

Antonella Spinazzola

List of Publications by Year in descending order

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Version: 2024-02-01

62
papers

4,847
citations

101543

36
h-index

138484

58
g-index

70
all docs

70
docs citations

70
times ranked

4541
citing authors

#	ARTICLE	IF	CITATIONS
1	2 deoxy-D-glucose augments the mitochondrial respiratory chain in heart. <i>Scientific Reports</i> , 2022, 12, 6890.	3.3	5
2	Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE): Position paper on diagnosis, prognosis, and treatment by the <scp>MNGIE</scp> International Network. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 376-387.	3.6	47
3	Uniparental isodisomy of chromosome 2 causing MRPL44-related multisystem mitochondrial disease. <i>Molecular Biology Reports</i> , 2021, 48, 2093-2104.	2.3	1
4	2-Deoxy-D-glucose couples mitochondrial DNA replication with mitochondrial fitness and promotes the selection of wild-type over mutant mitochondrial DNA. <i>Nature Communications</i> , 2021, 12, 6997.	12.8	12
5	Identification of a novel heterozygous guanosine monophosphate reductase (<i>GMPR</i>) variant in a patient with a late-onset disorder of mitochondrial DNA maintenance. <i>Clinical Genetics</i> , 2020, 97, 276-286.	2.0	7
6	Mechanisms of onset and accumulation of mtDNA mutations. , 2020, , 195-219.		0
7	Recurrent De Novo NAHR Reciprocal Duplications in the ATAD3 Gene Cluster Cause a Neurogenetic Trait with Perturbed Cholesterol and Mitochondrial Metabolism. <i>American Journal of Human Genetics</i> , 2020, 106, 272-279.	6.2	33
8	Parkinsonism and spastic paraplegia type 7: Expanding the spectrum of mitochondrial Parkinsonism. <i>Movement Disorders</i> , 2019, 34, 1547-1561.	3.9	44
9	Beyond the unwinding: role of TOP1MT in mitochondrial translation. <i>Cell Cycle</i> , 2019, 18, 2377-2384.	2.6	11
10	MRPS25 mutations impair mitochondrial translation and cause encephalomyopathy. <i>Human Molecular Genetics</i> , 2019, 28, 2711-2719.	2.9	33
11	Reply to: "Mitochondrial Parkinsonism due to <i>SPG7/Paraplegin</i> variants with secondary mtDNA depletion". <i>Movement Disorders</i> , 2019, 34, 1932-1933.	3.9	0
12	The mitochondrial type IB topoisomerase drives mitochondrial translation and carcinogenesis. <i>Nature Communications</i> , 2019, 10, 83.	12.8	38
13	Transcript availability dictates the balance between strand-asynchronous and strand-coupled mitochondrial DNA replication. <i>Nucleic Acids Research</i> , 2018, 46, 10771-10781.	14.5	20
14	<scp>LETM</scp> 1 couples mitochondrial <scp>DNA</scp> metabolism and nutrient preference. <i>EMBO Molecular Medicine</i> , 2018, 10, .	6.9	41
15	Mitochondrial ribosomal protein S25 (MRPS25) mutations impair ribosomal assembly and cause mitochondrial encephalomyopathy with partial agenesis of the corpus callosum. <i>Neuromuscular Disorders</i> , 2018, 28, S30-S31.	0.6	0
16	Clinicopathologic and molecular spectrum of <i>RNASEH1</i> -related mitochondrial disease. <i>Neurology: Genetics</i> , 2017, 3, e149.	1.9	19
17	Aberrant ribonucleotide incorporation and multiple deletions in mitochondrial DNA of the murine MPV17 disease model. <i>Nucleic Acids Research</i> , 2017, 45, 12808-12815.	14.5	43
18	Reply: Genotype-phenotype correlation in ATAD3A deletions: not just of scientific relevance. <i>Brain</i> , 2017, 140, e67-e67.	7.6	9

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19	ATAD3 gene cluster deletions cause cerebellar dysfunction associated with altered mitochondrial DNA and cholesterol metabolism. <i>Brain</i> , 2017, 140, 1595-1610.	7.6	105
20	Pathological ribonuclease H1 causes R-loop depletion and aberrant DNA segregation in mitochondria. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, E4276-85.	7.1	48
21	Mitochondrial quality control: Cell-type-dependent responses to pathological mutant mitochondrial DNA. <i>Autophagy</i> , 2016, 12, 2098-2112.	9.1	21
22	MPV17 Loss Causes Deoxynucleotide Insufficiency and Slow DNA Replication in Mitochondria. <i>PLoS Genetics</i> , 2016, 12, e1005779.	3.5	67
23	Amino Acid Starvation Has Opposite Effects on Mitochondrial and Cytosolic Protein Synthesis. <i>PLoS ONE</i> , 2014, 9, e93597.	2.5	48
24	MPV17L2 is required for ribosome assembly in mitochondria. <i>Nucleic Acids Research</i> , 2014, 42, 8500-8515.	14.5	56
25	Mitochondrial diseases: Translation matters. <i>Molecular and Cellular Neurosciences</i> , 2013, 55, 1-12.	2.2	62
26	Mitochondrial nucleoid interacting proteins support mitochondrial protein synthesis. <i>Nucleic Acids Research</i> , 2012, 40, 6109-6121.	14.5	195
27	Mitochondrial DNA mutations and depletion in pediatric medicine. <i>Seminars in Fetal and Neonatal Medicine</i> , 2011, 16, 190-196.	2.3	40
28	EFNS guidelines for the molecular diagnosis of neurogenetic disorders: motoneuron, peripheral nerve and muscle disorders. <i>European Journal of Neurology</i> , 2011, 18, 207-217.	3.3	29
29	EFNS guidelines on the molecular diagnosis of ataxias and spastic paraplegias. <i>European Journal of Neurology</i> , 2010, 17, 179-188.	3.3	49
30	EFNS guidelines on the molecular diagnosis of channelopathies, epilepsies, migraine, stroke, and dementias. <i>European Journal of Neurology</i> , 2010, 17, 641-648.	3.3	10
31	Early-onset liver mtDNA depletion and late-onset proteinuric nephropathy in Mpv17 knockout mice. <i>Human Molecular Genetics</i> , 2009, 18, 12-26.	2.9	87
32	Identification of novel mutations in five patients with mitochondrial encephalomyopathy. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , 2009, 1787, 491-501.	1.0	83
33	Clinical and molecular features of mitochondrial DNA depletion syndromes. <i>Journal of Inherited Metabolic Disease</i> , 2009, 32, 143-158.	3.6	161
34	Disorders from perturbations of nuclear-mitochondrial intergenomic cross-talk. <i>Journal of Internal Medicine</i> , 2009, 265, 174-192.	6.0	64
35	EFNS guidelines on the molecular diagnosis of neurogenetic disorders: general issues, Huntington's disease, Parkinson's disease and dystonias. <i>European Journal of Neurology</i> , 2009, 16, 777-785.	3.3	51
36	EFNS guidelines on the molecular diagnosis of mitochondrial disorders. <i>European Journal of Neurology</i> , 2009, 16, 1255-1264.	3.3	55

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37	Collated mutations in mitochondrial DNA (mtDNA) depletion syndrome (excluding the mitochondrial) Tj ETQq1 1 0.784314 rgBT /Ove 1109-1112.	3.8	41
38	Glucose metabolism and diet-based prevention of liver dysfunction in MPV17 mutant patients. Journal of Hepatology, 2009, 50, 215-221.	3.7	44
39	Mitochondrial Diseases: A Cross-Talk Between Mitochondrial and Nuclear Genomes. Advances in Experimental Medicine and Biology, 2009, 652, 69-84.	1.6	27
40	Lack of founder effect for an identical mtDNA depletion syndrome (MDS)-associated MPV17 mutation shared by Navajos and Italians. Neuromuscular Disorders, 2008, 18, 315-318.	0.6	17
41	Hepatocerebral Form of Mitochondrial DNA Depletion Syndrome. Archives of Neurology, 2008, 65, 1108-13.	4.5	68
42	Disorders of Nuclear-Mitochondrial Intergenomic Communication. Bioscience Reports, 2007, 27, 39-51.	2.4	61
43	MPV17 encodes an inner mitochondrial membrane protein and is mutated in infantile hepatic mitochondrial DNA depletion. Nature Genetics, 2006, 38, 570-575.	21.4	380
44	Systematic identification of human mitochondrial disease genes through integrative genomics. Nature Genetics, 2006, 38, 576-582.	21.4	321
45	Disorders of nuclear-mitochondrial intergenomic signaling. Gene, 2005, 354, 162-168.	2.2	108
46	Definitive Diagnosis of Mitochondrial Neurogastrointestinal Encephalomyopathy by Biochemical Assays. Clinical Chemistry, 2004, 50, 120-124.	3.2	107
47	Thymidine Phosphorylase Deficiency Causes MNGIE: An Autosomal Recessive Mitochondrial Disorder. Nucleosides, Nucleotides and Nucleic Acids, 2004, 23, 1217-1225.	1.1	24
48	Mitochondrial myopathy and ophthalmoplegia in a sporadic patient with the 5698Gâ†’A mitochondrial DNA mutation. Neuromuscular Disorders, 2004, 14, 815-817.	0.6	12
49	Mitochondrial disorders. Current Neurology and Neuroscience Reports, 2003, 3, 423-432.	4.2	79
50	Nuclear genes in mitochondrial disorders. Current Opinion in Genetics and Development, 2003, 13, 262-270.	3.3	82
51	Altered Thymidine Metabolism Due to Defects of Thymidine Phosphorylase. Journal of Biological Chemistry, 2002, 277, 4128-4133.	3.4	209
52	Mitochondrial neurogastrointestinal encephalomyopathy and thymidine metabolism: results and hypotheses. Mitochondrion, 2002, 2, 143-147.	3.4	10
53	MNGIE: from nuclear DNA to mitochondrial DNA. Neuromuscular Disorders, 2001, 11, 7-10.	0.6	64
54	Assay of mitochondrial ATP synthesis in animal cells. Methods in Cell Biology, 2001, 65, 133-145.	1.1	30

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55	Coenzyme Q ₁₀ reverses pathological phenotype and reduces apoptosis in familial CoQ ₁₀ deficiency. <i>Neurology</i> , 2001, 57, 515-518.	1.1	157
56	Mitochondrial neurogastrointestinal encephalomyopathy: An autosomal recessive disorder due to thymidine phosphorylase mutations. <i>Annals of Neurology</i> , 2000, 47, 792-800.	5.3	324
57	Mitochondrial neurogastrointestinal encephalomyopathy: an autosomal recessive disorder due to thymidine phosphorylase mutations. <i>Annals of Neurology</i> , 2000, 47, 792-800.	5.3	81
58	A distinctive autosomal dominant vacuolar neuromyopathy linked to 19p13. <i>Neurology</i> , 1999, 53, 830-830.	1.1	36
59	Oligomycin Induces a Decrease in the Cellular Content of a Pathogenic Mutation in the Human Mitochondrial ATPase 6 Gene. <i>Journal of Biological Chemistry</i> , 1999, 274, 9386-9391.	3.4	90
60	Thymidine Phosphorylase Gene Mutations in MNGIE, a Human Mitochondrial Disorder. <i>Science</i> , 1999, 283, 689-692.	12.6	827
61	A Novel Mitochondrial DNA Point Mutation in the tRNA ^{Ala} Gene Is Associated with Progressive External Ophthalmoplegia. <i>Biochemical and Biophysical Research Communications</i> , 1996, 220, 623-627.	2.1	54
62	Morphological observations in mitochondrial diseases. <i>Progress in Cell Research</i> , 1995, , 217-221.	0.3	0