Anna Wredenberg

List of Publications by Year
in descending order

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43 papers	4,263 citations	377584 21 h-index	312153 41 g-index
45 all docs	45 docs citations	45 times ranked	7241 citing authors

#	Article	IF	Citations
1	Pathogenic SLC25A26 variants impair SAH transport activity causing mitochondrial disease. Human Molecular Genetics, 2022, 31, 2049-2062.	1.4	3
2	Human GTPBP5 is involved in the late stage of mitoribosome large subunit assembly. Nucleic Acids Research, 2021, 49, 354-370.	6.5	21
3	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.	0.9	6
4	The one-carbon pool controls mitochondrial energy metabolism via complex I and iron-sulfur clusters. Science Advances, 2021, 7, .	4.7	23
5	Severe congenital lactic acidosis and hypertrophic cardiomyopathy caused by an intronic variant in NDUFB7. Human Mutation, 2021, 42, 378-384.	1.1	8
6	Case Report: A Novel Mutation in the Mitochondrial MT-ND5 Gene Is Associated With Leber Hereditary Optic Neuropathy (LHON). Frontiers in Neurology, 2021, 12, 652590.	1.1	4
7	Novel Mutation m.10372A>G in MT-ND3 Causing Sensorimotor Axonal Polyneuropathy. Neurology: Genetics, 2021, 7, e566.	0.9	3
8	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.	3.6	116
9	Protocol for the derivation, culturing, and differentiation of human iPS-cell-derived neuroepithelial stem cells to study neural differentiation inÂvitro. STAR Protocols, 2021, 2, 100528.	0.5	11
10	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.	2.5	6
11	Quantitative Proteomics in Drosophila with Holidic Stable-Isotope Labeling of Amino Acids in Fruit Flies (SILAF). Methods in Molecular Biology, 2021, 2192, 75-87.	0.4	2
12	Metabolic regulation of neurodifferentiation in the adult brain. Cellular and Molecular Life Sciences, 2020, 77, 2483-2496.	2.4	46
13	<scp>FBXL</scp> 4 deficiency increases mitochondrial removal by autophagy. EMBO Molecular Medicine, 2020, 12, e11659.	3.3	44
14	SLC12A2 mutations cause NKCC1 deficiency with encephalopathy and impaired secretory epithelia. Neurology: Genetics, 2020, 6, e478.	0.9	20
15	C6orf203 is an RNA-binding protein involved in mitochondrial protein synthesis. Nucleic Acids Research, 2019, 47, 9386-9399.	6.5	26
16	Defects of mitochondrial RNA turnover lead to the accumulation of double-stranded RNA in vivo. PLoS Genetics, 2019, 15, e1008240.	1.5	40
17	SQSTM1/p62-Directed Metabolic Reprogramming Is Essential for Normal Neurodifferentiation. Stem Cell Reports, 2019, 12, 696-711.	2.3	32
18	Myoglobinopathy is an adult-onset autosomal dominant myopathy with characteristic sarcoplasmic inclusions. Nature Communications, 2019, 10, 1396.	5.8	11

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19	Mutations in the mitochondrial tryptophanylâ€tRNA synthetase cause growth retardation and progressive leukoencephalopathy. Molecular Genetics & Genomic Medicine, 2019, 7, e654.	0.6	13
20	Absence of TXNIP in Humans Leads to Lactic Acidosis and Low Serum Methionine Linked to Deficient Respiration on Pyruvate. Diabetes, 2019, 68, 709-723.	0.3	22
21	RNA modification landscape of the human mitochondrial tRNALys regulates protein synthesis. Nature Communications, 2018, 9, 3966.	5.8	61
22	Chorea, psychosis, acanthocytosis, and prolonged survival associated with ELAC2 mutations. Neurology, 2018, 91, 710-712.	1.5	8
23	Mitochondrial RNA Turnover in Metazoa. Nucleic Acids and Molecular Biology, 2018, , 17-46.	0.2	1
24	A multi-systemic mitochondrial disorder due to a dominant p.Y955H disease variant in DNA polymerase gamma. Human Molecular Genetics, 2017, 26, 2515-2525.	1.4	12
25	Detection of 6-demethoxyubiquinone in CoQ10 deficiency disorders: Insights into enzyme interactions and identification of potential therapeutics. Molecular Genetics and Metabolism, 2017, 121, 216-223.	0.5	25
26	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.	1.2	20
27	J10â€…Chorea, psychotic symptoms and long survival in a subject with ELAC2 mutations. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, A78.3-A79.	0.9	0
28	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.	2.6	99
29	Mitochondrial Polyadenylation Is a One-Step Process Required for mRNA Integrity and tRNA Maturation. PLoS Genetics, 2016, 12, e1006028.	1.5	43
30	Complementation between polymerase- and exonuclease-deficient mitochondrial DNA polymerase mutants in genomically engineered flies. Nature Communications, 2015, 6, 8808.	5.8	48
31	Cyclophilin D, a target for counteracting skeletal muscle dysfunction in mitochondrial myopathy. Human Molecular Genetics, 2015, 24, 6580-6587.	1.4	16
32	SUV3 helicase is required for correct processing of mitochondrial transcripts. Nucleic Acids Research, 2015, 43, 7398-7413.	6.5	20
33	Rescue of primary ubiquinone deficiency due to a novelCOQ7defect using 2,4â€“dihydroxybensoic acid. Journal of Medical Genetics, 2015, 52, 779-783.	1.5	94
34	Intra-mitochondrial Methylation Deficiency Due to Mutations in SLC25A26. American Journal of Human Genetics, 2015, 97, 761-768.	2.6	58
35	Rapid pulsed whole genome sequencing for comprehensive acute diagnostics of inborn errors of metabolism. BMC Genomics, 2014, 15, 1090.	1.2	54
36	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, 285-293.	2.6	110

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37	MTERF3 Regulates Mitochondrial Ribosome Biogenesis in Invertebrates and Mammals. PLoS Genetics, 2013, 9, e1003178.	1.5	85
38	LRPPRC is necessary for polyadenylation and coordination of translation of mitochondrial mRNAs. EMBO Journal, 2012, 31, 443-456.	3.5	264
39	The Bicoid Stability Factor Controls Polyadenylation and Expression of Specific Mitochondrial mRNAs in Drosophila melanogaster. PLoS Genetics, 2011, 7, e1002324.	1.5	55
40	Respiratory chain dysfunction in skeletal muscle does not cause insulin resistance. Biochemical and Biophysical Research Communications, 2006, 350, 202-207.	1.0	134
41	Premature ageing in mice expressing defective mitochondrial DNA polymerase. Nature, 2004, 429, 417-423.	13.7	2,318
42	Increased mitochondrial mass in mitochondrial myopathy mice. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 15066-15071.	3.3	262
43	Complex genetic counselling and prenatal analysis in a woman with external ophthalmoplegia and deleted mtDNA. , 2000, 20, 426-431.		18