

# Leonardo Caporali

## List of Publications by Year in descending order

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73  
papers

2,993  
citations

218592

26  
h-index

175177

52  
g-index

78  
all docs

78  
docs citations

78  
times ranked

4648  
citing authors

#	ARTICLE	IF	CITATIONS
1	Multi-system neurological disease is common in patients with OPA1 mutations. <i>Brain</i> , 2010, 133, 771-786.	3.7	385
2	Efficient mitochondrial biogenesis drives incomplete penetrance in Leber's hereditary optic neuropathy. <i>Brain</i> , 2014, 137, 335-353.	3.7	229
3	Pharmacological Inhibition of Necroptosis Protects from Dopaminergic Neuronal Cell Death in Parkinson's Disease Models. <i>Cell Reports</i> , 2018, 22, 2066-2079.	2.9	167
4	Mutations in SLC25A46, encoding a UGO1-like protein, cause an optic atrophy spectrum disorder. <i>Nature Genetics</i> , 2015, 47, 926-932.	9.4	166
5	OPA1 Isoforms in the Hierarchical Organization of Mitochondrial Functions. <i>Cell Reports</i> , 2017, 19, 2557-2571.	2.9	158
6	Syndromic parkinsonism and dementia associated with OPA1 missense mutations. <i>Annals of Neurology</i> , 2015, 78, 21-38.	2.8	154
7	Infant and Adult Gut Microbiome and Metabolome in Rural Bassa and Urban Settlers from Nigeria. <i>Cell Reports</i> , 2018, 23, 3056-3067.	2.9	128
8	Impaired complex I repair causes recessive Leber's hereditary optic neuropathy. <i>Journal of Clinical Investigation</i> , 2021, 131, .	3.9	89
9	OPA1-related auditory neuropathy: site of lesion and outcome of cochlear implantation. <i>Brain</i> , 2015, 138, 563-576.	3.7	86
10	Liver transplantation for mitochondrial neurogastrointestinal encephalomyopathy. <i>Annals of Neurology</i> , 2016, 80, 448-455.	2.8	81
11	The optic nerve: A 'mito-window' on mitochondrial neurodegeneration. <i>Molecular and Cellular Neurosciences</i> , 2013, 55, 62-76.	1.0	78
12	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. <i>Journal of Clinical Investigation</i> , 2019, 130, 108-125.	3.9	65
13	Mitochondria: Biogenesis and mitophagy balance in segregation and clonal expansion of mitochondrial DNA mutations. <i>International Journal of Biochemistry and Cell Biology</i> , 2015, 63, 21-24.	1.2	63
14	Idebenone treatment in patients with OPA1-mutant dominant optic atrophy. <i>Brain</i> , 2013, 136, e231-e231.	3.7	62
15	Dna Methyltransferase 1 Mutations and Mitochondrial Pathology: Is Mtdna Methylated?. <i>Frontiers in Genetics</i> , 2015, 6, 90.	1.1	62
16	Somatic mutation profiling of hobnail variant of papillary thyroid carcinoma. <i>Endocrine-Related Cancer</i> , 2017, 24, 107-117.	1.6	58
17	Early Macular Retinal Ganglion Cell Loss in Dominant Optic Atrophy: Genotype-Phenotype Correlation. <i>American Journal of Ophthalmology</i> , 2014, 158, 628-636.e3.	1.7	56
18	Incomplete penetrance in mitochondrial optic neuropathies. <i>Mitochondrion</i> , 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. <i>Brain</i> , 2016, 139, e17-e17.	3.7	51
20	Behr syndrome™ with OPA1 compound heterozygote mutations. <i>Brain</i> , 2015, 138, e321-e321.	3.7	50
21	Peculiar combinations of individually non-pathogenic missense mitochondrial DNA variants cause low penetrance Leber™s hereditary optic neuropathy. <i>PLoS Genetics</i> , 2018, 14, e1007210.	1.5	47
22	A Novel in-Frame 18-bp Microdeletion in <i>MT-CYB</i> Causes a Multisystem Disorder with Prominent Exercise Intolerance. <i>Human Mutation</i> , 2014, 35, 954-958.	1.1	38
23	Deciphering OPA1 mutations pathogenicity by combined analysis of human, mouse and yeast cell models. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2018, 1864, 3496-3514.	1.8	36
24	Mitochondrial diseases in adults. <i>Journal of Internal Medicine</i> , 2020, 287, 592-608.	2.7	33
25	Calcium mishandling in absence of primary mitochondrial dysfunction drives cellular pathology in Wolfram Syndrome. <i>Scientific Reports</i> , 2020, 10, 4785.	1.6	33
26	ATPase Domain <i>AFG3L2</i> Mutations Alter OPA1 Processing and Cause Optic Neuropathy. <i>Annals of Neurology</i> , 2020, 88, 18-32.	2.8	31
27	Mutations in the m-AAA proteases <i>AFG3L2</i> and <i>SPG7</i> are causing isolated dominant optic atrophy. <i>Neurology: Genetics</i> , 2020, 6, e428.	0.9	31
28	Whole Mitogenomes Reveal the History of Swamp Buffalo: Initially Shaped by Glacial Periods and Eventually Modelled by Domestication. <i>Scientific Reports</i> , 2017, 7, 4708.	1.6	30
29	Association of the mtDNA m.4171C>A/MT-ND1 mutation with both optic neuropathy and bilateral brainstem lesions. <i>BMC Neurology</i> , 2014, 14, 116.	0.8	28
30	Diffusion Tensor Imaging Mapping of Brain White Matter Pathology in Mitochondrial Optic Neuropathies. <i>American Journal of Neuroradiology</i> , 2015, 36, 1259-1265.	1.2	28
31	Liver transplant reverses biochemical imbalance in mitochondrial neurogastrointestinal encephalomyopathy. <i>Mitochondrion</i> , 2017, 34, 101-102.	1.6	23
32	Haplogroup J mitogenomes are the most sensitive to the pesticide rotenone: Relevance for human diseases. <i>Neurobiology of Disease</i> , 2018, 114, 129-139.	2.1	22
33	DNMT1 mutations leading to neurodegeneration paradoxically reflect on mitochondrial metabolism. <i>Human Molecular Genetics</i> , 2020, 29, 1864-1881.	1.4	19
34	Identification of rare genetic variants in Italian patients with dementia by targeted gene sequencing. <i>Neurobiology of Aging</i> , 2018, 66, 180.e23-180.e31.	1.5	18
35	Cerebral Mitochondrial Microangiopathy Leads to Leukoencephalopathy in Mitochondrial Neurogastrointestinal Encephalopathy. <i>American Journal of Neuroradiology</i> , 2018, 39, 427-434.	1.2	18
36	Cybrid studies establish the causal link between the mtDNA m.3890G>A/MT-ND1 mutation and optic atrophy with bilateral brainstem lesions. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2013, 1832, 445-452.	1.8	17

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37	Liver transplantation in mitochondrial neurogastrointestinal encephalomyopathy (MNGIE): clinical long-term follow-up and pathogenic implications. <i>Journal of Neurology</i> , 2020, 267, 3702-3710.	1.8	17
38	Dominant <i>ACO2</i> mutations are a frequent cause of isolated optic atrophy. <i>Brain Communications</i> , 2021, 3, fcab063.	1.5	16
39	Genetic Basis of Mitochondrial Optic Neuropathies. <i>Current Molecular Medicine</i> , 2014, 14, 985-992.	0.6	16
40	Oncocytic glioblastoma: a glioblastoma showing oncocytic changes and increased mitochondrial DNA copy number. <i>Human Pathology</i> , 2013, 44, 1867-1876.	1.1	15
41	DGUOK recessive mutations in patients with CPEO, mitochondrial myopathy, parkinsonism and mtDNA deletions. <i>Brain</i> , 2018, 141, e3-e3.	3.7	15
42	First TMEM126A missense mutation in an Italian proband with optic atrophy and deafness. <i>Neurology: Genetics</i> , 2019, 5, e329.	0.9	14
43	Revisiting the issue of mitochondrial DNA content in optic mitochondriopathies. <i>Neurology</i> , 2012, 79, 1517-1519.	1.5	13
44	ITA-MNGIE: an Italian regional and national survey for mitochondrial neuro-gastro-intestinal encephalomyopathy. <i>Neurological Sciences</i> , 2016, 37, 1149-1151.	0.9	13
45	Novel mutations in DNA2 associated with myopathy and mtDNA instability. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1893-1899.	1.7	13
46	Leber's Hereditary Optic Neuropathy: A Report on Novel mtDNA Pathogenic Variants. <i>Frontiers in Neurology</i> , 2021, 12, 657317.	1.1	13
47	HPLC-UV analysis of thymidine and deoxyuridine in plasma of patients with thymidine phosphorylase deficiency. <i>Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences</i> , 2014, 949-950, 58-62.	1.2	10
48	Brain MRS correlates with mitochondrial dysfunction biomarkers in MELAS-associated mtDNA mutations. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 1200-1211.	1.7	10
49	Epilepsy in <i>MT-ATP6</i> -related milt/NARP: correlation of electroclinical features with heteroplasmy. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 704-710.	1.7	10
50	Papillary thyroid carcinoma tall cell variant shares accumulation of mitochondria, mitochondrial DNA mutations, and loss of oxidative phosphorylation complex I integrity with oncocytic tumors. <i>Journal of Pathology: Clinical Research</i> , 2022, 8, 155-168.	1.3	10
51	Combined Optic Atrophy and Rodâ€‘Cone Dystrophy Expands the RTN4IP1 (Optic Atrophy 10) Phenotype. <i>Journal of Neuro-Ophthalmology</i> , 2021, 41, e290-e292.	0.4	9
52	The relevance of mitochondrial DNA variants fluctuation during reprogramming and neuronal differentiation of human iPSCs. <i>Stem Cell Reports</i> , 2021, 16, 1953-1967.	2.3	8
53	Capturing the Pattern of Transition From Carrier to Affected in Leber Hereditary Optic Neuropathy. <i>American Journal of Ophthalmology</i> , 2022, 241, 71-79.	1.7	8
54	Terminal-restriction fragment length polymorphism analysis of biphenyl dioxygenase genes from a polychlorinated biphenyl-polluted soil. <i>Research in Microbiology</i> , 2009, 160, 742-750.	1.0	7

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55	Reply: Both mitochondrial DNA and mitonuclear gene mutations cause hearing loss through cochlear dysfunction. <i>Brain</i> , 2016, 139, e34-e34.	3.7	7
56	Complex II phosphorylation is triggered by unbalanced redox homeostasis in cells lacking complex III. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , 2018, 1859, 182-190.	0.5	7
57	Missense <i>PDSS1</i> mutations in CoenzymeQ10 synthesis cause optic atrophy and sensorineural deafness. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 247-251.	1.7	7
58	TYMP Variants Result in Late-Onset Mitochondrial Myopathy With Altered Muscle Mitochondrial DNA Homeostasis. <i>Frontiers in Genetics</i> , 2020, 11, 860.	1.1	6
59	Epilepsy with auditory features: Contribution of known genes in 112 patients. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2021, 85, 115-118.	0.9	6
60	Pharmacogenetics and Treatment Response in Narcolepsy Type 1. <i>Clinical Neuropharmacology</i> , 2016, 39, 18-23.	0.2	5
61	<i>SCN1A</i> mutations in focal epilepsy with auditory features: widening the spectrum of GEFS <i>plus</i> . <i>Epileptic Disorders</i> , 2019, 21, 185-191.	0.7	5
62	The Pattern of Retinal Ganglion Cell Loss in Wolfram Syndrome is Distinct From Mitochondrial Optic Neuropathies. <i>American Journal of Ophthalmology</i> , 2022, 241, 206-216.	1.7	5
63	The m.3890G>A/MT-ND1 mtDNA rare pathogenic variant: Expanding clinical and MRI phenotypes. <i>Mitochondrion</i> , 2021, 60, 142-149.	1.6	4
64	Variation in OPA1 does not explain the incomplete penetrance of Leber hereditary optic neuropathy. <i>Molecular Vision</i> , 2010, 16, 2760-4.	1.1	4
65	The Mitogenome Relationships and Phylogeography of Barn Swallows ( <i>Hirundo rustica</i> ). <i>Molecular Biology and Evolution</i> , 2022, 39, .	3.5	4
66	An increased burden of rare exonic variants in NRXN1 microdeletion carriers is likely to enhance the penetrance for autism spectrum disorder. <i>Journal of Cellular and Molecular Medicine</i> , 2021, 25, 2459-2470.	1.6	3
67	A novel <i>ALG14</i> missense variant in an alive child with myopathy, epilepsy, and progressive cerebral atrophy. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1918-1921.	0.7	3
68	Molecular biomarkers correlate with brain grey and white matter changes in patients with mitochondrial m.3243A>G mutation. <i>Molecular Genetics and Metabolism</i> , 2022, 135, 72-81.	0.5	3
69	The role of mtDNA haplogroups on metabolic features in narcolepsy type 1. <i>Mitochondrion</i> , 2022, 63, 37-42.	1.6	3
70	Case Report: Optic Atrophy and Nephropathy With m.13513G>A/MT-ND5 mtDNA Pathogenic Variant. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	3
71	Searching for genetic modifiers of Leber's hereditary optic neuropathy penetrance. <i>Mitochondrion</i> , 2012, 12, 572.	1.6	0
72	An Ugo1-like protein is associated with optic atrophy <i>plus</i> ™ disorders. <i>Mitochondrion</i> , 2015, 24, S16.	1.6	0

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73	Genetic landscape of Leber's hereditary optic neuropathy: reflection on pathogenic mechanisms. Acta Ophthalmologica, 2015, 93, n/a-n/a.	0.6	0