Antonella Spinazzola

List of Publications by Year
in descending order

62	4,847	101543 36	138484 58
papers	citations	h-index	g-index

70	70	70	4541
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Thymidine Phosphorylase Gene Mutations in MNGIE, a Human Mitochondrial Disorder. Science, 1999, 283, 689-692.	12.6	827
2	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
3	Mitochondrial neurogastrointestinal encephalomyopathy: An autosomal recessive disorder due to thymidine phosphorylase mutations. Annals of Neurology, 2000, 47, 792-800.	5.3	324
4	Systematic identification of human mitochondrial disease genes through integrative genomics. Nature Genetics, 2006, 38, 576-582.	21.4	321
5	Altered Thymidine Metabolism Due to Defects of Thymidine Phosphorylase. Journal of Biological Chemistry, 2002, 277, 4128-4133.	3.4	209
6	Mitochondrial nucleoid interacting proteins support mitochondrial protein synthesis. Nucleic Acids Research, 2012, 40, 6109-6121.	14.5	195
7	Clinical and molecular features of mitochondrial DNA depletion syndromes. Journal of Inherited Metabolic Disease, 2009, 32, 143-158.	3.6	161
8	Coenzyme Q ₁₀ reverses pathological phenotype and reduces apoptosis in familial CoQ ₁₀ deficiency. Neurology, 2001, 57, 515-518.	1.1	157
9	Disorders of nuclear-mitochondrial intergenomic signaling. Gene, 2005, 354, 162-168.	2.2	108
10	Definitive Diagnosis of Mitochondrial Neurogastrointestinal Encephalomyopathy by Biochemical Assays. Clinical Chemistry, 2004, 50, 120-124.	3.2	107
11	ATAD3 gene cluster deletions cause cerebellar dysfunction associated with altered mitochondrial DNA and cholesterol metabolism. Brain, 2017, 140, 1595-1610.	7.6	105
12	Oligomycin Induces a Decrease in the Cellular Content of a Pathogenic Mutation in the Human Mitochondrial ATPase 6 Gene. Journal of Biological Chemistry, 1999, 274, 9386-9391.	3.4	90
13	Early-onset liver mtDNA depletion and late-onset proteinuric nephropathy in Mpv17 knockout mice. Human Molecular Genetics, 2009, 18, 12-26.	2.9	87
14	Identification of novel mutations in five patients with mitochondrial encephalomyopathy. Biochimica Et Biophysica Acta - Bioenergetics, 2009, 1787, 491-501.	1.0	83
15	Nuclear genes in mitochondrial disorders. Current Opinion in Genetics and Development, 2003, 13, 262-270.	3.3	82
16	Mitochondrial neurogastrointestinal encephalomyopathy: an autosomal recessive disorder due to thymidine phosphorylase mutations. Annals of Neurology, 2000, 47, 792-800.	5.3	81
17	Mitochondrial disorders. Current Neurology and Neuroscience Reports, 2003, 3, 423-432.	4.2	79
18	Hepatocerebral Form of Mitochondrial DNA Depletion Syndrome. Archives of Neurology, 2008, 65, 1108-13.	4.5	68

#	Article	IF	Citations
19	MPV17 Loss Causes Deoxynucleotide Insufficiency and Slow DNA Replication in Mitochondria. PLoS Genetics, 2016, 12, e1005779.	3.5	67
20	MNGIE: from nuclear DNA to mitochondrial DNA. Neuromuscular Disorders, 2001, 11, 7-10.	0.6	64
21	Disorders from perturbations of nuclearâ€mitochondrial intergenomic crossâ€talk. Journal of Internal Medicine, 2009, 265, 174-192.	6.0	64
22	Mitochondrial diseases: Translation matters. Molecular and Cellular Neurosciences, 2013, 55, 1-12.	2.2	62
23	Disorders of Nuclear-Mitochondrial Intergenomic Communication. Bioscience Reports, 2007, 27, 39-51.	2.4	61
24	MPV17L2 is required for ribosome assembly in mitochondria. Nucleic Acids Research, 2014, 42, 8500-8515.	14.5	56
25	EFNS guidelines on the molecular diagnosis of mitochondrial disorders. European Journal of Neurology, 2009, 16, 1255-1264.	3.3	55
26	A Novel Mitochondrial DNA Point Mutation in the tRNAIleGene Is Associated with Progressive External Ophtalmoplegia. Biochemical and Biophysical Research Communications, 1996, 220, 623-627.	2.1	54
27	EFNS guidelines on the molecular diagnosis of neurogenetic disorders: general issues, Huntingtonâ€™s disease, Parkinsonâ€™s disease and dystonias. European Journal of Neurology, 2009, 16, 777-785.	3.3	51
28	EFNS guidelines on the molecular diagnosis of ataxias and spastic paraplegias. European Journal of Neurology, 2010, 17, 179-188.	3.3	49
29	Amino Acid Starvation Has Opposite Effects on Mitochondrial and Cytosolic Protein Synthesis. PLoS ONE, 2014, 9, e93597.	2.5	48
30	Pathological ribonuclease H1 causes R-loop depletion and aberrant DNA segregation in mitochondria. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E4276-85.	7.1	48
31	Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE): Position paper on diagnosis, prognosis, and treatment by the <scp>MNGIE</scp> International Network. Journal of Inherited Metabolic Disease, 2021, 44, 376-387.	3.6	47
32	Glucose metabolism and diet-based prevention of liver dysfunction in MPV17 mutant patients. Journal of Hepatology, 2009, 50, 215-221.	3.7	44
33	Parkinsonism and spastic paraplegia type 7: Expanding the spectrum of mitochondrial Parkinsonism. Movement Disorders, 2019, 34, 1547-1561.	3.9	44
34	Aberrant ribonucleotide incorporation and multiple deletions in mitochondrial DNA of the murine MPV17 disease model. Nucleic Acids Research, 2017, 45, 12808-12815.	14.5	43
35	Collated mutations in mitochondrial DNA (mtDNA) depletion syndrome (excluding the mitochondrial) Tj ETQq1 1 0.784314 rgBT /Ove 1109-1112.	3.8	41
36	<scp>LETM</scp> 1 couples mitochondrial <scp>DNA</scp> metabolism and nutrient preference. EMBO Molecular Medicine, 2018, 10, .	6.9	41

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37	Mitochondrial DNA mutations and depletion in pediatric medicine. Seminars in Fetal and Neonatal Medicine, 2011, 16, 190-196.	2.3	40
38	The mitochondrial type IB topoisomerase drives mitochondrial translation and carcinogenesis. Nature Communications, 2019, 10, 83.	12.8	38
39	A distinctive autosomal dominant vacuolar neuromyopathy linked to 19p13. Neurology, 1999, 53, 830-830.	1.1	36
40	MRPS25 mutations impair mitochondrial translation and cause encephalomyopathy. Human Molecular Genetics, 2019, 28, 2711-2719.	2.9	33
41	Recurrent De Novo NAHR Reciprocal Duplications in the ATAD3 Gene Cluster Cause a Neurogenetic Trait with Perturbed Cholesterol and Mitochondrial Metabolism. American Journal of Human Genetics, 2020, 106, 272-279.	6.2	33
42	Assay of mitochondrial ATP synthesis in animal cells. Methods in Cell Biology, 2001, 65, 133-145.	1.1	30
43	EFNS guidelines for the molecular diagnosis of neurogenetic disorders: motoneuron, peripheral nerve and muscle disorders. European Journal of Neurology, 2011, 18, 207-217.	3.3	29
44	Mitochondrial Diseases: A Cross-Talk Between Mitochondrial and Nuclear Genomes. Advances in Experimental Medicine and Biology, 2009, 652, 69-84.	1.6	27
45	Thymidine Phosphorylase Deficiency Causes MNGIE: An Autosomal Recessive Mitochondrial Disorder. Nucleosides, Nucleotides and Nucleic Acids, 2004, 23, 1217-1225.	1.1	24
46	Mitochondrial quality control: Cell-type-dependent responses to pathological mutant mitochondrial DNA. Autophagy, 2016, 12, 2098-2112.	9.1	21
47	Transcript availability dictates the balance between strand-asynchronous and strand-coupled mitochondrial DNA replication. Nucleic Acids Research, 2018, 46, 10771-10781.	14.5	20
48	Clinicopathologic and molecular spectrum of RNASEH1 -related mitochondrial disease. Neurology: Genetics, 2017, 3, e149.	1.9	19
49	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
50	Mitochondrial myopathy and ophthalmoplegia in a sporadic patient with the 5698Gâ†'A mitochondrial DNA mutation. Neuromuscular Disorders, 2004, 14, 815-817.	0.6	12
51	2-Deoxy-D-glucose couples mitochondrial DNA replication with mitochondrial fitness and promotes the selection of wild-type over mutant mitochondrial DNA. Nature Communications, 2021, 12, 6997.	12.8	12
52	Beyond the unwinding: role of TOP1MT in mitochondrial translation. Cell Cycle, 2019, 18, 2377-2384.	2.6	11
53	Mitochondrial neurogastrointestinal encephalomyopathy and thymidine metabolism: results and hypotheses. Mitochondrion, 2002, 2, 143-147.	3.4	10
54	EFNS guidelines on the molecular diagnosis of channelopathies, epilepsies, migraine, stroke, and dementias. European Journal of Neurology, 2010, 17, 641-648.	3.3	10

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55	Reply: Genotype-phenotype correlation in ATAD3A deletions: not just of scientific relevance. Brain, 2017, 140, e67-e67.	7.6	9
56	Identification of a novel heterozygous guanosine monophosphate reductase (GMPR) variant in a patient with a lateâ€onset disorder of mitochondrial DNA maintenance. Clinical Genetics, 2020, 97, 276-286.	2.0	7
57	2 deoxy-D-glucose augments the mitochondrial respiratory chain in heart. Scientific Reports, 2022, 12, 6890.	3.3	5
58	Uniparental isodisomy of chromosome 2 causing MRPL44-related multisystem mitochondrial disease. Molecular Biology Reports, 2021, 48, 2093-2104.	2.3	1
59	Mitochondrial ribosomal protein S25 (MRPS25) mutations impair ribosomal assembly and cause mitochondrial encephalomyopathy with partial agenesis of the corpus callosum. Neuromuscular Disorders, 2018, 28, S30-S31.	0.6	0
60	Reply to: â€œMitochondrial Parkinsonism due to SPG7/Paraplegin variants with secondary mtDNA depletionâ€• Movement Disorders, 2019, 34, 1932-1933.	3.9	0
61	Mechanisms of onset and accumulation of mtDNA mutations. , 2020, , 195-219.		0
62	Morphological observations in mitochondrial diseases. Progress in Cell Research, 1995, , 217-221.	0.3	0