Neuropathology and Experimental Neurology, 2005, 64, 123-128.	1.7	40