# Marcela Votruba

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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|---|---|---|---|
| 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 | A Perspective on Accelerated Aging Caused by the Genetic Deficiency of the Metabolic Protein, OPA1. *Frontiers in Neurology*, **2021**, 12, 641259 | 4.1 | |
| 98 | Opa1 Deficiency Promotes Development of Retinal Vascular Lesions in Diabetic Retinopathy. *International Journal of Molecular Sciences*, **2021**, 22, | 6.3 | 2 |
| 97 | New avenues for therapy in mitochondrial optic neuropathies. *Therapeutic Advances in Rare Disease* , **2021**, 2, 263300402110290 | 1.4 | |
| 96 | Nicotinamide provides neuroprotection in glaucoma by protecting against mitochondrial and metabolic dysfunction. *Redox Biology*, **2021**, 43, 101988 | 11.3 | 12 |
| 95 | 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 |
| 94 | Real-World Clinical Experience With Idebenone in the Treatment of Leber Hereditary Optic Neuropathy-Response to Dr. FinstererR Letter. *Journal of Neuro-Ophthalmology*, **2021**, | 2.6 | 2 |
| 93 | Opa1 Deficiency Leads to Diminished Mitochondrial Bioenergetics With Compensatory Increased Mitochondrial Motility **2020**, 61, 42 | | 11 |
| 92 | 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 |
| 91 | Mutations in the m-AAA proteases AFG3L2 and SPG7 are causing isolated dominant optic atrophy. *Neurology: Genetics*, **2020**, 6, e428 | 3.8 | 18 |
| 90 | 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 |
| 89 | OPA1 deficiency accelerates hippocampal synaptic remodelling and age-related deficits in learning and memory. *Brain Communications*, **2020**, 2, fcaa101 | 4.5 | 3 |
| 88 | 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 |
| 87 | Discovery of Novel 2-Aniline-1,4-naphthoquinones as Potential New Drug Treatment for LeberR Hereditary Optic Neuropathy (LHON). *Journal of Medicinal Chemistry*, **2020**, 63, 13638-13655 | 8.3 | 3 |
| 86 | Potential Therapeutic Benefit of NAD Supplementation for Glaucoma and Age-Related Macular Degeneration. *Nutrients*, **2020**, 12, | 6.7 | 10 |
| 85 | SSBP1 mutations in dominant optic atrophy with variable retinal degeneration. *Annals of Neurology*, **2019**, 86, 368-383 | 9.4 | 29 |
| 84 | The Relationship Between the Photopic Negative Response and Retinal Ganglion Cell Topography **2019**, 60, 1879-1887 | | 6 |
| 83 | 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 |

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| 82 | Peripapillary microcirculation in Leber hereditary optic neuropathy. *Acta Ophthalmologica*, **2019**, 97, e71-e76 | 3.7 | 12 |
| 81 | A novel NR2E3 gene mutation in autosomal recessive retinitis pigmentosa with cystic maculopathy. *Acta Ophthalmologica*, **2018**, 96, e535-e536 | 3.7 | 3 |
| 80 | 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 |
| 79 | Photostimulation of mitochondria as a treatment for retinal neurodegeneration. *Mitochondrion*, **2017**, 36, 85-95 | 4.9 | 12 |
| 78 | Childhood-onset Leber hereditary optic neuropathy. *British Journal of Ophthalmology*, **2017**, 101, 1505-1509 | 3.9 | 34 |
| 77 | OPA1 analysis in an international series of probands with bilateral optic atrophy. *Acta Ophthalmologica*, **2017**, 95, 363-369 | 3.7 | 5 |
| 76 | Dysregulated mitophagy and mitochondrial organization in optic atrophy due to OPA1 mutations. *Neurology*, **2017**, 88, 131-142 | 6.5 | 60 |
| 75 | 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 |
| 74 | The pattern of retinal ganglion cell dysfunction in Leber hereditary optic neuropathy. *Mitochondrion*, **2017**, 36, 138-149 | 4.9 | 19 |
| 73 | Can the retina be used to diagnose and plot the progression of Alzheimer's disease?. *Acta Ophthalmologica*, **2017**, 95, 768-777 | 3.7 | 23 |
| 72 | 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 |
| 71 | 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 |
| 70 | Disrupted mitochondrial function in the Opa3L122P mouse model for Costeff Syndrome impairs skeletal integrity. *Human Molecular Genetics*, **2016**, 25, 2404-2416 | 5.6 | 5 |
| 69 | 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 |
| 68 | Red Light Treatment in an Axotomy Model of Neurodegeneration. *Photochemistry and Photobiology* , **2016**, 92, 624-31 | 3.6 | 6 |
| 67 | A neurodegenerative perspective on mitochondrial optic neuropathies. *Acta Neuropathologica*, **2016**, 132, 789-806 | 14.3 | 90 |
| 66 | Clinical and molecular genetic findings in autosomal dominant OPA3-related optic neuropathy. *Neurogenetics*, **2015**, 16, 69-75 | 3 | 13 |
| 65 | Retinal changes in a patient with acquired partial lipodystrophy (Laignel-Lavastine and Viard Syndrome). *Acta Ophthalmologica*, **2015**, 93, e598-9 | 3.7 | 2 |

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| 64 | Electrophysiological ON and OFF Responses in Autosomal Dominant Optic Atrophy **2015**, 56, 7629-37 | | 10 |
| 63 | 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 |
| 62 | Treatment strategies for inherited optic neuropathies: past, present and future. *Eye*, **2014**, 28, 521-37 | 4.4 | 98 |
| 61 | 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 |
| 60 | Mean cellular volume in a patient with Leber's optic neuropathy and visual return on alcohol cessation. *Acta Ophthalmologica*, **2014**, 92, e77 | 3.7 | 1 |
| 59 | Visual dysfunction and pupillary responses are dissociated in the Opa3 mutant mouse with retinal degeneration. *Acta Ophthalmologica*, **2014**, 92, 0-0 | 3.7 | |
| 58 | Ganglion Cell Diseases **2014**, 427-440 | | |
| 57 | Inherited dominant optic neuropathy: from clinical studies to gene function and back again. *Drug Discovery Today: Disease Models*, **2013**, 10, e173-e180 | 1.3 | |
| 56 | Retinal ganglion cell dendritic degeneration in a mouse model of Alzheimer's disease. *Neurobiology of Aging*, **2013**, 34, 1799-806 | 5.6 | 69 |
| 55 | Visual and psychological morbidity among patients with autosomal dominant optic atrophy. *Acta Ophthalmologica*, **2013**, 91, e413-4 | 3.7 | 7 |
| 54 | Novel OPA1 missense mutation in a family with optic atrophy and severe widespread neurological disorder. *Acta Ophthalmologica*, **2013**, 91, e225-31 | 3.7 | 23 |
| 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 | 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 |
| 51 | Leber's optic neuropathy - visual return on alcohol cessation. *Acta Ophthalmologica*, **2012**, 90, e568 | 3.7 | 5 |
| 50 | Guidelines for the use and interpretation of assays for monitoring autophagy. *Autophagy*, **2012**, 8, 445-544 | 10.2 | 2783 |
| 49 | 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 |
| 48 | OPA1 mutation and late-onset cardiomyopathy: mitochondrial dysfunction and mtDNA instability. *Journal of the American Heart Association*, **2012**, 1, e003012 | 6 | 128 |
| 47 | 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 |

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| 46 | Opa1 is essential for retinal ganglion cell synaptic architecture and connectivity. *Brain*, **2012**, 135, 493-505 | 11.2 | 74 |
| 45 | Mitochondrial optic neuropathies. *Acta Ophthalmologica*, **2012**, 90, 0-0 | 3.7 | |
| 44 | Specific deficits in visual electrophysiology in a mouse model of dominant optic atrophy. *Experimental Eye Research*, **2011**, 93, 771-7 | 3.7 | 19 |
| 43 | 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 |
| 42 | Mitochondrial localization and ocular expression of mutant Opa3 in a mouse model of 3-methylglutaconicaciduria type III **2011**, 52, 4369-80 | | 14 |
| 41 | Changes in retinal neuronal connectivity in a mouse model of dominant optic atrophy. *Acta Ophthalmologica*, **2011**, 89, 0-0 | 3.7 | |
| 40 | Changes in corneal collagen architecture during mouse postnatal development **2010**, 51, 2936-42 | | 14 |
| 39 | Opa1 deficiency in a mouse model of dominant optic atrophy leads to retinal ganglion cell dendropathy. *Brain*, **2010**, 133, 2942-51 | 11.2 | 84 |
| 38 | Multi-system neurological disease is common in patients with OPA1 mutations. *Brain*, **2010**, 133, 771-86 | 11.2 | 314 |
| 37 | 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 | |
| 36 | No retinal apoptosis in mouse model of Opa1 dominant optic atrophy despite visual loss. *Acta Ophthalmologica*, **2010**, 88, 0-0 | 3.7 | |
| 35 | Dendritic pruning of retinal ganglion cells in a mouse model of dominant optic atrophy (ADOA). *Acta Ophthalmologica*, **2010**, 88, 0-0 | 3.7 | |
| 34 | Heterozygous B6;C3-Opa1Q285STOP mouse model of dominant optic atrophy displays subtle neuromuscular features with age. *Acta Ophthalmologica*, **2010**, 88, 0-0 | 3.7 | |
| 33 | Secondary mtDNA defects do not cause optic nerve dysfunction in a mouse model of dominant optic atrophy **2009**, 50, 4561-6 | | 19 |
| 32 | OPA1 deficiency associated with increased autophagy in retinal ganglion cells in a murine model of dominant optic atrophy **2009**, 50, 2567-71 | | 76 |
| 31 | A missense mutation in the murine Opa3 gene models human Costeff syndrome. *Brain*, **2008**, 131, 368-80 | 11.2 | 30 |
| 30 | Inherited Optic Neuropathies **2008**, 51-67 | | 0 |
| 29 | Comparative study of fibrillar collagen arrangement in the corneas of primates and other mammals. *Anatomical Record*, **2007**, 290, 1542-50 | 2.1 | 65 |

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| 28 | 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 |
| 27 | Focus on molecules: the OPA1 protein. *Experimental Eye Research*, **2006**, 83, 1003-4 | 3.7 | 24 |
| 26 | 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 |
| 25 | Molecular genetic basis of primary inherited optic neuropathies. *Eye*, **2004**, 18, 1126-32 | 4.4 | 45 |
| 24 | A review of primary hereditary optic neuropathies. *Journal of Inherited Metabolic Disease*, **2003**, 26, 209-27 | 3.4 | 23 |
| 23 | 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 |
| 22 | Optic disc morphology of patients with OPA1 autosomal dominant optic atrophy. *British Journal of Ophthalmology*, **2003**, 87, 48-53 | 5.5 | 71 |
| 21 | A major marker for normal tension glaucoma: association with polymorphisms in the OPA1 gene. *Human Genetics*, **2002**, 110, 52-6 | 6.3 | 101 |
| 20 | Primary monophasic synovial sarcoma of the conjunctiva. *British Journal of Ophthalmology*, **2002**, 86, 1453-4 | 5.5 | 8 |
| 19 | 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 |
| 18 | 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 |
| 17 | Neovascular age-related macular degeneration: present and future treatment options. *Eye*, **2001**, 15, 424-9 | 4.4 | 45 |
| 16 | LDL receptor-related protein 5 (LRP5) affects bone accrual and eye development. *Cell*, **2001**, 107, 513-23 | 56.2 | 1827 |
| 15 | The pupil in dominant optic atrophy. *Investigative Ophthalmology and Visual Science*, **2001**, 42, 675-8 | | 20 |
| 14 | 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 |
| 13 | MRI of the intraorbital optic nerve in patients with autosomal dominant optic atrophy. *Neuroradiology*, **2000**, 42, 180-3 | 3.2 | 22 |
| 12 | Association between autosomal dominant optic atrophy and Ewing sarcoma. *Journal of Pediatric Hematology/Oncology*, **1999**, 21, 557-9 | 1.2 | |
| 11 | 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 |

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|---|---|---|---|
| 10 | Dominant optic atrophy: exclusion and fine genetic mapping of the candidate gene, HRY. *Mammalian Genome*, **1998**, 9, 784-7 | 3.2 | 3 |
| 9 | 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 |
| 8 | Colour discrimination ellipses in patients with dominant optic atrophy. *Vision Research*, **1998**, 38, 3413-9 | 2.1 | 27 |
| 7 | Clinical features, molecular genetics, and pathophysiology of dominant optic atrophy. *Journal of Medical Genetics*, **1998**, 35, 793-800 | 5.8 | 91 |
| 6 | Clinical features in affected individuals from 21 pedigrees with dominant optic atrophy. *JAMA Ophthalmology*, **1998**, 116, 351-8 | | 127 |
| 5 | The management of solitary trichoepithelioma versus basal cell carcinoma. *Eye*, **1998**, 12 ( Pt 1), 43-6 | 4.4 | 15 |
| 4 | 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 |
| 3 | The case of the disappearing bullet. *Eye*, **1994**, 8 ( Pt 1), 143-4 | 4.4 | 1 |
| 2 | Thymoxamine in the treatment of traumatic mydriasis. *British Journal of Ophthalmology*, **1993**, 77, 681 | 5.5 | 4 |
| 1 | Nicotinamide provides neuroprotection in glaucoma by protecting against mitochondrial and metabolic dysfunction | | 2 |