Valerio Carelli

List of Publications by Year in Descending Order

316 papers	17,579 citations	77 h-index	119 g-index
334 ext. papers	20,131 ext. citations	6.5 avg, IF	6.35 L-index

#	Paper	IF	Citations
316	Longitudinal Study of Optic Disk Perfusion and Retinal Structure in Leber's Hereditary Optic Neuropathy. **2022**, 63, 43		0
315	The role of mtDNA haplogroups on metabolic features in narcolepsy type 1.. *Mitochondrion*, **2022**, 63, 37-42	4.9	0
314	The relevance of migraine in the clinical spectrum of mitochondrial disorders.. *Scientific Reports*, **2022**, 12, 4222	4.9	0
313	Capturing the pattern of transition from carrier to affected in Leber's hereditary optic neuropathy.. *American Journal of Ophthalmology*, **2022**,	4.9	2
312	Multishell Diffusion MR Tractography Yields Morphological and Microstructural Information of the Anterior Optic Pathway: A Proof-of-Concept Study in Patients with Leber's Hereditary Optic Neuropathy. *International Journal of Environmental Research and Public Health*, **2022**, 19, 6914	4.6	
311	Combined Optic Atrophy and Rod-Cone Dystrophy Expands the RTN4IP1 (Optic Atrophy 10) Phenotype. *Journal of Neuro-Ophthalmology*, **2021**, 41, e290-e292	2.6	5
310	Electrocochleography in Auditory Neuropathy Related to Mutations in the OTOF or OPA1 Gene.. *Audiology Research*, **2021**, 11, 639-652	1.5	1
309	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	2.6	3
308	Case Report: A Novel Mutation in the Mitochondrial Gene Is Associated With Leber Hereditary Optic Neuropathy (LHON). *Frontiers in Neurology*, **2021**, 12, 652590	4.1	2
307	Impaired complex I repair causes recessive Leber's hereditary optic neuropathy. *Journal of Clinical Investigation*, **2021**, 131,	15.9	27
306	Natural history of patients with Leber hereditary optic neuropathy-results from the REALITY study. *Eye*, **2021**,	4.4	7
305	Dominant mutations are a frequent cause of isolated optic atrophy. *Brain Communications*, **2021**, 3, fcab063	4.3	5
304	Biallelic variants in LIG3 cause a novel mitochondrial neurogastrointestinal encephalomyopathy. *Brain*, **2021**, 144, 1451-1466	11.2	8
303	Brain MRS correlates with mitochondrial dysfunction biomarkers in MELAS-associated mtDNA mutations. *Annals of Clinical and Translational Neurology*, **2021**, 8, 1200-1211	5.3	3
302	Efficacy and Safety of Intravitreal Gene Therapy for Leber Hereditary Optic Neuropathy Treated within 6 Months of Disease Onset. *Ophthalmology*, **2021**, 128, 649-660	7.3	28
301	Intravitreal Gene Therapy vs. Natural History in Patients With Leber Hereditary Optic Neuropathy Carrying the m.11778G>A Mutation: Systematic Review and Indirect Comparison. *Frontiers in Neurology*, **2021**, 12, 662838	4.1	6
300	Movement Disorders in Children with a Mitochondrial Disease: A Cross-Sectional Survey from the Nationwide Italian Collaborative Network of Mitochondrial Diseases. *Journal of Clinical Medicine*, **2021**, 10,	5.1	3

299	Evidence of enteric angiopathy and neuromuscular hypoxia in patients with mitochondrial neurogastrointestinal encephalomyopathy. *American Journal of Physiology - Renal Physiology*, **2021**, 320, G768-G779	5.1	2
298	Exploiting hiPSCs in Leber's Hereditary Optic Neuropathy (LHON): Present Achievements and Future Perspectives. *Frontiers in Neurology*, **2021**, 12, 648916	4.1	1
297	Leber's Hereditary Optic Neuropathy: A Report on Novel mtDNA Pathogenic Variants. *Frontiers in Neurology*, **2021**, 12, 657317	4.1	0
296	Dominant Optic Atrophy (DOA): Modeling the Kaleidoscopic Roles of OPA1 in Mitochondrial Homeostasis. *Frontiers in Neurology*, **2021**, 12, 681326	4.1	2
295	Adult-onset mitochondrial movement disorders: a national picture from the Italian Network. *Journal of Neurology*, **2021**, 1	5.5	2
294	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	2.6	0
293	Therapeutic Options in Hereditary Optic Neuropathies. *Drugs*, **2021**, 81, 57-86	12.1	9
292	Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE): Position paper on diagnosis, prognosis, and treatment by the MNGIE International Network. *Journal of Inherited Metabolic Disease*, **2021**, 44, 376-387	5.4	15
291	Drug repositioning as a therapeutic strategy for neurodegenerations associated with OPA1 mutations. *Human Molecular Genetics*, **2021**, 29, 3631-3645	5.6	9
290	Exploring Metabolic Adaptations to the Acidic Microenvironment of Osteosarcoma Cells Unveils Sphingosine 1-Phosphate as a Valuable Therapeutic Target. *Cancers*, **2021**, 13,	6.6	8
289	Retina and melanopsin neurons. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2021**, 179, 315-329	3	2
288	Gene Therapies for the Treatment of Leber Hereditary Optic Neuropathy. *International Ophthalmology Clinics*, **2021**, 61, 195-208	1.7	1
287	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	5.6	1
286	Molecular Mechanisms behind Inherited Neurodegeneration of the Optic Nerve. *Biomolecules*, **2021**, 11,	5.9	4
285	The mitochondrial single-stranded DNA binding protein is essential for initiation of mtDNA replication. *Science Advances*, **2021**, 7,	14.3	9
284	The relevance of mitochondrial DNA variants fluctuation during reprogramming and neuronal differentiation of human iPSCs. *Stem Cell Reports*, **2021**, 16, 1953-1967	8	1
283	The m.3890G>A/MT-ND1 mtDNA rare pathogenic variant: Expanding clinical and MRI phenotypes. *Mitochondrion*, **2021**, 60, 142-149	4.9	1
282	Brain functional MRI responses to blue light stimulation in Leber's hereditary optic neuropathy. *Biochemical Pharmacology*, **2021**, 191, 114488	6	2

281	Biodistribution of intravitreal nolparvovec gene therapy in nonhuman primates. *Molecular Therapy - Methods and Clinical Development*, **2021**, 23, 307-318	6.4	3
280	Mitochondrial Retinopathies.. *International Journal of Molecular Sciences*, **2021**, 23,	6.3	4
279	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	2.6	21
278	Bilateral visual improvement with unilateral gene therapy injection for Leber hereditary optic neuropathy. *Science Translational Medicine*, **2020**, 12,	17.5	50
277	Idebenone increases chance of stabilization/recovery of visual acuity in OPA1-dominant optic atrophy. *Annals of Clinical and Translational Neurology*, **2020**, 7, 590-594	5.3	16
276	Mitochondrial diseases in adults. *Journal of Internal Medicine*, **2020**, 287, 592-608	10.8	16
275	Autozygosity-driven genetic diagnosis in consanguineous families from Italy and the Greater Middle East. *Human Genetics*, **2020**, 139, 1429-1441	6.3	4
274	Calcium mishandling in absence of primary mitochondrial dysfunction drives cellular pathology in Wolfram Syndrome. *Scientific Reports*, **2020**, 10, 4785	4.9	16
273	Metabolomics hallmarks OPA1 variants correlating with their in vitro phenotype and predicting clinical severity. *Human Molecular Genetics*, **2020**, 29, 1319-1329	5.6	9
272	Interaction Between Mitochondrial DNA Variants and Mitochondria/Endoplasmic Reticulum Contact Sites: A Perspective Review. *DNA and Cell Biology*, **2020**, 39, 1431-1443	3.6	1
271	DNMT1 mutations leading to neurodegeneration paradoxically reflect on mitochondrial metabolism. *Human Molecular Genetics*, **2020**, 29, 1864-1881	5.6	10
270	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. *Journal of Clinical Investigation*, **2020**, 130, 108-125	15.9	49
269	Mutations in the m-AAA proteases AFG3L2 and SPG7 are causing isolated dominant optic atrophy. *Neurology: Genetics*, **2020**, 6, e428	3.8	18
268	Cataplexy and ataxia: red flags for the diagnosis of DNA methyltransferase 1 mutation. *Journal of Clinical Sleep Medicine*, **2020**, 16, 143-147	3.1	3
267	Fine-tuning of the respiratory complexes stability and supercomplexes assembly in cells defective of complex III. *Biochimica Et Biophysica Acta - Bioenergetics*, **2020**, 1861, 148133	4.6	10
266	Exploring metabolic reprogramming in melanoma via acquired resistance to the oxidative phosphorylation inhibitor phenformin. *Melanoma Research*, **2020**, 30, 1-13	3.3	3
265	Mitochondrial epilepsy: a cross-sectional nationwide Italian survey. *Neurogenetics*, **2020**, 21, 87-96	3	9
264	Liver transplantation in mitochondrial neurogastrointestinal encephalomyopathy (MNGIE): clinical long-term follow-up and pathogenic implications. *Journal of Neurology*, **2020**, 267, 3702-3710	5.5	9

263	Inhibition of autophagy curtails visual loss in a model of autosomal dominant optic atrophy. *Nature Communications*, **2020**, 11, 4029	17.4	28
262	Expanding and validating the biomarkers for mitochondrial diseases. *Journal of Molecular Medicine*, **2020**, 98, 1467-1478	5.5	19
261	Variants Result in Late-Onset Mitochondrial Myopathy With Altered Muscle Mitochondrial DNA Homeostasis. *Frontiers in Genetics*, **2020**, 11, 860	4.5	2
260	Primary mitochondrial myopathy: Clinical features and outcome measures in 118 cases from Italy. *Neurology: Genetics*, **2020**, 6, e519	3.8	3
259	ATPase Domain AFG3L2 Mutations Alter OPA1 Processing and Cause Optic Neuropathy. *Annals of Neurology*, **2020**, 88, 18-32	9.4	18
258	Impaired Ganglion Cell Function Objectively Assessed by the Photopic Negative Response in Affected and Asymptomatic Members From Brazilian Families With Leber's Hereditary Optic Neuropathy. *Frontiers in Neurology*, **2020**, 11, 628014	4.1	1
257	Muscle pain in mitochondrial diseases: a picture from the Italian network. *Journal of Neurology*, **2019**, 266, 953-959	5.5	8
256	First missense mutation in an Italian proband with optic atrophy and deafness. *Neurology: Genetics*, **2019**, 5, e329	3.8	10
255	Hearing Dysfunction in a Large Family Affected by Dominant Optic Atrophy (OPA8-Related DOA): A Human Model of Hidden Auditory Neuropathy. *Frontiers in Neuroscience*, **2019**, 13, 501	5.1	3
254	Mitochondrial Optic Neuropathies **2019**, 125-139		1
253	Functional Changes of Retinal Ganglion Cells and Visual Pathways in Patients with Chronic Leber's Hereditary Optic Neuropathy during One Year of Follow-up. *Ophthalmology*, **2019**, 126, 1033-1044	7.3	9
252	Pharmacological Inhibition of Necroptosis Protects from Dopaminergic Neuronal Cell Death in Parkinson's Disease Models. *Cell Reports*, **2018**, 22, 2066-2079	10.6	101
251	Haplogroup J mitogenomes are the most sensitive to the pesticide rotenone: Relevance for human diseases. *Neurobiology of Disease*, **2018**, 114, 129-139	7.5	16
250	The idebenone metabolite QS10 restores electron transfer in complex I and coenzyme Q defects. *Biochimica Et Biophysica Acta - Bioenergetics*, **2018**, 1859, 901-908	4.6	19
249	Rewiring of Glutamine Metabolism Is a Bioenergetic Adaptation of Human Cells with Mitochondrial DNA Mutations. *Cell Metabolism*, **2018**, 27, 1007-1025.e5	24.6	78
248	OPA1: How much do we know to approach therapy?. *Pharmacological Research*, **2018**, 131, 199-210	10.2	29
247	Eight human OPA1 isoforms, long and short: What are they for?. *Biochimica Et Biophysica Acta - Bioenergetics*, **2018**, 1859, 263-269	4.6	70
246	Cerebral Mitochondrial Microangiopathy Leads to Leukoencephalopathy in Mitochondrial Neurogastrointestinal Encephalopathy. *American Journal of Neuroradiology*, **2018**, 39, 427-434	4.4	13

245	Complex II phosphorylation is triggered by unbalanced redox homeostasis in cells lacking complex III. *Biochimica Et Biophysica Acta - Bioenergetics*, **2018**, 1859, 182-190	4.6	6
244	DGUOK recessive mutations in patients with CPEO, mitochondrial myopathy, parkinsonism and mtDNA deletions. *Brain*, **2018**, 141, e3	11.2	9
243	Retinal dysfunction characterizes subtypes of dominant optic atrophy. *Acta Ophthalmologica*, **2018**, 96, e156-e163	3.7	10
242	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	6.9	23
241	Clonal expansion of mtDNA deletions: different disease models assessed by digital droplet PCR in single muscle cells. *Scientific Reports*, **2018**, 8, 11682	4.9	16
240	Clinical syndromes associated with mtDNA mutations: where we stand after 30 years. *Essays in Biochemistry*, **2018**, 62, 235-254	7.6	19
239	Infant and Adult Gut Microbiome and Metabolome in Rural Bassa and Urban Settlers from Nigeria. *Cell Reports*, **2018**, 23, 3056-3067	10.6	84
238	Peculiar combinations of individually non-pathogenic missense mitochondrial DNA variants cause low penetrance Leber's hereditary optic neuropathy. *PLoS Genetics*, **2018**, 14, e1007210	6	24
237	Melanopsin Retinal Ganglion Cells and Pupil: Clinical Implications for Neuro-Ophthalmology. *Frontiers in Neurology*, **2018**, 9, 1047	4.1	25
236	Peripapillary vessel density changes in Leber's hereditary optic neuropathy: a new biomarker. *Clinical and Experimental Ophthalmology*, **2018**, 46, 1055-1062	2.4	33
235	Combined Cerebellar Proton MR Spectroscopy and DWI Study of Patients with Friedreich's Ataxia. *Cerebellum*, **2017**, 16, 82-88	4.3	10
234	Liver transplant reverses biochemical imbalance in mitochondrial neurogastrointestinal encephalomyopathy. *Mitochondrion*, **2017**, 34, 101-102	4.9	22
233	Natural History of Conversion of Leber's Hereditary Optic Neuropathy: A Prospective Case Series. *Ophthalmology*, **2017**, 124, 843-850	7.3	43
232	Mitochondrial DNA and primary mitochondrial dysfunction in Parkinson's disease. *Movement Disorders*, **2017**, 32, 346-363	7	92
231	The glutamate/cystine xCT antiporter antagonizes glutamine metabolism and reduces nutrient flexibility. *Nature Communications*, **2017**, 8, 15074	17.4	126
230	Melanopsin-expressing retinal ganglion cells are resistant to cell injury, but not always. *Mitochondrion*, **2017**, 36, 77-84	4.9	14
229	OPA1 Isoforms in the Hierarchical Organization of Mitochondrial Functions. *Cell Reports*, **2017**, 19, 2557-2571	25.1	103
228	Optical coherence tomography angiography of the peripapillary retina and optic nerve head in dominant optic atrophy. *Mitochondrion*, **2017**, 36, 60-65	4.9	18

227	Optic neuropathies: the tip of the neurodegeneration iceberg. *Human Molecular Genetics*, **2017**, 26, R139-R150	5.6	55
226	The Photopic Negative Response: An Objective Measure of Retinal Ganglion Cell Function in Patients With Leber's Hereditary Optic Neuropathy **2017**, 58, BIO300-BIO306		20
225	International Workshop:: Outcome measures and clinical trial readiness in primary mitochondrial myopathies in children and adults. Consensus recommendations. 16-18 November 2016, Rome, Italy. *Neuromuscular Disorders*, **2017**, 27, 1126-1137	2.9	42
224	Incomplete penetrance in mitochondrial optic neuropathies. *Mitochondrion*, **2017**, 36, 130-137	4.9	38
223	Revisiting mitochondrial ocular myopathies: a study from the Italian Network. *Journal of Neurology*, **2017**, 264, 1777-1784	5.5	23
222	Retinal Ganglion Cells and Circadian Rhythms in Alzheimer's Disease, Parkinson's Disease, and Beyond. *Frontiers in Neurology*, **2017**, 8, 162	4.1	62
221	Patterns of Retinal Ganglion Cell Damage in Neurodegenerative Disorders: Parvocellular vs Magnocellular Degeneration in Optical Coherence Tomography Studies. *Frontiers in Neurology*, **2017**, 8, 710	4.1	57
220	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
219	Reply: Mitochondrial DNA copy number differentiates the Leber's hereditary optic neuropathy affected individuals from the unaffected mutation carriers. *Brain*, **2016**, 139, e2	11.2	3
218	Melanopsin retinal ganglion cell loss in Alzheimer disease. *Annals of Neurology*, **2016**, 79, 90-109	9.4	215
217	Liver transplantation for mitochondrial neurogastrointestinal encephalomyopathy. *Annals of Neurology*, **2016**, 80, 448-55	9.4	68
216	Changes in Choroidal Thickness follow the RNFL Changes in Leber's Hereditary Optic Neuropathy. *Scientific Reports*, **2016**, 6, 37332	4.9	22
215	Parsing the differences in affected with LHON: genetic versus environmental triggers of disease conversion. *Brain*, **2016**, 139, e17	11.2	35
214	ITA-MNGIE: an Italian regional and national survey for mitochondrial neuro-gastro-intestinal encephalomyopathy. *Neurological Sciences*, **2016**, 37, 1149-51	3.5	9
213	Reply: Both mitochondrial DNA and mitonuclear gene mutations cause hearing loss through cochlear dysfunction. *Brain*, **2016**, 139, e34	11.2	4
212	"Mitochondrial neuropathies": A survey from the large cohort of the Italian Network. *Neuromuscular Disorders*, **2016**, 26, 272-6	2.9	32
211	Re: Pilat et al.: High-resolution imaging of the optic nerve and retina in optic nerve hypoplasia (Ophthalmology 2015;122:1330-9). *Ophthalmology*, **2016**, 123, e19-20	7.3	1
210	Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. *Human Molecular Genetics*, **2016**, 25, 1031-41	5.6	44

209	Clinical and molecular study in a long-surviving patient with MLASA syndrome due to novel PUS1 mutations. *Neurogenetics*, **2016**, 17, 65-70	3	19
208	Macular nerve fibre and ganglion cell layer changes in acute Leber's hereditary optic neuropathy. *British Journal of Ophthalmology*, **2016**, 100, 1232-7	5.5	52
207	A neurodegenerative perspective on mitochondrial optic neuropathies. *Acta Neuropathologica*, **2016**, 132, 789-806	14.3	90
206	Mutations in SLC25A46, encoding a UGO1-like protein, cause an optic atrophy spectrum disorder. *Nature Genetics*, **2015**, 47, 926-32	36.3	131
205	Diffusion Tensor Imaging Mapping of Brain White Matter Pathology in Mitochondrial Optic Neuropathies. *American Journal of Neuroradiology*, **2015**, 36, 1259-65	4.4	23
204	DNA methyltransferase 1 mutations and mitochondrial pathology: is mtDNA methylated?. *Frontiers in Genetics*, **2015**, 6, 90	4.5	48
203	Keeping in shape the dogma of mitochondrial DNA maternal inheritance. *PLoS Genetics*, **2015**, 11, e1005679	6	16
202	Syndromic parkinsonism and dementia associated with OPA1 missense mutations. *Annals of Neurology*, **2015**, 78, 21-38	9.4	119
201	Multifocal VEP provide electrophysiological evidence of predominant dysfunction of the optic nerve fibers derived from the central retina in Leber's hereditary optic neuropathy. *Graefeis Archive for Clinical and Experimental Ophthalmology*, **2015**, 253, 1591-600	3.8	11
200	Redefining phenotypes associated with mitochondrial DNA single deletion. *Journal of Neurology*, **2015**, 262, 1301-9	5.5	53
199	Recessive Mutations in RTN4IP1 Cause Isolated and Syndromic Optic Neuropathies. *American Journal of Human Genetics*, **2015**, 97, 754-60	11	42
198	Targeting estrogen receptor as preventive therapeutic strategy for Leber's hereditary optic neuropathy. *Human Molecular Genetics*, **2015**, 24, 6921-31	5.6	47
197	OPA1-related auditory neuropathy: site of lesion and outcome of cochlear implantation. *Brain*, **2015**, 138, 563-76	11.2	58
196	Disturbed mitochondrial dynamics and neurodegenerative disorders. *Nature Reviews Neurology*, **2015**, 11, 11-24	15	425
195	Cigarette toxicity triggers Leber's hereditary optic neuropathy by affecting mtDNA copy number, oxidative phosphorylation and ROS detoxification pathways. *Cell Death and Disease*, **2015**, 6, e2021	9.8	73
194	Homozygous NOTCH3 null mutation and impaired NOTCH3 signaling in recessive early-onset arteriopathy and cavitating leukoencephalopathy. *EMBO Molecular Medicine*, **2015**, 7, 848-58	12	33
193	Reactive Oxygen Species in Mitochondrial Optic Neuropathies: Comment. *Journal of Neuro-Ophthalmology*, **2015**, 35, 445-6	2.6	2
192	'Behr syndrome' with OPA1 compound heterozygote mutations. *Brain*, **2015**, 138, e321	11.2	40

191	Mitochondria: Biogenesis and mitophagy balance in segregation and clonal expansion of mitochondrial DNA mutations. *International Journal of Biochemistry and Cell Biology*, **2015**, 63, 21-4	5.6	49
190	Macular Microcysts in Mitochondrial Optic Neuropathies: Prevalence and Retinal Layer Thickness Measurements. *PLoS ONE*, **2015**, 10, e0127906	3.7	19
189	Proteolytic cleavage of Opa1 stimulates mitochondrial inner membrane fusion and couples fusion to oxidative phosphorylation. *Cell Metabolism*, **2014**, 19, 630-41	24.6	257
188	Nocturnal melatonin regulation in post-traumatic vegetative state: a possible role for melatonin supplementation?. *Chronobiology International*, **2014**, 31, 741-5	3.6	17
187	The m.3243A>G mitochondrial DNA mutation and related phenotypes. A matter of gender?. *Journal of Neurology*, **2014**, 261, 504-10	5.5	91
186	Efficient mitochondrial biogenesis drives incomplete penetrance in Leber's hereditary optic neuropathy. *Brain*, **2014**, 137, 335-53	11.2	186
185	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	3.2	9
184	Association of the mtDNA m.4171C>A/MT-ND1 mutation with both optic neuropathy and bilateral brainstem lesions. *BMC Neurology*, **2014**, 14, 116	3.1	18
183	Proteolytic Cleavage of Opa1 Stimulates Mitochondrial Inner Membrane Fusion and Couples Fusion to Oxidative Phosphorylation. *Cell Metabolism*, **2014**, 19, 891	24.6	4
182	A novel in-frame 18-bp microdeletion in MT-CYB causes a multisystem disorder with prominent exercise intolerance. *Human Mutation*, **2014**, 35, 954-8	4.7	31
181	Myoclonus in mitochondrial disorders. *Movement Disorders*, **2014**, 29, 722-8	7	27
180	Early macular retinal ganglion cell loss in dominant optic atrophy: genotype-phenotype correlation. *American Journal of Ophthalmology*, **2014**, 158, 628-36.e3	4.9	45
179	Polysomnographic and neurometabolic features may mark preclinical autosomal dominant cerebellar ataxia, deafness, and narcolepsy due to a mutation in the DNA (cytosine-5-)-methyltransferase gene, DNMT1. *Sleep Medicine*, **2014**, 15, 582-5	4.6	4
178	Liver as a source for thymidine phosphorylase replacement in mitochondrial neurogastrointestinal encephalomyopathy. *PLoS ONE*, **2014**, 9, e96692	3.7	35
177	Different mtDNA mutations modify tumor progression in dependence of the degree of respiratory complex I impairment. *Human Molecular Genetics*, **2014**, 23, 1453-66	5.6	73
176	Narcolepsy is a common phenotype in HSAN IE and ADCA-DN. *Brain*, **2014**, 137, 1643-55	11.2	36
175	Pure and syndromic optic atrophy explained by deep intronic OPA1 mutations and an intralocus modifier. *Brain*, **2014**, 137, 2164-77	11.2	54
174	Mitochondrial DNA: impacting central and peripheral nervous systems. *Neuron*, **2014**, 84, 1126-42	13.9	77

173	Mitochondrial optic neuropathies: additional facts and concepts - response. *Clinical and Experimental Ophthalmology*, **2014**, 42, 207-8	2.4	5
172	Medical management of hereditary optic neuropathies. *Frontiers in Neurology*, **2014**, 5, 141	4.1	46
171	A wide range of 3243A>G/tRNALeu(UUR) (MELAS) mutation loads may segregate in offspring through the female germline bottleneck. *PLoS ONE*, **2014**, 9, e96663	3.7	9
170	Genetic Basis of Mitochondrial Optic Neuropathies. *Current Molecular Medicine*, **2014**, 14, 985-992	2.5	14
169	Loss of temporal retinal nerve fibers in Parkinson disease: a mitochondrial pattern?. *European Journal of Neurology*, **2013**, 20, 198-201	6	76
168	The clinical maze of mitochondrial neurology. *Nature Reviews Neurology*, **2013**, 9, 429-44	15	245
167	Respiratory complex I is essential to induce a Warburg profile in mitochondria-defective tumor cells. *Cancer & Metabolism*, **2013**, 1, 11	5.4	58
166	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, 445-52	6.9	15
165	Oncocytic glioblastoma: a glioblastoma showing oncocytic changes and increased mitochondrial DNA copy number. *Human Pathology*, **2013**, 44, 1867-76	3.7	12
164	The optic nerve: a "mito-window" on mitochondrial neurodegeneration. *Molecular and Cellular Neurosciences*, **2013**, 55, 62-76	4.8	66
163	Phenotypic heterogeneity of the 8344A>G mtDNA "MERRF" mutation. *Neurology*, **2013**, 80, 2049-54	6.5	135
162	Optic nerve histopathology in a case of Wolfram Syndrome: a mitochondrial pattern of axonal loss. *Mitochondrion*, **2013**, 13, 841-5	4.9	24
161	New treatments for mitochondrial disease-no time to drop our standards. *Nature Reviews Neurology*, **2013**, 9, 474-81	15	133
160	Gamma rays induce a p53-independent mitochondrial biogenesis that is counter-regulated by HIF1*Cell Death and Disease*, **2013**, 4, e663	9.8	29
159	Acute rhabdomyolysis induced by tonic-clonic epileptic seizures in a patient with glucose-6-phosphate dehydrogenase deficiency. *Journal of Neurology*, **2013**, 260, 2669-71	5.5	4
158	Microcystic macular degeneration from optic neuropathy: not inflammatory, not trans-synaptic degeneration. *Brain*, **2013**, 136, e239	11.2	65
157	Mitochondrial dysfunction in optic neuropathies: animal models and therapeutic options. *Current Opinion in Neurology*, **2013**, 26, 52-8	7.1	33
156	Retinal function and neural conduction along the visual pathways in affected and unaffected carriers with Leber's hereditary optic neuropathy **2013**, 54, 6893-901		31

155	The pupil light reflex in Leber's hereditary optic neuropathy: evidence for preservation of melanopsin-expressing retinal ganglion cells **2013**, 54, 4471-7		60
154	Mitochondrial optic neuropathies: our travels from bench to bedside and back again. *Clinical and Experimental Ophthalmology*, **2013**, 41, 702-12	2.4	23
153	Idebenone treatment in patients with OPA1-mutant dominant optic atrophy. *Brain*, **2013**, 136, e231	11.2	53
152	Melanopsin retinal ganglion cells and circadian dysfunction in Alzheimer's disease. *Acta Ophthalmologica*, **2013**, 91, 0-0	3.7	5
151	A novel null homozygous mutation confirms CACNA2D2 as a gene mutated in epileptic encephalopathy. *PLoS ONE*, **2013**, 8, e82154	3.7	55
150	The effects of idebenone on mitochondrial bioenergetics. *Biochimica Et Biophysica Acta - Bioenergetics*, **2012**, 1817, 363-9	4.6	87
149	Mouse mtDNA mutant model of Leber hereditary optic neuropathy. *Proceedings of the National Academy of Sciences of the United States of America*, **2012**, 109, 20065-70	11.5	149
148	Fatigue and exercise intolerance in mitochondrial diseases. Literature revision and experience of the Italian Network of mitochondrial diseases. *Neuromuscular Disorders*, **2012**, 22 Suppl 3, S226-9	2.9	23
147	Rare primary mitochondrial DNA mutations and probable synergistic variants in Leber's hereditary optic neuropathy. *PLoS ONE*, **2012**, 7, e42242	3.7	60
146	Retinal nerve fiber layer thickness variability in Leber hereditary optic neuropathy carriers. *European Journal of Ophthalmology*, **2012**, 22, 985-91	1.9	28
145	Neuron-specific enolase is elevated in asymptomatic carriers of Leber's hereditary optic neuropathy **2012**, 53, 6389-92		12
144	Mathematically modeling the involvement of axons in Leber's hereditary optic neuropathy **2012**, 53, 7608-17		88
143	Mitochondrial DNA signals of late glacial recolonization of Europe from near eastern refugia. *American Journal of Human Genetics*, **2012**, 90, 915-24	11	123
142	Effect of EPI-743 on the clinical course of the mitochondrial disease Leber hereditary optic neuropathy. *Archives of Neurology*, **2012**, 69, 331-8		135
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