Patrick Chinnery

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/5777212/publications.pdf

Version: 2024-02-01

291 papers	31,113 citations	4370 86 h-index	5364 164 g-index
303 all docs	303 docs citations	303 times ranked	28334 citing authors

#	Article	IF	Citations
1	Reanalysis and revision of the Cambridge reference sequence for human mitochondrial DNA. Nature Genetics, 1999, 23, 147-147.	9.4	2,800
2	Mitochondrial diseases. Nature Reviews Disease Primers, 2016, 2, 16080.	18.1	1,001
3	Prevalence of nuclear and mitochondrial <scp>DNA</scp> mutations related to adult mitochondrial disease. Annals of Neurology, 2015, 77, 753-759.	2.8	706
4	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	6.5	699
5	The dynamics of mitochondrial DNA heteroplasmy: implications for human health and disease. Nature Reviews Genetics, 2015, 16, 530-542.	7.7	679
6	A Unique Gene Regulatory Network Resets the Human Germline Epigenome for Development. Cell, 2015, 161, 1453-1467.	13.5	556
7	Prevalence of mitochondrial DNA disease in adults. Annals of Neurology, 2008, 63, 35-39.	2.8	540
8	Pathogenic Mitochondrial DNA Mutations Are Common in the General Population. American Journal of Human Genetics, 2008, 83, 254-260.	2.6	534
9	Disturbed mitochondrial dynamics and neurodegenerative disorders. Nature Reviews Neurology, 2015, 11, 11-24.	4.9	533
10	Mitochondrial optic neuropathies â€“ Disease mechanisms and therapeutic strategies. Progress in Retinal and Eye Research, 2011, 30, 81-114.	7.3	514
11	A randomized placebo-controlled trial of idebenone in Leberâ€™s hereditary optic neuropathy. Brain, 2011, 134, 2677-2686.	3.7	461
12	Mitochondrial DNA mutations in human colonic crypt stem cells. Journal of Clinical Investigation, 2003, 112, 1351-1360.	3.9	454
13	A reduction of mitochondrial DNA molecules during embryogenesis explains the rapid segregation of genotypes. Nature Genetics, 2008, 40, 249-254.	9.4	438
14	Pronuclear transfer in human embryos to prevent transmission of mitochondrial DNA disease. Nature, 2010, 465, 82-85.	13.7	421
15	Mammalian mitochondrial genetics: heredity, heteroplasmy and disease. Trends in Genetics, 1997, 13, 450-455.	2.9	415
16	The Epidemiology of Leber Hereditary Optic Neuropathy in the North East of England. American Journal of Human Genetics, 2003, 72, 333-339.	2.6	404
17	Leber hereditary optic neuropathy. Journal of Medical Genetics, 2002, 39, 162-169.	1.5	393
18	Multi-system neurological disease is common in patients with OPA1 mutations. Brain, 2010, 133, 771-786.	3.7	385

#	Article	IF	Citations
19	The epidemiology of pathogenic mitochondrial DNA mutations. Annals of Neurology, 2000, 48, 188-193.	2.8	381
20	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, 329-337.	3.7	381
21	Characterizing mild cognitive impairment in incident Parkinson disease. Neurology, 2014, 82, 308-316.	1.5	359
22	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880.	13.9	352
23	Universal heteroplasmy of human mitochondrial DNA. Human Molecular Genetics, 2013, 22, 384-390.	1.4	344
24	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	13.7	338
25	Clinical Expression of Leber Hereditary Optic Neuropathy Is Affected by the Mitochondrial DNAâ€"Haplogroup Background. American Journal of Human Genetics, 2007, 81, 228-233.	2.6	331
26	The epidemiology of mitochondrial disordersâ€"past, present and future. Biochimica Et Biophysica Acta - Bioenergetics, 2004, 1659, 115-120.	0.5	325
27	Molecular pathology of MELAS and MERRF. The relationship between mutation load and clinical phenotypes. Brain, 1997, 120, 1713-1721.	3.7	319
28	Origins and functional consequences of somatic mitochondrial DNA mutations in human cancer. ELife, 2014, 3, .	2.8	318
29	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.	3.8	304
30	Random Intracellular Drift Explains the Clonal Expansion of Mitochondrial DNA Mutations with Age. American Journal of Human Genetics, 2001, 68, 802-806.	2.6	289
31	Mitochondrial genetics. British Medical Bulletin, 2013, 106, 135-159.	2.7	275
32	Mitochondrial dysfunction in aging: Much progress but many unresolved questions. Biochimica Et Biophysica Acta - Bioenergetics, 2015, 1847, 1347-1353.	0.5	267
33	Selective inhibition of mutant human mitochondrial DNA replication in vitro by peptide nucleic acids. Nature Genetics, 1997, 15, 212-215.	9.4	252
34	Treatment for mitochondrial disorders. The Cochrane Library, 2012, , CD004426.	1.5	244
35	Efficient mitochondrial biogenesis drives incomplete penetrance in Leberâ€™s hereditary optic neuropathy. Brain, 2014, 137, 335-353.	3.7	229
36	The inheritance of mitochondrial DNA heteroplasmy: random drift, selection or both?. Trends in Genetics, 2000, 16, 500-505.	2.9	227

#	Article	IF	Citations
37	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, 659-670.	2.6	213
38	Accumulation of mitochondrial DNA mutations in ageing, cancer, and mitochondrial disease: is there a common mechanism?. Lancet, The, 2002, 360, 1323-1325.	6.3	203
39	Risk of developing a mitochondrial DNA deletion disorder. Lancet, The, 2004, 364, 592-596.	6.3	201
40	Mitochondrial aging is accelerated by anti-retroviral therapy through the clonal expansion of mtDNA mutations. Nature Genetics, 2011, 43, 806-810.	9.4	201
41	Segregation of mitochondrial DNA heteroplasmy through a developmental genetic bottleneck in human embryos. Nature Cell Biology, 2018, 20, 144-151.	4.6	182
42	Germline selection shapes human mitochondrial DNA diversity. Science, 2019, 364, .	6.0	178
43	Peptide nucleic acid delivery to human mitochondria. Gene Therapy, 1999, 6, 1919-1928.	2.3	176
44	Patient care standards for primary mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. Genetics in Medicine, 2017, 19, 1380-1397.	1.1	173
45	Relaxed Replication of mtDNA: A Model with Implications for the Expression of Disease. American Journal of Human Genetics, 1999, 64, 1158-1165.	2.6	167
46	The mitochondrial ND6 gene is a hot spot for mutations that cause Leber's hereditary optic neuropathy. Brain, 2001, 124, 209-218.	3.7	167
47	Mitochondrial DNA and survival after sepsis: a prospective study. Lancet, The, 2005, 366, 2118-2121.	6.3	162
48	The Prevalence and Natural History of Dominant Optic Atrophy Due to OPA1 Mutations. Ophthalmology, 2010, 117, 1538-1546.e1.	2.5	162
49	New treatments for mitochondrial diseaseâ€”no time to drop our standards. Nature Reviews Neurology, 2013, 9, 474-481.	4.9	157
50	Mutations in the SPG7 gene cause chronic progressive external ophthalmoplegia through disordered mitochondrial DNA maintenance. Brain, 2014, 137, 1323-1336.	3.7	151
51	Random Genetic Drift Determines the Level of Mutant mtDNA in Human Primary Oocytes. American Journal of Human Genetics, 2001, 68, 533-536.	2.6	147
52	Epigenetics, epidemiology and mitochondrial DNA diseases. International Journal of Epidemiology, 2012, 41, 177-187.	0.9	146
53	Mitochondria. Journal of Neurology, Neurosurgery and Psychiatry, 2003, 74, 1188-1199.	0.9	142
54	Extreme heterogeneity of human mitochondrial DNA from organelles to populations. Nature Reviews Genetics, 2021, 22, 106-118.	7.7	139

#	Article	IF	Citations
55	Can mitochondrial DNA mutations cause sperm dysfunction?. Molecular Human Reproduction, 2002, 8, 719-721.	1.3	138
56	The spectrum of hearing loss due to mitochondrial DNA defects. Brain, 2000, 123, 82-92.	3.7	132
57	Recent Mitochondrial DNA Mutations Increase the Risk of Developing Common Late-Onset Human Diseases. PLoS Genetics, 2014, 10, e1004369.	1.5	131
58	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, 235-243.	2.4	130
59	Two-stage association study and meta-analysis of mitochondrial DNA variants in Parkinson disease. Neurology, 2013, 80, 2042-2048.	1.5	129
60	Variation in germline mtDNA heteroplasmy is determined prenatally but modified during subsequent transmission. Nature Genetics, 2012, 44, 1282-1285.	9.4	128
61	Allogeneic haematopoietic stem cell transplantation for mitochondrial neurogastrointestinal encephalomyopathy. Brain, 2015, 138, 2847-2858.	3.7	128
62	Treatment strategies for inherited optic neuropathies: past, present and future. Eye, 2014, 28, 521-537.	1.1	127
63	Genetic impact on cognition and brain function in newly diagnosed Parkinsonâ€™s disease: ICICLE-PD study. Brain, 2014, 137, 2743-2758.	3.7	127
64	Mitochondrial disease in adults: A scale to monitor progression and treatment. Neurology, 2006, 66, 1932-1934.	1.5	125
65	Mutations in GTPBP3 Cause a Mitochondrial Translation Defect Associated with Hypertrophic Cardiomyopathy, Lactic Acidosis, and Encephalopathy. American Journal of Human Genetics, 2014, 95, 708-720.	2.6	123
66	EXOSC8 mutations alter mRNA metabolism and cause hypomyelination with spinal muscular atrophy and cerebellar hypoplasia. Nature Communications, 2014, 5, 4287.	5.8	120
67	Mitochondrial DNA mutations in neurodegeneration. Biochimica Et Biophysica Acta - Bioenergetics, 2015, 1847, 1401-1411.	0.5	120
68	Exome sequencing in undiagnosed inherited and sporadic ataxias. Brain, 2015, 138, 276-283.	3.7	120
69	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.	2.6	118
70	Analysis of European mtDNAs for Recombination. American Journal of Human Genetics, 2001, 68, 145-153.	2.6	116
71	Mitochondrial DNA and disease. Lancet, The, 1999, 354, S17-S21.	6.3	114
72	Titin mutation segregates with hereditary myopathy with early respiratory failure. Brain, 2012, 135, 1695-1713.	3.7	113

#	Article	IF	Citations
73	Selection against Pathogenic mtDNA Mutations in a Stem Cell Population Leads to the Loss of the 3243Aâ†'G Mutation in Blood. American Journal of Human Genetics, 2008, 82, 333-343.	2.6	112
74	Molecular basis of infantile reversible cytochrome c oxidase deficiency myopathy. Brain, 2009, 132, 3165-3174.	3.7	112
75	Defective i6A37 Modification of Mitochondrial and Cytosolic tRNAs Results from Pathogenic Mutations in TRIT1 and Its Substrate tRNA. PLoS Genetics, 2014, 10, e1004424.	1.5	112
76	Mitochondrial disease in adults: what's old and what's new?. EMBO Molecular Medicine, 2015, 7, 1503-1512.	3.3	110
77	Reduced cerebrospinal fluid mitochondrial DNA is a biomarker for earlyâ€stage Parkinson's disease. Annals of Neurology, 2015, 78, 1000-1004.	2.8	106
78	The epidemiology of pathogenic mitochondrial DNA mutations. Annals of Neurology, 2000, 48, 188-93.	2.8	103
79	Mitochondrial DNA haplogroups and susceptibility to AD and dementia with Lewy bodies. Neurology, 2000, 55, 302-304.	1.5	100
80	The Power to Detect Disease Associations with Mitochondrial DNA Haplogroups. American Journal of Human Genetics, 2006, 78, 713-720.	2.6	100
81	Clinical features of MS associated with Leber hereditary optic neuropathy mtDNA mutations. Neurology, 2013, 81, 2073-2081.	1.5	100
82	PGD and heteroplasmic mitochondrial DNA point mutations: a systematic review estimating the chance of healthy offspring. Human Reproduction Update, 2012, 18, 341-349.	5.2	96
83	Synaptotagmin 2 Mutations Cause an Autosomal-Dominant Form of Lambert-Eaton Myasthenic Syndrome and Nonprogressive Motor Neuropathy. American Journal of Human Genetics, 2014, 95, 332-339.	2.6	96
84	OPA1 mutations cause cytochrome c oxidase deficiency due to loss of wild-type mtDNA molecules. Human Molecular Genetics, 2010, 19, 3043-3052.	1.4	95
85	Nonrandom tissue distribution of mutant mtDNA. American Journal of Medical Genetics Part A, 1999, 85, 498-501.	2.4	90
86	Point Mutations of the mtDNA Control Region in Normal and Neurodegenerative Human Brains. American Journal of Human Genetics, 2001, 68, 529-532.	2.6	90
87	HIV Treatment and Associated Mitochondrial Pathology. Toxicologic Pathology, 2014, 42, 811-822.	0.9	90
88	Recessive Mutations in TRMT10C Cause Defects in Mitochondrial RNA Processing and Multiple Respiratory Chain Deficiencies. American Journal of Human Genetics, 2016, 98, 993-1000.	2.6	89
89	Mitochondrial DNA haplogroups and risk of transient ischaemic attack and ischaemic stroke: a genetic association study. Lancet Neurology, The, 2010, 9, 498-503.	4.9	88
90	Quality of Life in Patients with Leber Hereditary Optic Neuropathy. , 2009, 50, 3112.		87

#	Article	IF	Citations
91	SPG7 mutations are a common cause of undiagnosed ataxia. Neurology, 2015, 84, 1174-1176.	1.5	87
92	Late-onset axial jerky dystonia due to the DYT1 deletion. Movement Disorders, 2002, 17, 196-198.	2.2	86
93	Homozygous deletion in MICU1 presenting with fatigue and lethargy in childhood. Neurology: Genetics, 2016, 2, e59.	0.9	86
94	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578.	3.7	85
95	Amyloid-Î² accumulation in the CNS in human growth hormone recipients in the UK. Acta Neuropathologica, 2017, 134, 221-240.	3.9	85
96	The mitochondrial DNA genetic bottleneck: inheritance and beyond. Essays in Biochemistry, 2018, 62, 225-234.	2.1	85
97	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, 319-328.	2.6	83
98	Mitochondrial DNA point mutations and relative copy number in 1363 disease and control human brains. Acta Neuropathologica Communications, 2017, 5, 13.	2.4	83
99	Genetic heterogeneity of motor neuropathies. Neurology, 2017, 88, 1226-1234.	1.5	81
100	Molecular pathogenesis of polymerase gammaâ€“related neurodegeneration. Annals of Neurology, 2014, 76, 66-81.	2.8	77
101	Genotypes from patients indicate no paternal mitochondrial DNA contribution. Annals of Neurology, 2003, 54, 521-524.	2.8	76
102	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, 388-396.	0.5	76
103	Nuclear-mitochondrial DNA segments resemble paternally inherited mitochondrial DNA in humans. Nature Communications, 2020, 11, 1740.	5.8	75
104	TIA1 variant drives myodegeneration in multisystem proteinopathy with SQSTM1 mutations. Journal of Clinical Investigation, 2018, 128, 1164-1177.	3.9	75
105	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.	4.9	74
106	Retrospective natural history of thymidine kinase 2 deficiency. Journal of Medical Genetics, 2018, 55, 515-521.	1.5	73
107	Mitochondrial Diseases: A Diagnostic Revolution. Trends in Genetics, 2020, 36, 702-717.	2.9	73
108	Depletion of mitochondrial DNA in leucocytes harbouring the 3243A->G mtDNA mutation. Journal of Medical Genetics, 2006, 44, 69-74.	1.5	72

#	Article	IF	Citations
109	The Distribution of Mitochondrial DNA Heteroplasmy Due to Random Genetic Drift. American Journal of Human Genetics, 2008, 83, 582-593.	2.6	72
110	An atlas of mitochondrial DNA genotypeâ€“phenotype associations in the UK Biobank. Nature Genetics, 2021, 53, 982-993.	9.4	72
111	Titin founder mutation is a common cause of myofibrillar myopathy with early respiratory failure. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 331-338.	0.9	71
112	Adults with RRM2B-related mitochondrial disease have distinct clinical and molecular characteristics. Brain, 2012, 135, 3392-3403.	3.7	70
113	Clinical, Genetic, and Radiological Features of Extrapyramidal Movement Disorders in Mitochondrial Disease. JAMA Neurology, 2016, 73, 668.	4.5	69
114	Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency. Brain, 2011, 134, 183-195.	3.7	66
115	155th ENMC workshop: Polymerase gamma and disorders of mitochondrial DNA synthesis, 21â€“23 September 2007, Naarden, The Netherlands. Neuromuscular Disorders, 2008, 18, 259-267.	0.3	65
116	Accurate mitochondrial DNA sequencing using off-target reads provides a single test to identify pathogenic point mutations. Genetics in Medicine, 2014, 16, 962-971.	1.1	64
117	Clinical, pathological and functional characterization of riboflavin-responsive neuropathy. Brain, 2017, 140, 2820-2837.	3.7	64
118	Neuroferritinopathy in a French family with late onset dominant dystonia. Journal of Medical Genetics, 2003, 40, 69e-69.	1.5	63
119	Fall in circulating mononuclear cell mitochondrial DNA content in human sepsis. Intensive Care Medicine, 2010, 36, 956-962.	3.9	62
120	Extreme-Depth Re-sequencing of Mitochondrial DNA Finds No Evidence of Paternal Transmission in Humans. PLoS Genetics, 2015, 11, e1005040.	1.5	62
121	Childhood-onset Leber hereditary optic neuropathy. British Journal of Ophthalmology, 2017, 101, 1505-1509.	2.1	62
122	The Challenges of Mitochondrial Replacement. PLoS Genetics, 2014, 10, e1004315.	1.5	61
123	Somatic mtDNA variation is an important component of Parkinson's disease. Neurobiology of Aging, 2016, 38, 217.e1-217.e6.	1.5	61
124	Treatment of mitochondrial disease. Journal of Bioenergetics and Biomembranes, 1997, 29, 195-205.	1.0	59
125	Emerging therapies for mitochondrial disorders. Brain, 2016, 139, 1633-1648.	3.7	59
126	Mitochondrial medicine. QJM - Monthly Journal of the Association of Physicians, 1997, 90, 657-667.	0.2	59

#	Article	IF	Citations
127	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.	2.6	58
128	Dominant Optic Atrophy: Novel OPA1 Mutations and Revised Prevalence Estimates. Ophthalmology, 2013, 120, 1712-1712.e1.	2.5	57
129	Mitochondrial heteroplasmy beyond the oocyte bottleneck. Seminars in Cell and Developmental Biology, 2020, 97, 156-166.	2.3	57
130	Assessing mitochondrial heteroplasmy using next generation sequencing: A note of caution. Mitochondrion, 2019, 46, 302-306.	1.6	56
131	Recessive Mutations in RTN4IP1 Cause Isolated and Syndromic Optic Neuropathies. American Journal of Human Genetics, 2015, 97, 754-760.	2.6	54
132	High prevalence of focal and multi-focal somatic genetic variants in the human brain. Nature Communications, 2018, 9, 4257.	5.8	54
133	Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. Human Molecular Genetics, 2016, 25, 1031-1041.	1.4	53
134	Mitochondrial DNA haplogroups and type 2 diabetes: a study of 897 cases and 1010 controls. Journal of Medical Genetics, 2007, 44, e80-e80.	1.5	52
135	Altered 2-thiouridylation impairs mitochondrial translation in reversible infantile respiratory chain deficiency. Human Molecular Genetics, 2013, 22, 4602-4615.	1.4	52
136	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, 115-124.	1.7	52
137	Mutations in mitochondrial DNA causing tubulointerstitial kidney disease. PLoS Genetics, 2017, 13, e1006620.	1.5	52
138	A novel mechanism causing imbalance of mitochondrial fusion and fission in human myopathies. Human Molecular Genetics, 2018, 27, 1186-1195.	1.4	52
139	Mitochondria in neuroinflammation â€“ Multiple sclerosis (MS), leber hereditary optic neuropathy (LHON) and LHON-MS. Neuroscience Letters, 2019, 710, 132932.	1.0	52
140	Population Screening for Variant Creutzfeldt-Jakob Disease Using a Novel Blood Test. JAMA Neurology, 2014, 71, 421.	4.5	51
141	Somatic variants in autosomal dominant genes are a rare cause of sporadic Alzheimer's disease. Alzheimer's and Dementia, 2018, 14, 1632-1639.	0.4	51
142	Normokalemic periodic paralysis revisited: Does it exist?. Annals of Neurology, 2002, 52, 251-252.	2.8	50
143	Oldies but Goldies mtDNA Population Variants and Neurodegenerative Diseases. Frontiers in Neuroscience, 2018, 12, 682.	1.4	50
144	A novel mitochondrial tRNA isoleucine gene mutation causing chronic progressive external ophthalmoplegia. Neurology, 1997, 49, 1166-1168.	1.5	49

#	Article	IF	Citations
145	Background sequence characteristics influence the occurrence and severity of disease-causing mtDNA mutations. PLoS Genetics, 2017, 13, e1007126.	1.5	49
146	Mitochondrial DNA Heteroplasmy and Purifying Selection in the Mammalian Female Germ Line. Development Growth and Differentiation, 2018, 60, 21-32.	0.6	49
147	First-line genomic diagnosis of mitochondrial disorders. Nature Reviews Genetics, 2018, 19, 399-400.	7.7	49
148	Clinical mitochondrial genetics. Journal of Medical Genetics, 1999, 36, 425-36.	1.5	48
149	MT-ND5 Mutation Exhibits Highly Variable Neurological Manifestations at Low Mutant Load. EBioMedicine, 2018, 30, 86-93.	2.7	47
150	A novel mitochondrial tRNA phenylalanine mutation presenting with acute rhabdomyolysis. Annals of Neurology, 1997, 41, 408-410.	2.8	46
151	Inheritance of mitochondrial DNA in humans: implications for rare and common diseases. Journal of Internal Medicine, 2020, 287, 634-644.	2.7	46
152	Mitochondria and Hypoxia: Metabolic Crosstalk in Cell-Fate Decisions. Trends in Endocrinology and Metabolism, 2018, 29, 249-259.	3.1	45
153	Exposure of Monocytic Cells to Lipopolysaccharide Induces Coordinated Endotoxin Tolerance, Mitochondrial Biogenesis, Mitophagy, and Antioxidant Defenses. Frontiers in Immunology, 2018, 9, 2217.	2.2	45
154	Metabolic effects of bezafibrate in mitochondrial disease. EMBO Molecular Medicine, 2020, 12, e11589.	3.3	45
155	In vitro supplementation with dAMP/dGMP leads to partial restoration of mtDNA levels in mitochondrial depletion syndromes. Human Molecular Genetics, 2009, 18, 1590-1599.	1.4	44
156	Subtle neurological and metabolic abnormalities in an Opa1 mouse model of autosomal dominant optic atrophy. Experimental Neurology, 2009, 220, 404-409.	2.0	44
157	Hereditary mtDNA Heteroplasmy: A Baseline for Aging?. Cell Metabolism, 2013, 18, 463-464.	7.2	44
158	Genetic compendium of 1511 human brains available through the UK Medical Research Council Brain Banks Network Resource. Genome Research, 2017, 27, 165-173.	2.4	44
159	Monitoring clinical progression with mitochondrial disease biomarkers. Brain, 2017, 140, 2530-2540.	3.7	44
160	Age-Related Mitochondrial DNA Depletion and the Impact on Pancreatic Beta Cell Function. PLoS ONE, 2014, 9, e115433.	1.1	44
161	Adult-onset spinocerebellar ataxia syndromes due toMTATP6mutations. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 883-886.	0.9	42
162	MSeqDR: A Centralized Knowledge Repository and Bioinformatics Web Resource to Facilitate Genomic Investigations in Mitochondrial Disease. Human Mutation, 2016, 37, 540-548.	1.1	42

#	Article	IF	Citations
163	Diagnosis of â€˜possibleâ€™ mitochondrial disease: an existential crisis. Journal of Medical Genetics, 2019, 56, 123-130.	1.5	42
164	Use of whole genome sequencing to determine genetic basis of suspected mitochondrial disorders: cohort study. BMJ, The, 2021, 375, e066288.	3.0	42
165	Clonal Expansion of Secondary Mitochondrial DNA Deletions Associated With Spinocerebellar Ataxia Type 28. JAMA Neurology, 2015, 72, 106.	4.5	41
166	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.	2.3	41
167	Valproic acid triggers increased mitochondrial biogenesis in POLG-deficient fibroblasts. Molecular Genetics and Metabolism, 2014, 112, 57-63.	0.5	40
168	Mitochondrial DNA variants modulate N-formylmethionine, proteostasis and risk of late-onset human diseases. Nature Medicine, 2021, 27, 1564-1575.	15.2	40
169	No evidence of an association between mitochondrial DNA variants and osteoarthritis in 7393 cases and 5122 controls. Annals of the Rheumatic Diseases, 2013, 72, 136-139.	0.5	39
170	SCP2 mutations and neurodegeneration with brain iron accumulation. Neurology, 2015, 85, 1909-1911.	1.5	39
171	Multisystem fatal infantile disease caused by a novel homozygous EARS2 mutation. Brain, 2013, 136, e228-e228.	3.7	38
172	The pattern of retinal ganglion cell dysfunction in Leber hereditary optic neuropathy. Mitochondrion, 2017, 36, 138-149.	1.6	38
173	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, 540-550.	2.6	37
174	Mitochondrial pathology in progressive cerebellar ataxia. Cerebellum and Ataxias, 2015, 2, 16.	1.9	37
175	Mutation-specific effects in germline transmission of pathogenic mtDNA variants. Human Reproduction, 2018, 33, 1331-1341.	0.4	36
176	Exome sequencing in dementia with Lewy bodies. Translational Psychiatry, 2016, 6, e728-e728.	2.4	35
177	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, 769-772.	1.1	34
178	Cardiomyopathy is common in patients with the mitochondrial DNA m.3243A>G mutation and correlates with mutation load. Neuromuscular Disorders, 2012, 22, 592-596.	0.3	34
179	116th ENMC international workshop: the treatment of mitochondrial disorders, 14thâ€“16th March 2003, Naarden, The Netherlands. Neuromuscular Disorders, 2003, 13, 757-764.	0.3	33
180	POLG mutations cause decreased mitochondrial DNA repopulation rates following induced depletion in human fibroblasts. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2011, 1812, 321-325.	1.8	33

#	Article	IF	Citations
181	Infantile hereditary spastic paraparesis due to codominant mutations in the spastin gene. Neurology, 2004, 63, 710-712.	1.5	32
182	Mitochondrial enteropathy: the primary pathology may not be within the gastrointestinal tract. Gut, 2001, 48, 121-124.	6.1	31
183	Mitochondrial oxodicarboxylate carrier deficiency is associated with mitochondrial DNA depletion and spinal muscular atrophyâ€“like disease. Genetics in Medicine, 2018, 20, 1224-1235.	1.1	31
184	The Contribution of the Cerebellum to Cognition in Spinocerebellar Ataxia Type 6. Behavioural Neurology, 2010, 23, 3-15.	1.1	31
185	Concentric hypertrophic remodelling and subendocardial dysfunction in mitochondrial DNA point mutation carriersâ€. European Heart Journal Cardiovascular Imaging, 2013, 14, 650-658.	0.5	30
186	Two recurrent mutations are associated with GNE myopathy in the North of Britain. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 1359-1365.	0.9	30
187	Abnormal cardiac energetics in patients carrying the A3243G mtDNA mutation measured in vivo using phosphorus MR spectroscopy. Biochimica Et Biophysica Acta - Bioenergetics, 2004, 1657, 146-150.	0.5	27
188	Phenotypic variability of TRPV4 related neuropathies. Neuromuscular Disorders, 2015, 25, 516-521.	0.3	27
189	A multiple sclerosisâ€like disorder in patients with OPA1 mutations. Annals of Clinical and Translational Neurology, 2016, 3, 723-729.	1.7	27
190	Nonrandom tissue distribution of mutant mtDNA. American Journal of Medical Genetics Part A, 1999, 85, 498-501.	2.4	27
191	Prevalence of neurogenetic disorders in the North of England. Neurology, 2015, 85, 1195-1201.	1.5	26
192	Chronic pain is common in mitochondrial disease. Neuromuscular Disorders, 2020, 30, 413-419.	0.3	26
193	Metabolic shift underlies recovery in reversible infantile respiratory chain deficiency. EMBO Journal, 2020, 39, e105364.	3.5	26
194	Clinical and functional characterisation of the combined respiratory chain defect in two sisters due to autosomal recessive mutations in MTFMT. Mitochondrion, 2013, 13, 743-748.	1.6	25
195	A genome-wide association study of mitochondrial DNA copy number in two population-based cohorts. Human Genomics, 2019, 13, 6.	1.4	25
196	Mitochondrial Replacement in the Clinic. New England Journal of Medicine, 2020, 382, 1855-1857.	13.9	25
197	Mitochondrial DNA analysis: polymorphisms and pathogenicity. Journal of Medical Genetics, 1999, 36, 505-10.	1.5	25
198	No correlation between muscle A3243G mutation load and mitochondrial function in vivo. Neurology, 2001, 56, 1101-1104.	1.5	24

#	Article	IF	Citations
199	Novel HSPB1 mutation causes both motor neuronopathy and distal myopathy. Neurology: Genetics, 2016, 2, e110.	0.9	24
200	A multi-center comparison of diagnostic methods for the biochemical evaluation of suspected mitochondrial disorders. Mitochondrion, 2013, 13, 36-43.	1.6	23
201	Respiratory chain deficiency in nonmitochondrial disease. Neurology: Genetics, 2015, 1, e6.	0.9	23
202	Lethal Neonatal LTBL Associated with Biallelic EARS2 Variants: Case Report and Review of the Reported Neuroradiological Features. JIMD Reports, 2016, 33, 61-68.	0.7	23
203	Mitochondrially-targeted APOBEC1 is a potent mtDNA mutator affecting mitochondrial function and organismal fitness in Drosophila. Nature Communications, 2019, 10, 3280.	5.8	23
204	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.	1.1	22
205	Mitochondrial DNA heteroplasmy is modulated during oocyte development propagating mutation transmission. Science Advances, 2021, 7, eabi5657.	4.7	22
206	Heteroplasmic mitochondrial DNA mutations in frontotemporal lobar degeneration. Acta Neuropathologica, 2022, 143, 687-695.	3.9	22
207	Herpes simplex encephalitis is linked with selective mitochondrial damage; a post-mortem and in vitro study. Acta Neuropathologica, 2016, 132, 433-451.	3.9	21
208	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.	1.2	21
209	Cell reprogramming shapes the mitochondrial DNA landscape. Nature Communications, 2021, 12, 5241.	5.8	21
210	Non-Random mtDNA Segregation Patterns Indicate a Metastable Heteroplasmic Segregation Unit in m.3243A>G Cybrid Cells. PLoS ONE, 2012, 7, e52080.	1.1	21
211	Behr's Syndrome is Typically Associated with Disturbed Mitochondrial Translation and Mutations in the C12orf65 Gene. Journal of Neuromuscular Diseases, 2014, 1, 55-63.	1.1	20
212	Frequency of rare recessive mutations in unexplained late onset cerebellar ataxia. Journal of Neurology, 2015, 262, 1822-1827.	1.8	20
213	Frequency and signature of somatic variants in 1461 human brain exomes. Genetics in Medicine, 2019, 21, 904-912.	1.1	20
214	Effects of thyroid hormone on mitochondria and metabolism of human preimplantation embryos. Stem Cells, 2020, 38, 369-381.	1.4	20
215	Adult-onset Mendelian PEO Associated with Mitochondrial Disease. Journal of Neuromuscular Diseases, 2014, 1, 119-133.	1.1	19
216	Lamination of the Outer Plexiform Layer in Optic Atrophy Caused by Dominant WFS1 Mutations. Ophthalmology, 2016, 123, 1624-1626.	2.5	19

#	Article	IF	Citations
217	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.	3.7	19
218	Molecular genetic investigations identify new clinical phenotypes associated with BCS1L-related mitochondrial disease. Human Molecular Genetics, 2019, 28, 3766-3776.	1.4	19
219	Heteroplasmic mitochondrial DNA variants in cardiovascular diseases. PLoS Genetics, 2022, 18, e1010068.	1.5	19
220	Phenotypic convergence of Menkes and Wilson disease. Neurology: Genetics, 2016, 2, e119.	0.9	18
221	Recurrent horizontal transfer identifies mitochondrial positive selection in a transmissible cancer. Nature Communications, 2020, 11, 3059.	5.8	18
222	Single-molecule mitochondrial DNA sequencing shows no evidence of CpG methylation in human cells and tissues. Nucleic Acids Research, 2021, 49, 12757-12768.	6.5	18
223	Clinical progression of mitochondrial myopathy is associated with the random accumulation of cytochrome c oxidase negative skeletal muscle fibres. Journal of the Neurological Sciences, 2003, 211, 63-66.	0.3	17
224	Very low levels of the mtDNA A3243G mutation associated with mitochondrial dysfunction in vivo. Annals of Neurology, 2000, 47, 381-4.	2.8	17
225	New approaches to the treatment of mitochondrial disorders. Reproductive BioMedicine Online, 2004, 8, 16-23.	1.1	16
226	Associating Mitochondrial DNA Variation with Complex Traits. American Journal of Human Genetics, 2007, 80, 378-382.	2.6	16
227	Both mitochondrial DNA and mitonuclear gene mutations cause hearing loss through cochlear dysfunction. Brain, 2016, 139, e33-e33.	3.7	15
228	Recurrent strokes in a 34-year-old man. Lancet, The, 1997, 350, 560.	6.3	14
229	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.	4.5	14
230	Clinical heterogeneity of primary familial brain calcification due to a novel mutation in PDGFB. Neurology, 2015, 84, 1818-1820.	1.5	14
231	The frequency of the m.1555Aâ€‰›â€‰G (MTRNR1) variant in UK patients with suspected mitochondrial deafness. Hearing, Balance and Communication, 2016, 14, 101-102.	0.1	14
232	Visualizing, quantifying, and manipulating mitochondrial DNA in vivo. Journal of Biological Chemistry, 2020, 295, 17588-17601.	1.6	14
233	MitoPhen database: a human phenotype ontology-based approach to identify mitochondrial DNA diseases. Nucleic Acids Research, 2021, 49, 9686-9695.	6.5	14
234	High diagnostic rate of trio exome sequencing in consanguineous families with neurogenetic diseases. Brain, 2022, 145, 1507-1518.	3.7	14

#	Article	IF	Citations
235	The p.Ser107Leu inBICD2is a mutation â€˜hot spotâ€™ causing distal spinal muscular atrophy. Brain, 2015, 138, e391-e391.	3.7	13
236	Reply: Parsing the differences in affected with LHON: genetic versus environmental triggers of disease conversion. Brain, 2016, 139, e18-e18.	3.7	13
237	Cardiac involvement in hereditary myopathy with early respiratory failure. Neurology, 2016, 87, 1031-1035.	1.5	12
238	Biparental inheritance of mitochondrial DNA revisited. Nature Reviews Genetics, 2021, 22, 477-478.	7.7	12
239	De novo CTBP1 variant is associated with decreased mitochondrial respiratory chain activities. Neurology: Genetics, 2017, 3, e187.	0.9	11
240	Oxygen in mitochondrial disease: can there be too much of a good thing?. Journal of Inherited Metabolic Disease, 2018, 41, 761-763.	1.7	11
241	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.	1.1	11
242	In Vivo Mitochondrial Function in HIV-Infected Persons Treated with Contemporary Anti-Retroviral Therapy: A Magnetic Resonance Spectroscopy Study. PLoS ONE, 2014, 9, e84678.	1.1	10
243	A new disease allele for the p.C30071R mutation in titin causing hereditary myopathy with early respiratory failure. Neuromuscular Disorders, 2014, 24, 241-244.	0.3	10
244	Use of stereotypical mutational motifs to define resolution limits for the ultra-deep resequencing of mitochondrial DNA. European Journal of Human Genetics, 2015, 23, 413-415.	1.4	10
245	Epigenetic regulation in the pathophysiology of Lewy body dementia. Progress in Neurobiology, 2020, 192, 101822.	2.8	10
246	Mutations in SUCLA2: a tandem ride back to the Krebs cycle. Brain, 2007, 130, 606-609.	3.7	9
247	Defining neurogenetic phenotypes (or how to compare needles in haystacks). Brain, 2010, 133, 649-651.	3.7	9
248	Identification of UBAP1 mutations in juvenile hereditary spastic paraplegia in the 100,000 Genomes Project. European Journal of Human Genetics, 2020, 28, 1763-1768.	1.4	9
249	Adult-onset Mendelian PEO Associated with Mitochondrial Disease. Journal of Neuromuscular Diseases, 2014, 1, 119-133.	1.1	9
250	Mitochondrial Causes of Epilepsy: Evaluation, Diagnosis, and Treatment. Seminars in Neurology, 2015, 35, 300-309.	0.5	8
251	Mitochondrial Matchmaking. New England Journal of Medicine, 2016, 375, 1894-1896.	13.9	8
252	Behr syndrome and hypertrophic cardiomyopathy in a family with a novel UCHL1 deletion. Journal of Neurology, 2020, 267, 3643-3649.	1.8	8

#	Article	IF	Citations
253	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.	0.7	8
254	WFS1-Associated Optic Neuropathy: Genotype-Phenotype Correlations and Disease Progression. American Journal of Ophthalmology, 2022, 241, 9-27.	1.7	8
255	Leber hereditary optic neuropathy â€“ Therapeutic challenges and early promise. Taiwan Journal of Ophthalmology, 2011, 1, 12-15.	0.3	7
256	Identification of a novel heterozygous guanosine monophosphate reductase (GMPR) variant in a patient with a lateâ€onset disorder of mitochondrial DNA maintenance. Clinical Genetics, 2020, 97, 276-286.	1.0	7
257	[11C]PK11195-PET Brain Imaging of the Mitochondrial Translocator Protein in Mitochondrial Disease. Neurology, 2021, 96, e2761-e2773.	1.5	7
258	Oxygen tension modulates the mitochondrial genetic bottleneck and influences the segregation of a heteroplasmic mtDNA variant in vitro. Communications Biology, 2021, 4, 584.	2.0	7
259	Annotation: Mitochondrial genotype and clinical phenotype. Journal of Inherited Metabolic Disease, 1998, 21, 321-325.	1.7	6
260	Is There Alteration in Aortic Stiffness in Leber Hereditary Optic Neuropathy?. European Journal of Ophthalmology, 2008, 18, 309-312.	0.7	6
261	Reply: Early-onset Behr syndrome due to compound heterozygous mutations in OPA1. Brain, 2014, 137, e302-e302.	3.7	6
262	Reply: Hereditary myopathy with early respiratory failure is caused by mutations in the titin FN3 119 domain. Brain, 2014, 137, e280-e280.	3.7	6
263	Modifying Mitochondrial tRNAs: Delivering What the Cell Needs. Cell Metabolism, 2015, 21, 351-352.	7.2	6
264	Mitochondrial DNA depletion induces innate immune dysfunction rescued by IFN-Î³. Journal of Allergy and Clinical Immunology, 2017, 140, 1461-1464.e8.	1.5	5
265	De-fusing mitochondria defuses the mtDNA time-bomb. Cell Research, 2019, 29, 781-782.	5.7	5
266	Implications of mitochondrial DNA mutations in human induced pluripotent stem cells. Nature Reviews Genetics, 2022, 23, 69-70.	7.7	5
267	Multifocal demyelinating motor neuropathy and hamartoma syndrome associated with a de novo PTEN mutation. Neurology, 2018, 90, e1842-e1848.	1.5	4
268	Voxel-based analysis in neuroferritinopathy expands the phenotype and determines radiological correlates of disease severity. Journal of Neurology, 2015, 262, 2232-2240.	1.8	3
269	Clinical and Pathological Features of Mitochondrial DNA Deletion Disease Following Antiretroviral Treatment. JAMA Neurology, 2015, 72, 603.	4.5	3
270	Response to Newman et al.. Genetics in Medicine, 2017, 19, 1380-1380.	1.1	3

#	Article	IF	Citations
271	Editorial: Mitochondrial medicine special issue. Journal of Inherited Metabolic Disease, 2021, 44, 289-291.	1.7	3
272	Nuclear-mitochondrial proteins: too much to process?: Figure 1. Brain, 2015, 138, 1451-1453.	3.7	2
273	Reply: Evaluation of exome sequencing variation in undiagnosed ataxias. Brain, 2015, 138, e384-e384.	3.7	2
274	The Effect of Neurological Genomics and Personalized Mitochondrial Medicine. JAMA Neurology, 2017, 74, 11.	4.5	2
275	Mitochondrial Haplogroup and the Risk of Acute Kidney Injury Following Cardiac Bypass Surgery. Scientific Reports, 2019, 9, 2279.	1.6	2
276	Isolated homozygous R217X OPTN mutation causes knock-out of functional C-terminal optineurin domains and associated oligodendrogliopathy-dominant ALSâ€“TDP. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 1022-1024.	0.9	2
277	Shortening the diagnostic odysseyâ€”the impact of whole genome sequencing in the NHS. BMJ, The, 2021, 375, n2683.	3.0	2
278	The age of single-gene neurological disorders is not dead. Brain, 2010, 133, 1865-1868.	3.7	1
279	A Woman With Intellectual Disability, Amenorrhoea, Seizures, and Balance Problems. JAMA Neurology, 2016, 73, 1494.	4.5	1
280	Opening One's Eyes to Mosaicism in Progressive External Ophthalmoplegia. Neurology: Genetics, 2017, 3, e202.	0.9	1
281	Nonrandom tissue distribution of mutant mtDNA. , 1999, 85, 498.		1
282	Teaching Video Neuro Images : Muscle cramps and a raised creatine kinase. Neurology, 2014, 82, e220-1.	1.5	0
283	Reply: â€˜Behr syndromeâ€™ with OPA1 compound heterozygote mutations. Brain, 2015, 138, e322-e322.	3.7	0
284	Inherited mtDNA variations are not strong risk factors in human prion disease. Neurobiology of Aging, 2015, 36, 2908.e1-2908.e3.	1.5	0
285	PLP1 mutations and central demyelination. Neurology: Clinical Practice, 2017, 7, 451-454.	0.8	0
286	Response to Simon et al.,. Acta Neuropathologica Communications, 2017, 5, 34.	2.4	0
287	Clinical Management of Mitochondrial Diseases. , 0, , 59-68.		0
288	Diagnostic Approach to Mitochondrial Diseases. , 2019, , 281-287.		0

#	Article	IF	Citations
289	Heredity and segregation of mtDNA. , 2020, , 87-107.		0
290	Cracking the enigma of mitochondrial-DNA variants and cancer. Nature Metabolism, 2020, 2, 221-222.	5.1	0
291	Mitochondrial disorders due to mutations in the nuclear genome. , 2020, , 415-425.		0