Valerio Carelli

List of Publications by Citations

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	Mitochondrial dysfunction as a cause of optic neuropathies. *Progress in Retinal and Eye Research*, **2004**, 23, 53-89	20.5	577
315	Disturbed mitochondrial dynamics and neurodegenerative disorders. *Nature Reviews Neurology*, **2015**, 11, 11-24	15	425
314	OPA1 mutations induce mitochondrial DNA instability and optic atrophy 'plus' phenotypes. *Brain*, **2008**, 131, 338-51	11.2	394
313	The molecular dissection of mtDNA haplogroup H confirms that the Franco-Cantabrian glacial refuge was a major source for the European gene pool. *American Journal of Human Genetics*, **2004**, 75, 910-8	11	364
312	Multi-system neurological disease is common in patients with OPA1 mutations. *Brain*, **2010**, 133, 771-86	11.2	314
311	Haplotype and phylogenetic analyses suggest that one European-specific mtDNA background plays a role in the expression of Leber hereditary optic neuropathy by increasing the penetrance of the primary mutations 11778 and 14484. *American Journal of Human Genetics*, **1997**, 60, 1107-21	11	313
310	Clinical expression of Leber hereditary optic neuropathy is affected by the mitochondrial DNA-haplogroup background. *American Journal of Human Genetics*, **2007**, 81, 228-33	11	280
309	Proteolytic cleavage of Opa1 stimulates mitochondrial inner membrane fusion and couples fusion to oxidative phosphorylation. *Cell Metabolism*, **2014**, 19, 630-41	24.6	257
308	OPA1 mutations associated with dominant optic atrophy impair oxidative phosphorylation and mitochondrial fusion. *Brain*, **2008**, 131, 352-67	11.2	256
307	The clinical maze of mitochondrial neurology. *Nature Reviews Neurology*, **2013**, 9, 429-44	15	245
306	Complex I deficiency primes Bax-dependent neuronal apoptosis through mitochondrial oxidative damage. *Proceedings of the National Academy of Sciences of the United States of America*, **2005**, 102, 19126-31	11.5	245
305	Disruptive mitochondrial DNA mutations in complex I subunits are markers of oncocytic phenotype in thyroid tumors. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 9001-6	11.5	217
304	Melanopsin retinal ganglion cell loss in Alzheimer disease. *Annals of Neurology*, **2016**, 79, 90-109	9.4	215
303	Rescue of a mitochondrial deficiency causing Leber Hereditary Optic Neuropathy. *Annals of Neurology*, **2002**, 52, 534-42	9.4	215
302	Optic nerve degeneration and mitochondrial dysfunction: genetic and acquired optic neuropathies. *Neurochemistry International*, **2002**, 40, 573-84	4.4	214
301	Efficient mitochondrial biogenesis drives incomplete penetrance in Leber's hereditary optic neuropathy. *Brain*, **2014**, 137, 335-53	11.2	186
300	Retinal nerve fiber layer evaluation by optical coherence tomography in Leber's hereditary optic neuropathy. *Ophthalmology*, **2005**, 112, 120-6	7.3	180

299	Defective oxidative phosphorylation in thyroid oncocytic carcinoma is associated with pathogenic mitochondrial DNA mutations affecting complexes I and III. *Cancer Research*, **2006**, 66, 6087-96	10.1	178
298	Mitochondrial DNA haplogroup K is associated with a lower risk of Parkinson's disease in Italians. *European Journal of Human Genetics*, **2005**, 13, 748-52	5.3	169
297	Oestrogens ameliorate mitochondrial dysfunction in Leber's hereditary optic neuropathy. *Brain*, **2011**, 134, 220-34	11.2	166
296	Retinal ganglion cell neurodegeneration in mitochondrial inherited disorders. *Biochimica Et Biophysica Acta - Bioenergetics*, **2009**, 1787, 518-28	4.6	163
295	OPA1 links human mitochondrial genome maintenance to mtDNA replication and distribution. *Genome Research*, **2011**, 21, 12-20	9.7	162
294	Haplogroup effects and recombination of mitochondrial DNA: novel clues from the analysis of Leber hereditary optic neuropathy pedigrees. *American Journal of Human Genetics*, **2006**, 78, 564-74	11	156
293	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
292	Idebenone treatment in Leber's hereditary optic neuropathy. *Brain*, **2011**, 134, e188	11.2	149
291	Natural history of Leber's hereditary optic neuropathy: longitudinal analysis of the retinal nerve fiber layer by optical coherence tomography. *Ophthalmology*, **2010**, 117, 623-7	7.3	148
290	Melanopsin retinal ganglion cells are resistant to neurodegeneration in mitochondrial optic neuropathies. *Brain*, **2010**, 133, 2426-38	11.2	140
289	Leber's hereditary optic neuropathy (LHON) pathogenic mutations induce mitochondrial-dependent apoptotic death in transmitochondrial cells incubated with galactose medium. *Journal of Biological Chemistry*, **2003**, 278, 4145-50	5.4	139
288	Correlation between retinal nerve fibre layer thickness and optic nerve head size: an optical coherence tomography study. *British Journal of Ophthalmology*, **2005**, 89, 489-92	5.5	137
287	Phenotypic heterogeneity of the 8344A>G mtDNA "MERRF" mutation. *Neurology*, **2013**, 80, 2049-54	6.5	135
286	Effect of EPI-743 on the clinical course of the mitochondrial disease Leber hereditary optic neuropathy. *Archives of Neurology*, **2012**, 69, 331-8		135
285	New treatments for mitochondrial disease-no time to drop our standards. *Nature Reviews Neurology*, **2013**, 9, 474-81	15	133
284	Mutations in SLC25A46, encoding a UGO1-like protein, cause an optic atrophy spectrum disorder. *Nature Genetics*, **2015**, 47, 926-32	36.3	131
283	Mitochondrial DNA background modulates the assembly kinetics of OXPHOS complexes in a cellular model of mitochondrial disease. *Human Molecular Genetics*, **2008**, 17, 4001-11	5.6	129
282	Severe impairment of complex I-driven adenosine triphosphate synthesis in leber hereditary optic neuropathy cybrids. *Archives of Neurology*, **2005**, 62, 730-6		129

281	The glutamate/cystine xCT antiporter antagonizes glutamine metabolism and reduces nutrient flexibility. *Nature Communications*, **2017**, 8, 15074	17.4	126
280	Biochemical features of mtDNA 14484 (ND6/m64V) point mutation associated with Leber's hereditary optic neuropathy. *Annals of Neurology*, **1999**, 45, 320-328	9.4	126
279	Pathogenic expression of homoplasmic mtDNA mutations needs a complex nuclear-mitochondrial interaction. *Trends in Genetics*, **2003**, 19, 257-62	8.5	124
278	Mitochondrial DNA signals of late glacial recolonization of Europe from near eastern refugia. *American Journal of Human Genetics*, **2012**, 90, 915-24	11	123
277	Deficit of in vivo mitochondrial ATP production in OPA1-related dominant optic atrophy. *Annals of Neurology*, **2004**, 56, 719-23	9.4	122
276	Extensive investigation of a large Brazilian pedigree of 11778/haplogroup J Leber hereditary optic neuropathy. *American Journal of Ophthalmology*, **2003**, 136, 231-8	4.9	122
275	Leber's hereditary optic neuropathy differentially affects smaller axons in the optic nerve. *Transactions of the American Ophthalmological Society*, **2000**, 98, 223-32; discussion 232-5		122
274	Syndromic parkinsonism and dementia associated with OPA1 missense mutations. *Annals of Neurology*, **2015**, 78, 21-38	9.4	119
273	Visual system involvement in patients with Friedreich's ataxia. *Brain*, **2009**, 132, 116-23	11.2	117
272	Detection and quantification of retinal nerve fiber layer thickness in optic disc edema using stratus OCT. *JAMA Ophthalmology*, **2006**, 124, 1111-7		113
271	Retinal nerve fiber layer evaluation by optical coherence tomography in unaffected carriers with Leber's hereditary optic neuropathy mutations. *Ophthalmology*, **2005**, 112, 127-31	7.3	113
270	Leber's hereditary optic neuropathy: biochemical effect of 11778/ND4 and 3460/ND1 mutations and correlation with the mitochondrial genotype. *Neurology*, **1997**, 48, 1623-32	6.5	109
269	MtDNA mutations associated with Leber's hereditary optic neuropathy: studies on cytoplasmic hybrid (cybrid) cells. *Biochemical and Biophysical Research Communications*, **1995**, 210, 880-8	3.4	108
268	Caspase-independent death of Leber's hereditary optic neuropathy cybrids is driven by energetic failure and mediated by AIF and Endonuclease G. *Apoptosis: an International Journal on Programmed Cell Death*, **2005**, 10, 997-1007	5.4	105
267	OPA1 Isoforms in the Hierarchical Organization of Mitochondrial Functions. *Cell Reports*, **2017**, 19, 2557-2571	25.1	103
266	Functional alterations of the mitochondrially encoded ND4 subunit associated with Leber's hereditary optic neuropathy. *FEBS Letters*, **1994**, 352, 375-9	3.8	103
265	Pharmacological Inhibition of Necroptosis Protects from Dopaminergic Neuronal Cell Death in Parkinson's Disease Models. *Cell Reports*, **2018**, 22, 2066-2079	10.6	101
264	Mitochondrial disorders. *Current Opinion in Neurology*, **2007**, 20, 564-71	7.1	101

263	The genetic and metabolic signature of oncocytic transformation implicates HIF1alpha destabilization. *Human Molecular Genetics*, **2010**, 19, 1019-32	5.6	100
262	Cells bearing mutations causing Leber's hereditary optic neuropathy are sensitized to Fas-Induced apoptosis. *Journal of Biological Chemistry*, **2002**, 277, 5810-5	5.4	95
261	The interaction of Q analogs, particularly hydroxydecyl benzoquinone (idebenone), with the respiratory complexes of heart mitochondria. *Archives of Biochemistry and Biophysics*, **1996**, 330, 395-400	4.1	94
260	Mitochondrial DNA and primary mitochondrial dysfunction in Parkinson's disease. *Movement Disorders*, **2017**, 32, 346-363	7	92
259	Leber's Hereditary Optic Neuropathy. *Current Treatment Options in Neurology*, **2011**, 13, 109-17	4.4	92
258	Leber's hereditary optic neuropathy with childhood onset. *Investigative Ophthalmology and Visual Science*, **2006**, 47, 5303-9		92
257	The m.3243A>G mitochondrial DNA mutation and related phenotypes. A matter of gender?. *Journal of Neurology*, **2014**, 261, 504-10	5.5	91
256	A neurodegenerative perspective on mitochondrial optic neuropathies. *Acta Neuropathologica*, **2016**, 132, 789-806	14.3	90
255	Evidence for a novel x-linked modifier locus for leber hereditary optic neuropathy. *Ophthalmic Genetics*, **2008**, 29, 17-24	1.2	89
254	Mathematically modeling the involvement of axons in Leber's hereditary optic neuropathy **2012**, 53, 7608-17		88
253	Leber hereditary optic neuropathy mtDNA mutations disrupt glutamate transport in cybrid cell lines. *Brain*, **2004**, 127, 2183-92	11.2	88
252	The ND1 gene of complex I is a mutational hot spot for Leber's hereditary optic neuropathy. *Annals of Neurology*, **2004**, 56, 631-41	9.4	88
251	The effects of idebenone on mitochondrial bioenergetics. *Biochimica Et Biophysica Acta - Bioenergetics*, **2012**, 1817, 363-9	4.6	87
250	Gastrointestinal dysmotility in mitochondrial neurogastrointestinal encephalomyopathy is caused by mitochondrial DNA depletion. *American Journal of Pathology*, **2008**, 173, 1120-8	5.8	87
249	Inefficient coupling between proton transport and ATP synthesis may be the pathogenic mechanism for NARP and Leigh syndrome resulting from the T8993G mutation in mtDNA. *Biochemical Journal*, **2006**, 395, 493-500	3.8	86
248	Respiratory complex I dysfunction due to mitochondrial DNA mutations shifts the voltage threshold for opening of the permeability transition pore toward resting levels. *Journal of Biological Chemistry*, **2009**, 284, 2045-52	5.4	85
247	Infant and Adult Gut Microbiome and Metabolome in Rural Bassa and Urban Settlers from Nigeria. *Cell Reports*, **2018**, 23, 3056-3067	10.6	84
246	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

245	Antioxidant defences in cybrids harboring mtDNA mutations associated with Leber's hereditary optic neuropathy. *FEBS Journal*, **2005**, 272, 1124-35	5.7	83
244	Catalytic activities of mitochondrial ATP synthase in patients with mitochondrial DNA T8993G mutation in the ATPase 6 gene encoding subunit a. *Journal of Biological Chemistry*, **2000**, 275, 4177-82	5.4	83
243	Bioenergetics of mitochondrial diseases associated with mtDNA mutations. *Biochimica Et Biophysica Acta - Bioenergetics*, **2004**, 1658, 89-94	4.6	82
242	Bioenergetics shapes cellular death pathways in Leber's hereditary optic neuropathy: a model of mitochondrial neurodegeneration. *Biochimica Et Biophysica Acta - Bioenergetics*, **2004**, 1658, 172-9	4.6	82
241	Biochemical phenotypes associated with the mitochondrial ATP6 gene mutations at nt8993. *Biochimica Et Biophysica Acta - Bioenergetics*, **2007**, 1767, 913-9	4.6	79
240	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
239	Mitochondrial DNA: impacting central and peripheral nervous systems. *Neuron*, **2014**, 84, 1126-42	13.9	77
238	A mutation threshold distinguishes the antitumorigenic effects of the mitochondrial gene MTND1, an oncojanus function. *Cancer Research*, **2011**, 71, 6220-9	10.1	77
237	Loss of temporal retinal nerve fibers in Parkinson disease: a mitochondrial pattern?. *European Journal of Neurology*, **2013**, 20, 198-201	6	76
236	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
235	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
234	Eight human OPA1 isoforms, long and short: What are they for?. *Biochimica Et Biophysica Acta - Bioenergetics*, **2018**, 1859, 263-269	4.6	70
233	Clinical and brain bioenergetics improvement with idebenone in a patient with Leber's hereditary optic neuropathy: a clinical and 31P-MRS study. *Journal of the Neurological Sciences*, **1997**, 148, 25-31	3.2	70
232	Retinal nerve fiber layer thickness in nonarteritic anterior ischemic optic neuropathy: OCT characterization of the acute and resolving phases. *Graefeis Archive for Clinical and Experimental Ophthalmology*, **2008**, 246, 641-7	3.8	70
231	Nuclear genes in mitochondrial disorders. *Current Opinion in Genetics and Development*, **2003**, 13, 262-70	4.9	69
230	Ophthalmologic findings in a large pedigree of 11778/Haplogroup J Leber hereditary optic neuropathy. *American Journal of Ophthalmology*, **2004**, 137, 271-7	4.9	69
229	Liver transplantation for mitochondrial neurogastrointestinal encephalomyopathy. *Annals of Neurology*, **2016**, 80, 448-55	9.4	68
228	The optic nerve: a "mito-window" on mitochondrial neurodegeneration. *Molecular and Cellular Neurosciences*, **2013**, 55, 62-76	4.8	66

227	Microcystic macular degeneration from optic neuropathy: not inflammatory, not trans-synaptic degeneration. *Brain*, **2013**, 136, e239	11.2	65
226	The background of mitochondrial DNA haplogroup J increases the sensitivity of Leber's hereditary optic neuropathy cells to 2,5-hexanedione toxicity. *PLoS ONE*, **2009**, 4, e7922	3.7	65
225	Melanopsin-expressing retinal ganglion cells: implications for human diseases. *Vision Research*, **2011**, 51, 296-302	2.1	64
224	Association of optic disc size with development and prognosis of Leber's hereditary optic neuropathy **2009**, 50, 1666-74		63
223	Mitochondrial optic neuropathies: how two genomes may kill the same cell type?. *Bioscience Reports*, **2007**, 27, 173-84	4.1	63
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	Biochemical-clinical correlation in patients with different loads of the mitochondrial DNA T8993G mutation. *Archives of Neurology*, **2002**, 59, 264-70		62
220	Testing models for genetic determination in migraine. *Cephalalgia*, **1993**, 13, 389-94	6.1	62
219	Rare primary mitochondrial DNA mutations and probable synergistic variants in Leber's hereditary optic neuropathy. *PLoS ONE*, **2012**, 7, e42242	3.7	60
218	The pupil light reflex in Leber's hereditary optic neuropathy: evidence for preservation of melanopsin-expressing retinal ganglion cells **2013**, 54, 4471-7		60
217	Retinal nerve fiber layer thickness in dominant optic atrophy measurements by optical coherence tomography and correlation with age. *Ophthalmology*, **2011**, 118, 2076-80	7.3	60
216	Inhibition of mitochondrial function induces an integrated stress response in oligodendroglia. *Neurobiology of Disease*, **2009**, 34, 357-65	7.5	59
215	OPA1-related auditory neuropathy: site of lesion and outcome of cochlear implantation. *Brain*, **2015**, 138, 563-76	11.2	58
214	Respiratory complex I is essential to induce a Warburg profile in mitochondria-defective tumor cells. *Cancer & Metabolism*, **2013**, 1, 11	5.4	58
213	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
212	Rare mtDNA variants in Leber hereditary optic neuropathy families with recurrence of myoclonus. *Neurology*, **2008**, 70, 762-70	6.5	57
211	Mitochondrial neurogastrointestinal encephalomyopathy: evidence of mitochondrial DNA depletion in the small intestine. *Gastroenterology*, **2006**, 130, 893-901	13.3	57
210	Optic neuropathies: the tip of the neurodegeneration iceberg. *Human Molecular Genetics*, **2017**, 26, R139-R150	5.8	55

209	A novel null homozygous mutation confirms CACNA2D2 as a gene mutated in epileptic encephalopathy. *PLoS ONE*, **2013**, 8, e82154	3.7	55
208	Pure and syndromic optic atrophy explained by deep intronic OPA1 mutations and an intralocus modifier. *Brain*, **2014**, 137, 2164-77	11.2	54
207	Thymidine and deoxyuridine accumulate in tissues of patients with mitochondrial neurogastrointestinal encephalomyopathy (MNGIE). *FEBS Letters*, **2007**, 581, 3410-4	3.8	54
206	Redefining phenotypes associated with mitochondrial DNA single deletion. *Journal of Neurology*, **2015**, 262, 1301-9	5.5	53
205	Idebenone treatment in patients with OPA1-mutant dominant optic atrophy. *Brain*, **2013**, 136, e231	11.2	53
204	Male prevalence of acquired color vision defects in asymptomatic carriers of Leber's hereditary optic neuropathy. *Investigative Ophthalmology and Visual Science*, **2007**, 48, 2362-70		53
203	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
202	OPA1 mutations associated with dominant optic atrophy influence optic nerve head size. *Ophthalmology*, **2010**, 117, 1547-53	7.3	51
201	Bilateral visual improvement with unilateral gene therapy injection for Leber hereditary optic neuropathy. *Science Translational Medicine*, **2020**, 12,	17.5	50
200	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
199	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. *Journal of Clinical Investigation*, **2020**, 130, 108-125	15.9	49
198	DNA methyltransferase 1 mutations and mitochondrial pathology: is mtDNA methylated?. *Frontiers in Genetics*, **2015**, 6, 90	4.5	48
197	An inherited mitochondrial DNA disruptive mutation shifts to homoplasmy in oncocytic tumor cells. *Human Mutation*, **2009**, 30, 391-6	4.7	48
196	Respiratory function in cybrid cell lines carrying European mtDNA haplogroups: implications for Leber's hereditary optic neuropathy. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2002**, 1588, 7-14	6.9	48
195	Targeting estrogen receptor as preventive therapeutic strategy for Leber's hereditary optic neuropathy. *Human Molecular Genetics*, **2015**, 24, 6921-31	5.6	47
194	Medical management of hereditary optic neuropathies. *Frontiers in Neurology*, **2014**, 5, 141	4.1	46
193	Mitochondrial DNA nucleotide changes C14482G and C14482A in the ND6 gene are pathogenic for Leber's hereditary optic neuropathy. *Annals of Neurology*, **2002**, 51, 774-8	9.4	46
192	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

191	Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. *Human Molecular Genetics*, **2016**, 25, 1031-41	5.6	44
190	Brain diffusion-weighted imaging in Friedreich's ataxia. *Movement Disorders*, **2011**, 26, 705-12	7	44
189	'Secondary' 4216/ND1 and 13708/ND5 Leber's hereditary optic neuropathy mitochondrial DNA mutations do not further impair in vivo mitochondrial oxidative metabolism when associated with the 11778/ND4 mitochondrial DNA mutation. *Brain*, **2000**, 123 (Pt 9), 1896-902	11.2	44
188	Natural History of Conversion of Leber's Hereditary Optic Neuropathy: A Prospective Case Series. *Ophthalmology*, **2017**, 124, 843-850	7.3	43
187	Mitochondrial disease activates transcripts of the unfolded protein response and cell cycle and inhibits vesicular secretion and oligodendrocyte-specific transcripts. *Mitochondrion*, **2006**, 6, 161-75	4.9	43
186	Recessive Mutations in RTN4IP1 Cause Isolated and Syndromic Optic Neuropathies. *American Journal of Human Genetics*, **2015**, 97, 754-60	11	42
185	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
184	Isolation of transcriptomal changes attributable to LHON mutations and the cybridization process. *Brain*, **2005**, 128, 1026-37	11.2	42
183	Mitochondrial disorders. *Current Opinion in Neurology*, **2003**, 16, 585-594	7.1	42
182	Leber's Hereditary Optic Neuropathy (LHON) with 14484/ND6 mutation in a North African patient. *Journal of the Neurological Sciences*, **1998**, 160, 183-8	3.2	41
181	'Behr syndrome' with OPA1 compound heterozygote mutations. *Brain*, **2015**, 138, e321	11.2	40
180	Grand rounds: could occupational exposure to n-hexane and other solvents precipitate visual failure in leber hereditary optic neuropathy?. *Environmental Health Perspectives*, **2007**, 115, 113-5	8.4	40
179	Subclinical carriers and conversions in Leber hereditary optic neuropathy: a prospective psychophysical study. *Transactions of the American Ophthalmological Society*, **2006**, 104, 51-61		39
178	Incomplete penetrance in mitochondrial optic neuropathies. *Mitochondrion*, **2017**, 36, 130-137	4.9	38
177	Low brain intracellular free magnesium in mitochondrial cytopathies. *Journal of Cerebral Blood Flow and Metabolism*, **1999**, 19, 528-32	7.3	38
176	Narcolepsy is a common phenotype in HSAN IE and ADCA-DN. *Brain*, **2014**, 137, 1643-55	11.2	36
175	Protection against oxidant-induced apoptosis by exogenous glutathione in Leber hereditary optic neuropathy cybrids. *Investigative Ophthalmology and Visual Science*, **2008**, 49, 671-6		36
174	Phosphorus MR spectroscopy shows a tissue specific in vivo distribution of biochemical expression of the G3460A mutation in Leber's hereditary optic neuropathy. *Journal of Neurology, Neurosurgery and Psychiatry*, **2002**, 72, 805-7	5.5	36

173	Parsing the differences in affected with LHON: genetic versus environmental triggers of disease conversion. *Brain*, **2016**, 139, e17	11.2	35
172	Liver as a source for thymidine phosphorylase replacement in mitochondrial neurogastrointestinal encephalomyopathy. *PLoS ONE*, **2014**, 9, e96692	3.7	35
171	A clinically complex form of dominant optic atrophy (OPA8) maps on chromosome 16. *Human Molecular Genetics*, **2011**, 20, 1893-905	5.6	35
170	Chromatic and luminance contrast sensitivities in asymptomatic carriers from a large Brazilian pedigree of 11778 Leber hereditary optic neuropathy. *Investigative Ophthalmology and Visual Science*, **2005**, 46, 4809-14		35
169	Biochemical features of mtDNA 14484 (ND6/M64V) point mutation associated with Leber's hereditary optic neuropathy. *Annals of Neurology*, **1999**, 45, 320-8	9.4	34
168	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
167	Mitochondrial dysfunction in optic neuropathies: animal models and therapeutic options. *Current Opinion in Neurology*, **2013**, 26, 52-8	7.1	33
166	A very large Brazilian pedigree with 11778 Leber's hereditary optic neuropathy. *Transactions of the American Ophthalmological Society*, **2002**, 100, 169-78; discussion 178-9		33
165	Peripapillary vessel density changes in Leber's hereditary optic neuropathy: a new biomarker. *Clinical and Experimental Ophthalmology*, **2018**, 46, 1055-1062	2.4	33
164	"Mitochondrial neuropathies": A survey from the large cohort of the Italian Network. *Neuromuscular Disorders*, **2016**, 26, 272-6	2.9	32
163	Defective mitochondrial adenosine triphosphate production in skeletal muscle from patients with dominant optic atrophy due to OPA1 mutations. *Archives of Neurology*, **2011**, 68, 67-73		32
162	Apoptotic cell death of cybrid cells bearing Leber's hereditary optic neuropathy mutations is caspase independent. *Annals of the New York Academy of Sciences*, **2003**, 1010, 213-7	6.5	32
161	X-inactivation patterns in female Leber's hereditary optic neuropathy patients do not support a strong X-linked determinant. *American Journal of Medical Genetics Part A*, **1996**, 61, 356-62		32
160	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
159	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
158	Lack of association between mitochondrial tRNA(Leu(UUR)) point mutation and cluster headache. *Lancet, The*, **1995**, 345, 1120-1	40	31
157	OPA1: How much do we know to approach therapy?. *Pharmacological Research*, **2018**, 131, 199-210	10.2	29
156	Gamma rays induce a p53-independent mitochondrial biogenesis that is counter-regulated by HIF1 *Cell Death and Disease*, **2013**, 4, e663	9.8	29

155	Colour vision defects in asymptomatic carriers of the Leber's hereditary optic neuropathy (LHON) mtDNA 11778 mutation from a large Brazilian LHON pedigree: a case-control study. *British Journal of Ophthalmology*, **2006**, 90, 150-3	5.5	29
154	Retinal nerve fiber layer thickness variability in Leber hereditary optic neuropathy carriers. *European Journal of Ophthalmology*, **2012**, 22, 985-91	1.9	28
153	Inhibition of autophagy curtails visual loss in a model of autosomal dominant optic atrophy. *Nature Communications*, **2020**, 11, 4029	17.4	28
152	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
151	Myoclonus in mitochondrial disorders. *Movement Disorders*, **2014**, 29, 722-8	7	27
150	Impaired complex I repair causes recessive Leber's hereditary optic neuropathy. *Journal of Clinical Investigation*, **2021**, 131,	15.9	27
149	Differential cerebro spinal fluid proteome investigation of Leber hereditary optic neuropathy (LHON) and multiple sclerosis. *Journal of Neuroimmunology*, **2008**, 193, 156-60	3.5	25
148	Leber hereditary optic neuropathy possibly triggered by exposure to tire fire. *Journal of Neuro-Ophthalmology*, **2006**, 26, 268-72	2.6	25
147	Secondary post-geniculate involvement in Leber's hereditary optic neuropathy. *PLoS ONE*, **2012**, 7, e50230	3.7	25
146	Melanopsin Retinal Ganglion Cells and Pupil: Clinical Implications for Neuro-Ophthalmology. *Frontiers in Neurology*, **2018**, 9, 1047	4.1	25
145	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
144	Optic nerve histopathology in a case of Wolfram Syndrome: a mitochondrial pattern of axonal loss. *Mitochondrion*, **2013**, 13, 841-5	4.9	24
143	Severe CMT type 2 with fatal encephalopathy associated with a novel MFN2 splicing mutation. *Neurology*, **2010**, 74, 1919-21	6.5	24
142	Phosphorylated neurofilament heavy chain is a marker of neurodegeneration in Leber hereditary optic neuropathy (LHON). *Molecular Vision*, **2008**, 14, 2443-50	2.3	24
141	Diffusion Tensor Imaging Mapping of Brain White Matter Pathology in Mitochondrial Optic Neuropathies. *American Journal of Neuroradiology*, **2015**, 36, 1259-65	4.4	23
140	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
139	Revisiting mitochondrial ocular myopathies: a study from the Italian Network. *Journal of Neurology*, **2017**, 264, 1777-1784	5.5	23
138	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

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