Patrick Chinnery

List of Publications by Year in Descending Order

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	Development and evaluation of rapid data-enabled access to routine clinical information to enhance early recruitment to the national clinical platform trial of COVID-19 community treatments.. *Trials*, **2022**, 23, 62	2.8	0
289	Whole genome sequencing for the diagnosis of neurological repeat expansion disorders in the UK: a retrospective diagnostic accuracy and prospective clinical validation study.. *Lancet Neurology, The*, **2022**, 21, 234-245	24.1	6
288	Heteroplasmic mitochondrial DNA variants in cardiovascular diseases.. *PLoS Genetics*, **2022**, 18, e1010068	8	2
287	Heteroplasmic mitochondrial DNA mutations in frontotemporal lobar degeneration.. *Acta Neuropathologica*, **2022**, 143, 687-695	14.3	1
286	Shortening the diagnostic odyssey-the impact of whole genome sequencing in the NHS. *BMJ, The*, **2021**, 375, n2683	5.9	2
285	Use of whole genome sequencing to determine genetic basis of suspected mitochondrial disorders: cohort study. *BMJ, The*, **2021**, 375, e066288	5.9	5
284	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
283	Single-molecule mitochondrial DNA sequencing shows no evidence of CpG methylation in human cells and tissues. *Nucleic Acids Research*, **2021**,	20.1	4
282	High diagnostic rate of trio exome sequencing in consanguineous families with neurogenetic diseases. *Brain*, **2021**,	11.2	1
281	Isolated homozygous R217X mutation causes knock-out of functional C-terminal optineurin domains and associated oligodendrogliopathy-dominant ALS-TDP. *Journal of Neurology, Neurosurgery and Psychiatry*, **2021**, 92, 1022-1024	5.5	0
280	[11C]PK11195-PET Brain Imaging of the Mitochondrial Translocator Protein in Mitochondrial Disease. *Neurology*, **2021**,	6.5	3
279	Oxygen tension modulates the mitochondrial genetic bottleneck and influences the segregation of a heteroplasmic mtDNA variant in vitro. *Communications Biology*, **2021**, 4, 584	6.7	2
278	An atlas of mitochondrial DNA genotype-phenotype associations in the UK Biobank. *Nature Genetics*, **2021**, 53, 982-993	36.3	12
277	Biparental inheritance of mitochondrial DNA revisited. *Nature Reviews Genetics*, **2021**, 22, 477-478	30.1	3
276	Extreme heterogeneity of human mitochondrial DNA from organelles to populations. *Nature Reviews Genetics*, **2021**, 22, 106-118	30.1	44
275	MitoPhen database: a human phenotype ontology-based approach to identify mitochondrial DNA diseases. *Nucleic Acids Research*, **2021**, 49, 9686-9695	20.1	5
274	Mitochondrial DNA variants modulate N-formylmethionine, proteostasis and risk of late-onset human diseases. *Nature Medicine*, **2021**, 27, 1564-1575	50.5	4

273	Cell reprogramming shapes the mitochondrial DNA landscape. *Nature Communications*, **2021**, 12, 5241	17.4	3
272	Mitochondrial DNA heteroplasmy is modulated during oocyte development propagating mutation transmission. *Science Advances*, **2021**, 7, eabi5657	14.3	0
271	Mitochondrial Replacement in the Clinic. *New England Journal of Medicine*, **2020**, 382, 1855-1857	59.2	8
270	Epigenetic regulation in the pathophysiology of Lewy body dementia. *Progress in Neurobiology*, **2020**, 192, 101822	10.9	4
269	Whole-genome sequencing of patients with rare diseases in a national health system. *Nature*, **2020**, 583, 96-102	50.4	139
268	Recurrent horizontal transfer identifies mitochondrial positive selection in a transmissible cancer. *Nature Communications*, **2020**, 11, 3059	17.4	5
267	Inheritance of mitochondrial DNA in humans: implications for rare and common diseases. *Journal of Internal Medicine*, **2020**, 287, 634-644	10.8	16
266	Nuclear-mitochondrial DNA segments resemble paternally inherited mitochondrial DNA in humans. *Nature Communications*, **2020**, 11, 1740	17.4	32
265	Chronic pain is common in mitochondrial disease. *Neuromuscular Disorders*, **2020**, 30, 413-419	2.9	8
264	Cracking the enigma of mitochondrial-DNA variants and cancer. *Nature Metabolism*, **2020**, 2, 221-222	14.6	
263	Metabolic effects of bezafibrate in mitochondrial disease. *EMBO Molecular Medicine*, **2020**, 12, e11589	12	23
262	Metabolic shift underlies recovery in reversible infantile respiratory chain deficiency. *EMBO Journal*, **2020**, 39, e105364	13	15
261	Mitochondrial disorders due to mutations in the nuclear genome **2020**, 415-425		
260	Identification of a novel heterozygous guanosine monophosphate reductase (GMPR) variant in a patient with a late-onset disorder of mitochondrial DNA maintenance. *Clinical Genetics*, **2020**, 97, 276-286	4	4
259	Effects of thyroid hormone on mitochondria and metabolism of human preimplantation embryos. *Stem Cells*, **2020**, 38, 369-381	5.8	11
258	Visualizing, quantifying, and manipulating mitochondrial DNA in vivo. *Journal of Biological Chemistry*, **2020**, 295, 17588-17601	5.4	10
257	Mitochondrial Diseases: A Diagnostic Revolution. *Trends in Genetics*, **2020**, 36, 702-717	8.5	30
256	Behr syndrome and hypertrophic cardiomyopathy in a family with a novel UCHL1 deletion. *Journal of Neurology*, **2020**, 267, 3643-3649	5.5	3

255	Heredity and segregation of mtDNA **2020**, 87-107		
254	A guide to writing systematic reviews of rare disease treatments to generate FAIR-compliant datasets: building a Treatabolome. *Orphanet Journal of Rare Diseases*, **2020**, 15, 206	4.2	8
253	Identification of UBAP1 mutations in juvenile hereditary spastic paraplegia in the 100,000 Genomes Project. *European Journal of Human Genetics*, **2020**, 28, 1763-1768	5.3	5
252	Mitochondrial heteroplasmy beyond the oocyte bottleneck. *Seminars in Cell and Developmental Biology*, **2020**, 97, 156-166	7.5	26
251	Molecular genetic investigations identify new clinical phenotypes associated with BCS1L-related mitochondrial disease. *Human Molecular Genetics*, **2019**, 28, 3766-3776	5.6	14
250	Diagnosis of QpossibleQmitochondrial disease: an existential crisis. *Journal of Medical Genetics*, **2019**, 56, 123-130	5.8	27
249	Germline selection shapes human mitochondrial DNA diversity. *Science*, **2019**, 364,	33.3	105
248	Diagnostic Approach to Mitochondrial Diseases **2019**, 281-287		
247	A genome-wide association study of mitochondrial DNA copy number in two population-based cohorts. *Human Genomics*, **2019**, 13, 6	6.8	15
246	Assessing mitochondrial heteroplasmy using next generation sequencing: A note of caution. *Mitochondrion*, **2019**, 46, 302-306	4.9	39
245	De-fusing mitochondria defuses the mtDNA time-bomb. *Cell Research*, **2019**, 29, 781-782	24.7	3
244	Mitochondrially-targeted APOBEC1 is a potent mtDNA mutator affecting mitochondrial function and organismal fitness in Drosophila. *Nature Communications*, **2019**, 10, 3280	17.4	10
243	Mitochondrial Haplogroup and the Risk of Acute Kidney Injury Following Cardiac Bypass Surgery. *Scientific Reports*, **2019**, 9, 2279	4.9	2
242	Frequency and signature of somatic variants in 1461 human brain exomes. *Genetics in Medicine*, **2019**, 21, 904-912	8.1	14
241	Mitochondria in neuroinflammation - Multiple sclerosis (MS), leber hereditary optic neuropathy (LHON) and LHON-MS. *Neuroscience Letters*, **2019**, 710, 132932	3.3	29
240	MT-ND5 Mutation Exhibits Highly Variable Neurological Manifestations at Low Mutant Load. *EBioMedicine*, **2018**, 30, 86-93	8.8	33
239	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
238	Heterozygous SSBP1 start loss mutation co-segregates with hearing loss and the m.1555A>G mtDNA variant in a large multigenerational family. *Brain*, **2018**, 141, 55-62	11.2	15

237	Mitochondria and Hypoxia: Metabolic Crosstalk in Cell-Fate Decisions. *Trends in Endocrinology and Metabolism*, **2018**, 29, 249-259	8.8	25
236	A novel mechanism causing imbalance of mitochondrial fusion and fission in human myopathies. *Human Molecular Genetics*, **2018**, 27, 1186-1195	5.6	40
235	Mitochondrial DNA Heteroplasmy and Purifying Selection in the Mammalian Female Germ Line. *Development Growth and Differentiation*, **2018**, 60, 21-32	3	37
234	Segregation of mitochondrial DNA heteroplasmy through a developmental genetic bottleneck in human embryos. *Nature Cell Biology*, **2018**, 20, 144-151	23.4	110
233	Multifocal demyelinating motor neuropathy and hamartoma syndrome associated with a de novo mutation. *Neurology*, **2018**, 90, e1842-e1848	6.5	3
232	Retrospective natural history of thymidine kinase 2 deficiency. *Journal of Medical Genetics*, **2018**, 55, 515-521	5.8	56
231	The mitochondrial DNA genetic bottleneck: inheritance and beyond. *Essays in Biochemistry*, **2018**, 62, 225-234	7.6	50
230	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
229	Oxygen in mitochondrial disease: can there be too much of a good thing?. *Journal of Inherited Metabolic Disease*, **2018**, 41, 761-763	5.4	8
228	TIA1 variant drives myodegeneration in multisystem proteinopathy with SQSTM1 mutations. *Journal of Clinical Investigation*, **2018**, 128, 1164-1177	15.9	53
227	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
226	High prevalence of focal and multi-focal somatic genetic variants in the human brain. *Nature Communications*, **2018**, 9, 4257	17.4	33
225	mtDNA Population Variants and Neurodegenerative Diseases. *Frontiers in Neuroscience*, **2018**, 12, 682	5.1	31
224	First-line genomic diagnosis of mitochondrial disorders. *Nature Reviews Genetics*, **2018**, 19, 399-400	30.1	27
223	Mutation-specific effects in germline transmission of pathogenic mtDNA variants. *Human Reproduction*, **2018**, 33, 1331-1341	5.7	21
222	The Human Phenotype Ontology in 2017. *Nucleic Acids Research*, **2017**, 45, D865-D876	20.1	507
221	Mitochondrial DNA point mutations and relative copy number in 1363 disease and control human brains. *Acta Neuropathologica Communications*, **2017**, 5, 13	7.3	55
220	Genetic heterogeneity of motor neuropathies. *Neurology*, **2017**, 88, 1226-1234	6.5	54

219	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. *Brain*, **2017**, 140, 1561-1578	11.2	58
218	Mitochondrial DNA depletion induces innate immune dysfunction rescued by IFN-□ *Journal of Allergy and Clinical Immunology*, **2017**, 140, 1461-1464.e8	11.5	4
217	Amyloid-□ accumulation in the CNS in human growth hormone recipients in the UK. *Acta Neuropathologica*, **2017**, 134, 221-240	14.3	67
216	Childhood-onset Leber hereditary optic neuropathy. *British Journal of Ophthalmology*, **2017**, 101, 1505-1509	3.9	34
215	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
214	De novo variant is associated with decreased mitochondrial respiratory chain activities. *Neurology: Genetics*, **2017**, 3, e187	3.8	6
213	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
212	mutations and central demyelination: Evidence from electrophysiologic phenotyping in female manifesting carriers. *Neurology: Clinical Practice*, **2017**, 7, 451-454	1.7	
211	Monitoring clinical progression with mitochondrial disease biomarkers. *Brain*, **2017**, 140, 2530-2540	11.2	38
210	Clinical, pathological and functional characterization of riboflavin-responsive neuropathy. *Brain*, **2017**, 140, 2820-2837	11.2	40
209	Mutations in mitochondrial DNA causing tubulointerstitial kidney disease. *PLoS Genetics*, **2017**, 13, e1006620	6	35
208	Background sequence characteristics influence the occurrence and severity of disease-causing mtDNA mutations. *PLoS Genetics*, **2017**, 13, e1007126	6	24
207	Opening One□s Eyes to Mosaicism in Progressive External Ophthalmoplegia. *Neurology: Genetics*, **2017**, 3, e202	3.8	1
206	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
205	Patient care standards for primary mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. *Genetics in Medicine*, **2017**, 19,	8.1	113
204	The pattern of retinal ganglion cell dysfunction in Leber hereditary optic neuropathy. *Mitochondrion*, **2017**, 36, 138-149	4.9	19
203	Response to Newman et al. *Genetics in Medicine*, **2017**, 19,	8.1	2
202	Response to Simon et al. *Acta Neuropathologica Communications*, **2017**, 5, 34	7.3	

201	The Effect of Neurological Genomics and Personalized Mitochondrial Medicine. *JAMA Neurology*, **2017**, 74, 11-13	17.2	2
200	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
199	Cardiac involvement in hereditary myopathy with early respiratory failure: A cohort study. *Neurology*, **2016**, 87, 1031-5	6.5	11
198	Phenotypic convergence of Menkes and Wilson disease. *Neurology: Genetics*, **2016**, 2, e119	3.8	14
197	Mitochondrial Matchmaking. *New England Journal of Medicine*, **2016**, 375, 1894-1896	59.2	7
196	A multiple sclerosis-like disorder in patients with OPA1 mutations. *Annals of Clinical and Translational Neurology*, **2016**, 3, 723-9	5.3	21
195	A Woman With Intellectual Disability, Amenorrhoea, Seizures, and Balance Problems. *JAMA Neurology*, **2016**, 73, 1494-1495	17.2	1
194	Mitochondrial diseases. *Nature Reviews Disease Primers*, **2016**, 2, 16080	51.1	585
193	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
192	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
191	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
190	Both mitochondrial DNA and mitonuclear gene mutations cause hearing loss through cochlear dysfunction. *Brain*, **2016**, 139, e33	11.2	9
189	Somatic mtDNA variation is an important component of Parkinson's disease. *Neurobiology of Aging*, **2016**, 38, 217.e1-217.e6	5.6	43
188	Reply: Parsing the differences in affected with LHON: genetic versus environmental triggers of disease conversion. *Brain*, **2016**, 139, e18	11.2	10
187	Exome sequencing in dementia with Lewy bodies. *Translational Psychiatry*, **2016**, 6, e728	8.6	30
186	Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. *Human Molecular Genetics*, **2016**, 25, 1031-41	5.6	44
185	Novel mutation causes both motor neuronopathy and distal myopathy. *Neurology: Genetics*, **2016**, 2, e110	3.8	18
184	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

183	The frequency of the m.1555A>G () variant in UK patients with suspected mitochondrial deafness. *Hearing, Balance and Communication*, **2016**, 14, 101-102	0.7	1
182	Homozygous deletion in MICU1 presenting with fatigue and lethargy in childhood. *Neurology: Genetics*, **2016**, 2, e59	3.8	64
181	Lamination of the Outer Plexiform Layer in Optic Atrophy Caused by Dominant WFS1 Mutations. *Ophthalmology*, **2016**, 123, 1624-6	7.3	15
180	Emerging therapies for mitochondrial disorders. *Brain*, **2016**, 139, 1633-48	11.2	42
179	Clinical, Genetic, and Radiological Features of Extrapyramidal Movement Disorders in Mitochondrial Disease. *JAMA Neurology*, **2016**, 73, 668-74	17.2	46
178	Herpes simplex encephalitis is linked with selective mitochondrial damage; a post-mortem and in vitro study. *Acta Neuropathologica*, **2016**, 132, 433-51	14.3	15
177	Inherited mtDNA variations are not strong risk factors in human prion disease. *Neurobiology of Aging*, **2015**, 36, 2908.e1-3	5.6	
176	Reply: Evaluation of exome sequencing variation in undiagnosed ataxias. *Brain*, **2015**, 138, e384	11.2	2
175	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
174	Use of stereotypical mutational motifs to define resolution limits for the ultra-deep resequencing of mitochondrial DNA. *European Journal of Human Genetics*, **2015**, 23, 413-5	5.3	10
173	Voxel-based analysis in neuroferritinopathy expands the phenotype and determines radiological correlates of disease severity. *Journal of Neurology*, **2015**, 262, 2232-40	5.5	2
172	Clinical and pathological features of mitochondrial DNA deletion disease following antiretroviral treatment. *JAMA Neurology*, **2015**, 72, 603-5	17.2	2
171	Extreme-Depth Re-sequencing of Mitochondrial DNA Finds No Evidence of Paternal Transmission in Humans. *PLoS Genetics*, **2015**, 11, e1005040	6	50
170	SPG7 mutations are a common cause of undiagnosed ataxia. *Neurology*, **2015**, 84, 1174-6	6.5	72
169	Phenotypic variability of TRPV4 related neuropathies. *Neuromuscular Disorders*, **2015**, 25, 516-21	2.9	22
168	Modifying mitochondrial tRNAs: delivering what the cell needs. *Cell Metabolism*, **2015**, 21, 351-2	24.6	5
167	Clinical heterogeneity of primary familial brain calcification due to a novel mutation in PDGFB. *Neurology*, **2015**, 84, 1818-20	6.5	11
166	Mitochondrial Causes of Epilepsy: Evaluation, Diagnosis, and Treatment. *Seminars in Neurology*, **2015**, 35, 300-9	3.2	8

165	The p.Ser107Leu in BICD2 is a mutation ‘hot spot’ causing distal spinal muscular atrophy. *Brain*, **2015**, 138, e391	11.2	12
164	Allogeneic haematopoietic stem cell transplantation for mitochondrial neurogastrointestinal encephalomyopathy. *Brain*, **2015**, 138, 2847-58	11.2	107
163	SCP2 mutations and neurodegeneration with brain iron accumulation. *Neurology*, **2015**, 85, 1909-11	6.5	29
162	Recessive Mutations in RTN4IP1 Cause Isolated and Syndromic Optic Neuropathies. *American Journal of Human Genetics*, **2015**, 97, 754-60	11	42
161	The dynamics of mitochondrial DNA heteroplasmy: implications for human health and disease. *Nature Reviews Genetics*, **2015**, 16, 530-42	30.1	430
160	Exome sequencing in undiagnosed inherited and sporadic ataxias. *Brain*, **2015**, 138, 276-83	11.2	97
159	Prevalence of neurogenetic disorders in the North of England. *Neurology*, **2015**, 85, 1195-201	6.5	18
158	Disturbed mitochondrial dynamics and neurodegenerative disorders. *Nature Reviews Neurology*, **2015**, 11, 11-24	15	425
157	Mitochondrial disease in adults: what’s old and what’s new?. *EMBO Molecular Medicine*, **2015**, 7, 1503-12	12	88
156	Mitochondrial pathology in progressive cerebellar ataxia. *Cerebellum and Ataxias*, **2015**, 2, 16	1.7	31
155	Reduced cerebrospinal fluid mitochondrial DNA is a biomarker for early-stage Parkinson’s disease. *Annals of Neurology*, **2015**, 78, 1000-4	9.4	67
154	Adult Onset Leigh Syndrome in the Intensive Care Setting: A Novel Presentation of a C12orf65 Related Mitochondrial Disease. *Journal of Neuromuscular Diseases*, **2015**, 2, 409-419	5	15
153	Nuclear-mitochondrial proteins: too much to process?. *Brain*, **2015**, 138, 1451-3	11.2	2
152	Mitochondrial dysfunction in aging: Much progress but many unresolved questions. *Biochimica Et Biophysica Acta - Bioenergetics*, **2015**, 1847, 1347-53	4.6	202
151	Prevalence of nuclear and mitochondrial DNA mutations related to adult mitochondrial disease. *Annals of Neurology*, **2015**, 77, 753-9	9.4	483
150	Frequency of rare recessive mutations in unexplained late onset cerebellar ataxia. *Journal of Neurology*, **2015**, 262, 1822-7	5.5	18
149	A Unique Gene Regulatory Network Resets the Human Germline Epigenome for Development. *Cell*, **2015**, 161, 1453-67	56.2	417
148	Mitochondrial DNA mutations in neurodegeneration. *Biochimica Et Biophysica Acta - Bioenergetics*, **2015**, 1847, 1401-11	4.6	84

147	Clonal expansion of secondary mitochondrial DNA deletions associated with spinocerebellar ataxia type 28. *JAMA Neurology*, **2015**, 72, 106-11	17.2	33
146	Reply: Behr syndrome with OPA1 compound heterozygote mutations. *Brain*, **2015**, 138, e322	11.2	
145	Respiratory chain deficiency in nonmitochondrial disease. *Neurology: Genetics*, **2015**, 1, e6	3.8	19
144	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
143	Molecular pathogenesis of polymerase related neurodegeneration. *Annals of Neurology*, **2014**, 76, 66-81	9.4	61
142	Efficient mitochondrial biogenesis drives incomplete penetrance in Leber hereditary optic neuropathy. *Brain*, **2014**, 137, 335-53	11.2	186
141	Mutations in the SPG7 gene cause chronic progressive external ophthalmoplegia through disordered mitochondrial DNA maintenance. *Brain*, **2014**, 137, 1323-36	11.2	124
140	Teaching video neuroimages: muscle cramps and a raised creatine kinase. *Neurology*, **2014**, 82, e220-1	6.5	
139	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
138	Valproic acid triggers increased mitochondrial biogenesis in POLG-deficient fibroblasts. *Molecular Genetics and Metabolism*, **2014**, 112, 57-63	3.7	33
137	A new disease allele for the p.C30071R mutation in titin causing hereditary myopathy with early respiratory failure. *Neuromuscular Disorders*, **2014**, 24, 241-4	2.9	10
136	Behr Syndrome is Typically Associated with Disturbed Mitochondrial Translation and Mutations in the Gene. *Journal of Neuromuscular Diseases*, **2014**, 1, 55-63	5	18
135	In vivo mitochondrial function in HIV-infected persons treated with contemporary anti-retroviral therapy: a magnetic resonance spectroscopy study. *PLoS ONE*, **2014**, 9, e84678	3.7	6
134	Origins and functional consequences of somatic mitochondrial DNA mutations in human cancer. *ELife*, **2014**, 3,	8.9	229
133	Adult-onset Mendelian PEO Associated with Mitochondrial Disease. *Journal of Neuromuscular Diseases*, **2014**, 1, 119-133	5	15
132	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
131	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
130	Characterizing mild cognitive impairment in incident Parkinson disease: the ICICLE-PD study. *Neurology*, **2014**, 82, 308-16	6.5	288

129	Treatment strategies for inherited optic neuropathies: past, present and future. *Eye*, **2014**, 28, 521-37	4.4	98
128	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
127	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
126	The challenges of mitochondrial replacement. *PLoS Genetics*, **2014**, 10, e1004315	6	49
125	Recent mitochondrial DNA mutations increase the risk of developing common late-onset human diseases. *PLoS Genetics*, **2014**, 10, e1004369	6	102
124	Reply: Early-onset Behr syndrome due to compound heterozygous mutations in OPA1. *Brain*, **2014**, 137, e302	11.2	6
123	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
122	EXOSC8 mutations alter mRNA metabolism and cause hypomyelination with spinal muscular atrophy and cerebellar hypoplasia. *Nature Communications*, **2014**, 5, 4287	17.4	80
121	Genetic impact on cognition and brain function in newly diagnosed Parkinson disease: ICICLE-PD study. *Brain*, **2014**, 137, 2743-58	11.2	109
120	Reply: Hereditary myopathy with early respiratory failure is caused by mutations in the titin FN3 119 domain. *Brain*, **2014**, 137, e280	11.2	5
119	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
118	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
117	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
116	Reactive oxygen species production and mitochondrial dysfunction in white blood cells are not valid biomarkers of ageing in the very old. *PLoS ONE*, **2014**, 9, e91005	3.7	11
115	Age-related mitochondrial DNA depletion and the impact on pancreatic Beta cell function. *PLoS ONE*, **2014**, 9, e115433	3.7	33
114	Author response: Origins and functional consequences of somatic mitochondrial DNA mutations in human cancer **2014**,		3
113	Adult-onset Mendelian PEO Associated with Mitochondrial Disease. *Journal of Neuromuscular Diseases*, **2014**, 1, 119-133	5	8
112	Dominant optic atrophy: novel OPA1 mutations and revised prevalence estimates. *Ophthalmology*, **2013**, 120, 1712-1712.e1	7.3	46

111	Hereditary mtDNA heteroplasmy: a baseline for aging?. *Cell Metabolism*, **2013**, 18, 463-4	24.6	25
110	A multi-center comparison of diagnostic methods for the biochemical evaluation of suspected mitochondrial disorders. *Mitochondrion*, **2013**, 13, 36-43	4.9	19
109	Clinical features of MS associated with Leber hereditary optic neuropathy mtDNA mutations. *Neurology*, **2013**, 81, 2073-81	6.5	73
108	Universal heteroplasmy of human mitochondrial DNA. *Human Molecular Genetics*, **2013**, 22, 384-90	5.6	278
107	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
106	Multisystem fatal infantile disease caused by a novel homozygous EARS2 mutation. *Brain*, **2013**, 136, e228	11.2	33
105	New treatments for mitochondrial disease-no time to drop our standards. *Nature Reviews Neurology*, **2013**, 9, 474-81	15	133
104	Mitochondrial genetics. *British Medical Bulletin*, **2013**, 106, 135-59	5.4	219
103	Concentric hypertrophic remodelling and subendocardial dysfunction in mitochondrial DNA point mutation carriers. *European Heart Journal Cardiovascular Imaging*, **2013**, 14, 650-8	4.1	20
102	Two-stage association study and meta-analysis of mitochondrial DNA variants in Parkinson disease. *Neurology*, **2013**, 80, 2042-8	6.5	94
101	Near-identical segregation of mtDNA heteroplasmy in blood, muscle, urinary epithelium, and hair follicles in twins with optic atrophy, ptosis, and intractable epilepsy. *JAMA Neurology*, **2013**, 70, 1552-5	17.2	11
100	Altered 2-thiouridylation impairs mitochondrial translation in reversible infantile respiratory chain deficiency. *Human Molecular Genetics*, **2013**, 22, 4602-15	5.6	44
99	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
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