Patrick Yu-Wai-Man

List of Publications by Year
in descending order

120	8,053	81839	53190
papers	citations	39 h-index	85 g-index

128	128	128	9044
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Prevalence of nuclear and mitochondrial <scp>DNA</scp> mutations related to adult mitochondrial disease. Annals of Neurology, 2015, 77, 753-759.	2.8	706
2	Disturbed mitochondrial dynamics and neurodegenerative disorders. Nature Reviews Neurology, 2015, 11, 11-24.	4.9	533
3	Mitochondrial optic neuropathies â€“ Disease mechanisms and therapeutic strategies. Progress in Retinal and Eye Research, 2011, 30, 81-114.	7.3	514
4	A randomized placebo-controlled trial of idebenone in Leberâ€™s hereditary optic neuropathy. Brain, 2011, 134, 2677-2686.	3.7	461
5	A mitochondrial origin for frontotemporal dementia and amyotrophic lateral sclerosis through CHCHD10 involvement. Brain, 2014, 137, 2329-2345.	3.7	377
6	Universal heteroplasmy of human mitochondrial DNA. Human Molecular Genetics, 2013, 22, 384-390.	1.4	344
7	Clinical Expression of Leber Hereditary Optic Neuropathy Is Affected by the Mitochondrial DNAâ€“Haplogroup Background. American Journal of Human Genetics, 2007, 81, 228-233.	2.6	331
8	Geneâ€“environment interactions in Leber hereditary optic neuropathy. Brain, 2009, 132, 2317-2326.	3.7	307
9	Efficient mitochondrial biogenesis drives incomplete penetrance in Leberâ€™s hereditary optic neuropathy. Brain, 2014, 137, 335-353.	3.7	229
10	Artificial Intelligence to Detect Papilledema from Ocular Fundus Photographs. New England Journal of Medicine, 2020, 382, 1687-1695.	13.9	214
11	OPA1 links human mitochondrial genome maintenance to mtDNA replication and distribution. Genome Research, 2011, 21, 12-20.	2.4	207
12	The Prevalence and Natural History of Dominant Optic Atrophy Due to OPA1 Mutations. Ophthalmology, 2010, 117, 1538-1546.e1.	2.5	162
13	International Consensus Statement on the Clinical and Therapeutic Management of Leber Hereditary Optic Neuropathy. Journal of Neuro-Ophthalmology, 2017, 37, 371-381.	0.4	156
14	<scp>CHCHD</scp> 10 mutations promote loss of mitochondrial cristae junctions with impaired mitochondrial genome maintenance and inhibition of apoptosis. EMBO Molecular Medicine, 2016, 8, 58-72.	3.3	143
15	A neurodegenerative perspective on mitochondrial optic neuropathies. Acta Neuropathologica, 2016, 132, 789-806.	3.9	135
16	Genetic Basis of Inherited Retinal Disease in a Molecularly Characterized Cohort of More Than 3000 Families from the United Kingdom. Ophthalmology, 2020, 127, 1384-1394.	2.5	131
17	Bilateral visual improvement with unilateral gene therapy injection for Leber hereditary optic neuropathy. Science Translational Medicine, 2020, 12, .	5.8	128
18	Exome sequencing in undiagnosed inherited and sporadic ataxias. Brain, 2015, 138, 276-283.	3.7	120

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19	Clinical features of MS associated with Leber hereditary optic neuropathy mtDNA mutations. Neurology, 2013, 81, 2073-2081.	1.5	100
20	APOSTEL 2.0 Recommendations for Reporting Quantitative Optical Coherence Tomography Studies. Neurology, 2021, 97, 68-79.	1.5	96
21	OPA1 mutations cause cytochrome c oxidase deficiency due to loss of wild-type mtDNA molecules. Human Molecular Genetics, 2010, 19, 3043-3052.	1.4	95
22	Genetic Counselling for Maternally Inherited Mitochondrial Disorders. Molecular Diagnosis and Therapy, 2017, 21, 419-429.	1.6	93
23	Fatal infantile mitochondrial encephalomyopathy, hypertrophic cardiomyopathy and optic atrophy associated with a homozygousOPA1mutation. Journal of Medical Genetics, 2016, 53, 127-131.	1.5	91
24	Quality of Life in Patients with Leber Hereditary Optic Neuropathy. , 2009, 50, 3112.		87
25	Efficacy and Safety of Intravitreal Gene Therapy for Leber Hereditary Optic Neuropathy Treated within 6 Months of Disease Onset. Ophthalmology, 2021, 128, 649-660.	2.5	87
26	Homozygous deletion in MICU1 presenting with fatigue and lethargy in childhood. Neurology: Genetics, 2016, 2, e59.	0.9	86
27	Dysregulated mitophagy and mitochondrial organization in optic atrophy due to OPA1 mutations. Neurology, 2017, 88, 131-142.	1.5	81
28	Steroids for traumatic optic neuropathy. The Cochrane Library, 2013, , CD006032.	1.5	80
29	Visual Outcomes in Leber Hereditary Optic Neuropathy Patients With the m.11778G>A (MTND4) Mitochondrial DNA Mutation. Journal of Neuro-Ophthalmology, 2020, 40, 547-557.	0.4	66
30	Childhood-onset Leber hereditary optic neuropathy. British Journal of Ophthalmology, 2017, 101, 1505-1509.	2.1	62
31	Traumatic optic neuropathyâ€"Clinical features and management issues. Taiwan Journal of Ophthalmology, 2015, 5, 3-8.	0.3	61
32	metabolic profiling of Parkinson's disease and mild cognitive impairment. Movement Disorders, 2017, 32, 927-932.	2.2	58
33	Dominant Optic Atrophy: Novel OPA1 Mutations and Revised Prevalence Estimates. Ophthalmology, 2013, 120, 1712-1712.e1.	2.5	57
34	A novel CISD2 mutation associated with a classical Wolfram syndrome phenotype alters Ca2+ homeostasis and ER-mitochondria interactions. Human Molecular Genetics, 2017, 26, 1599-1611.	1.4	57
35	Surgery for traumatic optic neuropathy. The Cochrane Library, 2013, 6, CD005024.	1.5	55
36	Recessive Mutations in RTN4IP1 Cause Isolated and Syndromic Optic Neuropathies. American Journal of Human Genetics, 2015, 97, 754-760.	2.6	54

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37	Retinal Ganglion Cellsâ€"Diversity of Cell Types and Clinical Relevance. Frontiers in Neurology, 2021, 12, 661938.	1.1	53
38	Evaluating the therapeutic potential of idebenone and related quinone analogues in Leber hereditary optic neuropathy. Mitochondrion, 2017, 36, 36-42.	1.6	50
39	Somatic Mitochondrial DNA Deletions Accumulate to High Levels in Aging Human Extraocular Muscles. , 2010, 51, 3347.		48
40	Leber hereditary optic neuropathy. Current Opinion in Ophthalmology, 2017, 28, 403-409.	1.3	48
41	Dominant optic atrophy: Culprit mitochondria in the optic nerve. Progress in Retinal and Eye Research, 2021, 83, 100935.	7.3	48
42	Monogenic diabetes syndromes: Locus-specific databases for AlstrÃ¶m, Wolfram, and Thiamine-responsive megaloblastic anemia. Human Mutation, 2017, 38, 764-777.	1.1	47
43	Subtle neurological and metabolic abnormalities in an Opa1 mouse model of autosomal dominant optic atrophy. Experimental Neurology, 2009, 220, 404-409.	2.0	44
44	Inherited eye-related disorders due to mitochondrial dysfunction. Human Molecular Genetics, 2017, 26, R12-R20.	1.4	43
45	Intravitreal Gene Therapy vs. Natural History in Patients With Leber Hereditary Optic Neuropathy Carrying the m.11778G>A ND4 Mutation: Systematic Review and Indirect Comparison. Frontiers in Neurology, 2021, 12, 662838.	1.1	42
46	SSBP1 mutations in dominant optic atrophy with variable retinal degeneration. Annals of Neurology, 2019, 86, 368-383.	2.8	41
47	Natural history of patients with Leber hereditary optic neuropathyâ€"results from the REALITY study. Eye, 2022, 36, 818-826.	1.1	37
48	Genetic manipulation for inherited neurodegenerative diseases: myth or reality?. British Journal of Ophthalmology, 2016, 100, 1322-1331.	2.1	36
49	The m.13051G>A mitochondrial DNA mutation results in variable neurology and activated mitophagy. Neurology, 2016, 86, 1921-1923.	1.5	35
50	Vertigo and vestibular abnormalities in spinocerebellar ataxia type 6. Journal of Neurology, 2009, 256, 78-82.	1.8	34
51	OPA1 mutations impair mitochondrial function in both pure and complicated dominant optic atrophy. Brain, 2011, 134, e164-e164.	3.7	34
52	Highâ€throughput screening identifies suppressors of mitochondrial fragmentation in OPA1 fibroblasts. EMBO Molecular Medicine, 2021, 13, e13579.	3.3	33
53	Mutations in the m-AAA proteases AFG3L2 and SPG7 are causing isolated dominant optic atrophy. Neurology: Genetics, 2020, 6, e428.	0.9	31
54	Long-Term Follow-Up After Unilateral Intravitreal Gene Therapy for Leber Hereditary Optic Neuropathy: The RESTORE Study. Journal of Neuro-Ophthalmology, 2021, 41, 309-315.	0.4	30

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55	Clinical utility gene card for: inherited optic neuropathies including next-generation sequencing-based approaches. European Journal of Human Genetics, 2019, 27, 494-502.	1.4	29
56	Monoamine oxidase-A promotes protective autophagy in human SH-SY5Y neuroblastoma cells through Bcl-2 phosphorylation. Redox Biology, 2019, 20, 167-181.	3.9	29
57	A multiple sclerosisâ€like disorder in patients with OPA1 mutations. Annals of Clinical and Translational Neurology, 2016, 3, 723-729.	1.7	27
58	Treatment strategies for Leber hereditary optic neuropathy. Current Opinion in Neurology, 2019, 32, 99-104.	1.8	27
59	Prevalence of neurogenetic disorders in the North of England. Neurology, 2015, 85, 1195-1201.	1.5	26
60	Steroids for traumatic optic neuropathy. , 2011, , CD006032.		25
61	Abnormal retinal thickening is a common feature among patients with ARSACS-related phenotypes: TableÂ1. British Journal of Ophthalmology, 2014, 98, 711-713.	2.1	25
62	Leber hereditary optic neuropathyâ€"new insights and old challenges. Graefe's Archive for Clinical and Experimental Ophthalmology, 2021, 259, 2461-2472.	1.0	24
63	Therapeutic Approaches to Inherited Optic Neuropathies. Seminars in Neurology, 2015, 35, 578-586.	0.5	23
64	Clonal expansion of mtDNA deletions: different disease models assessed by digital droplet PCR in single muscle cells. Scientific Reports, 2018, 8, 11682.	1.6	21
65	CRISPR-Cas9 correction of OPA1 c.1334G>A: p.R445H restores mitochondrial homeostasis in dominant optic atrophy patient-derived iPSCs. Molecular Therapy - Nucleic Acids, 2021, 26, 432-443.	2.3	21
66	Secondary mtDNA Defects Do Not Cause Optic Nerve Dysfunction in a Mouse Model of Dominant Optic Atrophy. , 2009, 50, 4561.		20
67	Behr's Syndrome is Typically Associated with Disturbed Mitochondrial Translation and Mutations in the C12orf65 Gene. Journal of Neuromuscular Diseases, 2014, 1, 55-63.	1.1	20
68	Biodistribution of intravitreal lenadogene nolparvovec gene therapy in nonhuman primates. Molecular Therapy - Methods and Clinical Development, 2021, 23, 307-318.	1.8	20
69	Lamination of the Outer Plexiform Layer in Optic Atrophy Caused by Dominant WFS1 Mutations. Ophthalmology, 2016, 123, 1624-1626.	2.5	19
70	Leber Hereditary Optic Neuropathyâ€"Light at the End of the Tunnel?. Asia-Pacific Journal of Ophthalmology, 2019, 7, 242-245.	1.3	18
71	Cysteine Supplementation May be Beneficial in a Subgroup of Mitochondrial Translation Deficiencies. Journal of Neuromuscular Diseases, 2016, 3, 363-379.	1.1	17
72	Human iPSC disease modelling reveals functional and structural defects in retinal pigment epithelial cells harbouring the m.3243Aâ€‰>â€‰G mitochondrial DNA mutation. Scientific Reports, 2017, 7, 12320.	1.6	17

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73	DominantACO2mutations are a frequent cause of isolated optic atrophy. Brain Communications, 2021, 3, fcab063.	1.5	16
74	Both mitochondrial DNA and mitonuclear gene mutations cause hearing loss through cochlear dysfunction. Brain, 2016, 139, e33-e33.	3.7	15
75	Common data elements for clinical research in mitochondrial disease: a National Institute for Neurological Disorders and Stroke project. Journal of Inherited Metabolic Disease, 2017, 40, 403-414.	1.7	15
76	Multirater Validation of Peripapillary Hyperreflective Ovoid Mass-like Structures (PHOMS). Neuro-Ophthalmology, 2020, 44, 413-414.	0.4	15
77	OPA1 Modulates Mitochondrial Ca2+ Uptake Through ER-Mitochondria Coupling. Frontiers in Cell and Developmental Biology, 2021, 9, 774108.	1.8	15
78	Near-Identical Segregation of mtDNA Heteroplasmy in Blood, Muscle, Urinary Epithelium, and Hair Follicles in Twins With Optic Atrophy, Ptosis, and Intractable Epilepsy. JAMA Neurology, 2013, 70, 1552-5.	4.5	14
79	Gene Therapies for the Treatment of Leber Hereditary Optic Neuropathy. International Ophthalmology Clinics, 2021, 61, 195-208.	0.3	14
80	Investigation of auditory dysfunction in Leber hereditary optic neuropathy. Acta Ophthalmologica, 2008, 86, 630-633.	0.6	13
81	Reply: Parsing the differences in affected with LHON: genetic versus environmental triggers of disease conversion. Brain, 2016, 139, e18-e18.	3.7	13
82	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.		13
83	Pathogenic NR2F1 variants cause a developmental ocular phenotype recapitulated in a mutant mouse model. Brain Communications, 2021, 3, fcab162.	1.5	13
84	Optical Coherence Tomography Angiography Reveals Distinct Retinal Structural and Microvascular Abnormalities in Cerebrovascular Disease. Frontiers in Neuroscience, 2020, 14, 588515.	1.4	12
85	OPA1 mutations induce mtDNA proliferation in leukocytes of patients with dominant optic atrophy. Neurology, 2012, 79, 1515-1517.	1.5	11
86	Spatial Technology Assessment of Green Space Exposure andÂMyopia. Ophthalmology, 2022, 129, 113-117.	2.5	11
87	Reply: Are CHCHD10 mutations indeed associated with familial amyotrophic lateral sclerosis?. Brain, 2014, 137, e314-e314.	3.7	9
88	Modelling autosomal dominant optic atrophy associated with OPA1 variants in iPSC-derived retinal ganglion cells. Human Molecular Genetics, 2022, 31, 3478-3493.	1.4	9
89	WFS1-Associated Optic Neuropathy: Genotype-Phenotype Correlations and Disease Progression. American Journal of Ophthalmology, 2022, 241, 9-27.	1.7	8
90	Leber hereditary optic neuropathy â€“ Therapeutic challenges and early promise. Taiwan Journal of Ophthalmology, 2011, 1, 12-15.	0.3	7

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91	Identification of a novel heterozygous guanosine monophosphate reductase (GMPR) variant in a patient with a lateâ€onset disorder of mitochondrial DNA maintenance. Clinical Genetics, 2020, 97, 276-286.	1.0	7
92	Comparison of macular structural and vascular changes in neuromyelitis optica spectrum disorder and primary open angle glaucoma: a cross-sectional study. British Journal of Ophthalmology, 2021, 105, 354-360.	2.1	7
93	[11C]PK11195-PET Brain Imaging of the Mitochondrial Translocator Protein in Mitochondrial Disease. Neurology, 2021, 96, e2761-e2773.	1.5	7
94	Mitochondrial Dysfunction in Glaucomaâ€"Closing the Loop. , 2012, 53, 2438.		6
95	Reply: Early-onset Behr syndrome due to compound heterozygous mutations in OPA1. Brain, 2014, 137, e302-e302.	3.7	6
96	Clinical utility gene card for: Wolfram syndrome. European Journal of Human Genetics, 2016, 24, 1-4.	1.4	6
97	Associations Between Regional Environment and Cornea-Related Morphology of the Eye in Young Adults: A Large-Scale Multicenter Cross-Sectional Study. , 2021, 62, 35.		6
98	Mitochondrial Disorders and the Eye: A New Era for Diagnosis. Ophthalmology, 2021, 128, 632-633.	2.5	5
99	Cross-Sectional Analysis of Baseline Visual Parameters in Subjects Recruited Into the RESCUE and REVERSE ND4-LHON Gene Therapy Studies. Journal of Neuro-Ophthalmology, 2021, 41, 298-308.	0.4	5
100	Reply: Two novel mutations in conserved codons indicate that CHCHD10 is a gene associated with motor neuron disease. Brain, 2014, 137, e310-e310.	3.7	4
101	Management of ophthalmologic manifestations of mitochondrial diseases. Genetics in Medicine, 2017, 19, 1380-1380.	1.1	4
102	Absence of lenadogene nolparvovec DNA in a brain tumor biopsy from a patient in the REVERSE clinical study, a case report. BMC Neurology, 2022, 22, .	0.8	4
103	Reply: Mutations in the CHCHD10 gene are a common cause of familial amyotrophic lateral sclerosis. Brain, 2014, 137, e312-e312.	3.7	3
104	Reply: IsCHCHD10Pro34Ser pathogenic for frontotemporal dementia and amyotrophic lateral sclerosis?. Brain, 2015, 138, e386-e386.	3.7	3
105	Harnessing the Power of Genetic Engineering for Patients With Mitochondrial Eye Diseases. Journal of Neuro-Ophthalmology, 2017, 37, 56-64.	0.4	3
106	Reply: A distinct clinical phenotype in a German kindred with motor neuron disease carrying aCHCHD10mutation: Table 1. Brain, 2015, 138, e377-e377.	3.7	2
107	OPA1 GTPase and GE Domain-Specific Mutations Differentially Alter Mitochondrial Fusion Dynamics and Calcium Homeostasis. Biophysical Journal, 2020, 118, 184a.	0.2	2
108	Quantifying inter-organelle membrane contact sites using proximity ligation assay in fixed optic nerve sections. Experimental Eye Research, 2021, 213, 108793.	1.2	2

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109	Capturing the experiences of patients with inherited optic neuropathies: a systematic review of patient-reported outcome measures (PROMs) and qualitative studies. Graefe's Archive for Clinical and Experimental Ophthalmology, 2022, , 1.	1.0	2
110	Characterisation of a novel OPA1 splice variant resulting in cryptic splice site activation and mitochondrial dysfunction. European Journal of Human Genetics, 2022, 30, 848-855.	1.4	2
111	Reply: Spastic paraplegia in 'dominant optic atrophy plus' phenotype due to OPA1 mutation. Brain, 2011, 134, e196-e196.	3.7	1
112	Reply:CHCHD10mutations in Italian patients with sporadic amyotrophic lateral sclerosis. Brain, 2015, 138, e373-e373.	3.7	1
113	Wolfram syndrome: new pathophysiological insights and therapeutic strategies. Therapeutic Advances in Rare Disease, 2021, 2, 263300402110395.	0.3	1
114	Reply: â€˜Behr syndromeâ€™ with OPA1 compound heterozygote mutations. Brain, 2015, 138, e322-e322.	3.7	0
115	Reply: High prevalence ofCHCHD10mutations in patients with frontotemporal dementia from China: Table 1. Brain, 2016, 139, e22-e22.	3.7	0
116	Treatment of Leber Hereditary Optic Neuropathy. , 2019, , 201-207.		0
117	Neuro-Ophthalmology in the United Kingdom. Journal of Neuro-Ophthalmology, 2015, 35, e17-e19.	0.4	0
118	Delayed Optochiasmal Arachnoiditis following Intervention for a Subarachnoid Haemorrhage. Ophthalmology International, 2013, 8, 85-86.	0.0	0
119	Hereditary Optic Neuropathies. , 2022, , 4575-4607.		0
120	A wolf in sheepâ€™s clothing. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, A13.3-A14.	0.9	0