Patrick Chinnery

List of Publications by Year
in descending order

291	31,113	5126	6177
papers	citations	86 h-index	164 g-index

303	303	303	30773
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	High diagnostic rate of trio exome sequencing in consanguineous families with neurogenetic diseases. Brain, 2022, 145, 1507-1518.	3.7	14
2	Implications of mitochondrial DNA mutations in human induced pluripotent stem cells. Nature Reviews Genetics, 2022, 23, 69-70.	7.7	5
3	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
4	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
5	Heteroplasmic mitochondrial DNA variants in cardiovascular diseases. PLoS Genetics, 2022, 18, e1010068.	1.5	19
6	WFS1-Associated Optic Neuropathy: Genotype-Phenotype Correlations and Disease Progression. American Journal of Ophthalmology, 2022, 241, 9-27.	1.7	8
7	Heteroplasmic mitochondrial DNA mutations in frontotemporal lobar degeneration. Acta Neuropathologica, 2022, 143, 687-695.	3.9	22
8	Extreme heterogeneity of human mitochondrial DNA from organelles to populations. Nature Reviews Genetics, 2021, 22, 106-118.	7.7	139
9	Editorial: Mitochondrial medicine special issue. Journal of Inherited Metabolic Disease, 2021, 44, 289-291.	1.7	3
10	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
11	[11C]PK11195-PET Brain Imaging of the Mitochondrial Translocator Protein in Mitochondrial Disease. Neurology, 2021, 96, e2761-e2773.	1.5	7
12	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
13	An atlas of mitochondrial DNA genotypeâ€“phenotype associations in the UK Biobank. Nature Genetics, 2021, 53, 982-993.	9.4	72
14	Biparental inheritance of mitochondrial DNA revisited. Nature Reviews Genetics, 2021, 22, 477-478.	7.7	12
15	MitoPhen database: a human phenotype ontology-based approach to identify mitochondrial DNA diseases. Nucleic Acids Research, 2021, 49, 9686-9695.	6.5	14
16	Mitochondrial DNA variants modulate N-formylmethionine, proteostasis and risk of late-onset human diseases. Nature Medicine, 2021, 27, 1564-1575.	15.2	40
17	Cell reprogramming shapes the mitochondrial DNA landscape. Nature Communications, 2021, 12, 5241.	5.8	21
18	Shortening the diagnostic odysseyâ€”the impact of whole genome sequencing in the NHS. BMJ, The, 2021, 375, n2683.	3.0	2

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19	Use of whole genome sequencing to determine genetic basis of suspected mitochondrial disorders: cohort study. BMJ, The, 2021, 375, e066288.	3.0	42
20	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
21	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
22	Mitochondrial DNA heteroplasmy is modulated during oocyte development propagating mutation transmission. Science Advances, 2021, 7, eabi5657.	4.7	22
23	Mitochondrial heteroplasmy beyond the oocyte bottleneck. Seminars in Cell and Developmental Biology, 2020, 97, 156-166.	2.3	57
24	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
25	Effects of thyroid hormone on mitochondria and metabolism of human preimplantation embryos. Stem Cells, 2020, 38, 369-381.	1.4	20
26	Visualizing, quantifying, and manipulating mitochondrial DNA in vivo. Journal of Biological Chemistry, 2020, 295, 17588-17601.	1.6	14
27	Mitochondrial Diseases: A Diagnostic Revolution. Trends in Genetics, 2020, 36, 702-717.	2.9	73
28	Behr syndrome and hypertrophic cardiomyopathy in a family with a novel UCHL1 deletion. Journal of Neurology, 2020, 267, 3643-3649.	1.8	8
29	Heredity and segregation of mtDNA. , 2020, , 87-107.		0
30	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
31	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
32	Mitochondrial Replacement in the Clinic. New England Journal of Medicine, 2020, 382, 1855-1857.	13.9	25
33	Epigenetic regulation in the pathophysiology of Lewy body dementia. Progress in Neurobiology, 2020, 192, 101822.	2.8	10
34	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	13.7	338
35	Recurrent horizontal transfer identifies mitochondrial positive selection in a transmissible cancer. Nature Communications, 2020, 11, 3059.	5.8	18
36	Inheritance of mitochondrial DNA in humans: implications for rare and common diseases. Journal of Internal Medicine, 2020, 287, 634-644.	2.7	46

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37	Nuclear-mitochondrial DNA segments resemble paternally inherited mitochondrial DNA in humans. Nature Communications, 2020, 11, 1740.	5.8	75
38	Chronic pain is common in mitochondrial disease. Neuromuscular Disorders, 2020, 30, 413-419.	0.3	26
39	Cracking the enigma of mitochondrial-DNA variants and cancer. Nature Metabolism, 2020, 2, 221-222.	5.1	0
40	Metabolic effects of bezafibrate in mitochondrial disease. EMBO Molecular Medicine, 2020, 12, e11589.	3.3	45
41	Metabolic shift underlies recovery in reversible infantile respiratory chain deficiency. EMBO Journal, 2020, 39, e105364.	3.5	26
42	Mitochondrial disorders due to mutations in the nuclear genome. , 2020, , 415-425.		0
43	Assessing mitochondrial heteroplasmy using next generation sequencing: A note of caution. Mitochondrion, 2019, 46, 302-306.	1.6	56
44	De-fusing mitochondria defuses the mtDNA time-bomb. Cell Research, 2019, 29, 781-782.	5.7	5
45	Mitochondrially-targeted APOBEC1 is a potent mtDNA mutator affecting mitochondrial function and organismal fitness in Drosophila. Nature Communications, 2019, 10, 3280.	5.8	23
46	Molecular genetic investigations identify new clinical phenotypes associated with BCS1L-related mitochondrial disease. Human Molecular Genetics, 2019, 28, 3766-3776.	1.4	19
47	Diagnosis of â€˜possibleâ€™ mitochondrial disease: an existential crisis. Journal of Medical Genetics, 2019, 56, 123-130.	1.5	42
48	Germline selection shapes human mitochondrial DNA diversity. Science, 2019, 364, .	6.0	178
49	Diagnostic Approach to Mitochondrial Diseases. , 2019, , 281-287.		0
50	A genome-wide association study of mitochondrial DNA copy number in two population-based cohorts. Human Genomics, 2019, 13, 6.	1.4	25
51	Mitochondrial Haplogroup and the Risk of Acute Kidney Injury Following Cardiac Bypass Surgery. Scientific Reports, 2019, 9, 2279.	1.6	2
52	Frequency and signature of somatic variants in 1461 human brain exomes. Genetics in Medicine, 2019, 21, 904-912.	1.1	20
53	Mitochondria in neuroinflammation â€“ Multiple sclerosis (MS), leber hereditary optic neuropathy (LHON) and LHON-MS. Neuroscience Letters, 2019, 710, 132932.	1.0	52
54	MT-ND5 Mutation Exhibits Highly Variable Neurological Manifestations at Low Mutant Load. EBioMedicine, 2018, 30, 86-93.	2.7	47

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55	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
56	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
57	Mitochondria and Hypoxia: Metabolic Crosstalk in Cell-Fate Decisions. Trends in Endocrinology and Metabolism, 2018, 29, 249-259.	3.1	45
58	A novel mechanism causing imbalance of mitochondrial fusion and fission in human myopathies. Human Molecular Genetics, 2018, 27, 1186-1195.	1.4	52
59	Mitochondrial DNA Heteroplasmy and Purifying Selection in the Mammalian Female Germ Line. Development Growth and Differentiation, 2018, 60, 21-32.	0.6	49
60	Segregation of mitochondrial DNA heteroplasmy through a developmental genetic bottleneck in human embryos. Nature Cell Biology, 2018, 20, 144-151.	4.6	182
61	Multifocal demyelinating motor neuropathy and hamartoma syndrome associated with a de novo PTEN mutation. Neurology, 2018, 90, e1842-e1848.	1.5	4
62	Retrospective natural history of thymidine kinase 2 deficiency. Journal of Medical Genetics, 2018, 55, 515-521.	1.5	73
63	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
64	High prevalence of focal and multi-focal somatic genetic variants in the human brain. Nature Communications, 2018, 9, 4257.	5.8	54
65	Oldies but Goldies mtDNA Population Variants and Neurodegenerative Diseases. Frontiers in Neuroscience, 2018, 12, 682.	1.4	50
66	First-line genomic diagnosis of mitochondrial disorders. Nature Reviews Genetics, 2018, 19, 399-400.	7.7	49
67	Mutation-specific effects in germline transmission of pathogenic mtDNA variants. Human Reproduction, 2018, 33, 1331-1341.	0.4	36
68	The mitochondrial DNA genetic bottleneck: inheritance and beyond. Essays in Biochemistry, 2018, 62, 225-234.	2.1	85
69	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
70	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
71	TIA1 variant drives myodegeneration in multisystem proteinopathy with SQSTM1 mutations. Journal of Clinical Investigation, 2018, 128, 1164-1177.	3.9	75
72	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	6.5	699

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73	Mitochondrial DNA point mutations and relative copy number in 1363 disease and control human brains. Acta Neuropathologica Communications, 2017, 5, 13.	2.4	83
74	Genetic heterogeneity of motor neuropathies. Neurology, 2017, 88, 1226-1234.	1.5	81
75	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578.	3.7	85
76	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
77	Amyloid-Î² accumulation in the CNS in human growth hormone recipients in the UK. Acta Neuropathologica, 2017, 134, 221-240.	3.9	85
78	Childhood-onset Leber hereditary optic neuropathy. British Journal of Ophthalmology, 2017, 101, 1505-1509.	2.1	62
79	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
80	De novo CTBP1 variant is associated with decreased mitochondrial respiratory chain activities. Neurology: Genetics, 2017, 3, e187.	0.9	11
81	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
82	PLP1 mutations and central demyelination. Neurology: Clinical Practice, 2017, 7, 451-454.	0.8	0
83	Monitoring clinical progression with mitochondrial disease biomarkers. Brain, 2017, 140, 2530-2540.	3.7	44
84	Clinical, pathological and functional characterization of riboflavin-responsive neuropathy. Brain, 2017, 140, 2820-2837.	3.7	64
85	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
86	The pattern of retinal ganglion cell dysfunction in Leber hereditary optic neuropathy. Mitochondrion, 2017, 36, 138-149.	1.6	38
87	Response to Newman et al.. Genetics in Medicine, 2017, 19, 1380-1380.	1.1	3
88	Response to Simon et al.,. Acta Neuropathologica Communications, 2017, 5, 34.	2.4	0
89	The Effect of Neurological Genomics and Personalized Mitochondrial Medicine. JAMA Neurology, 2017, 74, 11.	4.5	2
90	Mutations in mitochondrial DNA causing tubulointerstitial kidney disease. PLoS Genetics, 2017, 13, e1006620.	1.5	52

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91	Background sequence characteristics influence the occurrence and severity of disease-causing mtDNA mutations. PLoS Genetics, 2017, 13, e1007126.	1.5	49
92	Opening One's Eyes to Mosaicism in Progressive External Ophthalmoplegia. Neurology: Genetics, 2017, 3, e202.	0.9	1
93	Novel HSPB1 mutation causes both motor neuronopathy and distal myopathy. Neurology: Genetics, 2016, 2, e110.	0.9	24
94	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
95	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
96	Homozygous deletion in MICU1 presenting with fatigue and lethargy in childhood. Neurology: Genetics, 2016, 2, e59.	0.9	86
97	Lamination of the Outer Plexiform Layer in Optic Atrophy Caused by Dominant WFS1 Mutations. Ophthalmology, 2016, 123, 1624-1626.	2.5	19
98	Emerging therapies for mitochondrial disorders. Brain, 2016, 139, 1633-1648.	3.7	59
99	Clinical, Genetic, and Radiological Features of Extrapyramidal Movement Disorders in Mitochondrial Disease. JAMA Neurology, 2016, 73, 668.	4.5	69
100	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
101	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
102	Cardiac involvement in hereditary myopathy with early respiratory failure. Neurology, 2016, 87, 1031-1035.	1.5	12
103	Phenotypic convergence of Menkes and Wilson disease. Neurology: Genetics, 2016, 2, e119.	0.9	18
104	Mitochondrial Matchmaking. New England Journal of Medicine, 2016, 375, 1894-1896.	13.9	8
105	A multiple sclerosisâ€like disorder in patients with OPA1 mutations. Annals of Clinical and Translational Neurology, 2016, 3, 723-729.	1.7	27
106	A Woman With Intellectual Disability, Amenorrhoea, Seizures, and Balance Problems. JAMA Neurology, 2016, 73, 1494.	4.5	1
107	Mitochondrial diseases. Nature Reviews Disease Primers, 2016, 2, 16080.	18.1	1,001
108	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

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109	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
110	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
111	Both mitochondrial DNA and mitonuclear gene mutations cause hearing loss through cochlear dysfunction. Brain, 2016, 139, e33-e33.	3.7	15
112	Somatic mtDNA variation is an important component of Parkinson's disease. Neurobiology of Aging, 2016, 38, 217.e1-217.e6.	1.5	61
113	Reply: Parsing the differences in affected with LHON: genetic versus environmental triggers of disease conversion. Brain, 2016, 139, e18-e18.	3.7	13
114	Exome sequencing in dementia with Lewy bodies. Translational Psychiatry, 2016, 6, e728-e728.	2.4	35
115	Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. Human Molecular Genetics, 2016, 25, 1031-1041.	1.4	53
116	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
117	Mitochondrial disease in adults: what's old and what's new?. EMBO Molecular Medicine, 2015, 7, 1503-1512.	3.3	110
118	Mitochondrial pathology in progressive cerebellar ataxia. Cerebellum and Ataxias, 2015, 2, 16.	1.9	37
119	Reduced cerebrospinal fluid mitochondrial DNA is a biomarker for earlyâ€stage Parkinson's disease. Annals of Neurology, 2015, 78, 1000-1004.	2.8	106
120	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
121	Nuclear-mitochondrial proteins: too much to process?: Figure 1. Brain, 2015, 138, 1451-1453.	3.7	2
122	Mitochondrial dysfunction in aging: Much progress but many unresolved questions. Biochimica Et Biophysica Acta - Bioenergetics, 2015, 1847, 1347-1353.	0.5	267
123	Prevalence of nuclear and mitochondrial <scp>DNA</scp> mutations related to adult mitochondrial disease. Annals of Neurology, 2015, 77, 753-759.	2.8	706
124	Frequency of rare recessive mutations in unexplained late onset cerebellar ataxia. Journal of Neurology, 2015, 262, 1822-1827.	1.8	20
125	A Unique Gene Regulatory Network Resets the Human Germline Epigenome for Development. Cell, 2015, 161, 1453-1467.	13.5	556
126	Mitochondrial DNA mutations in neurodegeneration. Biochimica Et Biophysica Acta - Bioenergetics, 2015, 1847, 1401-1411.	0.5	120

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127	Clonal Expansion of Secondary Mitochondrial DNA Deletions Associated With Spinocerebellar Ataxia Type 28. JAMA Neurology, 2015, 72, 106.	4.5	41
128	Reply: â€˜Behr syndromeâ€™ with OPA1 compound heterozygote mutations. Brain, 2015, 138, e322-e322.	3.7	0
129	Respiratory chain deficiency in nonmitochondrial disease. Neurology: Genetics, 2015, 1, e6.	0.9	23
130	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
131	Inherited mtDNA variations are not strong risk factors in human prion disease. Neurobiology of Aging, 2015, 36, 2908.e1-2908.e3.	1.5	0
132	Reply: Evaluation of exome sequencing variation in undiagnosed ataxias. Brain, 2015, 138, e384-e384.	3.7	2
133	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
134	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
135	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
136	Clinical and Pathological Features of Mitochondrial DNA Deletion Disease Following Antiretroviral Treatment. JAMA Neurology, 2015, 72, 603.	4.5	3
137	Extreme-Depth Re-sequencing of Mitochondrial DNA Finds No Evidence of Paternal Transmission in Humans. PLoS Genetics, 2015, 11, e1005040.	1.5	62
138	SPG7 mutations are a common cause of undiagnosed ataxia. Neurology, 2015, 84, 1174-1176.	1.5	87
139	Phenotypic variability of TRPV4 related neuropathies. Neuromuscular Disorders, 2015, 25, 516-521.	0.3	27
140	Modifying Mitochondrial tRNAs: Delivering What the Cell Needs. Cell Metabolism, 2015, 21, 351-352.	7.2	6
141	Clinical heterogeneity of primary familial brain calcification due to a novel mutation in PDGFB. Neurology, 2015, 84, 1818-1820.	1.5	14
142	Mitochondrial Causes of Epilepsy: Evaluation, Diagnosis, and Treatment. Seminars in Neurology, 2015, 35, 300-309.	0.5	8
143	The p.Ser107Leu inBICD2is a mutation â€˜hot spotâ€™ causing distal spinal muscular atrophy. Brain, 2015, 138, e391-e391.	3.7	13
144	Allogeneic haematopoietic stem cell transplantation for mitochondrial neurogastrointestinal encephalomyopathy. Brain, 2015, 138, 2847-2858.	3.7	128

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145	SCP2 mutations and neurodegeneration with brain iron accumulation. Neurology, 2015, 85, 1909-1911.	1.5	39
146	Recessive Mutations in RTN4IP1 Cause Isolated and Syndromic Optic Neuropathies. American Journal of Human Genetics, 2015, 97, 754-760.	2.6	54
147	The dynamics of mitochondrial DNA heteroplasmy: implications for human health and disease. Nature Reviews Genetics, 2015, 16, 530-542.	7.7	679
148	Exome sequencing in undiagnosed inherited and sporadic ataxias. Brain, 2015, 138, 276-283.	3.7	120
149	Prevalence of neurogenetic disorders in the North of England. Neurology, 2015, 85, 1195-1201.	1.5	26
150	Disturbed mitochondrial dynamics and neurodegenerative disorders. Nature Reviews Neurology, 2015, 11, 11-24.	4.9	533
151	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
152	Origins and functional consequences of somatic mitochondrial DNA mutations in human cancer. ELife, 2014, 3, .	2.8	318
153	Adult-onset Mendelian PEO Associated with Mitochondrial Disease. Journal of Neuromuscular Diseases, 2014, 1, 119-133.	1.1	19
154	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
155	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
156	Characterizing mild cognitive impairment in incident Parkinson disease. Neurology, 2014, 82, 308-316.	1.5	359
157	Treatment strategies for inherited optic neuropathies: past, present and future. Eye, 2014, 28, 521-537.	1.1	127
158	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
159	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
160	The Challenges of Mitochondrial Replacement. PLoS Genetics, 2014, 10, e1004315.	1.5	61
161	Recent Mitochondrial DNA Mutations Increase the Risk of Developing Common Late-Onset Human Diseases. PLoS Genetics, 2014, 10, e1004369.	1.5	131
162	Reply: Early-onset Behr syndrome due to compound heterozygous mutations in OPA1. Brain, 2014, 137, e302-e302.	3.7	6

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163	Population Screening for Variant Creutzfeldt-Jakob Disease Using a Novel Blood Test. JAMA Neurology, 2014, 71, 421.	4.5	51
164	EXOSC8 mutations alter mRNA metabolism and cause hypomyelination with spinal muscular atrophy and cerebellar hypoplasia. Nature Communications, 2014, 5, 4287.	5.8	120
165	Genetic impact on cognition and brain function in newly diagnosed Parkinsonâ€™s disease: ICICLE-PD study. Brain, 2014, 137, 2743-2758.	3.7	127
166	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
167	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
168	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
169	HIV Treatment and Associated Mitochondrial Pathology. Toxicologic Pathology, 2014, 42, 811-822.	0.9	90
170	Molecular pathogenesis of polymerase gammaâ€“related neurodegeneration. Annals of Neurology, 2014, 76, 66-81.	2.8	77
171	Efficient mitochondrial biogenesis drives incomplete penetrance in Leberâ€™s hereditary optic neuropathy. Brain, 2014, 137, 335-353.	3.7	229
172	Mutations in the SPG7 gene cause chronic progressive external ophthalmoplegia through disordered mitochondrial DNA maintenance. Brain, 2014, 137, 1323-1336.	3.7	151
173	Teaching Video Neuro Images : Muscle cramps and a raised creatine kinase. Neurology, 2014, 82, e220-1.	1.5	0
174	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
175	Valproic acid triggers increased mitochondrial biogenesis in POLG-deficient fibroblasts. Molecular Genetics and Metabolism, 2014, 112, 57-63.	0.5	40
176	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
177	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
178	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
179	Age-Related Mitochondrial DNA Depletion and the Impact on Pancreatic Beta Cell Function. PLoS ONE, 2014, 9, e115433.	1.1	44
180	Adult-onset Mendelian PEO Associated with Mitochondrial Disease. Journal of Neuromuscular Diseases, 2014, 1, 119-133.	1.1	9

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181	Dominant Optic Atrophy: Novel OPA1 Mutations and Revised Prevalence Estimates. Ophthalmology, 2013, 120, 1712-1712.e1.	2.5	57
182	Hereditary mtDNA Heteroplasmy: A Baseline for Aging?. Cell Metabolism, 2013, 18, 463-464.	7.2	44
183	A multi-center comparison of diagnostic methods for the biochemical evaluation of suspected mitochondrial disorders. Mitochondrion, 2013, 13, 36-43.	1.6	23
184	Clinical features of MS associated with Leber hereditary optic neuropathy mtDNA mutations. Neurology, 2013, 81, 2073-2081.	1.5	100
185	Universal heteroplasmy of human mitochondrial DNA. Human Molecular Genetics, 2013, 22, 384-390.	1.4	344
186	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
187	Multisystem fatal infantile disease caused by a novel homozygous EARS2 mutation. Brain, 2013, 136, e228-e228.	3.7	38
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