Christoph Freyer

List of Publications by Citations

42 papers	1,982 citations	20 h-index	43 g-index
43 ext. papers	2,378 ext. citations	11.4 avg, IF	4.27 L-index

#	Paper	IF	Citations
42	Strong purifying selection in transmission of mammalian mitochondrial DNA. *PLoS Biology*, **2008**, 6, e10	9.7	352
41	Germline mitochondrial DNA mutations aggravate ageing and can impair brain development. *Nature*, **2013**, 501, 412-5	50.4	188
40	Ultra-deep sequencing of mouse mitochondrial DNA: mutational patterns and their origins. *PLoS Genetics*, **2011**, 7, e1002028	6	136
39	Purifying selection of mtDNA and its implications for understanding evolution and mitochondrial disease. *Nature Reviews Genetics*, **2008**, 9, 657-62	30.1	129
38	Respiratory chain dysfunction in skeletal muscle does not cause insulin resistance. *Biochemical and Biophysical Research Communications*, **2006**, 350, 202-7	3.4	122
37	TWINKLE is an essential mitochondrial helicase required for synthesis of nascent D-loop strands and complete mtDNA replication. *Human Molecular Genetics*, **2013**, 22, 1983-93	5.6	114
36	Variation in germline mtDNA heteroplasmy is determined prenatally but modified during subsequent transmission. *Nature Genetics*, **2012**, 44, 1282-5	36.3	102
35	Neu-Laxova syndrome is a heterogeneous metabolic disorder caused by defects in enzymes of the L-serine biosynthesis pathway. *American Journal of Human Genetics*, **2014**, 95, 285-93	11	82
34	A Phenotype-Driven Approach to Generate Mouse Models with Pathogenic mtDNA Mutations Causing Mitochondrial Disease. *Cell Reports*, **2016**, 16, 2980-2990	10.6	72
33	MTERF3 regulates mitochondrial ribosome biogenesis in invertebrates and mammals. *PLoS Genetics* , **2013**, 9, e1003178	6	70
32	Absence of the Autophagy Adaptor SQSTM1/p62 Causes Childhood-Onset Neurodegeneration with Ataxia, Dystonia, and Gaze Palsy. *American Journal of Human Genetics*, **2016**, 99, 735-743	11	69
31	Rescue of primary ubiquinone deficiency due to a novel COQ7 defect using 2,4-dihydroxybensoic acid. *Journal of Medical Genetics*, **2015**, 52, 779-83	5.8	67
30	No recombination of mtDNA after heteroplasmy for 50 generations in the mouse maternal germline. *Nucleic Acids Research*, **2014**, 42, 1111-6	20.1	63
29	Regulation of DNA replication at the end of the mitochondrial D-loop involves the helicase TWINKLE and a conserved sequence element. *Nucleic Acids Research*, **2015**, 43, 9262-75	20.1	54
28	Intra-mitochondrial Methylation Deficiency Due to Mutations in SLC25A26. *American Journal of Human Genetics*, **2015**, 97, 761-8	11	44
27	MtDNA mutations are a common cause of severe disease phenotypes in children with Leigh syndrome. *Biochimica Et Biophysica Acta - Bioenergetics*, **2009**, 1787, 484-90	4.6	38
26	Maintenance of respiratory chain function in mouse hearts with severely impaired mtDNA transcription. *Nucleic Acids Research*, **2010**, 38, 6577-88	20.1	29

25	Mitochondrial Polyadenylation Is a One-Step Process Required for mRNA Integrity and tRNA Maturation. *PLoS Genetics*, **2016**, 12, e1006028	6	25
24	Metabolic regulation of neurodifferentiation in the adult brain. *Cellular and Molecular Life Sciences*, **2020**, 77, 2483-2496	10.3	22
23	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. *Genome Medicine*, **2021**, 13, 40	14.4	22
22	SQSTM1/p62-Directed Metabolic Reprogramming Is Essential for Normal Neurodifferentiation. *Stem Cell Reports*, **2019**, 12, 696-711	8	20
21	Defects of mitochondrial RNA turnover lead to the accumulation of double-stranded RNA in vivo. *PLoS Genetics*, **2019**, 15, e1008240	6	19
20	Respiratory chain complex III deficiency due to mutated BCS1L: a novel phenotype with encephalomyopathy, partially phenocopied in a Bcs1l mutant mouse model. *Orphanet Journal of Rare Diseases*, **2017**, 12, 73	4.2	18
19	Absence of TXNIP in Humans Leads to Lactic Acidosis and Low Serum Methionine Linked to Deficient Respiration on Pyruvate. *Diabetes*, **2019**, 68, 709-723	0.9	17
18	Detection of 6-demethoxyubiquinone in CoQ deficiency disorders: Insights into enzyme interactions and identification of potential therapeutics. *Molecular Genetics and Metabolism*, **2017**, 121, 216-223	3.7	15
17	SUV3 helicase is required for correct processing of mitochondrial transcripts. *Nucleic Acids Research* , **2015**, 43, 7398-413	20.1	15
16	C6orf203 is an RNA-binding protein involved in mitochondrial protein synthesis. *Nucleic Acids Research*, **2019**, 47, 9386-9399	20.1	14
15	Cyclophilin D, a target for counteracting skeletal muscle dysfunction in mitochondrial myopathy. *Human Molecular Genetics*, **2015**, 24, 6580-7	5.6	11
14	Is energy deficiency good in moderation?. *Cell*, **2007**, 131, 448-50	56.2	11
13	Myoglobinopathy is an adult-onset autosomal dominant myopathy with characteristic sarcoplasmic inclusions. *Nature Communications*, **2019**, 10, 1396	17.4	7
12	Chorea, psychosis, acanthocytosis, and prolonged survival associated with mutations. *Neurology*, **2018**, 91, 710-712	6.5	7
11	Mutations in the mitochondrial tryptophanyl-tRNA synthetase cause growth retardation and progressive leukoencephalopathy. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e654	2.3	6
10	The one-carbon pool controls mitochondrial energy metabolism via complex I and iron-sulfur clusters. *Science Advances*, **2021**, 7,	14.3	6
9	A multi-systemic mitochondrial disorder due to a dominant p.Y955H disease variant in DNA polymerase gamma. *Human Molecular Genetics*, **2017**, 26, 2515-2525	5.6	5
8	Protocol for the derivation, culturing, and differentiation of human iPS-cell-derived neuroepithelial stem cells to study neural differentiation in vitro. *STAR Protocols*, **2021**, 2, 100528	1.4	4

7	Severe congenital lactic acidosis and hypertrophic cardiomyopathy caused by an intronic variant in NDUFB7. *Human Mutation*, **2021**, 42, 378-384	4.7	3
6	Novel Mutation m.10372A>G in Causing Sensorimotor Axonal Polyneuropathy. *Neurology: Genetics*, **2021**, 7, e566	3.8	1
5	Clinical Presentation, Genetic Etiology, and Coenzyme Q10 Levels in 55 Children with Combined Enzyme Deficiencies of the Mitochondrial Respiratory Chain. *Journal of Pediatrics*, **2021**, 228, 240-251.e2	3.6	1
4	Stable Isotope Labeling of Amino Acids in Flies (SILAF) Reveals Differential Phosphorylation of Mitochondrial Proteins Upon Loss of OXPHOS Subunits. *Molecular and Cellular Proteomics*, **2021**, 20, 100065	7.6	1
3	Quantitative Proteomics in Drosophila with Holidic Stable-Isotope Labeling of Amino Acids in Fruit Flies (SILAF). *Methods in Molecular Biology*, **2021**, 2192, 75-87	1.4	1
2	Mitochondrial RNA Turnover in Metazoa. *Nucleic Acids and Molecular Biology*, **2018**, 17-46		0
1	J10 Chorea, psychotic symptoms and long survival in a subject with ELAC2 mutations. *Journal of Neurology, Neurosurgery and Psychiatry*, **2016**, 87, A78.3-A79	5.5	