Pascal Reynier

List of Publications by Year in Descending Order

171 papers	7,134 citations	45 h-index	79 g-index
190 ext. papers	8,427 ext. citations	5.9 avg, IF	5.25 L-index

#	Paper	IF	Citations
171	Altered Mitochondrial Opa1-Related Fusion in Mouse Promotes Endothelial Cell Dysfunction and Atherosclerosis. *Antioxidants*, **2022**, 11, 1078	7.1	1
170	Metabolic Profile and Pathological Alterations in the Muscle of Patients with Early-Stage Amyotrophic Lateral Sclerosis. *Biomedicines*, **2022**, 10, 1307	4.8	1
169	Dominant optic atrophy: Culprit mitochondria in the optic nerve. *Progress in Retinal and Eye Research*, **2021**, 83, 100935	20.5	11
168	Use of Next-Generation Sequencing for the Molecular Diagnosis of 1,102 Patients With a Autosomal Optic Neuropathy. *Frontiers in Neurology*, **2021**, 12, 602979	4.1	2
167	Secondary coenzyme Q deficiency in neurological disorders. *Free Radical Biology and Medicine*, **2021** , 165, 203-218	7.8	1
166	Metabolomic Sexual Dimorphism of the Mouse Brain is Predominantly Abolished by Gonadectomy with a Higher Impact on Females. *Journal of Proteome Research*, **2021**, 20, 2772-2779	5.6	0
165	Ocular growth and metabolomics are dependent upon the spectral content of ambient white light. *Scientific Reports*, **2021**, 11, 7586	4.9	7
164	Preliminary Metabolomic Profiling of the Vitreous Humor from Hypothermia Fatalities. *Journal of Proteome Research*, **2021**, 20, 2390-2396	5.6	1
163	Dominant mutations are a frequent cause of isolated optic atrophy. *Brain Communications*, **2021**, 3, fcab063	4.5	5
162	High-throughput screening identifies suppressors of mitochondrial fragmentation in OPA1 fibroblasts. *EMBO Molecular Medicine*, **2021**, 13, e13579	12	6
161	Improved detection of mitochondrial DNA instability in mitochondrial genome maintenance disorders. *Genetics in Medicine*, **2021**, 23, 1769-1778	8.1	0
160	Protective role of the mitochondrial fusion protein OPA1 in hypertension. *FASEB Journal*, **2021**, 35, e21678	4.9	2
159	Towards personalized medicine for amyotrophic lateral sclerosis. *Trends in Endocrinology and Metabolism*, **2021**, 32, 839-841	8.8	
158	A plasma metabolomic signature of Leber hereditary optic neuropathy showing taurine and nicotinamide deficiencies. *Human Molecular Genetics*, **2021**, 30, 21-29	5.6	3
157	Embryo and Its Mitochondria. *Antioxidants*, **2021**, 10,	7.1	11
156	Mitochondrial Dysfunction in Mitochondrial Medicine: Current Limitations, Pitfalls, and Tomorrow. *Methods in Molecular Biology*, **2021**, 2276, 1-29	1.4	0
155	Mitochondria: their role in spermatozoa and in male infertility. *Human Reproduction Update*, **2021**, 27, 697-719	15.8	16

154	Dominant mutations in MIEF1 affect mitochondrial dynamics and cause a singular late onset optic neuropathy. *Molecular Neurodegeneration*, **2021**, 16, 12	19	10
153	Optic neuropathy linked to ACAD9 pathogenic variants: A potentially riboflavin-responsive disorder?. *Mitochondrion*, **2021**, 59, 169-174	4.9	1
152	ACO2 clinicobiological dataset with extensive phenotype ontology annotation. *Scientific Data*, **2021**, 8, 205	8.2	0
151	Late-onset argininosuccinic aciduria in a 72-year-old man presenting with fatal hyperammonemia. *JIMD Reports*, **2021**, 62, 44-48	1.9	
150	Tear metabolomics highlights new potential biomarkers for differentiating between Sjögren's syndrome and other causes of dry eye. *Ocular Surface*, **2021**, 22, 110-116	6.5	1
149	Acetoacetate protects macrophages from lactic acidosis-induced mitochondrial dysfunction by metabolic reprograming. *Nature Communications*, **2021**, 12, 7115	17.4	2
148	Elevated Levels of Monocyte Chemotactic Protein-1 in the Follicular Fluid Reveals Different Populations among Women with Severe Endometriosis. *Journal of Clinical Medicine*, **2020**, 9,	5.1	6
147	Metabolomic Profiling of Plasma and Erythrocytes in Sickle Mice Points to Altered Nociceptive Pathways. *Cells*, **2020**, 9,	7.9	1
146	Sexual Dimorphism of Metabolomic Profile in Arterial Hypertension. *Scientific Reports*, **2020**, 10, 7517	4.9	6
145	Metabolomics hallmarks OPA1 variants correlating with their in vitro phenotype and predicting clinical severity. *Human Molecular Genetics*, **2020**, 29, 1319-1329	5.6	9
144	A Plasma Metabolomic Profiling of Exudative Age-Related Macular Degeneration Showing Carnosine and Mitochondrial Deficiencies. *Journal of Clinical Medicine*, **2020**, 9,	5.1	6
143	A Data Mining Metabolomics Exploration of Glaucoma. *Metabolites*, **2020**, 10,	5.6	9
142	Tryptophane-kynurenine pathway in the remote ischemic conditioning mechanism. *Basic Research in Cardiology*, **2020**, 115, 13	11.8	11
141	Increased Protein -Glutathionylation in Leber's Hereditary Optic Neuropathy (LHON). *International Journal of Molecular Sciences*, **2020**, 21,	6.3	3
140	Mutations in aARS genes revealed by targeted next-generation sequencing in patients with mitochondrial diseases. *Molecular Biology Reports*, **2020**, 47, 3779-3787	2.8	2
139	Mutations in the m-AAA proteases AFG3L2 and SPG7 are causing isolated dominant optic atrophy. *Neurology: Genetics*, **2020**, 6, e428	3.8	18
138	Oxidative stress contributes differentially to the pathophysiology of Charcot-Marie-Tooth disease type 2K. *Experimental Neurology*, **2020**, 323, 113069	5.7	13
137	Familial Dysalbuminemic Hyperthyroxinemia: An Underdiagnosed Entity. *Journal of Clinical Medicine*, **2020**, 9,	5.1	2

136	Endometriosis Lowers the Cumulative Live Birth Rates in IVF by Decreasing the Number of Embryos but Not Their Quality. *Journal of Clinical Medicine*, **2020**, 9,	5.1	10
135	Mitochondrial DNA Parameters in Blood of Infants Receiving Lopinavir/Ritonavir or Lamivudine Prophylaxis to Prevent Breastfeeding Transmission of HIV-1. *Journal of Clinical Medicine*, **2020**, 9,	5.1	1
134	Metabolomics reveals highly regional specificity of cerebral sexual dimorphism in mice. *Progress in Neurobiology*, **2020**, 184, 101698	10.9	6
133	Sickle Cell Disease: Metabolomic Profiles of Vaso-Occlusive Crisis in Plasma and Erythrocytes. *Journal of Clinical Medicine*, **2020**, 9,	5.1	3
132	OPA1: 516 unique variants and 831 patients registered in an updated centralized Variome database. *Orphanet Journal of Rare Diseases*, **2019**, 14, 214	4.2	21
131	Metabolomics shows no impairment of the microenvironment of the cumulus-oocyte complex in women with isolated endometriosis. *Reproductive BioMedicine Online*, **2019**, 39, 885-892	4	5
130	Metabolomic Profiling of Aqueous Humor in Glaucoma Points to Taurine and Spermine Deficiency: Findings from the Eye-D Study. *Journal of Proteome Research*, **2019**, 18, 1307-1315	5.6	20
129	Metabo-lipidomics of Fibroblasts and Mitochondrial-Endoplasmic Reticulum Extracts from ALS Patients Shows Alterations in Purine, Pyrimidine, Energetic, and Phospholipid Metabolisms. *Molecular Neurobiology*, **2019**, 56, 5780-5791	6.2	18
128	Nicotinamide Deficiency in Primary Open-Angle Glaucoma **2019**, 60, 2509-2514		29
127	Maternal ageing impairs mitochondrial DNA kinetics during early embryogenesis in mice. *Human Reproduction*, **2019**, 34, 1313-1324	5.7	7
126	Lipidomics Reveals Triacylglycerol Accumulation Due to Impaired Fatty Acid Flux in Opa1-Disrupted Fibroblasts. *Journal of Proteome Research*, **2019**, 18, 2779-2790	5.6	4
125	Warburg-like effect is a hallmark of complex I assembly defects. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2019**, 1865, 2475-2489	6.9	2
124	The Metabolomic Signature of Opa1 Deficiency in Rat Primary Cortical Neurons Shows Aspartate/Glutamate Depletion and Phospholipids Remodeling. *Scientific Reports*, **2019**, 9, 6107	4.9	5
123	A serum metabolomics signature of hypothermia fatalities involving arginase activity, tryptophan content, and phosphatidylcholine saturation. *International Journal of Legal Medicine*, **2019**, 133, 889-898	3.1	6
122	Next generation sequencing in family with MNGIE syndrome associated to optic atrophy: Novel homozygous POLG mutation in the C-terminal sub-domain leading to mtDNA depletion. *Clinica Chimica Acta*, **2019**, 488, 104-110	6.2	7
121	Metabolomic Approach in STEMI-Patients Undergoing Left Ventricular Remodeling. *International Journal of Molecular Sciences*, **2019**, 20,	6.3	2
120	The mitochondrial DNA content of cumulus cells may help predict embryo implantation. *Journal of Assisted Reproduction and Genetics*, **2019**, 36, 223-228	3.4	15
119	eKLIPse: a sensitive tool for the detection and quantification of mitochondrial DNA deletions from next-generation sequencing data. *Genetics in Medicine*, **2019**, 21, 1407-1416	8.1	20

118	The accumulation of assembly intermediates of the mitochondrial complex I matrix arm is reduced by limiting glucose uptake in a neuronal-like model of MELAS syndrome. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2018**, 1864, 1596-1608	6.9	9
117	Current mechanistic insights into the CCCP-induced cell survival response. *Biochemical Pharmacology*, **2018**, 148, 100-110	6	33
116	Cyclosporine A does not prevent second-eye involvement in Leber's hereditary optic neuropathy. *Orphanet Journal of Rare Diseases*, **2018**, 13, 33	4.2	10
115	Updated review of postmortem biochemical exploration of hypothermia with a presentation of standard strategy of sampling and analyses. *Clinical Chemistry and Laboratory Medicine*, **2018**, 56, 1819-1827	5.9	6
114	Reply: The expanding neurological phenotype of DNM1L-related disorders. *Brain*, **2018**, 141, e29	11.2	3
113	The combination of four analytical methods to explore skeletal muscle metabolomics: Better coverage of metabolic pathways or a marketing argument?. *Journal of Pharmaceutical and Biomedical Analysis*, **2018**, 148, 273-279	3.5	14
112	A Plasma Metabolomic Signature of the Exfoliation Syndrome Involves Amino Acids, Acylcarnitines, and Polyamines **2018**, 59, 1025-1032		10
111	Mitochondrial complex I defect resulting from exercise-induced lower limb ischemia in patients with peripheral arterial disease. *Journal of Applied Physiology*, **2018**, 125, 938-946	3.7	4
110	The Metabolomic Bioenergetic Signature of Opa1-Disrupted Mouse Embryonic Fibroblasts Highlights Aspartate Deficiency. *Scientific Reports*, **2018**, 8, 11528	4.9	12
109	Primary antiphospholipid syndrome and antiphospholipid syndrome associated to systemic lupus: Are they different entities?. *Autoimmunity Reviews*, **2018**, 17, 739-745	13.6	15
108	Study of mitochondrial function in placental insufficiency. *Placenta*, **2018**, 67, 1-7	3.4	7
107	Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. *JAMA Neurology*, **2018**, 75, 105-113	17.2	20
106	Metabolomics and Lipidomics Profiling of a Combined Mitochondrial Plus Endoplasmic Reticulum Fraction of Human Fibroblasts: A Robust Tool for Clinical Studies. *Journal of Proteome Research*, **2018**, 17, 745-750	5.6	12
105	Bioinformatics Tools and Databases to Assess the Pathogenicity of Mitochondrial DNA Variants in the Field of Next Generation Sequencing. *Frontiers in Genetics*, **2018**, 9, 632	4.5	30
104	A Plasma Metabolomic Signature Involving Purine Metabolism in Human Optic Atrophy 1 (OPA1)-Related Disorders **2018**, 59, 185-195		15
103	A Metabolomics Profiling of Glaucoma Points to Mitochondrial Dysfunction, Senescence, and Polyamines Deficiency **2018**, 59, 4355-4361		24
102	The mitochondrial DNA content of cumulus granulosa cells is linked to embryo quality. *Human Reproduction*, **2017**, 32, 607-614	5.7	44
101	CLUH couples mitochondrial distribution to the energetic and metabolic status. *Journal of Cell Science*, **2017**, 130, 1940-1951	5.3	22

100	A randomized, double-blind, placebo-controlled trial evaluating cysteamine in Huntington's disease. *Movement Disorders*, **2017**, 32, 932-936	7	22
99	Autophagy controls the pathogenicity of OPA1 mutations in dominant optic atrophy. *Journal of Cellular and Molecular Medicine*, **2017**, 21, 2284-2297	5.6	24
98	A Nontargeted UHPLC-HRMS Metabolomics Pipeline for Metabolite Identification: Application to Cardiac Remote Ischemic Preconditioning. *Analytical Chemistry*, **2017**, 89, 2138-2146	7.8	29
97	Multiethnic involvement in autosomal-dominant optic atrophy in Singapore. *Eye*, **2017**, 31, 475-480	4.4	5
96	5-Oxoprolinuria in hyperammonemic encephalopathy: Coincidence or worsening factor?. *Clinical Biochemistry*, **2017**, 50, 1115-1117	3.5	2
95	Mutations in DNM1L, as in OPA1, result in dominant optic atrophy despite opposite effects on mitochondrial fusion and fission. *Brain*, **2017**, 140, 2586-2596	11.2	79
94	Deep sequencing shows that oocytes are not prone to accumulate mtDNA heteroplasmic mutations during ovarian ageing. *Human Reproduction*, **2017**, 32, 2101-2109	5.7	20
93	The addition of ketone bodies alleviates mitochondrial dysfunction by restoring complex I assembly in a MELAS cellular model. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2017**, 1863, 284-291	6.9	28
92	Letter in response to remote ischaemic conditioning provides humoral cross-species cardioprotection through glycine receptor activation. *Cardiovascular Research*, **2017**, 113, 562	9.9	0
91	Lipidomics Reveals Cerebrospinal-Fluid Signatures of ALS. *Scientific Reports*, **2017**, 7, 17652	4.9	72
90	Novel gene mutation in an atypical late-onset mitochondrial form of multifocal dystonia. *Neurology: Genetics*, **2017**, 3, e205	3.8	5
89	Targeted Metabolomics Reveals Early Dominant Optic Atrophy Signature in Optic Nerves of Opa1delTTAG/+ Mice **2017**, 58, 812-820		15
88	Ovarian ageing: the role of mitochondria in oocytes and follicles. *Human Reproduction Update*, **2016** , 22, 725-743	15.8	201
87	Biallelic Variants in UBA5 Reveal that Disruption of the UFM1 Cascade Can Result in Early-Onset Encephalopathy. *American Journal of Human Genetics*, **2016**, 99, 695-703	11	47
86	The metabolomic signature of Leber's hereditary optic neuropathy reveals endoplasmic reticulum stress. *Brain*, **2016**, 139, 2864-2876	11.2	28
85	Increased mitochondrial fusion in a autosomal recessive CMT2A family with mitochondrial GTPase mitofusin 2 mutations. *Journal of the Peripheral Nervous System*, **2016**, 21, 365-369	4.7	9
84	OPA1-related disorders: Diversity of clinical expression, modes of inheritance and pathophysiology. *Neurobiology of Disease*, **2016**, 90, 20-6	7.5	39
83	Increase in Cardiac Ischemia-Reperfusion Injuries in Opa1+/- Mouse Model. *PLoS ONE*, **2016**, 11, e0164066	3.7	37

82	Increased steroidogenesis promotes early-onset and severe vision loss in females with OPA1 dominant optic atrophy. *Human Molecular Genetics*, **2016**, 25, 2539-2551	5.6	11
81	Loss of function of in mice induces deafness and cochlear outer hair cells' degeneration. *Cell Death Discovery*, **2016**, 2, 16017	6.9	7
80	Recurrent Isolated Neonatal Hemolytic Anemia: Think About Glutathione Synthetase Deficiency. *Pediatrics*, **2016**, 138,	7.4	6
79	Neuropathies optiques d□rigine mitochondriale. *Pratique Neurologique - FMC*, **2016**, 7, 100-116	0	
78	Metabolic Signature of Remote Ischemic Preconditioning Involving a Cocktail of Amino Acids and Biogenic Amines. *Journal of the American Heart Association*, **2016**, 5,	6	25
77	Loss of functional OPA1 unbalances redox state: implications in dominant optic atrophy pathogenesis. *Annals of Clinical and Translational Neurology*, **2016**, 3, 408-21	5.3	24
76	Recessive Mutations in RTN4IP1 Cause Isolated and Syndromic Optic Neuropathies. *American Journal of Human Genetics*, **2015**, 97, 754-60	11	42
75	Mitochondrial energetic defects in muscle and brain of a Hmbs-/- mouse model of acute intermittent porphyria. *Human Molecular Genetics*, **2015**, 24, 5015-23	5.6	25
74	Improved locus-specific database for OPA1 mutations allows inclusion of advanced clinical data. *Human Mutation*, **2015**, 36, 20-5	4.7	35
73	Relationship between diminished ovarian reserve and mitochondrial biogenesis in cumulus cells. *Human Reproduction*, **2015**, 30, 1653-64	5.7	75
72	Assembly defects induce oxidative stress in inherited mitochondrial complex I deficiency. *International Journal of Biochemistry and Cell Biology*, **2015**, 65, 91-103	5.6	26
71	Resveratrol Directly Binds to Mitochondrial Complex I and Increases Oxidative Stress in Brain Mitochondria of Aged Mice. *PLoS ONE*, **2015**, 10, e0144290	3.7	53
70	Acute intermittent porphyria causes hepatic mitochondrial energetic failure in a mouse model. *International Journal of Biochemistry and Cell Biology*, **2014**, 51, 93-101	5.6	36
69	Pro-oxidant effect of ALA is implicated in mitochondrial dysfunction of HepG2 cells. *Biochimie*, **2014** , 106, 157-66	4.6	18
68	Perspectives of drug-based neuroprotection targeting mitochondria. *Revue Neurologique*, **2014**, 170, 390-400	3	47
67	Early-onset Behr syndrome due to compound heterozygous mutations in OPA1. *Brain*, **2014**, 137, e301	11.2	52
66	Mitochondrial dysfunction affecting visual pathways. *Revue Neurologique*, **2014**, 170, 344-54	3	10
65	Resveratrol induces a mitochondrial complex I-dependent increase in NADH oxidation responsible for sirtuin activation in liver cells. *Journal of Biological Chemistry*, **2013**, 288, 36662-75	5.4	91

64	Relative frequencies of inherited retinal dystrophies and optic neuropathies in Southern France: assessment of 21-year data management. *Ophthalmic Epidemiology*, **2013**, 20, 13-25	1.9	36
63	Prevalence of rare mitochondrial DNA mutations in mitochondrial disorders. *Journal of Medical Genetics*, **2013**, 50, 704-14	5.8	76
62	Sensorineural hearing loss in OPA1-linked disorders. *Brain*, **2013**, 136, e236	11.2	23
61	Metabolically induced heteroplasmy shifting and l-arginine treatment reduce the energetic defect in a neuronal-like model of MELAS. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2012**, 1822, 1019-29	6.9	30
60	Standardized mitochondrial analysis gives new insights into mitochondrial dynamics and OPA1 function. *International Journal of Biochemistry and Cell Biology*, **2012**, 44, 980-8	5.6	31
59	Dominant optic atrophy. *Orphanet Journal of Rare Diseases*, **2012**, 7, 46	4.2	152
58	Rare primary mitochondrial DNA mutations and probable synergistic variants in Leber's hereditary optic neuropathy. *PLoS ONE*, **2012**, 7, e42242	3.7	60
57	The human OPA1delTTAG mutation induces premature age-related systemic neurodegeneration in mouse. *Brain*, **2012**, 135, 3599-613	11.2	78
56	Comparison of spheroids formed by rat glioma stem cells and neural stem cells reveals differences in glucose metabolism and promising therapeutic applications. *Journal of Biological Chemistry*, **2012** , 287, 33664-74	5.4	49
55	Cataract as a phenotypic marker for a mutation in WFS1, the Wolfram syndrome gene. *European Journal of Ophthalmology*, **2012**, 22, 254-8	1.9	8
54	Neurological Diseases Associated with Mutations in the Mitochondrial Fusion Machinery **2011**, 169-196		
53	Mitochondrial dysfunction and pathophysiology of Charcot-Marie-Tooth disease involving GDAP1 mutations. *Experimental Neurology*, **2011**, 227, 31-41	5.7	70
52	A locus-specific database for mutations in GDAP1 allows analysis of genotype-phenotype correlations in Charcot-Marie-Tooth diseases type 4A and 2K. *Orphanet Journal of Rare Diseases*, **2011**, 6, 87	4.2	27
51	Idebenone increases mitochondrial complex I activity in fibroblasts from LHON patients while producing contradictory effects on respiration. *BMC Research Notes*, **2011**, 4, 557	2.3	45
50	Adenine nucleotide translocase 2 is a key mitochondrial protein in cancer metabolism. *Biochimica Et Biophysica Acta - Bioenergetics*, **2011**, 1807, 562-7	4.6	113
49	Hereditary spastic paraplegia-like disorder due to a mitochondrial ATP6 gene point mutation. *Mitochondrion*, **2011**, 11, 70-5	4.9	63
48	Heterozygous OPA1 mutations in Behr syndrome. *Brain*, **2011**, 134, e169; author reply e170	11.2	28
47	Never too old to harbour a young man's disease?. *British Journal of Ophthalmology*, **2011**, 95, 887, 896-7	5.5	18

46	OPA1 links human mitochondrial genome maintenance to mtDNA replication and distribution. *Genome Research*, **2011**, 21, 12-20	9.7	162
45	Bioenergetic defect associated with mKATP channel opening in a mouse model carrying a mitofusin 2 mutation. *FASEB Journal*, **2011**, 25, 1618-27	0.9	33
44	Simultaneous MFN2 and GDAP1 mutations cause major mitochondrial defects in a patient with CMT. *Neurology*, **2011**, 76, 1524-6	6.5	27
43	Ethambutol-induced optic neuropathy linked to OPA1 mutation and mitochondrial toxicity. *Mitochondrion*, **2010**, 10, 115-24	4.9	36
42	Multi-system neurological disease is common in patients with OPA1 mutations. *Brain*, **2010**, 133, 771-86	11.2	314
41	Genetically determined optic neuropathies. *Current Opinion in Neurology*, **2010**, 23, 24-8	7.1	25
40	Adenine nucleotide translocase is involved in a mitochondrial coupling defect in MFN2-related Charcot-Marie-Tooth type 2A disease. *Neurogenetics*, **2010**, 11, 127-33	3	31
39	Molecular screening of 980 cases of suspected hereditary optic neuropathy with a report on 77 novel OPA1 mutations. *Human Mutation*, **2009**, 30, E692-705	4.7	120
38	Mitochondrial bioenergetic background confers a survival advantage to HepG2 cells in response to chemotherapy. *Molecular Carcinogenesis*, **2009**, 48, 733-41	5	20
37	Mitochondrial complex I deficiency in GDAP1-related autosomal dominant Charcot-Marie-Tooth disease (CMT2K). *Neurogenetics*, **2009**, 10, 145-50	3	64
36	OPA1-related dominant optic atrophy is not strongly influenced by mitochondrial DNA background. *BMC Medical Genetics*, **2009**, 10, 70	2.1	11
35	OPA1-associated disorders: phenotypes and pathophysiology. *International Journal of Biochemistry and Cell Biology*, **2009**, 41, 1855-65	5.6	105
34	OPA1 functions in mitochondria and dysfunctions in optic nerve. *International Journal of Biochemistry and Cell Biology*, **2009**, 41, 1866-74	5.6	59
33	Acute and late-onset optic atrophy due to a novel OPA1 mutation leading to a mitochondrial coupling defect. *Molecular Vision*, **2009**, 15, 598-608	2.3	33
32	Mitochondrial DNA A3243G mutation involved in familial diabetes, chronic intestinal pseudo-obstruction and recurrent pancreatitis. *Diabetes and Metabolism*, **2008**, 34, 620-6	5.4	22
31	Mutation of OPA1 causes dominant optic atrophy with external ophthalmoplegia, ataxia, deafness and multiple mitochondrial DNA deletions: a novel disorder of mtDNA maintenance. *Brain*, **2008**, 131, 329-37	11.2	331
30	OPA1 mutations induce mitochondrial DNA instability and optic atrophy 'plus' phenotypes. *Brain*, **2008**, 131, 338-51	11.2	394
29	Multiple sclerosis-like disorder in OPA1-related autosomal dominant optic atrophy. *Neurology*, **2008**, 70, 1152-3	6.5	48

28	New evidence of a mitochondrial genetic background paradox: impact of the J haplogroup on the A3243G mutation. *BMC Medical Genetics*, **2008**, 9, 41	2.1	18
27	Reversible optic neuropathy with OPA1 exon 5b mutation. *Annals of Neurology*, **2008**, 63, 667-71	9.4	30
26	Hereditary optic neuropathies share a common mitochondrial coupling defect. *Annals of Neurology*, **2008**, 63, 794-8	9.4	102
25	Mitochondrial coupling defect in Charcot-Marie-Tooth type 2A disease. *Annals of Neurology*, **2007**, 61, 315-23	9.4	109
24	Effects of OPA1 mutations on mitochondrial morphology and apoptosis: relevance to ADOA pathogenesis. *Journal of Cellular Physiology*, **2007**, 211, 423-30	7	117
23	Mitochondrial DNA in the oocyte and the developing embryo. *Current Topics in Developmental Biology*, **2007**, 77, 51-83	5.3	120
22	Mitochondrial dynamics and disease, OPA1. *Biochimica Et Biophysica Acta - Molecular Cell Research*, **2006**, 1763, 500-9	4.9	162
21	mtDNA controls expression of the Death Associated Protein 3. *Experimental Cell Research*, **2006**, 312, 737-45	4.2	11
20	Low oocyte mitochondrial DNA content in ovarian insufficiency. *Human Reproduction*, **2005**, 20, 593-7	5.7	208
19	Clinical Heterogeneity of Hereditary Optic Atrophy in a Turkish Family. *Neuro-Ophthalmology*, **2005**, 29, 9-15	0.9	
18	A novel mutation in the mitochondrial tRNA Asn gene associated with a lethal disease. *Biochemical and Biophysical Research Communications*, **2005**, 329, 1152-4	3.4	14
17	Increase of mitochondrial DNA content and transcripts in early bovine embryogenesis associated with upregulation of mtTFA and NRF1 transcription factors. *Reproductive Biology and Endocrinology* , **2005**, 3, 65	5	120
16	Sporadic optic atrophy due to synonymous codon change altering mRNA splicing of OPA1. *Clinical Genetics*, **2005**, 67, 102-3	4	9
15	eOPA1: an online database for OPA1 mutations. *Human Mutation*, **2005**, 25, 423-8	4.7	101
14	OPA1 R445H mutation in optic atrophy associated with sensorineural deafness. *Annals of Neurology* , **2005**, 58, 958-63	9.4	135
13	Maternal smoking is associated with mitochondrial DNA depletion and respiratory chain complex III deficiency in placenta. *American Journal of Physiology - Endocrinology and Metabolism*, **2005**, 288, E171-7	6	66
12	In vitro embryo production efficiency in cattle and its association with oocyte adenosine triphosphate content, quantity of mitochondrial DNA, and mitochondrial DNA haplogroup. *Biology of Reproduction*, **2004**, 71, 697-704	3.9	88
11	OPA3 gene mutations responsible for autosomal dominant optic atrophy and cataract. *Journal of Medical Genetics*, **2004**, 41, e110	5.8	111

10	Fourteen novel OPA1 mutations in autosomal dominant optic atrophy including two de novo mutations in sporadic optic atrophy. *Human Mutation*, **2003**, 21, 656	4.7	44
9	The association of autosomal dominant optic atrophy and moderate deafness may be due to the R445H mutation in the OPA1 gene. *American Journal of Ophthalmology*, **2003**, 136, 1170-1	4.9	51
8	Increased sperm mitochondrial DNA content in male infertility. *Human Reproduction*, **2003**, 18, 550-6	5.7	117
7	Leigh-like encephalopathy complicating Leber's hereditary optic neuropathy. *Annals of Neurology*, **2002**, 52, 374-7	9.4	49
6	Analysis of Tg transcripts by real-time RT-PCR in the blood of thyroid cancer patients. *Journal of Clinical Endocrinology and Metabolism*, **2002**, 87, 635-9	5.6	43
5	Structure and chromosomal distribution of human mitochondrial pseudogenes. *Genomics*, **2002**, 80, 71-7	4.3	113
4	Mitochondrial DNA content affects the fertilizability of human oocytes. *Molecular Human Reproduction*, **2001**, 7, 425-9	4.4	379
3	mtDNA haplogroup J: a contributing factor of optic neuritis. *European Journal of Human Genetics*, **1999**, 7, 404-6	5.3	45
2	Long PCR analysis of human gamete mtDNA suggests defective mitochondrial maintenance in spermatozoa and supports the bottleneck theory for oocytes. *Biochemical and Biophysical Research Communications*, **1998**, 252, 373-7	3.4	48
1	Oligoasthenospermia associated with multiple mitochondrial DNA rearrangements. *Molecular Human Reproduction*, **1997**, 3, 811-4	4.4	73