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List of Publications by Year in descending order

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Version: 2024-02-01

27
papers

7,284
citations

257450

24
h-index

552781

26
g-index

29
all docs

29
docs citations

29
times ranked

12357
citing authors

#	ARTICLE	IF	CITATIONS
1	Mitochondrial DNA variation across 56,434 individuals in gnomAD. <i>Genome Research</i> , 2022, 32, 569-582.	5.5	59
2	Combinatorial GxGxE CRISPR screen identifies SLC25A39 in mitochondrial glutathione transport linking iron homeostasis to OXPHOS. <i>Nature Communications</i> , 2022, 13, 2483.	12.8	31
3	Fatal Perinatal Mitochondrial Cardiac Failure Caused by Recurrent De Novo Duplications in the ATAD3 Locus. <i>Med</i> , 2021, 2, 49-73.e10.	4.4	33
4	MitoCarta3.0: an updated mitochondrial proteome now with sub-organelle localization and pathway annotations. <i>Nucleic Acids Research</i> , 2021, 49, D1541-D1547.	14.5	760
5	Loss of LUC7L2 and U1 snRNP subunits shifts energy metabolism from glycolysis to OXPHOS. <i>Molecular Cell</i> , 2021, 81, 1905-1919.e12.	9.7	33
6	Genetic Screen for Cell Fitness in High or Low Oxygen Highlights Mitochondrial and Lipid Metabolism. <i>Cell</i> , 2020, 181, 716-727.e11.	28.9	126
7	Hypoxia Rescues Frataxin Loss by Restoring Iron Sulfur Cluster Biogenesis. <i>Cell</i> , 2019, 177, 1507-1521.e16.	28.9	80
8	A patient with homozygous nonsense variants in two Leigh syndrome disease genes: Distinguishing a dual diagnosis from a hypomorphic proteinâ€truncating variant. <i>Human Mutation</i> , 2019, 40, 893-898.	2.5	8
9	Spatiotemporal compartmentalization of hepatic NADH and NADPH metabolism. <i>Journal of Biological Chemistry</i> , 2018, 293, 7508-7516.	3.4	81
10	Widespread Chromosomal Losses and Mitochondrial DNA Alterations as Genetic Drivers in HÃ¼rthle Cell Carcinoma. <i>Cancer Cell</i> , 2018, 34, 242-255.e5.	16.8	185
11	Early loss of mitochondrial complex I and rewiring of glutathione metabolism in renal oncocytoma. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, E6283-E6290.	7.1	70
12	Comparative Analysis of Mitochondrial N-Termini from Mouse, Human, and Yeast. <i>Molecular and Cellular Proteomics</i> , 2017, 16, 512-523.	3.8	71
13	Biallelic C1QBP Mutations Cause Severe Neonatal-, Childhood-, or Later-Onset Cardiomyopathy Associated with Combined Respiratory-Chain Deficiencies. <i>American Journal of Human Genetics</i> , 2017, 101, 525-538.	6.2	58
14	Biallelic Mutations in MRPS34 Lead to Instability of the Small Mitoribosomal Subunit and Leigh Syndrome. <i>American Journal of Human Genetics</i> , 2017, 101, 239-254.	6.2	83
15	CLIC, a tool for expanding biological pathways based on co-expression across thousands of datasets. <i>PLoS Computational Biology</i> , 2017, 13, e1005653.	3.2	30
16	A Genome-wide CRISPR Death Screen Identifies Genes Essential for Oxidative Phosphorylation. <i>Cell Metabolism</i> , 2016, 24, 875-885.	16.2	244
17	MitoCarta2.0: an updated inventory of mammalian mitochondrial proteins. <i>Nucleic Acids Research</i> , 2016, 44, D1251-D1257.	14.5	1,170
18	CLYBL is a polymorphic human enzyme with malate synthase and Î² ² -methylmalate synthase activity. <i>Human Molecular Genetics</i> , 2014, 23, 2313-2323.	2.9	29

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19	Expansion of Biological Pathways Based on Evolutionary Inference. <i>Cell</i> , 2014, 158, 213-225.	28.9	107
20	EMRE Is an Essential Component of the Mitochondrial Calcium Uniporter Complex. <i>Science</i> , 2013, 342, 1379-1382.	12.6	537
21	Targeted exome sequencing of suspected mitochondrial disorders. <i>Neurology</i> , 2013, 80, 1762-1770.	1.1	155
22	Molecular Diagnosis of Infantile Mitochondrial Disease with Targeted Next-Generation Sequencing. <i>Science Translational Medicine</i> , 2012, 4, 118ra10.	12.4	406
23	Megaloblastic Anemia and Mitochondriopathy Caused by a Homozygous Mutation in Sideroflexin-4. <i>Blood</i> , 2012, 120, 79-79.	1.4	0
24	High-throughput, pooled sequencing identifies mutations in NUBPL and FOXRED1 in human complex I deficiency. <i>Nature Genetics</i> , 2010, 42, 851-858.	21.4	332
25	The Mitochondrial Proteome and Human Disease. <i>Annual Review of Genomics and Human Genetics</i> , 2010, 11, 25-44.	6.2	497
26	A Mitochondrial Protein Compendium Elucidates Complex I Disease Biology. <i>Cell</i> , 2008, 134, 112-123.	28.9	1,766
27	Systematic identification of human mitochondrial disease genes through integrative genomics. <i>Nature Genetics</i> , 2006, 38, 576-582.	21.4	321