Carl Fratter

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/1017347/publications.pdf

Version: 2024-02-01

20 papers	878 citations	471509 17 h-index	713466 21 g-index
22 all docs	22 docs citations	22 times ranked	3675 citing authors

#	Article	IF	Citations
1	OMA1 mediates local and global stress responses against protein misfolding in CHCHD10 mitochondrial myopathy. Journal of Clinical Investigation, 2022, 132, .	8.2	24
2	The natural history of infantile mitochondrial DNA depletion syndrome due to RRM2B deficiency. Genetics in Medicine, 2020, 22, 199-209.	2.4	14
3	EMQN best practice guidelines for genetic testing in dystrophinopathies. European Journal of Human Genetics, 2020, 28, 1141-1159.	2.8	35
4	Nuclear-mitochondrial DNA segments resemble paternally inherited mitochondrial DNA in humans. Nature Communications, 2020, 11, 1740.	12.8	75
5	Pathogenic variants in MTâ€ATP6: A United Kingdomâ€“based mitochondrial disease cohort study. Annals of Neurology, 2019, 86, 310-315.	5.3	33
6	Retrospective natural history of thymidine kinase 2 deficiency. Journal of Medical Genetics, 2018, 55, 515-521.	3.2	73
7	Rare NaV1.7 variants associated with painful diabetic peripheral neuropathy. Pain, 2018, 159, 469-480.	4.2	116
8	Bi-allelic Mutations in NDUFA6 Establish Its Role in Early-Onset Isolated Mitochondrial Complex I Deficiency. American Journal of Human Genetics, 2018, 103, 592-601.	6.2	41
9	De novo DNM1L mutation associated with mitochondrial epilepsy syndrome with fever sensitivity. Neurology: Genetics, 2018, 4, e258.	1.9	27
10	The Novel Activity of Carbamazepine as an Activation Modulator Extends from Na_V1.7 Mutations to the Na_V1.8-S242T Mutant Channel from a Patient with Painful Diabetic Neuropathy. Molecular Pharmacology, 2018, 94, 1256-1269.	2.3	24
11	Dysregulated mitophagy and mitochondrial organization in optic atrophy due to OPA1 mutations. Neurology, 2017, 88, 131-142.	1.1	81
12	Pigmentary retinopathy, rodâ€“cone dysfunction and sensorineural deafness associated with a rare mitochondrial tRNALys(m.8340G>A) gene variant. British Journal of Ophthalmology, 2017, 101, 1298-1302.	3.9	8
13	Use of FGF-21 as a Biomarker of Mitochondrial Disease in Clinical Practice. Journal of Clinical Medicine, 2017, 6, 80.	2.4	50
14	A Clinical, Neuropathological and Genetic Study of Homozygous A467T POLG-Related Mitochondrial Disease. PLoS ONE, 2016, 11, e0145500.	2.5	36
15	Clinical features of the pathogenic m.5540G>A mitochondrial transfer RNA tryptophan gene mutation. Neuromuscular Disorders, 2016, 26, 702-705.	0.6	6
16	The m.13051G>A mitochondrial DNA mutation results in variable neurology and activated mitophagy. Neurology, 2016, 86, 1921-1923.	1.1	35
17	A novel quantitative assay of mitophagy: Combining high content fluorescence microscopy and mitochondrial DNA load to quantify mitophagy and identify novel pharmacological tools against pathogenic heteroplasmic mtDNA. Pharmacological Research, 2015, 100, 24-35.	7.1	47
18	Unexplained gastrointestinal symptoms: Think mitochondrial disease. Digestive and Liver Disease, 2014, 46, 1-8.	0.9	29

#	Article	IF	Citations
19	Adults with RRM2B-related mitochondrial disease have distinct clinical and molecular characteristics. Brain, 2012, 135, 3392-3403.	7.6	70
20	Kearns-Sayre syndrome caused by defective R1/p53R2 assembly. Journal of Medical Genetics, 2011, 48, 610-617.	3.2	49