

# Christoph Freyer

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	Novel Mutation m.10372A>G in Causing Sensorimotor Axonal Polyneuropathy. <i>Neurology: Genetics</i> , <b>2021</b> , 7, e566	3.8	1
41	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. <i>Genome Medicine</i> , <b>2021</b> , 13, 40	14.4	22
40	Protocol for the derivation, culturing, and differentiation of human iPS-cell-derived neuroepithelial stem cells to study neural differentiation in vitro. <i>STAR Protocols</i> , <b>2021</b> , 2, 100528	1.4	4
39	Clinical Presentation, Genetic Etiology, and Coenzyme Q10 Levels in 55 Children with Combined Enzyme Deficiencies of the Mitochondrial Respiratory Chain. <i>Journal of Pediatrics</i> , <b>2021</b> , 228, 240-251.e23.6	3.6	1
38	The one-carbon pool controls mitochondrial energy metabolism via complex I and iron-sulfur clusters. <i>Science Advances</i> , <b>2021</b> , 7,	14.3	6
37	Severe congenital lactic acidosis and hypertrophic cardiomyopathy caused by an intronic variant in NDUFB7. <i>Human Mutation</i> , <b>2021</b> , 42, 378-384	4.7	3
36	Stable Isotope Labeling of Amino Acids in Flies (SILAF) Reveals Differential Phosphorylation of Mitochondrial Proteins Upon Loss of OXPHOS Subunits. <i>Molecular and Cellular Proteomics</i> , <b>2021</b> , 20, 100065	7.6	1
35	Quantitative Proteomics in Drosophila with Holidic Stable-Isotope Labeling of Amino Acids in Fruit Flies (SILAF). <i>Methods in Molecular Biology</i> , <b>2021</b> , 2192, 75-87	1.4	1
34	Metabolic regulation of neurodifferentiation in the adult brain. <i>Cellular and Molecular Life Sciences</i> , <b>2020</b> , 77, 2483-2496	10.3	22
33	SQSTM1/p62-Directed Metabolic Reprogramming Is Essential for Normal Neurodifferentiation. <i>Stem Cell Reports</i> , <b>2019</b> , 12, 696-711	8	20
32	Myoglobinopathy is an adult-onset autosomal dominant myopathy with characteristic sarcoplasmic inclusions. <i>Nature Communications</i> , <b>2019</b> , 10, 1396	17.4	7
31	Mutations in the mitochondrial tryptophanyl-tRNA synthetase cause growth retardation and progressive leukoencephalopathy. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2019</b> , 7, e654	2.3	6
30	Absence of TXNIP in Humans Leads to Lactic Acidosis and Low Serum Methionine Linked to Deficient Respiration on Pyruvate. <i>Diabetes</i> , <b>2019</b> , 68, 709-723	0.9	17
29	C6orf203 is an RNA-binding protein involved in mitochondrial protein synthesis. <i>Nucleic Acids Research</i> , <b>2019</b> , 47, 9386-9399	20.1	14
28	Defects of mitochondrial RNA turnover lead to the accumulation of double-stranded RNA in vivo. <i>PLoS Genetics</i> , <b>2019</b> , 15, e1008240	6	19
27	Mitochondrial RNA Turnover in Metazoa. <i>Nucleic Acids and Molecular Biology</i> , <b>2018</b> , 17-46		0
26	Chorea, psychosis, acanthocytosis, and prolonged survival associated with mutations. <i>Neurology</i> , <b>2018</b> , 91, 710-712	6.5	7

25	A multi-systemic mitochondrial disorder due to a dominant p.Y955H disease variant in DNA polymerase gamma. <i>Human Molecular Genetics</i> , <b>2017</b> , 26, 2515-2525	5.6	5
24	Detection of 6-demethoxyubiquinone in CoQ deficiency disorders: Insights into enzyme interactions and identification of potential therapeutics. <i>Molecular Genetics and Metabolism</i> , <b>2017</b> , 121, 216-223	3.7	15
23	Respiratory chain complex III deficiency due to mutated BCS1L: a novel phenotype with encephalomyopathy, partially phenocopied in a Bcs1l mutant mouse model. <i>Orphanet Journal of Rare Diseases</i> , <b>2017</b> , 12, 73	4.2	18
22	Absence of the Autophagy Adaptor SQSTM1/p62 Causes Childhood-Onset Neurodegeneration with Ataxia, Dystonia, and Gaze Palsy. <i>American Journal of Human Genetics</i> , <b>2016</b> , 99, 735-743	11	69
21	Mitochondrial Polyadenylation Is a One-Step Process Required for mRNA Integrity and tRNA Maturation. <i>PLoS Genetics</i> , <b>2016</b> , 12, e1006028	6	25
20	J10 Chorea, psychotic symptoms and long survival in a subject with ELAC2 mutations. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2016</b> , 87, A78.3-A79	5.5	
19	A Phenotype-Driven Approach to Generate Mouse Models with Pathogenic mtDNA Mutations Causing Mitochondrial Disease. <i>Cell Reports</i> , <b>2016</b> , 16, 2980-2990	10.6	72
18	Cyclophilin D, a target for counteracting skeletal muscle dysfunction in mitochondrial myopathy. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 6580-7	5.6	11
17	SUV3 helicase is required for correct processing of mitochondrial transcripts. <i>Nucleic Acids Research</i> , <b>2015</b> , 43, 7398-413	20.1	15
16	Rescue of primary ubiquinone deficiency due to a novel COQ7 defect using 2,4-dihydroxybenzoic acid. <i>Journal of Medical Genetics</i> , <b>2015</b> , 52, 779-83	5.8	67
15	Intra-mitochondrial Methylation Deficiency Due to Mutations in SLC25A26. <i>American Journal of Human Genetics</i> , <b>2015</b> , 97, 761-8	11	44
14	Regulation of DNA replication at the end of the mitochondrial D-loop involves the helicase TWINKLE and a conserved sequence element. <i>Nucleic Acids Research</i> , <b>2015</b> , 43, 9262-75	20.1	54
13	Neu-Laxova syndrome is a heterogeneous metabolic disorder caused by defects in enzymes of the L-serine biosynthesis pathway. <i>American Journal of Human Genetics</i> , <b>2014</b> , 95, 285-93	11	82
12	No recombination of mtDNA after heteroplasmy for 50 generations in the mouse maternal germline. <i>Nucleic Acids Research</i> , <b>2014</b> , 42, 1111-6	20.1	63
11	Germline mitochondrial DNA mutations aggravate ageing and can impair brain development. <i>Nature</i> , <b>2013</b> , 501, 412-5	50.4	188
10	MTERF3 regulates mitochondrial ribosome biogenesis in invertebrates and mammals. <i>PLoS Genetics</i> , <b>2013</b> , 9, e1003178	6	70
9	TWINKLE is an essential mitochondrial helicase required for synthesis of nascent D-loop strands and complete mtDNA replication. <i>Human Molecular Genetics</i> , <b>2013</b> , 22, 1983-93	5.6	114
8	Variation in germline mtDNA heteroplasmy is determined prenatally but modified during subsequent transmission. <i>Nature Genetics</i> , <b>2012</b> , 44, 1282-5	36.3	102

7	Ultra-deep sequencing of mouse mitochondrial DNA: mutational patterns and their origins. <i>PLoS Genetics</i> , <b>2011</b> , 7, e1002028	6	136
6	Maintenance of respiratory chain function in mouse hearts with severely impaired mtDNA transcription. <i>Nucleic Acids Research</i> , <b>2010</b> , 38, 6577-88	20.1	29
5	MtDNA mutations are a common cause of severe disease phenotypes in children with Leigh syndrome. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , <b>2009</b> , 1787, 484-90	4.6	38
4	Purifying selection of mtDNA and its implications for understanding evolution and mitochondrial disease. <i>Nature Reviews Genetics</i> , <b>2008</b> , 9, 657-62	30.1	129
3	Strong purifying selection in transmission of mammalian mitochondrial DNA. <i>PLoS Biology</i> , <b>2008</b> , 6, e10	9.7	352
2	Is energy deficiency good in moderation?. <i>Cell</i> , <b>2007</b> , 131, 448-50	56.2	11
1	Respiratory chain dysfunction in skeletal muscle does not cause insulin resistance. <i>Biochemical and Biophysical Research Communications</i> , <b>2006</b> , 350, 202-7	3.4	122