Patrick Chinnery

List of Publications by Citations

290 papers	23,481 citations	78 h-index	147 g-index
303 ext. papers	27,502 ext. citations	11 avg, IF	6.94 L-index

#	Paper	IF	Citations
290	Reanalysis and revision of the Cambridge reference sequence for human mitochondrial DNA. *Nature Genetics*, **1999**, 23, 147	36.3	2349
289	Mitochondrial diseases. *Nature Reviews Disease Primers*, **2016**, 2, 16080	51.1	585
288	The Human Phenotype Ontology in 2017. *Nucleic Acids Research*, **2017**, 45, D865-D876	20.1	507
287	Prevalence of nuclear and mitochondrial DNA mutations related to adult mitochondrial disease. *Annals of Neurology*, **2015**, 77, 753-9	9.4	483
286	Prevalence of mitochondrial DNA disease in adults. *Annals of Neurology*, **2008**, 63, 35-9	9.4	474
285	Pathogenic mitochondrial DNA mutations are common in the general population. *American Journal of Human Genetics*, **2008**, 83, 254-60	11	431
284	The dynamics of mitochondrial DNA heteroplasmy: implications for human health and disease. *Nature Reviews Genetics*, **2015**, 16, 530-42	30.1	430
283	Disturbed mitochondrial dynamics and neurodegenerative disorders. *Nature Reviews Neurology*, **2015**, 11, 11-24	15	425
282	A Unique Gene Regulatory Network Resets the Human Germline Epigenome for Development. *Cell*, **2015**, 161, 1453-67	56.2	417
281	Mitochondrial optic neuropathies - disease mechanisms and therapeutic strategies. *Progress in Retinal and Eye Research*, **2011**, 30, 81-114	20.5	392
280	Mitochondrial DNA mutations in human colonic crypt stem cells. *Journal of Clinical Investigation*, **2003**, 112, 1351-60	15.9	389
279	A reduction of mitochondrial DNA molecules during embryogenesis explains the rapid segregation of genotypes. *Nature Genetics*, **2008**, 40, 249-54	36.3	362
278	Mammalian mitochondrial genetics: heredity, heteroplasmy and disease. *Trends in Genetics*, **1997**, 13, 450-5	8.5	356
277	The epidemiology of pathogenic mitochondrial DNA mutations. *Annals of Neurology*, **2000**, 48, 188-193	9.4	352
276	Pronuclear transfer in human embryos to prevent transmission of mitochondrial DNA disease. *Nature*, **2010**, 465, 82-5	50.4	341
275	A randomized placebo-controlled trial of idebenone in Leber's hereditary optic neuropathy. *Brain*, **2011**, 134, 2677-86	11.2	334
274	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

273	The epidemiology of Leber hereditary optic neuropathy in the North East of England. *American Journal of Human Genetics*, **2003**, 72, 333-9	11	329
272	Multi-system neurological disease is common in patients with OPA1 mutations. *Brain*, **2010**, 133, 771-86	11.2	314
271	Leber hereditary optic neuropathy. *Journal of Medical Genetics*, **2002**, 39, 162-9	5.8	290
270	Characterizing mild cognitive impairment in incident Parkinson disease: the ICICLE-PD study. *Neurology*, **2014**, 82, 308-16	6.5	288
269	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
268	Universal heteroplasmy of human mitochondrial DNA. *Human Molecular Genetics*, **2013**, 22, 384-90	5.6	278
267	The epidemiology of mitochondrial disorders--past, present and future. *Biochimica Et Biophysica Acta - Bioenergetics*, **2004**, 1659, 115-20	4.6	276
266	Molecular pathology of MELAS and MERRF. The relationship between mutation load and clinical phenotypes. *Brain*, **1997**, 120 (Pt 10), 1713-21	11.2	251
265	Random intracellular drift explains the clonal expansion of mitochondrial DNA mutations with age. *American Journal of Human Genetics*, **2001**, 68, 802-6	11	251
264	Use of whole-exome sequencing to determine the genetic basis of multiple mitochondrial respiratory chain complex deficiencies. *JAMA - Journal of the American Medical Association*, **2014**, 312, 68-77	27.4	244
263	Origins and functional consequences of somatic mitochondrial DNA mutations in human cancer. *ELife*, **2014**, 3,	8.9	229
262	Selective inhibition of mutant human mitochondrial DNA replication in vitro by peptide nucleic acids. *Nature Genetics*, **1997**, 15, 212-5	36.3	223
261	Mitochondrial genetics. *British Medical Bulletin*, **2013**, 106, 135-59	5.4	219
260	Mitochondrial dysfunction in aging: Much progress but many unresolved questions. *Biochimica Et Biophysica Acta - Bioenergetics*, **2015**, 1847, 1347-53	4.6	202
259	The inheritance of mitochondrial DNA heteroplasmy: random drift, selection or both?. *Trends in Genetics*, **2000**, 16, 500-5	8.5	198
258	Efficient mitochondrial biogenesis drives incomplete penetrance in Leber hereditary optic neuropathy. *Brain*, **2014**, 137, 335-53	11.2	186
257	The pedigree rate of sequence divergence in the human mitochondrial genome: there is a difference between phylogenetic and pedigree rates. *American Journal of Human Genetics*, **2003**, 72, 659-70	11	185
256	Accumulation of mitochondrial DNA mutations in ageing, cancer, and mitochondrial disease: is there a common mechanism?. *Lancet, The*, **2002**, 360, 1323-5	40	181

255	Treatment for mitochondrial disorders. *The Cochrane Library*, **2012**, CD004426	5.2	175
254	Risk of developing a mitochondrial DNA deletion disorder. *Lancet, The*, **2004**, 364, 592-6	40	161
253	Mitochondrial aging is accelerated by anti-retroviral therapy through the clonal expansion of mtDNA mutations. *Nature Genetics*, **2011**, 43, 806-10	36.3	160
252	Peptide nucleic acid delivery to human mitochondria. *Gene Therapy*, **1999**, 6, 1919-28	4	156
251	The mitochondrial ND6 gene is a hot spot for mutations that cause Leber hereditary optic neuropathy. *Brain*, **2001**, 124, 209-18	11.2	153
250	Relaxed replication of mtDNA: A model with implications for the expression of disease. *American Journal of Human Genetics*, **1999**, 64, 1158-65	11	147
249	Whole-genome sequencing of patients with rare diseases in a national health system. *Nature*, **2020**, 583, 96-102	50.4	139
248	Mitochondrial DNA and survival after sepsis: a prospective study. *Lancet, The*, **2005**, 366, 2118-21	40	139
247	New treatments for mitochondrial disease-no time to drop our standards. *Nature Reviews Neurology*, **2013**, 9, 474-81	15	133
246	The prevalence and natural history of dominant optic atrophy due to OPA1 mutations. *Ophthalmology*, **2010**, 117, 1538-46, 1546.e1	7.3	131
245	Mutations in the SPG7 gene cause chronic progressive external ophthalmoplegia through disordered mitochondrial DNA maintenance. *Brain*, **2014**, 137, 1323-36	11.2	124
244	Epigenetics, epidemiology and mitochondrial DNA diseases. *International Journal of Epidemiology*, **2012**, 41, 177-87	7.8	124
243	Can mitochondrial DNA mutations cause sperm dysfunction?. *Molecular Human Reproduction*, **2002**, 8, 719-21	4.4	121
242	Random genetic drift determines the level of mutant mtDNA in human primary oocytes. *American Journal of Human Genetics*, **2001**, 68, 533-6	11	121
241	Mitochondria. *Journal of Neurology, Neurosurgery and Psychiatry*, **2003**, 74, 1188-99	5.5	120
240	Patient care standards for primary mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. *Genetics in Medicine*, **2017**, 19,	8.1	113
239	Segregation of mitochondrial DNA heteroplasmy through a developmental genetic bottleneck in human embryos. *Nature Cell Biology*, **2018**, 20, 144-151	23.4	110
238	Leber hereditary optic neuropathy: Does heteroplasmy influence the inheritance and expression of the G11778A mitochondrial DNA mutation?. *American Journal of Medical Genetics Part A*, **2001**, 98, 235-43		110

237	Genetic impact on cognition and brain function in newly diagnosed Parkinson's disease: ICICLE-PD study. *Brain*, **2014**, 137, 2743-58	11.2	109
236	The spectrum of hearing loss due to mitochondrial DNA defects. *Brain*, **2000**, 123 (Pt 1), 82-92	11.2	108
235	Allogeneic haematopoietic stem cell transplantation for mitochondrial neurogastrointestinal encephalomyopathy. *Brain*, **2015**, 138, 2847-58	11.2	107
234	Mitochondrial disease in adults: a scale to monitor progression and treatment. *Neurology*, **2006**, 66, 1932-4	6.5	106
233	Germline selection shapes human mitochondrial DNA diversity. *Science*, **2019**, 364,	33.3	105
232	Recent mitochondrial DNA mutations increase the risk of developing common late-onset human diseases. *PLoS Genetics*, **2014**, 10, e1004369	6	102
231	Variation in germline mtDNA heteroplasmy is determined prenatally but modified during subsequent transmission. *Nature Genetics*, **2012**, 44, 1282-5	36.3	102
230	Analysis of European mtDNAs for recombination. *American Journal of Human Genetics*, **2001**, 68, 145-153	11	102
229	Treatment strategies for inherited optic neuropathies: past, present and future. *Eye*, **2014**, 28, 521-37	4.4	98
228	Titin mutation segregates with hereditary myopathy with early respiratory failure. *Brain*, **2012**, 135, 1695-713	11.1	98
227	Exome sequencing in undiagnosed inherited and sporadic ataxias. *Brain*, **2015**, 138, 276-83	11.2	97
226	Riboflavin-Responsive and -Non-responsive Mutations in FAD Synthase Cause Multiple Acyl-CoA Dehydrogenase and Combined Respiratory-Chain Deficiency. *American Journal of Human Genetics*, **2016**, 98, 1130-1145	11	97
225	Mitochondrial DNA and disease. *Lancet, The*, **1999**, 354 Suppl 1, SI17-21	40	97
224	Molecular basis of infantile reversible cytochrome c oxidase deficiency myopathy. *Brain*, **2009**, 132, 3165-74	11.2	96
223	Mutations in GTPBP3 cause a mitochondrial translation defect associated with hypertrophic cardiomyopathy, lactic acidosis, and encephalopathy. *American Journal of Human Genetics*, **2014**, 95, 708-20	11	95
222	Two-stage association study and meta-analysis of mitochondrial DNA variants in Parkinson disease. *Neurology*, **2013**, 80, 2042-8	6.5	94
221	The epidemiology of pathogenic mitochondrial DNA mutations. *Annals of Neurology*, **2000**, 48, 188-93	9.4	91
220	Mitochondrial DNA haplogroups and susceptibility to AD and dementia with Lewy bodies. *Neurology*, **2000**, 55, 302-4	6.5	90

219	Defective i6A37 modification of mitochondrial and cytosolic tRNAs results from pathogenic mutations in TRIT1 and its substrate tRNA. *PLoS Genetics*, **2014**, 10, e1004424	6	89
218	Mitochondrial disease in adults: what's old and what's new?. *EMBO Molecular Medicine*, **2015**, 7, 1503-12	12	88
217	Mitochondrial DNA mutations in neurodegeneration. *Biochimica Et Biophysica Acta - Bioenergetics*, **2015**, 1847, 1401-11	4.6	84
216	Nonrandom tissue distribution of mutant mtDNA. *American Journal of Medical Genetics Part A*, **1999**, 85, 498-501		84
215	OPA1 mutations cause cytochrome c oxidase deficiency due to loss of wild-type mtDNA molecules. *Human Molecular Genetics*, **2010**, 19, 3043-52	5.6	83
214	Point mutations of the mtDNA control region in normal and neurodegenerative human brains. *American Journal of Human Genetics*, **2001**, 68, 529-32	11	82
213	EXOSC8 mutations alter mRNA metabolism and cause hypomyelination with spinal muscular atrophy and cerebellar hypoplasia. *Nature Communications*, **2014**, 5, 4287	17.4	80
212	The power to detect disease associations with mitochondrial DNA haplogroups. *American Journal of Human Genetics*, **2006**, 78, 713-20	11	78
211	Synaptotagmin 2 mutations cause an autosomal-dominant form of lambert-eaton myasthenic syndrome and nonprogressive motor neuropathy. *American Journal of Human Genetics*, **2014**, 95, 332-9	11	77
210	Selection against pathogenic mtDNA mutations in a stem cell population leads to the loss of the 3243A-->G mutation in blood. *American Journal of Human Genetics*, **2008**, 82, 333-43	11	76
209	HIV treatment and associated mitochondrial pathology: review of 25 years of in vitro, animal, and human studies. *Toxicologic Pathology*, **2014**, 42, 811-22	2.1	75
208	Mitochondrial DNA haplogroups and risk of transient ischaemic attack and ischaemic stroke: a genetic association study. *Lancet Neurology, The*, **2010**, 9, 498-503	24.1	74
207	Clinical features of MS associated with Leber hereditary optic neuropathy mtDNA mutations. *Neurology*, **2013**, 81, 2073-81	6.5	73
206	SPG7 mutations are a common cause of undiagnosed ataxia. *Neurology*, **2015**, 84, 1174-6	6.5	72
205	Quality of life in patients with leber hereditary optic neuropathy **2009**, 50, 3112-5		71
204	Genotypes from patients indicate no paternal mitochondrial DNA contribution. *Annals of Neurology*, **2003**, 54, 521-4	9.4	69
203	Amyloid-□accumulation in the CNS in human growth hormone recipients in the UK. *Acta Neuropathologica*, **2017**, 134, 221-240	14.3	67
202	Reduced cerebrospinal fluid mitochondrial DNA is a biomarker for early-stage Parkinson's disease. *Annals of Neurology*, **2015**, 78, 1000-4	9.4	67

201	PGD and heteroplasmic mitochondrial DNA point mutations: a systematic review estimating the chance of healthy offspring. *Human Reproduction Update*, **2012**, 18, 341-9	15.8	64
200	Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency. *Brain*, **2011**, 134, 183-95	11.2	64
199	Homozygous deletion in MICU1 presenting with fatigue and lethargy in childhood. *Neurology: Genetics*, **2016**, 2, e59	3.8	64
198	TRMT5 Mutations Cause a Defect in Post-transcriptional Modification of Mitochondrial tRNA Associated with Multiple Respiratory-Chain Deficiencies. *American Journal of Human Genetics*, **2015**, 97, 319-28	11	62
197	Molecular pathogenesis of polymerase Ɣ-related neurodegeneration. *Annals of Neurology*, **2014**, 76, 66-81	9.4	61
196	Adults with RRM2B-related mitochondrial disease have distinct clinical and molecular characteristics. *Brain*, **2012**, 135, 3392-403	11.2	59
195	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. *Brain*, **2017**, 140, 1561-1578	11.2	58
194	Recessive Mutations in TRMT10C Cause Defects in Mitochondrial RNA Processing and Multiple Respiratory Chain Deficiencies. *American Journal of Human Genetics*, **2016**, 98, 993-1000	11	58
193	Titin founder mutation is a common cause of myofibrillar myopathy with early respiratory failure. *Journal of Neurology, Neurosurgery and Psychiatry*, **2014**, 85, 331-8	5.5	58
192	The distribution of mitochondrial DNA heteroplasmy due to random genetic drift. *American Journal of Human Genetics*, **2008**, 83, 582-93	11	58
191	Retrospective natural history of thymidine kinase 2 deficiency. *Journal of Medical Genetics*, **2018**, 55, 515-521	5.8	56
190	Mitochondrial Disease Sequence Data Resource (MSeqDR): a global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities. *Molecular Genetics and Metabolism*, **2015**, 114, 388-96	3.7	56
189	155th ENMC workshop: polymerase gamma and disorders of mitochondrial DNA synthesis, 21-23 September 2007, Naarden, The Netherlands. *Neuromuscular Disorders*, **2008**, 18, 259-67	2.9	56
188	Mitochondrial DNA point mutations and relative copy number in 1363 disease and control human brains. *Acta Neuropathologica Communications*, **2017**, 5, 13	7.3	55
187	Genetic heterogeneity of motor neuropathies. *Neurology*, **2017**, 88, 1226-1234	6.5	54
186	Neuroferritinopathy in a French family with late onset dominant dystonia. *Journal of Medical Genetics*, **2003**, 40, e69	5.8	54
185	TIA1 variant drives myodegeneration in multisystem proteinopathy with SQSTM1 mutations. *Journal of Clinical Investigation*, **2018**, 128, 1164-1177	15.9	53
184	Fall in circulating mononuclear cell mitochondrial DNA content in human sepsis. *Intensive Care Medicine*, **2010**, 36, 956-62	14.5	51

183	Depletion of mitochondrial DNA in leucocytes harbouring the 3243A->G mtDNA mutation. *Journal of Medical Genetics*, **2007**, 44, 69-74	5.8	51
182	Extreme-Depth Re-sequencing of Mitochondrial DNA Finds No Evidence of Paternal Transmission in Humans. *PLoS Genetics*, **2015**, 11, e1005040	6	50
181	The mitochondrial DNA genetic bottleneck: inheritance and beyond. *Essays in Biochemistry*, **2018**, 62, 225-234	7.6	50
180	The challenges of mitochondrial replacement. *PLoS Genetics*, **2014**, 10, e1004315	6	49
179	Mitochondrial medicine. *QJM - Monthly Journal of the Association of Physicians*, **1997**, 90, 657-67	2.7	49
178	Treatment of mitochondrial disease. *Journal of Bioenergetics and Biomembranes*, **1997**, 29, 195-205	3.7	47
177	Dominant optic atrophy: novel OPA1 mutations and revised prevalence estimates. *Ophthalmology*, **2013**, 120, 1712-1712.e1	7.3	46
176	Accurate mitochondrial DNA sequencing using off-target reads provides a single test to identify pathogenic point mutations. *Genetics in Medicine*, **2014**, 16, 962-71	8.1	46
175	Clinical, Genetic, and Radiological Features of Extrapyramidal Movement Disorders in Mitochondrial Disease. *JAMA Neurology*, **2016**, 73, 668-74	17.2	46
174	A novel mitochondrial tRNA isoleucine gene mutation causing chronic progressive external ophthalmoplegia. *Neurology*, **1997**, 49, 1166-8	6.5	45
173	Mitochondrial DNA haplogroups and type 2 diabetes: a study of 897 cases and 1010 controls. *Journal of Medical Genetics*, **2007**, 44, e80	5.8	45
172	Biallelic C1QBP Mutations Cause Severe Neonatal-, Childhood-, or Later-Onset Cardiomyopathy Associated with Combined Respiratory-Chain Deficiencies. *American Journal of Human Genetics*, **2017**, 101, 525-538	11	44
171	Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. *Human Molecular Genetics*, **2016**, 25, 1031-41	5.6	44
170	Altered 2-thiouridylation impairs mitochondrial translation in reversible infantile respiratory chain deficiency. *Human Molecular Genetics*, **2013**, 22, 4602-15	5.6	44
169	A novel mitochondrial tRNA phenylalanine mutation presenting with acute rhabdomyolysis. *Annals of Neurology*, **1997**, 41, 408-10	9.4	44
168	Extreme heterogeneity of human mitochondrial DNA from organelles to populations. *Nature Reviews Genetics*, **2021**, 22, 106-118	30.1	44
167	Somatic mtDNA variation is an important component of Parkinson's disease. *Neurobiology of Aging*, **2016**, 38, 217.e1-217.e6	5.6	43
166	Population screening for variant Creutzfeldt-Jakob disease using a novel blood test: diagnostic accuracy and feasibility study. *JAMA Neurology*, **2014**, 71, 421-8	17.2	43

165	Normokalemic periodic paralysis revisited: does it exist?. *Annals of Neurology*, **2002**, 52, 251-2	9.4	43
164	Recessive Mutations in RTN4IP1 Cause Isolated and Syndromic Optic Neuropathies. *American Journal of Human Genetics*, **2015**, 97, 754-60	11	42
163	Emerging therapies for mitochondrial disorders. *Brain*, **2016**, 139, 1633-48	11.2	42
162	Clinical, pathological and functional characterization of riboflavin-responsive neuropathy. *Brain*, **2017**, 140, 2820-2837	11.2	40
161	A novel mechanism causing imbalance of mitochondrial fusion and fission in human myopathies. *Human Molecular Genetics*, **2018**, 27, 1186-1195	5.6	40
160	Clinical mitochondrial genetics. *Journal of Medical Genetics*, **1999**, 36, 425-36	5.8	40
159	Assessing mitochondrial heteroplasmy using next generation sequencing: A note of caution. *Mitochondrion*, **2019**, 46, 302-306	4.9	39
158	In vitro supplementation with dAMP/dGMP leads to partial restoration of mtDNA levels in mitochondrial depletion syndromes. *Human Molecular Genetics*, **2009**, 18, 1590-9	5.6	39
157	Monitoring clinical progression with mitochondrial disease biomarkers. *Brain*, **2017**, 140, 2530-2540	11.2	38
156	Mitochondrial DNA Heteroplasmy and Purifying Selection in the Mammalian Female Germ Line. *Development Growth and Differentiation*, **2018**, 60, 21-32	3	37
155	Genetic compendium of 1511 human brains available through the UK Medical Research Council Brain Banks Network Resource. *Genome Research*, **2017**, 27, 165-173	9.7	36
154	Mutations in mitochondrial DNA causing tubulointerstitial kidney disease. *PLoS Genetics*, **2017**, 13, e1006620	6	35
153	Subtle neurological and metabolic abnormalities in an Opa1 mouse model of autosomal dominant optic atrophy. *Experimental Neurology*, **2009**, 220, 404-9	5.7	35
152	Childhood-onset Leber hereditary optic neuropathy. *British Journal of Ophthalmology*, **2017**, 101, 1505-1509	3.9	34
151	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care - Preliminary Report. *New England Journal of Medicine*, **2021**, 385, 1868-1880	59.2	34
150	MSeqDR: A Centralized Knowledge Repository and Bioinformatics Web Resource to Facilitate Genomic Investigations in Mitochondrial Disease. *Human Mutation*, **2016**, 37, 540-548	4.7	34
149	MT-ND5 Mutation Exhibits Highly Variable Neurological Manifestations at Low Mutant Load. *EBioMedicine*, **2018**, 30, 86-93	8.8	33
148	Valproic acid triggers increased mitochondrial biogenesis in POLG-deficient fibroblasts. *Molecular Genetics and Metabolism*, **2014**, 112, 57-63	3.7	33

147	Multisystem fatal infantile disease caused by a novel homozygous EARS2 mutation. *Brain*, **2013**, 136, e228	11.2	33
146	Clonal expansion of secondary mitochondrial DNA deletions associated with spinocerebellar ataxia type 28. *JAMA Neurology*, **2015**, 72, 106-11	17.2	33
145	Age-related mitochondrial DNA depletion and the impact on pancreatic Beta cell function. *PLoS ONE*, **2014**, 9, e115433	3.7	33
144	High prevalence of focal and multi-focal somatic genetic variants in the human brain. *Nature Communications*, **2018**, 9, 4257	17.4	33
143	Clinical pattern, mutations and in vitro residual activity in 33 patients with severe 5, 10 methylenetetrahydrofolate reductase (MTHFR) deficiency. *Journal of Inherited Metabolic Disease*, **2016**, 39, 115-24	5.4	32
142	Nuclear-mitochondrial DNA segments resemble paternally inherited mitochondrial DNA in humans. *Nature Communications*, **2020**, 11, 1740	17.4	32
141	Somatic variants in autosomal dominant genes are a rare cause of sporadic Alzheimer's disease. *Alzheimer's and Dementia*, **2018**, 14, 1632-1639	1.2	32
140	Infantile hereditary spastic paraparesis due to codominant mutations in the spastin gene. *Neurology*, **2004**, 63, 710-2	6.5	32
139	Evaluation of bupivacaine-induced muscle regeneration in the treatment of ptosis in patients with chronic progressive external ophthalmoplegia and Kearns-Sayre syndrome. *Eye*, **1999**, 13 (Pt 6), 769-72	4.4	32
138	Mitochondrial pathology in progressive cerebellar ataxia. *Cerebellum and Ataxias*, **2015**, 2, 16	1.7	31
137	Adult-onset spinocerebellar ataxia syndromes due to MTATP6 mutations. *Journal of Neurology, Neurosurgery and Psychiatry*, **2012**, 83, 883-6	5.5	31
136	mtDNA Population Variants and Neurodegenerative Diseases. *Frontiers in Neuroscience*, **2018**, 12, 682	5.1	31
135	Exome sequencing in dementia with Lewy bodies. *Translational Psychiatry*, **2016**, 6, e728	8.6	30
134	Mitochondrial Diseases: A Diagnostic Revolution. *Trends in Genetics*, **2020**, 36, 702-717	8.5	30
133	SCP2 mutations and neurodegeneration with brain iron accumulation. *Neurology*, **2015**, 85, 1909-11	6.5	29
132	Previous estimates of mitochondrial DNA mutation level variance did not account for sampling error: comparing the mtDNA genetic bottleneck in mice and humans. *American Journal of Human Genetics*, **2010**, 86, 540-50	11	29
131	116th ENMC international workshop: the treatment of mitochondrial disorders, 14th-16th March 2003, Naarden, The Netherlands. *Neuromuscular Disorders*, **2003**, 13, 757-64	2.9	29
130	Mitochondria in neuroinflammation - Multiple sclerosis (MS), leber hereditary optic neuropathy (LHON) and LHON-MS. *Neuroscience Letters*, **2019**, 710, 132932	3.3	29

129	The Contribution of the Cerebellum to Cognition in Spinocerebellar Ataxia Type 6. *Behavioural Neurology*, **2010**, 23, 3-15	3	28
128	Diagnosis of ‘possible’ mitochondrial disease: an existential crisis. *Journal of Medical Genetics*, **2019**, 56, 123-130	5.8	27
127	The mitochondrial protein CHCHD2 primes the differentiation potential of human induced pluripotent stem cells to neuroectodermal lineages. *Journal of Cell Biology*, **2016**, 215, 187-202	7.3	27
126	POLG mutations cause decreased mitochondrial DNA repopulation rates following induced depletion in human fibroblasts. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2011**, 1812, 321-5	6.9	27
125	First-line genomic diagnosis of mitochondrial disorders. *Nature Reviews Genetics*, **2018**, 19, 399-400	30.1	27
124	Nonrandom tissue distribution of mutant mtDNA. *American Journal of Medical Genetics Part A*, **1999**, 85, 498-501		27
123	Cardiomyopathy is common in patients with the mitochondrial DNA m.3243A>G mutation and correlates with mutation load. *Neuromuscular Disorders*, **2012**, 22, 592-6	2.9	26
122	Mitochondrial heteroplasmy beyond the oocyte bottleneck. *Seminars in Cell and Developmental Biology*, **2020**, 97, 156-166	7.5	26
121	Mitochondria and Hypoxia: Metabolic Crosstalk in Cell-Fate Decisions. *Trends in Endocrinology and Metabolism*, **2018**, 29, 249-259	8.8	25
120	Hereditary mtDNA heteroplasmy: a baseline for aging?. *Cell Metabolism*, **2013**, 18, 463-4	24.6	25
119	No evidence of an association between mitochondrial DNA variants and osteoarthritis in 7393 cases and 5122 controls. *Annals of the Rheumatic Diseases*, **2013**, 72, 136-9	2.4	25
118	Background sequence characteristics influence the occurrence and severity of disease-causing mtDNA mutations. *PLoS Genetics*, **2017**, 13, e1007126	6	24
117	Metabolic effects of bezafibrate in mitochondrial disease. *EMBO Molecular Medicine*, **2020**, 12, e11589	12	23
116	Two recurrent mutations are associated with GNE myopathy in the North of Britain. *Journal of Neurology, Neurosurgery and Psychiatry*, **2014**, 85, 1359-65	5.5	23
115	Mitochondrial DNA analysis: polymorphisms and pathogenicity. *Journal of Medical Genetics*, **1999**, 36, 505-10	5.8	23
114	Exposure of Monocytic Cells to Lipopolysaccharide Induces Coordinated Endotoxin Tolerance, Mitochondrial Biogenesis, Mitophagy, and Antioxidant Defenses. *Frontiers in Immunology*, **2018**, 9, 2217	8.4	23
113	Phenotypic variability of TRPV4 related neuropathies. *Neuromuscular Disorders*, **2015**, 25, 516-21	2.9	22
112	Clinical and functional characterisation of the combined respiratory chain defect in two sisters due to autosomal recessive mutations in MTFMT. *Mitochondrion*, **2013**, 13, 743-8	4.9	22

111	No correlation between muscle A3243G mutation load and mitochondrial function in vivo. *Neurology*, **2001**, 56, 1101-4	6.5	22
110	Lethal Neonatal LTBL Associated with Biallelic EARS2 Variants: Case Report and Review of the Reported Neuroradiological Features. *JIMD Reports*, **2017**, 33, 61-68	1.9	21
109	A multiple sclerosis-like disorder in patients with OPA1 mutations. *Annals of Clinical and Translational Neurology*, **2016**, 3, 723-9	5.3	21
108	Abnormal cardiac energetics in patients carrying the A3243G mtDNA mutation measured in vivo using phosphorus MR spectroscopy. *Biochimica Et Biophysica Acta - Bioenergetics*, **2004**, 1657, 146-50	4.6	21
107	Mutation-specific effects in germline transmission of pathogenic mtDNA variants. *Human Reproduction*, **2018**, 33, 1331-1341	5.7	21
106	Concentric hypertrophic remodelling and subendocardial dysfunction in mitochondrial DNA point mutation carriers. *European Heart Journal Cardiovascular Imaging*, **2013**, 14, 650-8	4.1	20
105	Mitochondrial enteropathy: the primary pathology may not be within the gastrointestinal tract. *Gut* , **2001**, 48, 121-4	19.2	20
104	A multi-center comparison of diagnostic methods for the biochemical evaluation of suspected mitochondrial disorders. *Mitochondrion*, **2013**, 13, 36-43	4.9	19
103	The pattern of retinal ganglion cell dysfunction in Leber hereditary optic neuropathy. *Mitochondrion*, **2017**, 36, 138-149	4.9	19
102	Respiratory chain deficiency in nonmitochondrial disease. *Neurology: Genetics*, **2015**, 1, e6	3.8	19
101	Prevalence of neurogenetic disorders in the North of England. *Neurology*, **2015**, 85, 1195-201	6.5	18
100	Mitochondrial oxodicarboxylate carrier deficiency is associated with mitochondrial DNA depletion and spinal muscular atrophy-like disease. *Genetics in Medicine*, **2018**, 20, 1224-1235	8.1	18
99	Behr Syndrome is Typically Associated with Disturbed Mitochondrial Translation and Mutations in the Gene. *Journal of Neuromuscular Diseases*, **2014**, 1, 55-63	5	18
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