Alessandra Maresca

List of Publications by Year
in descending order

50 papers	2,477 citations	218381 26 h-index	205818 48 g-index
56 all docs	56 docs citations	56 times ranked	4019 citing authors

#	Article	IF	Citations
1	Efficient mitochondrial biogenesis drives incomplete penetrance in Leberâ€™s hereditary optic neuropathy. Brain, 2014, 137, 335-353.	3.7	229
2	Pharmacological Inhibition of Necroptosis Protects from Dopaminergic Neuronal Cell Death in Parkinsonâ€™s Disease Models. Cell Reports, 2018, 22, 2066-2079.	2.9	167
3	Mutations in SLC25A46, encoding a UGO1-like protein, cause an optic atrophy spectrum disorder. Nature Genetics, 2015, 47, 926-932.	9.4	166
4	OPA1 Isoforms in the Hierarchical Organization of Mitochondrial Functions. Cell Reports, 2017, 19, 2557-2571.	2.9	158
5	Syndromic parkinsonism and dementia associated with <scp>OPA</scp>1 missense mutations. Annals of Neurology, 2015, 78, 21-38.	2.8	154
6	Cigarette toxicity triggers Leberâ€™s hereditary optic neuropathy by affecting mtDNA copy number, oxidative phosphorylation and ROS detoxification pathways. Cell Death and Disease, 2015, 6, e2021-e2021.	2.7	107
7	Impaired complex I repair causes recessive Leberâ€™s hereditary optic neuropathy. Journal of Clinical Investigation, 2021, 131, .	3.9	89
8	Liver transplantation for mitochondrial neurogastrointestinal encephalomyopathy. Annals of Neurology, 2016, 80, 448-455.	2.8	81
9	The optic nerve: A â€œmito-windowâ€•on mitochondrial neurodegeneration. Molecular and Cellular Neurosciences, 2013, 55, 62-76.	1.0	78
10	Retinal Nerve Fiber Layer Thickness in Dominant Optic Atrophy. Ophthalmology, 2011, 118, 2076-2080.	2.5	71
11	Mitochondrial Optic Neuropathies: How Two Genomes may Kill the Same Cell Type?. Bioscience Reports, 2007, 27, 173-184.	1.1	70
12	A Novel Null Homozygous Mutation Confirms CACNA2D2 as a Gene Mutated in Epileptic Encephalopathy. PLoS ONE, 2013, 8, e82154.	1.1	67
13	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. Journal of Clinical Investigation, 2019, 130, 108-125.	3.9	65
14	Mitochondria: Biogenesis and mitophagy balance in segregation and clonal expansion of mitochondrial DNA mutations. International Journal of Biochemistry and Cell Biology, 2015, 63, 21-24.	1.2	63
15	Idebenone treatment in patients with OPA1-mutant dominant optic atrophy. Brain, 2013, 136, e231-e231.	3.7	62
16	Dna Methyltransferase 1 Mutations and Mitochondrial Pathology: Is Mtdna Methylated?. Frontiers in Genetics, 2015, 6, 90.	1.1	62
17	Targeting estrogen receptor Î² as preventive therapeutic strategy for Leber's hereditary optic neuropathy. Human Molecular Genetics, 2015, 24, ddv396.	1.4	62
18	Incomplete penetrance in mitochondrial optic neuropathies. Mitochondrion, 2017, 36, 130-137.	1.6	55

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19	Parsing the differences in affected with LHON: genetic versus environmental triggers of disease conversion. Brain, 2016, 139, e17-e17.	3.7	51
20	Homozygous <scp>NOTCH</scp> 3 null mutation and impaired <scp>NOTCH</scp> 3 signaling in recessive earlyâ€onset arteriopathy and cavitating leukoencephalopathy. EMBO Molecular Medicine, 2015, 7, 848-858.	3.3	48
21	Peculiar combinations of individually non-pathogenic missense mitochondrial DNA variants cause low penetrance Leberâ€™s hereditary optic neuropathy. PLoS Genetics, 2018, 14, e1007210.	1.5	47
22	Expanding and validating the biomarkers for mitochondrial diseases. Journal of Molecular Medicine, 2020, 98, 1467-1478.	1.7	44
23	A Novel in-Frame 18-bp Microdeletion inMT-CYBCauses a Multisystem Disorder with Prominent Exercise Intolerance. Human Mutation, 2014, 35, 954-958.	1.1	38
24	Deciphering OPA1 mutations pathogenicity by combined analysis of human, mouse and yeast cell models. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 3496-3514.	1.8	36
25	Mitochondrial diseases in adults. Journal of Internal Medicine, 2020, 287, 592-608.	2.7	33
26	Calcium mishandling in absence of primary mitochondrial dysfunction drives cellular pathology in Wolfram Syndrome. Scientific Reports, 2020, 10, 4785.	1.6	33
27	Gamma rays induce a p53-independent mitochondrial biogenesis that is counter-regulated by HIF1Î±. Cell Death and Disease, 2013, 4, e663-e663.	2.7	31
28	ATPase Domain <scp>AFG3L2</scp> Mutations Alter <scp>OPA1</scp> Processing and Cause Optic Neuropathy. Annals of Neurology, 2020, 88, 18-32.	2.8	31
29	Clinical and molecular study in a long-surviving patient with MLASA syndrome due to novel PUS1 mutations. Neurogenetics, 2016, 17, 65-70.	0.7	29
30	Biallelic variants in LIG3 cause a novel mitochondrial neurogastrointestinal encephalomyopathy. Brain, 2021, 144, 1451-1466.	3.7	28
31	Haplogroup J mitogenomes are the most sensitive to the pesticide rotenone: Relevance for human diseases. Neurobiology of Disease, 2018, 114, 129-139.	2.1	22
32	DNMT1 mutations leading to neurodegeneration paradoxically reflect on mitochondrial metabolism. Human Molecular Genetics, 2020, 29, 1864-1881.	1.4	19
33	Cybrid studies establish the causal link between the mtDNA m.3890G>A/MT-ND1 mutation and optic atrophy with bilateral brainstem lesions. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2013, 1832, 445-452.	1.8	17
34	Exploring Metabolic Adaptations to the Acidic Microenvironment of Osteosarcoma Cells Unveils Sphingosine 1-Phosphate as a Valuable Therapeutic Target. Cancers, 2021, 13, 311.	1.7	16
35	Genetic Basis of Mitochondrial Optic Neuropathies. Current Molecular Medicine, 2014, 14, 985-992.	0.6	16
36	Oncocytic glioblastoma: a glioblastoma showing oncocytic changes and increased mitochondrial DNA copy number. Human Pathology, 2013, 44, 1867-1876.	1.1	15

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37	DGUOK recessive mutations in patients with CPEO, mitochondrial myopathy, parkinsonism and mtDNA deletions. Brain, 2018, 141, e3-e3.	3.7	15
38	Revisiting the issue of mitochondrial DNA content in optic mitochondriopathies. Neurology, 2012, 79, 1517-1519.	1.5	13
39	Drug repositioning as a therapeutic strategy for neurodegenerations associated with OPA1 mutations. Human Molecular Genetics, 2021, 29, 3631-3645.	1.4	13
40	Generation of a human iPSC line, FINCBi001-A, carrying a homoplasmic m.G3460A mutation in MT-ND1 associated with Leberâ€™s Hereditary optic Neuropathy (LHON). Stem Cell Research, 2020, 48, 101939.	0.3	12
41	Molecular Mechanisms behind Inherited Neurodegeneration of the Optic Nerve. Biomolecules, 2021, 11, 496.	1.8	10
42	Brain MRS correlates with mitochondrial dysfunction biomarkers in MELASâ€associated mtDNA mutations. Annals of Clinical and Translational Neurology, 2021, 8, 1200-1211.	1.7	10
43	Mitochondrial Mass Assessment in a Selected Cell Line under Different Metabolic Conditions. Cells, 2019, 8, 1454.	1.8	8
44	The relevance of mitochondrial DNA variants fluctuation during reprogramming and neuronal differentiation of human iPSCs. Stem Cell Reports, 2021, 16, 1953-1967.	2.3	8
45	Exploiting hiPSCs in Leber's Hereditary Optic Neuropathy (LHON): Present Achievements and Future Perspectives. Frontiers in Neurology, 2021, 12, 648916.	1.1	7
46	Exploring metabolic reprogramming in melanoma via acquired resistance to the oxidative phosphorylation inhibitor phenformin. Melanoma Research, 2020, 30, 1-13.	0.6	6
47	An increased burden of rare exonic variants in NRXN1 microdeletion carriers is likely to enhance the penetrance for autism spectrum disorder. Journal of Cellular and Molecular Medicine, 2021, 25, 2459-2470.	1.6	3
48	Molecular biomarkers correlate with brain grey and white matter changes in patients with mitochondrial m.3243AÂ>ÂG mutation. Molecular Genetics and Metabolism, 2022, 135, 72-81.	0.5	3
49	Searching for genetic modifiers of Leber's hereditary optic neuropathy penetrance. Mitochondrion, 2012, 12, 572.	1.6	0
50	Genetic landscape of Leber's hereditary optic neuropathy: reflection on pathogenic mechanisms. Acta Ophthalmologica, 2015, 93, n/a-n/a.	0.6	0