

# Pascal Reynier

## List of Publications by Citations

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171  
papers

7,134  
citations

45  
h-index

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190  
ext. papers

8,427  
ext. citations

5.9  
avg, IF

5.25  
L-index

#	Paper	IF	Citations
171	OPA1 mutations induce mitochondrial DNA instability and optic atrophy 'plus' phenotypes. <i>Brain</i> , <b>2008</b> , 131, 338-51	11.2	394
170	Mitochondrial DNA content affects the fertilizability of human oocytes. <i>Molecular Human Reproduction</i> , <b>2001</b> , 7, 425-9	4.4	379
169	Mutation of OPA1 causes dominant optic atrophy with external ophthalmoplegia, ataxia, deafness and multiple mitochondrial DNA deletions: a novel disorder of mtDNA maintenance. <i>Brain</i> , <b>2008</b> , 131, 329-37	11.2	331
168	Multi-system neurological disease is common in patients with OPA1 mutations. <i>Brain</i> , <b>2010</b> , 133, 771-86	11.2	314
167	Low oocyte mitochondrial DNA content in ovarian insufficiency. <i>Human Reproduction</i> , <b>2005</b> , 20, 593-7	5.7	208
166	Ovarian ageing: the role of mitochondria in oocytes and follicles. <i>Human Reproduction Update</i> , <b>2016</b> , 22, 725-743	15.8	201
165	OPA1 links human mitochondrial genome maintenance to mtDNA replication and distribution. <i>Genome Research</i> , <b>2011</b> , 21, 12-20	9.7	162
164	Mitochondrial dynamics and disease, OPA1. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , <b>2006</b> , 1763, 500-9	4.9	162
163	Dominant optic atrophy. <i>Orphanet Journal of Rare Diseases</i> , <b>2012</b> , 7, 46	4.2	152
162	OPA1 R445H mutation in optic atrophy associated with sensorineural deafness. <i>Annals of Neurology</i> , <b>2005</b> , 58, 958-63	9.4	135
161	Molecular screening of 980 cases of suspected hereditary optic neuropathy with a report on 77 novel OPA1 mutations. <i>Human Mutation</i> , <b>2009</b> , 30, E692-705	4.7	120
160	Mitochondrial DNA in the oocyte and the developing embryo. <i>Current Topics in Developmental Biology</i> , <b>2007</b> , 77, 51-83	5.3	120
159	Increase of mitochondrial DNA content and transcripts in early bovine embryogenesis associated with upregulation of mtTFA and NRF1 transcription factors. <i>Reproductive Biology and Endocrinology</i> , <b>2005</b> , 3, 65	5	120
158	Effects of OPA1 mutations on mitochondrial morphology and apoptosis: relevance to ADOA pathogenesis. <i>Journal of Cellular Physiology</i> , <b>2007</b> , 211, 423-30	7	117
157	Increased sperm mitochondrial DNA content in male infertility. <i>Human Reproduction</i> , <b>2003</b> , 18, 550-6	5.7	117
156	Adenine nucleotide translocase 2 is a key mitochondrial protein in cancer metabolism. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , <b>2011</b> , 1807, 562-7	4.6	113
155	Structure and chromosomal distribution of human mitochondrial pseudogenes. <i>Genomics</i> , <b>2002</b> , 80, 71-74	4.3	113

154	OPA3 gene mutations responsible for autosomal dominant optic atrophy and cataract. <i>Journal of Medical Genetics</i> , <b>2004</b> , 41, e110	5.8	111
153	Mitochondrial coupling defect in Charcot-Marie-Tooth type 2A disease. <i>Annals of Neurology</i> , <b>2007</b> , 61, 315-23	9.4	109
152	OPA1-associated disorders: phenotypes and pathophysiology. <i>International Journal of Biochemistry and Cell Biology</i> , <b>2009</b> , 41, 1855-65	5.6	105
151	Hereditary optic neuropathies share a common mitochondrial coupling defect. <i>Annals of Neurology</i> , <b>2008</b> , 63, 794-8	9.4	102
150	eOPA1: an online database for OPA1 mutations. <i>Human Mutation</i> , <b>2005</b> , 25, 423-8	4.7	101
149	Resveratrol induces a mitochondrial complex I-dependent increase in NADH oxidation responsible for sirtuin activation in liver cells. <i>Journal of Biological Chemistry</i> , <b>2013</b> , 288, 36662-75	5.4	91
148	In vitro embryo production efficiency in cattle and its association with oocyte adenosine triphosphate content, quantity of mitochondrial DNA, and mitochondrial DNA haplogroup. <i>Biology of Reproduction</i> , <b>2004</b> , 71, 697-704	3.9	88
147	Mutations in DNM1L, as in OPA1, result in dominant optic atrophy despite opposite effects on mitochondrial fusion and fission. <i>Brain</i> , <b>2017</b> , 140, 2586-2596	11.2	79
146	The human OPA1delTTAG mutation induces premature age-related systemic neurodegeneration in mouse. <i>Brain</i> , <b>2012</b> , 135, 3599-613	11.2	78
145	Prevalence of rare mitochondrial DNA mutations in mitochondrial disorders. <i>Journal of Medical Genetics</i> , <b>2013</b> , 50, 704-14	5.8	76
144	Relationship between diminished ovarian reserve and mitochondrial biogenesis in cumulus cells. <i>Human Reproduction</i> , <b>2015</b> , 30, 1653-64	5.7	75
143	Oligoasthenospermia associated with multiple mitochondrial DNA rearrangements. <i>Molecular Human Reproduction</i> , <b>1997</b> , 3, 811-4	4.4	73
142	Lipidomics Reveals Cerebrospinal-Fluid Signatures of ALS. <i>Scientific Reports</i> , <b>2017</b> , 7, 17652	4.9	72
141	Mitochondrial dysfunction and pathophysiology of Charcot-Marie-Tooth disease involving GDAP1 mutations. <i>Experimental Neurology</i> , <b>2011</b> , 227, 31-41	5.7	70
140	Maternal smoking is associated with mitochondrial DNA depletion and respiratory chain complex III deficiency in placenta. <i>American Journal of Physiology - Endocrinology and Metabolism</i> , <b>2005</b> , 288, E171-7 <sup>6</sup>		66
139	Mitochondrial complex I deficiency in GDAP1-related autosomal dominant Charcot-Marie-Tooth disease (CMT2K). <i>Neurogenetics</i> , <b>2009</b> , 10, 145-50	3	64
138	Hereditary spastic paraplegia-like disorder due to a mitochondrial ATP6 gene point mutation. <i>Mitochondrion</i> , <b>2011</b> , 11, 70-5	4.9	63
137	Rare primary mitochondrial DNA mutations and probable synergistic variants in Leber's hereditary optic neuropathy. <i>PLoS ONE</i> , <b>2012</b> , 7, e42242	3.7	60

136	OPA1 functions in mitochondria and dysfunctions in optic nerve. <i>International Journal of Biochemistry and Cell Biology</i> , <b>2009</b> , 41, 1866-74	5.6	59
135	Resveratrol Directly Binds to Mitochondrial Complex I and Increases Oxidative Stress in Brain Mitochondria of Aged Mice. <i>PLoS ONE</i> , <b>2015</b> , 10, e0144290	3.7	53
134	Early-onset Behr syndrome due to compound heterozygous mutations in OPA1. <i>Brain</i> , <b>2014</b> , 137, e301	11.2	52
133	The association of autosomal dominant optic atrophy and moderate deafness may be due to the R445H mutation in the OPA1 gene. <i>American Journal of Ophthalmology</i> , <b>2003</b> , 136, 1170-1	4.9	51
132	Comparison of spheroids formed by rat glioma stem cells and neural stem cells reveals differences in glucose metabolism and promising therapeutic applications. <i>Journal of Biological Chemistry</i> , <b>2012</b> , 287, 33664-74	5.4	49
131	Leigh-like encephalopathy complicating Leber's hereditary optic neuropathy. <i>Annals of Neurology</i> , <b>2002</b> , 52, 374-7	9.4	49
130	Multiple sclerosis-like disorder in OPA1-related autosomal dominant optic atrophy. <i>Neurology</i> , <b>2008</b> , 70, 1152-3	6.5	48
129	Long PCR analysis of human gamete mtDNA suggests defective mitochondrial maintenance in spermatozoa and supports the bottleneck theory for oocytes. <i>Biochemical and Biophysical Research Communications</i> , <b>1998</b> , 252, 373-7	3.4	48
128	Biallelic Variants in UBA5 Reveal that Disruption of the UFM1 Cascade Can Result in Early-Onset Encephalopathy. <i>American Journal of Human Genetics</i> , <b>2016</b> , 99, 695-703	11	47
127	Perspectives of drug-based neuroprotection targeting mitochondria. <i>Revue Neurologique</i> , <b>2014</b> , 170, 390-400	3	47
126	Idebenone increases mitochondrial complex I activity in fibroblasts from LHON patients while producing contradictory effects on respiration. <i>BMC Research Notes</i> , <b>2011</b> , 4, 557	2.3	45
125	mtDNA haplogroup J: a contributing factor of optic neuritis. <i>European Journal of Human Genetics</i> , <b>1999</b> , 7, 404-6	5.3	45
124	The mitochondrial DNA content of cumulus granulosa cells is linked to embryo quality. <i>Human Reproduction</i> , <b>2017</b> , 32, 607-614	5.7	44
123	Fourteen novel OPA1 mutations in autosomal dominant optic atrophy including two de novo mutations in sporadic optic atrophy. <i>Human Mutation</i> , <b>2003</b> , 21, 656	4.7	44
122	Analysis of Tg transcripts by real-time RT-PCR in the blood of thyroid cancer patients. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2002</b> , 87, 635-9	5.6	43
121	Recessive Mutations in RTN4IP1 Cause Isolated and Syndromic Optic Neuropathies. <i>American Journal of Human Genetics</i> , <b>2015</b> , 97, 754-60	11	42
120	OPA1-related disorders: Diversity of clinical expression, modes of inheritance and pathophysiology. <i>Neurobiology of Disease</i> , <b>2016</b> , 90, 20-6	7.5	39
119	Increase in Cardiac Ischemia-Reperfusion Injuries in Opa1 <sup>+/-</sup> Mouse Model. <i>PLoS ONE</i> , <b>2016</b> , 11, e0164066	6.7	37

118	Acute intermittent porphyria causes hepatic mitochondrial energetic failure in a mouse model. <i>International Journal of Biochemistry and Cell Biology</i> , <b>2014</b> , 51, 93-101	5.6	36
117	Relative frequencies of inherited retinal dystrophies and optic neuropathies in Southern France: assessment of 21-year data management. <i>Ophthalmic Epidemiology</i> , <b>2013</b> , 20, 13-25	1.9	36
116	Ethambutol-induced optic neuropathy linked to OPA1 mutation and mitochondrial toxicity. <i>Mitochondrion</i> , <b>2010</b> , 10, 115-24	4.9	36
115	Improved locus-specific database for OPA1 mutations allows inclusion of advanced clinical data. <i>Human Mutation</i> , <b>2015</b> , 36, 20-5	4.7	35
114	Current mechanistic insights into the CCCP-induced cell survival response. <i>Biochemical Pharmacology</i> , <b>2018</b> , 148, 100-110	6	33
113	Bioenergetic defect associated with mKATP channel opening in a mouse model carrying a mitofusin 2 mutation. <i>FASEB Journal</i> , <b>2011</b> , 25, 1618-27	0.9	33
112	Acute and late-onset optic atrophy due to a novel OPA1 mutation leading to a mitochondrial coupling defect. <i>Molecular Vision</i> , <b>2009</b> , 15, 598-608	2.3	33
111	Standardized mitochondrial analysis gives new insights into mitochondrial dynamics and OPA1 function. <i>International Journal of Biochemistry and Cell Biology</i> , <b>2012</b> , 44, 980-8	5.6	31
110	Adenine nucleotide translocase is involved in a mitochondrial coupling defect in MFN2-related Charcot-Marie-Tooth type 2A disease. <i>Neurogenetics</i> , <b>2010</b> , 11, 127-33	3	31
109	Metabolically induced heteroplasmy shifting and l-arginine treatment reduce the energetic defect in a neuronal-like model of MELAS. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , <b>2012</b> , 1822, 1019-29	6.9	30
108	Reversible optic neuropathy with OPA1 exon 5b mutation. <i>Annals of Neurology</i> , <b>2008</b> , 63, 667-71	9.4	30
107	Bioinformatics Tools and Databases to Assess the Pathogenicity of Mitochondrial DNA Variants in the Field of Next Generation Sequencing. <i>Frontiers in Genetics</i> , <b>2018</b> , 9, 632	4.5	30
106	A Nontargeted UHPLC-HRMS Metabolomics Pipeline for Metabolite Identification: Application to Cardiac Remote Ischemic Preconditioning. <i>Analytical Chemistry</i> , <b>2017</b> , 89, 2138-2146	7.8	29
105	Nicotinamide Deficiency in Primary Open-Angle Glaucoma <b>2019</b> , 60, 2509-2514		29
104	The metabolomic signature of Leber's hereditary optic neuropathy reveals endoplasmic reticulum stress. <i>Brain</i> , <b>2016</b> , 139, 2864-2876	11.2	28
103	The addition of ketone bodies alleviates mitochondrial dysfunction by restoring complex I assembly in a MELAS cellular model. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , <b>2017</b> , 1863, 284-291	6.9	28
102	Heterozygous OPA1 mutations in Behr syndrome. <i>Brain</i> , <b>2011</b> , 134, e169; author reply e170	11.2	28
101	A locus-specific database for mutations in GDAP1 allows analysis of genotype-phenotype correlations in Charcot-Marie-Tooth diseases type 4A and 2K. <i>Orphanet Journal of Rare Diseases</i> , <b>2011</b> , 6, 87	4.2	27

100	Simultaneous MFN2 and GDAP1 mutations cause major mitochondrial defects in a patient with CMT. <i>Neurology</i> , <b>2011</b> , 76, 1524-6	6.5	27
99	Assembly defects induce oxidative stress in inherited mitochondrial complex I deficiency. <i>International Journal of Biochemistry and Cell Biology</i> , <b>2015</b> , 65, 91-103	5.6	26
98	Mitochondrial energetic defects in muscle and brain of a Hmbs <sup>-/-</sup> mouse model of acute intermittent porphyria. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 5015-23	5.6	25
97	Genetically determined optic neuropathies. <i>Current Opinion in Neurology</i> , <b>2010</b> , 23, 24-8	7.1	25
96	Metabolic Signature of Remote Ischemic Preconditioning Involving a Cocktail of Amino Acids and Biogenic Amines. <i>Journal of the American Heart Association</i> , <b>2016</b> , 5,	6	25
95	Autophagy controls the pathogenicity of OPA1 mutations in dominant optic atrophy. <i>Journal of Cellular and Molecular Medicine</i> , <b>2017</b> , 21, 2284-2297	5.6	24
94	Loss of functional OPA1 unbalances redox state: implications in dominant optic atrophy pathogenesis. <i>Annals of Clinical and Translational Neurology</i> , <b>2016</b> , 3, 408-21	5.3	24
93	A Metabolomics Profiling of Glaucoma Points to Mitochondrial Dysfunction, Senescence, and Polyamines Deficiency <b>2018</b> , 59, 4355-4361		24
92	Sensorineural hearing loss in OPA1-linked disorders. <i>Brain</i> , <b>2013</b> , 136, e236	11.2	23
91	CLUH couples mitochondrial distribution to the energetic and metabolic status. <i>Journal of Cell Science</i> , <b>2017</b> , 130, 1940-1951	5.3	22
90	A randomized, double-blind, placebo-controlled trial evaluating cysteamine in Huntington's disease. <i>Movement Disorders</i> , <b>2017</b> , 32, 932-936	7	22
89	Mitochondrial DNA A3243G mutation involved in familial diabetes, chronic intestinal pseudo-obstruction and recurrent pancreatitis. <i>Diabetes and Metabolism</i> , <b>2008</b> , 34, 620-6	5.4	22
88	OPA1: 516 unique variants and 831 patients registered in an updated centralized Variome database. <i>Orphanet Journal of Rare Diseases</i> , <b>2019</b> , 14, 214	4.2	21
87	Deep sequencing shows that oocytes are not prone to accumulate mtDNA heteroplasmic mutations during ovarian ageing. <i>Human Reproduction</i> , <b>2017</b> , 32, 2101-2109	5.7	20
86	Metabolomic Profiling of Aqueous Humor in Glaucoma Points to Taurine and Spermine Deficiency: Findings from the Eye-D Study. <i>Journal of Proteome Research</i> , <b>2019</b> , 18, 1307-1315	5.6	20
85	Mitochondrial bioenergetic background confers a survival advantage to HepG2 cells in response to chemotherapy. <i>Molecular Carcinogenesis</i> , <b>2009</b> , 48, 733-41	5	20
84	eKLIPse: a sensitive tool for the detection and quantification of mitochondrial DNA deletions from next-generation sequencing data. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 1407-1416	8.1	20
83	Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. <i>JAMA Neurology</i> , <b>2018</b> , 75, 105-113	17.2	20

82	Metabo-lipidomics of Fibroblasts and Mitochondrial-Endoplasmic Reticulum Extracts from ALS Patients Shows Alterations in Purine, Pyrimidine, Energetic, and Phospholipid Metabolisms. <i>Molecular Neurobiology</i> , <b>2019</b> , 56, 5780-5791	6.2	18
81	Pro-oxidant effect of ALA is implicated in mitochondrial dysfunction of HepG2 cells. <i>Biochimie</i> , <b>2014</b> , 106, 157-66	4.6	18
80	Never too old to harbour a young man's disease?. <i>British Journal of Ophthalmology</i> , <b>2011</b> , 95, 887, 896-7	5.5	18
79	New evidence of a mitochondrial genetic background paradox: impact of the J haplogroup on the A3243G mutation. <i>BMC Medical Genetics</i> , <b>2008</b> , 9, 41	2.1	18
78	Mutations in the m-AAA proteases AFG3L2 and SPG7 are causing isolated dominant optic atrophy. <i>Neurology: Genetics</i> , <b>2020</b> , 6, e428	3.8	18
77	Mitochondria: their role in spermatozoa and in male infertility. <i>Human Reproduction Update</i> , <b>2021</b> , 27, 697-719	15.8	16
76	Primary antiphospholipid syndrome and antiphospholipid syndrome associated to systemic lupus: Are they different entities?. <i>Autoimmunity Reviews</i> , <b>2018</b> , 17, 739-745	13.6	15
75	Targeted Metabolomics Reveals Early Dominant Optic Atrophy Signature in Optic Nerves of Opa1delTTAG/+ Mice <b>2017</b> , 58, 812-820		15
74	The mitochondrial DNA content of cumulus cells may help predict embryo implantation. <i>Journal of Assisted Reproduction and Genetics</i> , <b>2019</b> , 36, 223-228	3.4	15
73	A Plasma Metabolomic Signature Involving Purine Metabolism in Human Optic Atrophy 1 (OPA1)-Related Disorders <b>2018</b> , 59, 185-195		15
72	The combination of four analytical methods to explore skeletal muscle metabolomics: Better coverage of metabolic pathways or a marketing argument?. <i>Journal of Pharmaceutical and Biomedical Analysis</i> , <b>2018</b> , 148, 273-279	3.5	14
71	A novel mutation in the mitochondrial tRNA Asn gene associated with a lethal disease. <i>Biochemical and Biophysical Research Communications</i> , <b>2005</b> , 329, 1152-4	3.4	14
70	Oxidative stress contributes differentially to the pathophysiology of Charcot-Marie-Tooth disease type 2K. <i>Experimental Neurology</i> , <b>2020</b> , 323, 113069	5.7	13
69	The Metabolomic Bioenergetic Signature of Opa1-Disrupted Mouse Embryonic Fibroblasts Highlights Aspartate Deficiency. <i>Scientific Reports</i> , <b>2018</b> , 8, 11528	4.9	12
68	Metabolomics and Lipidomics Profiling of a Combined Mitochondrial Plus Endoplasmic Reticulum Fraction of Human Fibroblasts: A Robust Tool for Clinical Studies. <i>Journal of Proteome Research</i> , <b>2018</b> , 17, 745-750	5.6	12
67	Dominant optic atrophy: Culprit mitochondria in the optic nerve. <i>Progress in Retinal and Eye Research</i> , <b>2021</b> , 83, 100935	20.5	11
66	Tryptophane-kynurenine pathway in the remote ischemic conditioning mechanism. <i>Basic Research in Cardiology</i> , <b>2020</b> , 115, 13	11.8	11
65	OPA1-related dominant optic atrophy is not strongly influenced by mitochondrial DNA background. <i>BMC Medical Genetics</i> , <b>2009</b> , 10, 70	2.1	11

64	mtDNA controls expression of the Death Associated Protein 3. <i>Experimental Cell Research</i> , <b>2006</b> , 312, 737-45	4.2	11
63	Increased steroidogenesis promotes early-onset and severe vision loss in females with OPA1 dominant optic atrophy. <i>Human Molecular Genetics</i> , <b>2016</b> , 25, 2539-2551	5.6	11
62	Embryo and Its Mitochondria. <i>Antioxidants</i> , <b>2021</b> , 10,	7.1	11
61	Cyclosporine A does not prevent second-eye involvement in Leber's hereditary optic neuropathy. <i>Orphanet Journal of Rare Diseases</i> , <b>2018</b> , 13, 33	4.2	10
60	A Plasma Metabolomic Signature of the Exfoliation Syndrome Involves Amino Acids, Acylcarnitines, and Polyamines <b>2018</b> , 59, 1025-1032		10
59	Mitochondrial dysfunction affecting visual pathways. <i>Revue Neurologique</i> , <b>2014</b> , 170, 344-54	3	10
58	Endometriosis Lowers the Cumulative Live Birth Rates in IVF by Decreasing the Number of Embryos but Not Their Quality. <i>Journal of Clinical Medicine</i> , <b>2020</b> , 9,	5.1	10
57	Dominant mutations in MIEF1 affect mitochondrial dynamics and cause a singular late onset optic neuropathy. <i>Molecular Neurodegeneration</i> , <b>2021</b> , 16, 12	19	10
56	Metabolomics hallmarks OPA1 variants correlating with their in vitro phenotype and predicting clinical severity. <i>Human Molecular Genetics</i> , <b>2020</b> , 29, 1319-1329	5.6	9
55	A Data Mining Metabolomics Exploration of Glaucoma. <i>Metabolites</i> , <b>2020</b> , 10,	5.6	9
54	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. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , <b>2018</b> , 1864, 1596-1608	6.9	9
53	Increased mitochondrial fusion in a autosomal recessive CMT2A family with mitochondrial GTPase mitofusin 2 mutations. <i>Journal of the Peripheral Nervous System</i> , <b>2016</b> , 21, 365-369	4.7	9
52	Sporadic optic atrophy due to synonymous codon change altering mRNA splicing of OPA1. <i>Clinical Genetics</i> , <b>2005</b> , 67, 102-3	4	9
51	Cataract as a phenotypic marker for a mutation in WFS1, the Wolfram syndrome gene. <i>European Journal of Ophthalmology</i> , <b>2012</b> , 22, 254-8	1.9	8
50	Maternal ageing impairs mitochondrial DNA kinetics during early embryogenesis in mice. <i>Human Reproduction</i> , <b>2019</b> , 34, 1313-1324	5.7	7
49	Study of mitochondrial function in placental insufficiency. <i>Placenta</i> , <b>2018</b> , 67, 1-7	3.4	7
48	Ocular growth and metabolomics are dependent upon the spectral content of ambient white light. <i>Scientific Reports</i> , <b>2021</b> , 11, 7586	4.9	7
47	Loss of function of in mice induces deafness and cochlear outer hair cells' degeneration. <i>Cell Death Discovery</i> , <b>2016</b> , 2, 16017	6.9	7



46	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. <i>Clinica Chimica Acta</i> , <b>2019</b> , 488, 104-110	6.2	7
45	Elevated Levels of Monocyte Chemotactic Protein-1 in the Follicular Fluid Reveals Different Populations among Women with Severe Endometriosis. <i>Journal of Clinical Medicine</i> , <b>2020</b> , 9,	5.1	6
44	Sexual Dimorphism of Metabolomic Profile in Arterial Hypertension. <i>Scientific Reports</i> , <b>2020</b> , 10, 7517	4.9	6
43	A Plasma Metabolomic Profiling of Exudative Age-Related Macular Degeneration Showing Carnosine and Mitochondrial Deficiencies. <i>Journal of Clinical Medicine</i> , <b>2020</b> , 9,	5.1	6
42	Updated review of postmortem biochemical exploration of hypothermia with a presentation of standard strategy of sampling and analyses. <i>Clinical Chemistry and Laboratory Medicine</i> , <b>2018</b> , 56, 1819-1827	5.9	6
41	High-throughput screening identifies suppressors of mitochondrial fragmentation in OPA1 fibroblasts. <i>EMBO Molecular Medicine</i> , <b>2021</b> , 13, e13579	12	6
40	Recurrent Isolated Neonatal Hemolytic Anemia: Think About Glutathione Synthetase Deficiency. <i>Pediatrics</i> , <b>2016</b> , 138,	7.4	6
39	A serum metabolomics signature of hypothermia fatalities involving arginase activity, tryptophan content, and phosphatidylcholine saturation. <i>International Journal of Legal Medicine</i> , <b>2019</b> , 133, 889-898 <sup>3.1</sup>	3.1	6
38	Metabolomics reveals highly regional specificity of cerebral sexual dimorphism in mice. <i>Progress in Neurobiology</i> , <b>2020</b> , 184, 101698	10.9	6
37	Multiethnic involvement in autosomal-dominant optic atrophy in Singapore. <i>Eye</i> , <b>2017</b> , 31, 475-480	4.4	5
36	Metabolomics shows no impairment of the microenvironment of the cumulus-oocyte complex in women with isolated endometriosis. <i>Reproductive BioMedicine Online</i> , <b>2019</b> , 39, 885-892	4	5
35	The Metabolomic Signature of Opa1 Deficiency in Rat Primary Cortical Neurons Shows Aspartate/Glutamate Depletion and Phospholipids Remodeling. <i>Scientific Reports</i> , <b>2019</b> , 9, 6107	4.9	5
34	Novel gene mutation in an atypical late-onset mitochondrial form of multifocal dystonia. <i>Neurology: Genetics</i> , <b>2017</b> , 3, e205	3.8	5
33	Dominant mutations are a frequent cause of isolated optic atrophy. <i>Brain Communications</i> , <b>2021</b> , 3, fcb063	4.63	5
32	Lipidomics Reveals Triacylglycerol Accumulation Due to Impaired Fatty Acid Flux in Opa1-Disrupted Fibroblasts. <i>Journal of Proteome Research</i> , <b>2019</b> , 18, 2779-2790	5.6	4
31	Mitochondrial complex I defect resulting from exercise-induced lower limb ischemia in patients with peripheral arterial disease. <i>Journal of Applied Physiology</i> , <b>2018</b> , 125, 938-946	3.7	4
30	Increased Protein -Glutathionylation in Leber's Hereditary Optic Neuropathy (LHON). <i>International Journal of Molecular Sciences</i> , <b>2020</b> , 21,	6.3	3
29	Reply: The expanding neurological phenotype of DNMT1L-related disorders. <i>Brain</i> , <b>2018</b> , 141, e29	11.2	3

28	Sickle Cell Disease: Metabolomic Profiles of Vaso-Occlusive Crisis in Plasma and Erythrocytes. <i>Journal of Clinical Medicine</i> , <b>2020</b> , 9,	5.1	3
27	A plasma metabolomic signature of Leber hereditary optic neuropathy showing taurine and nicotinamide deficiencies. <i>Human Molecular Genetics</i> , <b>2021</b> , 30, 21-29	5.6	3
26	5-Oxoprolinuria in hyperammonemic encephalopathy: Coincidence or worsening factor?. <i>Clinical Biochemistry</i> , <b>2017</b> , 50, 1115-1117	3.5	2
25	Warburg-like effect is a hallmark of complex I assembly defects. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , <b>2019</b> , 1865, 2475-2489	6.9	2
24	Mutations in aARS genes revealed by targeted next-generation sequencing in patients with mitochondrial diseases. <i>Molecular Biology Reports</i> , <b>2020</b> , 47, 3779-3787	2.8	2
23	Familial Dysalbuminemic Hyperthyroxinemia: An Underdiagnosed Entity. <i>Journal of Clinical Medicine</i> , <b>2020</b> , 9,	5.1	2
22	Use of Next-Generation Sequencing for the Molecular Diagnosis of 1,102 Patients With a Autosomal Optic Neuropathy. <i>Frontiers in Neurology</i> , <b>2021</b> , 12, 602979	4.1	2
21	Protective role of the mitochondrial fusion protein OPA1 in hypertension. <i>FASEB Journal</i> , <b>2021</b> , 35, e21673	6.3	2
20	Metabolomic Approach in STEMI-Patients Undergoing Left Ventricular Remodeling. <i>International Journal of Molecular Sciences</i> , <b>2019</b> , 20,	6.3	2
19	Acetoacetate protects macrophages from lactic acidosis-induced mitochondrial dysfunction by metabolic reprogramming. <i>Nature Communications</i> , <b>2021</b> , 12, 7115	17.4	2
18	Metabolomic Profiling of Plasma and Erythrocytes in Sickle Mice Points to Altered Nociceptive Pathways. <i>Cells</i> , <b>2020</b> , 9,	7.9	1
17	Mitochondrial DNA Parameters in Blood of Infants Receiving Lopinavir/Ritonavir or Lamivudine Prophylaxis to Prevent Breastfeeding Transmission of HIV-1. <i>Journal of Clinical Medicine</i> , <b>2020</b> , 9,	5.1	1
16	Secondary coenzyme Q deficiency in neurological disorders. <i>Free Radical Biology and Medicine</i> , <b>2021</b> , 165, 203-218	7.8	1
15	Preliminary Metabolomic Profiling of the Vitreous Humor from Hypothermia Fatalities. <i>Journal of Proteome Research</i> , <b>2021</b> , 20, 2390-2396	5.6	1
14	Optic neuropathy linked to ACAD9 pathogenic variants: A potentially riboflavin-responsive disorder?. <i>Mitochondrion</i> , <b>2021</b> , 59, 169-174	4.9	1
13	Tear metabolomics highlights new potential biomarkers for differentiating between Sjögren's syndrome and other causes of dry eye. <i>Ocular Surface</i> , <b>2021</b> , 22, 110-116	6.5	1
12	Altered Mitochondrial Opa1-Related Fusion in Mouse Promotes Endothelial Cell Dysfunction and Atherosclerosis. <i>Antioxidants</i> , <b>2022</b> , 11, 1078	7.1	1
11	Metabolic Profile and Pathological Alterations in the Muscle of Patients with Early-Stage Amyotrophic Lateral Sclerosis. <i>Biomedicines</i> , <b>2022</b> , 10, 1307	4.8	1

10	Letter in response to remote ischaemic conditioning provides humoral cross-species cardioprotection through glycine receptor activation. <i>Cardiovascular Research</i> , <b>2017</b> , 113, 562	9.9	o
9	Metabolomic Sexual Dimorphism of the Mouse Brain is Predominantly Abolished by Gonadectomy with a Higher Impact on Females. <i>Journal of Proteome Research</i> , <b>2021</b> , 20, 2772-2779	5.6	o
8	Improved detection of mitochondrial DNA instability in mitochondrial genome maintenance disorders. <i>Genetics in Medicine</i> , <b>2021</b> , 23, 1769-1778	8.1	o
7	Mitochondrial Dysfunction in Mitochondrial Medicine: Current Limitations, Pitfalls, and Tomorrow. <i>Methods in Molecular Biology</i> , <b>2021</b> , 2276, 1-29	1.4	o
6	ACO2 clinicobiological dataset with extensive phenotype ontology annotation. <i>Scientific Data</i> , <b>2021</b> , 8, 205	8.2	o
5	Neurological Diseases Associated with Mutations in the Mitochondrial Fusion Machinery <b>2011</b> , 169-196		
4	Clinical Heterogeneity of Hereditary Optic Atrophy in a Turkish Family. <i>Neuro-Ophthalmology</i> , <b>2005</b> , 29, 9-15	0.9	
3	Towards personalized medicine for amyotrophic lateral sclerosis. <i>Trends in Endocrinology and Metabolism</i> , <b>2021</b> , 32, 839-841	8.8	
2	Neuropathies optiques d'origine mitochondriale. <i>Pratique Neurologique - FMC</i> , <b>2016</b> , 7, 100-116		o
1	Late-onset argininosuccinic aciduria in a 72-year-old man presenting with fatal hyperammonemia. <i>JIMD Reports</i> , <b>2021</b> , 62, 44-48	1.9	