Leonardo Caporali

List of Publications by Year
in descending order

73 papers	2,993 citations	218381 26 h-index	174990 52 g-index
78 all docs	78 docs citations	78 times ranked	4648 citing authors

#	Article	IF	Citations
1	Papillary thyroid carcinoma tall cell variant shares accumulation of mitochondria, mitochondrial DNA mutations, and loss of oxidative phosphorylation complex I integrity with oncocytic tumors. Journal of Pathology: Clinical Research, 2022, 8, 155-168.	1.3	10
2	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
3	The role of mtDNA haplogroups on metabolic features in narcolepsy type 1. Mitochondrion, 2022, 63, 37-42.	1.6	3
4	The Pattern of Retinal Ganglion Cell Loss in Wolfram Syndrome is Distinct From Mitochondrial Optic Neuropathies. American Journal of Ophthalmology, 2022, 241, 206-216.	1.7	5
5	Capturing the Pattern of Transition From Carrier to Affected in Leber Hereditary Optic Neuropathy. American Journal of Ophthalmology, 2022, 241, 71-79.	1.7	8
6	The Mitogenome Relationships and Phylogeography of Barn Swallows (Hirundo rustica). Molecular Biology and Evolution, 2022, 39, .	3.5	4
7	Missense PDSS1 mutations in CoenzymeQ10 synthesis cause optic atrophy and sensorineural deafness. Annals of Clinical and Translational Neurology, 2021, 8, 247-251.	1.7	7
8	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
9	Epilepsy with auditory features: Contribution of known genes in 112 patients. Seizure: the Journal of the British Epilepsy Association, 2021, 85, 115-118.	0.9	6
10	A novel <scp>ALG14</scp> missense variant in an alive child with myopathy, epilepsy, and progressive cerebral atrophy. American Journal of Medical Genetics, Part A, 2021, 185, 1918-1921.	0.7	3
11	Impaired complex I repair causes recessive Leberâ€™s hereditary optic neuropathy. Journal of Clinical Investigation, 2021, 131, .	3.9	89
12	DominantACO2mutations are a frequent cause of isolated optic atrophy. Brain Communications, 2021, 3, fcab063.	1.5	16
13	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
14	Leber's Hereditary Optic Neuropathy: A Report on Novel mtDNA Pathogenic Variants. Frontiers in Neurology, 2021, 12, 657317.	1.1	13
15	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
16	The m.3890G>A/MT-ND1 mtDNA rare pathogenic variant: Expanding clinical and MRI phenotypes. Mitochondrion, 2021, 60, 142-149.	1.6	4
17	Epilepsy in MTâ€ATP6 â€•related mils/NARP: correlation of elettroclinical features with heteroplasmy. Annals of Clinical and Translational Neurology, 2021, 8, 704-710.	1.7	10
18	Combined Optic Atrophy and Rodâ€“Cone Dystrophy Expands the RTN4IP1 (Optic Atrophy 10) Phenotype. Journal of Neuro-Ophthalmology, 2021, 41, e290-e292.	0.4	9

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19	Liver transplantation in mitochondrial neurogastrointestinal encephalomyopathy (MNGIE): clinical long-term follow-up and pathogenic implications. Journal of Neurology, 2020, 267, 3702-3710.	1.8	17
20	TYMP Variants Result in Late-Onset Mitochondrial Myopathy With Altered Muscle Mitochondrial DNA Homeostasis. Frontiers in Genetics, 2020, 11, 860.	1.1	6
21	Mitochondrial diseases in adults. Journal of Internal Medicine, 2020, 287, 592-608.	2.7	33
22	Calcium mishandling in absence of primary mitochondrial dysfunction drives cellular pathology in Wolfram Syndrome. Scientific Reports, 2020, 10, 4785.	1.6	33
23	DNMT1 mutations leading to neurodegeneration paradoxically reflect on mitochondrial metabolism. Human Molecular Genetics, 2020, 29, 1864-1881.	1.4	19
24	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
25	Mutations in the m-AAA proteases AFG3L2 and SPG7 are causing isolated dominant optic atrophy. Neurology: Genetics, 2020, 6, e428.	0.9	31
26	Novel mutations in DNA2 associated with myopathy and mtDNA instability. Annals of Clinical and Translational Neurology, 2019, 6, 1893-1899.	1.7	13
27	First TMEM126A missense mutation in an Italian proband with optic atrophy and deafness. Neurology: Genetics, 2019, 5, e329.	0.9	14
28	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. Journal of Clinical Investigation, 2019, 130, 108-125.	3.9	65
29	SCN1A mutations in focal epilepsy with auditory features: widening the spectrum of GEFS plus. Epileptic Disorders, 2019, 21, 185-191.	0.7	5
30	Pharmacological Inhibition of Necroptosis Protects from Dopaminergic Neuronal Cell Death in Parkinsonâ€™s Disease Models. Cell Reports, 2018, 22, 2066-2079.	2.9	167
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	Identification of rare genetic variants in Italian patients with dementia by targeted gene sequencing. Neurobiology of Aging, 2018, 66, 180.e23-180.e31.	1.5	18
33	Cerebral Mitochondrial Microangiopathy Leads to Leukoencephalopathy in Mitochondrial Neurogastrointestinal Encephalopathy. American Journal of Neuroradiology, 2018, 39, 427-434.	1.2	18
34	Complex II phosphorylation is triggered by unbalanced redox homeostasis in cells lacking complex III. Biochimica Et Biophysica Acta - Bioenergetics, 2018, 1859, 182-190.	0.5	7
35	DGUOK recessive mutations in patients with CPEO, mitochondrial myopathy, parkinsonism and mtDNA deletions. Brain, 2018, 141, e3-e3.	3.7	15
36	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

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37	Infant and Adult Gut Microbiome and Metabolome in Rural Bassa and Urban Settlers from Nigeria. Cell Reports, 2018, 23, 3056-3067.	2.9	128
38	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
39	Somatic mutation profiling of hobnail variant of papillary thyroid carcinoma. Endocrine-Related Cancer, 2017, 24, 107-117.	1.6	58
40	Liver transplant reverses biochemical imbalance in mitochondrial neurogastrointestinal encephalomyopathy. Mitochondrion, 2017, 34, 101-102.	1.6	23
41	OPA1 Isoforms in the Hierarchical Organization of Mitochondrial Functions. Cell Reports, 2017, 19, 2557-2571.	2.9	158
42	Incomplete penetrance in mitochondrial optic neuropathies. Mitochondrion, 2017, 36, 130-137.	1.6	55
43	Whole Mitogenomes Reveal the History of Swamp Buffalo: Initially Shaped by Glacial Periods and Eventually Modelled by Domestication. Scientific Reports, 2017, 7, 4708.	1.6	30
44	Pharmacogenetics and Treatment Response in Narcolepsy Type 1. Clinical Neuropharmacology, 2016, 39, 18-23.	0.2	5
45	Liver transplantation for mitochondrial neurogastrointestinal encephalomyopathy. Annals of Neurology, 2016, 80, 448-455.	2.8	81
46	Parsing the differences in affected with LHON: genetic versus environmental triggers of disease conversion. Brain, 2016, 139, e17-e17.	3.7	51
47	ITA-MNGIE: an Italian regional and national survey for mitochondrial neuro-gastro-intestinal encephalomyopathy. Neurological Sciences, 2016, 37, 1149-1151.	0.9	13
48	Reply: Both mitochondrial DNA and mitonuclear gene mutations cause hearing loss through cochlear dysfunction. Brain, 2016, 139, e34-e34.	3.7	7
49	â€˜Behr syndromeâ€™ with OPA1 compound heterozygote mutations. Brain, 2015, 138, e321-e321.	3.7	50
50	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
51	Mutations in SLC25A46, encoding a UGO1-like protein, cause an optic atrophy spectrum disorder. Nature Genetics, 2015, 47, 926-932.	9.4	166
52	Diffusion Tensor Imaging Mapping of Brain White Matter Pathology in Mitochondrial Optic Neuropathies. American Journal of Neuroradiology, 2015, 36, 1259-1265.	1.2	28
53	Dna Methyltransferase 1 Mutations and Mitochondrial Pathology: Is Mtdna Methylated?. Frontiers in Genetics, 2015, 6, 90.	1.1	62
54	Syndromic parkinsonism and dementia associated with <scp>OPA</scp>1 missense mutations. Annals of Neurology, 2015, 78, 21-38.	2.8	154

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55	OPA1-related auditory neuropathy: site of lesion and outcome of cochlear implantation. Brain, 2015, 138, 563-576.	3.7	86
56	An Ugo1-like protein is associated with optic atrophy â€˜plusâ€™ disorders. Mitochondrion, 2015, 24, S16.	1.6	0
57	Genetic landscape of Leber's hereditary optic neuropathy: reflection on pathogenic mechanisms. Acta Ophthalmologica, 2015, 93, n/a-n/a.	0.6	0
58	Efficient mitochondrial biogenesis drives incomplete penetrance in Leberâ€™s hereditary optic neuropathy. Brain, 2014, 137, 335-353.	3.7	229
59	HPLC-UV analysis of thymidine and deoxyuridine in plasma of patients with thymidine phosphorylase deficiency. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2014, 949-950, 58-62.	1.2	10
60	Association of the mtDNA m.4171C>A/MT-ND1 mutation with both optic neuropathy and bilateral brainstem lesions. BMC Neurology, 2014, 14, 116.	0.8	28
61	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
62	Early Macular Retinal Ganglion Cell Loss in Dominant Optic Atrophy: Genotype-Phenotype Correlation. American Journal of Ophthalmology, 2014, 158, 628-636.e3.	1.7	56
63	Genetic Basis of Mitochondrial Optic Neuropathies. Current Molecular Medicine, 2014, 14, 985-992.	0.6	16
64	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
65	Oncocytic glioblastoma: a glioblastoma showing oncocytic changes and increased mitochondrial DNA copy number. Human Pathology, 2013, 44, 1867-1876.	1.1	15
66	The optic nerve: A â€œmito-windowâ€•on mitochondrial neurodegeneration. Molecular and Cellular Neurosciences, 2013, 55, 62-76.	1.0	78
67	Idebenone treatment in patients with OPA1-mutant dominant optic atrophy. Brain, 2013, 136, e231-e231.	3.7	62
68	Revisiting the issue of mitochondrial DNA content in optic mitochondriopathies. Neurology, 2012, 79, 1517-1519.	1.5	13
69	Searching for genetic modifiers of Leber's hereditary optic neuropathy penetrance. Mitochondrion, 2012, 12, 572.	1.6	0
70	Multi-system neurological disease is common in patients with OPA1 mutations. Brain, 2010, 133, 771-786.	3.7	385
71	Variation in OPA1 does not explain the incomplete penetrance of Leber hereditary optic neuropathy. Molecular Vision, 2010, 16, 2760-4.	1.1	4
72	Terminal-restriction fragment length polymorphism analysis of biphenyl dioxygenase genes from a polychlorinated biphenyl-polluted soil. Research in Microbiology, 2009, 160, 742-750.	1.0	7

#	Article	IF	Citations
73	Case Report: Optic Atrophy and Nephropathy With m.13513G>A/MT-ND5 mtDNA Pathogenic Variant. Frontiers in Genetics, 0, 13, .	1.1	3