Marcela Votruba

List of Publications by Citations

99 papers	8,625 citations	29 h-index	92 g-index
115 ext. papers	9,484 ext. citations	5.5 avg, IF	4.96 L-index

#	Paper	IF	Citations
99	Guidelines for the use and interpretation of assays for monitoring autophagy. *Autophagy*, **2012**, 8, 445-544	10.2	2783
98	LDL receptor-related protein 5 (LRP5) affects bone accrual and eye development. *Cell*, **2001**, 107, 513-23	36.2	1827
97	OPA1, encoding a dynamin-related GTPase, is mutated in autosomal dominant optic atrophy linked to chromosome 3q28. *Nature Genetics*, **2000**, 26, 211-5	36.3	1039
96	Opa1 deficiency in a mouse model of autosomal dominant optic atrophy impairs mitochondrial morphology, optic nerve structure and visual function. *Human Molecular Genetics*, **2007**, 16, 1307-18	5.6	319
95	Multi-system neurological disease is common in patients with OPA1 mutations. *Brain*, **2010**, 133, 771-86	11.2	314
94	OPA1 mutation and late-onset cardiomyopathy: mitochondrial dysfunction and mtDNA instability. *Journal of the American Heart Association*, **2012**, 1, e003012	6	128
93	Clinical features in affected individuals from 21 pedigrees with dominant optic atrophy. *JAMA Ophthalmology*, **1998**, 116, 351-8		127
92	A major marker for normal tension glaucoma: association with polymorphisms in the OPA1 gene. *Human Genetics*, **2002**, 110, 52-6	6.3	101
91	Treatment strategies for inherited optic neuropathies: past, present and future. *Eye*, **2014**, 28, 521-37	4.4	98
90	Clinical features, molecular genetics, and pathophysiology of dominant optic atrophy. *Journal of Medical Genetics*, **1998**, 35, 793-800	5.8	91
89	A neurodegenerative perspective on mitochondrial optic neuropathies. *Acta Neuropathologica*, **2016**, 132, 789-806	14.3	90
88	Opa1 deficiency in a mouse model of dominant optic atrophy leads to retinal ganglion cell dendropathy. *Brain*, **2010**, 133, 2942-51	11.2	84
87	International Consensus Statement on the Clinical and Therapeutic Management of Leber Hereditary Optic Neuropathy. *Journal of Neuro-Ophthalmology*, **2017**, 37, 371-381	2.6	84
86	OPA1 deficiency associated with increased autophagy in retinal ganglion cells in a murine model of dominant optic atrophy **2009**, 50, 2567-71		76
85	Opa1 is essential for retinal ganglion cell synaptic architecture and connectivity. *Brain*, **2012**, 135, 493-505	11.2	74
84	Optic disc morphology of patients with OPA1 autosomal dominant optic atrophy. *British Journal of Ophthalmology*, **2003**, 87, 48-53	5.5	71
83	Retinal ganglion cell dendritic degeneration in a mouse model of Alzheimer's disease. *Neurobiology of Aging*, **2013**, 34, 1799-806	5.6	69

82	Comparative study of fibrillar collagen arrangement in the corneas of primates and other mammals. *Anatomical Record*, **2007**, 290, 1542-50	2.1	65
81	A comprehensive survey of mutations in the OPA1 gene in patients with autosomal dominant optic atrophy. *Investigative Ophthalmology and Visual Science*, **2002**, 43, 1715-24		62
80	Dysregulated mitophagy and mitochondrial organization in optic atrophy due to OPA1 mutations. *Neurology*, **2017**, 88, 131-142	6.5	60
79	A frameshift mutation in exon 28 of the OPA1 gene explains the high prevalence of dominant optic atrophy in the Danish population: evidence for a founder effect. *Human Genetics*, **2001**, 109, 498-502	6.3	54
78	Electrophysiological findings in dominant optic atrophy (DOA) linking to the OPA1 locus on chromosome 3q 28-qter. *Documenta Ophthalmologica*, **1998**, 95, 217-28	2.2	46
77	Molecular genetic basis of primary inherited optic neuropathies. *Eye*, **2004**, 18, 1126-32	4.4	45
76	Neovascular age-related macular degeneration: present and future treatment options. *Eye*, **2001**, 15, 424-9	4.4	45
75	Developmental expression profile of the optic atrophy gene product: OPA1 is not localized exclusively in the mammalian retinal ganglion cell layer. *Investigative Ophthalmology and Visual Science*, **2004**, 45, 1667-73		42
74	Childhood-onset Leber hereditary optic neuropathy. *British Journal of Ophthalmology*, **2017**, 101, 1505-1509	3.9	34
73	A missense mutation in the murine Opa3 gene models human Costeff syndrome. *Brain*, **2008**, 131, 368-80	11.2	30
72	SSBP1 mutations in dominant optic atrophy with variable retinal degeneration. *Annals of Neurology*, **2019**, 86, 368-383	9.4	29
71	The Sight Loss and Vision Priority Setting Partnership (SLV-PSP): overview and results of the research prioritisation survey process. *BMJ Open*, **2014**, 4, e004905	3	29
70	Colour discrimination ellipses in patients with dominant optic atrophy. *Vision Research*, **1998**, 38, 3413-9	2.1	27
69	Mouse models of dominant optic atrophy: what do they tell us about the pathophysiology of visual loss?. *Vision Research*, **2011**, 51, 229-34	2.1	26
68	Demonstration of a founder effect and fine mapping of dominant optic atrophy locus on 3q28-qter by linkage disequilibrium method: a study of 38 British Isles pedigrees. *Human Genetics*, **1998**, 102, 79-86	6.3	26
67	Focus on molecules: the OPA1 protein. *Experimental Eye Research*, **2006**, 83, 1003-4	3.7	24
66	Electrophysiology and ocular blood flow in a family with dominant optic nerve atrophy and a mutation in the OPA1 gene. *Ophthalmic Genetics*, **2003**, 24, 233-45	1.2	24
65	Real-World Clinical Experience With Idebenone in the Treatment of Leber Hereditary Optic Neuropathy. *Journal of Neuro-Ophthalmology*, **2020**, 40, 558-565	2.6	24

64	Can the retina be used to diagnose and plot the progression of Alzheimer's disease?. *Acta Ophthalmologica*, **2017**, 95, 768-777	3.7	23
63	Novel OPA1 missense mutation in a family with optic atrophy and severe widespread neurological disorder. *Acta Ophthalmologica*, **2013**, 91, e225-31	3.7	23
62	A review of primary hereditary optic neuropathies. *Journal of Inherited Metabolic Disease*, **2003**, 26, 209-27	3.4	23
61	Genetic refinement of dominant optic atrophy (OPA1) locus to within a 2 cM interval of chromosome 3q. *Journal of Medical Genetics*, **1997**, 34, 117-21	5.8	22
60	MRI of the intraorbital optic nerve in patients with autosomal dominant optic atrophy. *Neuroradiology*, **2000**, 42, 180-3	3.2	22
59	A randomized, placebo-controlled trial of the benzoquinone idebenone in a mouse model of OPA1-related dominant optic atrophy reveals a limited therapeutic effect on retinal ganglion cell dendropathy and visual function. *Neuroscience*, **2016**, 319, 92-106	3.9	21
58	Mitochondrial dysfunction in an Opa1(Q285STOP) mouse model of dominant optic atrophy results from Opa1 haploinsufficiency. *Cell Death and Disease*, **2016**, 7, e2309	9.8	20
57	The pupil in dominant optic atrophy. *Investigative Ophthalmology and Visual Science*, **2001**, 42, 675-8		20
56	The pattern of retinal ganglion cell dysfunction in Leber hereditary optic neuropathy. *Mitochondrion*, **2017**, 36, 138-149	4.9	19
55	Specific deficits in visual electrophysiology in a mouse model of dominant optic atrophy. *Experimental Eye Research*, **2011**, 93, 771-7	3.7	19
54	Secondary mtDNA defects do not cause optic nerve dysfunction in a mouse model of dominant optic atrophy **2009**, 50, 4561-6		19
53	Non-image-forming light driven functions are preserved in a mouse model of autosomal dominant optic atrophy. *PLoS ONE*, **2013**, 8, e56350	3.7	18
52	Opa3, a novel regulator of mitochondrial function, controls thermogenesis and abdominal fat mass in a mouse model for Costeff syndrome. *Human Molecular Genetics*, **2012**, 21, 4836-44	5.6	18
51	Mutations in the m-AAA proteases AFG3L2 and SPG7 are causing isolated dominant optic atrophy. *Neurology: Genetics*, **2020**, 6, e428	3.8	18
50	The management of solitary trichoepithelioma versus basal cell carcinoma. *Eye*, **1998**, 12 (Pt 1), 43-6	4.4	15
49	Clinical utility gene card for: inherited optic neuropathies including next-generation sequencing-based approaches. *European Journal of Human Genetics*, **2019**, 27, 494-502	5.3	15
48	Changes in corneal collagen architecture during mouse postnatal development **2010**, 51, 2936-42		14
47	Mitochondrial localization and ocular expression of mutant Opa3 in a mouse model of 3-methylglutaconicaciduria type III **2011**, 52, 4369-80		14

46	Clinical and molecular genetic findings in autosomal dominant OPA3-related optic neuropathy. *Neurogenetics*, **2015**, 16, 69-75	3	13
45	Symmetric arrangement of mitochondria:plasma membrane contacts between adjacent photoreceptor cells regulated by Opa1. *Proceedings of the National Academy of Sciences of the United States of America*, **2020**, 117, 15684-15693	11.5	13
44	Incidence and patterns of detection and management of childhood-onset hereditary retinal disorders in the UK. *British Journal of Ophthalmology*, **2012**, 96, 360-5	5.5	13
43	Photostimulation of mitochondria as a treatment for retinal neurodegeneration. *Mitochondrion*, **2017**, 36, 85-95	4.9	12
42	The Pattern of Retinal Ganglion Cell Loss in OPA1-Related Autosomal Dominant Optic Atrophy Inferred From Temporal, Spatial, and Chromatic Sensitivity Losses **2017**, 58, 502-516		12
41	Peripapillary microcirculation in Leber hereditary optic neuropathy. *Acta Ophthalmologica*, **2019**, 97, e71-e76	3.7	12
40	Nicotinamide provides neuroprotection in glaucoma by protecting against mitochondrial and metabolic dysfunction. *Redox Biology*, **2021**, 43, 101988	11.3	12
39	Opa1 Deficiency Leads to Diminished Mitochondrial Bioenergetics With Compensatory Increased Mitochondrial Motility **2020**, 61, 42		11
38	Electrophysiological ON and OFF Responses in Autosomal Dominant Optic Atrophy **2015**, 56, 7629-37		10
37	Potential Therapeutic Benefit of NAD Supplementation for Glaucoma and Age-Related Macular Degeneration. *Nutrients*, **2020**, 12,	6.7	10
36	The ying and yang of idebenone: Not too little, not too much - cell death in NQO1 deficient cells and the mouse retina. *Free Radical Biology and Medicine*, **2020**, 152, 551-560	7.8	9
35	Perceptions and understanding of genetics and genetic eye disease and attitudes to genetic testing and gene therapy in a primary eye care setting. *Ophthalmic Genetics*, **2015**, 36, 50-7	1.2	8
34	Primary monophasic synovial sarcoma of the conjunctiva. *British Journal of Ophthalmology*, **2002**, 86, 1453-4	5.5	8
33	Visual and psychological morbidity among patients with autosomal dominant optic atrophy. *Acta Ophthalmologica*, **2013**, 91, e413-4	3.7	7
32	The Relationship Between the Photopic Negative Response and Retinal Ganglion Cell Topography **2019**, 60, 1879-1887		6
31	Red Light Treatment in an Axotomy Model of Neurodegeneration. *Photochemistry and Photobiology* , **2016**, 92, 624-31	3.6	6
30	Validating the RedMIT/GFP-LC3 Mouse Model by Studying Mitophagy in Autosomal Dominant Optic Atrophy Due to the OPA1Q285STOP Mutation. *Frontiers in Cell and Developmental Biology*, **2018**, 6, 103	5.7	6
29	OPA1 analysis in an international series of probands with bilateral optic atrophy. *Acta Ophthalmologica*, **2017**, 95, 363-369	3.7	5

28	Disrupted mitochondrial function in the Opa3L122P mouse model for Costeff Syndrome impairs skeletal integrity. *Human Molecular Genetics*, **2016**, 25, 2404-2416	5.6	5
27	Leber optic neuropathy - visual return on alcohol cessation. *Acta Ophthalmologica*, **2012**, 90, e568	3.7	5
26	Thymoxamine in the treatment of traumatic mydriasis. *British Journal of Ophthalmology*, **1993**, 77, 681	5.5	4
25	Dominant optic atrophy: exclusion and fine genetic mapping of the candidate gene, HRY. *Mammalian Genome*, **1998**, 9, 784-7	3.2	3
24	OPA1 deficiency accelerates hippocampal synaptic remodelling and age-related deficits in learning and memory. *Brain Communications*, **2020**, 2, fcaa101	4.5	3
23	Discovery of Novel 2-Aniline-1,4-naphthoquinones as Potential New Drug Treatment for Leber Hereditary Optic Neuropathy (LHON). *Journal of Medicinal Chemistry*, **2020**, 63, 13638-13655	8.3	3
22	A novel NR2E3 gene mutation in autosomal recessive retinitis pigmentosa with cystic maculopathy. *Acta Ophthalmologica*, **2018**, 96, e535-e536	3.7	3
21	Retinal changes in a patient with acquired partial lipodystrophy (Laignel-Lavastine and Viard Syndrome). *Acta Ophthalmologica*, **2015**, 93, e598-9	3.7	2
20	Nicotinamide provides neuroprotection in glaucoma by protecting against mitochondrial and metabolic dysfunction		2
19	Opa1 Deficiency Promotes Development of Retinal Vascular Lesions in Diabetic Retinopathy. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	2
18	Red Light Irradiation In Vivo Upregulates DJ-1 in the Retinal Ganglion Cell Layer and Protects against Axotomy-Related Dendritic Pruning. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	2
17	Real-World Clinical Experience With Idebenone in the Treatment of Leber Hereditary Optic Neuropathy-Response to Dr. Finsterer Letter. *Journal of Neuro-Ophthalmology*, **2021**,	2.6	2
16	Mean cellular volume in a patient with Leber optic neuropathy and visual return on alcohol cessation. *Acta Ophthalmologica*, **2014**, 92, e77	3.7	1
15	The case of the disappearing bullet. *Eye*, **1994**, 8 (Pt 1), 143-4	4.4	1
14	The UK Sight Loss and Vision Priority Setting Partnership (SLV-PSP): Vision research questions prioritised by patients and health care professionals. *Acta Ophthalmologica*, **2013**, 91, 0-0	3.7	1
13	Inherited Optic Neuropathies **2008**, 51-67		0
12	Inherited dominant optic neuropathy: from clinical studies to gene function and back again. *Drug Discovery Today: Disease Models*, **2013**, 10, e173-e180	1.3	
11	Association between autosomal dominant optic atrophy and Ewing sarcoma. *Journal of Pediatric Hematology/Oncology*, **1999**, 21, 557-9	1.2	

List of Publications

10	Visual dysfunction and pupillary responses are dissociated in the Opa3 mutant mouse with retinal degeneration. *Acta Ophthalmologica*, **2014**, 92, 0-0	3.7
9	Specific deficits in visual electrophysiology without retinal cellular apoptosis in a mouse model of dominant optic atrophy. *Acta Ophthalmologica*, **2010**, 88, 0-0	3.7
8	No retinal apoptosis in mouse model of Opa1 dominant optic atrophy despite visual loss. *Acta Ophthalmologica*, **2010**, 88, 0-0	3.7
7	Dendritic pruning of retinal ganglion cells in a mouse model of dominant optic atrophy (ADOA). *Acta Ophthalmologica*, **2010**, 88, 0-0	3.7
6	Heterozygous B6;C3-Opa1Q285STOP mouse model of dominant optic atrophy displays subtle neuromuscular features with age. *Acta Ophthalmologica*, **2010**, 88, 0-0	3.7
5	Changes in retinal neuronal connectivity in a mouse model of dominant optic atrophy. *Acta Ophthalmologica*, **2011**, 89, 0-0	3.7
4	Mitochondrial optic neuropathies. *Acta Ophthalmologica*, **2012**, 90, 0-0	3.7
3	Ganglion Cell Diseases **2014**, 427-440	
2	A Perspective on Accelerated Aging Caused by the Genetic Deficiency of the Metabolic Protein, OPA1. *Frontiers in Neurology*, **2021**, 12, 641259	4.1
1	New avenues for therapy in mitochondrial optic neuropathies. *Therapeutic Advances in Rare Disease* , **2021**, 2, 263300402110290	1.4