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	Cardiomyopathy and Exercise Intolerance in Muscle Glycogen Storage Disease 0. New England Journal of Medicine, 2007, 357, 1507-1514.	27.0	123
2	Clinical manifestation and a new ISCU mutation in ironâ€“sulphur cluster deficiency myopathy. Brain, 2009, 132, 2170-2179.	7.6	91
3	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
4	POLG1Mutations Associated With Progressive Encephalopathy in Childhood. Journal of Neuropathology and Experimental Neurology, 2006, 65, 758-768.	1.7	86
5	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
6	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
7	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
8	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
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	Phenotypic and genotypic variability in Alpers syndrome. European Journal of Paediatric Neurology, 2012, 16, 379-389.	1.6	22
11	Transient restoration of succinate dehydrogenase activity after rhabdomyolysis in ironâ€“sulphur cluster deficiency myopathy. Neuromuscular Disorders, 2011, 21, 115-120.	0.6	20
12	Antisense oligonucleotide therapeutics for ironâ€“sulphur cluster deficiency myopathy. Neuromuscular Disorders, 2009, 19, 833-836.	0.6	17
13	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
14	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
15	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
16	The phenotypic variability and natural history of NARS2 associated disease. European Journal of Paediatric Neurology, 2021, 31, 31-37.	1.6	14
17	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
18	Functional analysis of a novel POLÎ³A mutation associated with a severe perinatal mitochondrial encephalomyopathy. Neuromuscular Disorders, 2021, 31, 348-358.	0.6	2

#	Article	IF	Citations
19	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