Joanna L Elson

List of Publications by Year
in descending order

77 papers	3,653 citations	186265 28 h-index	138484 58 g-index
81 all docs	81 docs citations	81 times ranked	4470 citing authors

#	Article	IF	Citations
1	Reduced-Median-Network Analysis of Complete Mitochondrial DNA Coding-Region Sequences for the Major African, Asian, and European Haplogroups. American Journal of Human Genetics, 2002, 70, 1152-1171.	6.2	482
2	Strong Purifying Selection in Transmission of Mammalian Mitochondrial DNA. PLoS Biology, 2008, 6, e10.	5.6	425
3	Mitochondrial DNA and survival after sepsis: a prospective study. Lancet, The, 2005, 366, 2118-2121.	13.7	162
4	Assigning pathogenicity to mitochondrial tRNA mutations: when â€˜definitely maybeâ€™ is not good enough. Trends in Genetics, 2004, 20, 591-596.	6.7	159
5	A comparative analysis approach to determining the pathogenicity of mitochondrial tRNA mutations. Human Mutation, 2011, 32, 1319-1325.	2.5	159
6	Purifying selection of mtDNA and its implications for understanding evolution and mitochondrial disease. Nature Reviews Genetics, 2008, 9, 657-662.	16.3	155
7	Mitochondrial tRNA mutations and disease. Wiley Interdisciplinary Reviews RNA, 2010, 1, 304-324.	6.4	145
8	Nature of Mitochondrial DNA Deletions in Substantia Nigra Neurons. American Journal of Human Genetics, 2008, 82, 228-235.	6.2	123
9	Clonal Expansion of Early to Mid-Life Mitochondrial DNA Point Mutations Drives Mitochondrial Dysfunction during Human Ageing. PLoS Genetics, 2014, 10, e1004620.	3.5	115
10	Cellular bioenergetics is impaired in patients with chronic fatigue syndrome. PLoS ONE, 2017, 12, e0186802.	2.5	106
11	Does the mitochondrial genome play a role in the etiology of Alzheimerâ€™s disease?. Human Genetics, 2006, 119, 241-254.	3.8	102
12	Deep-brain stimulation associates with improved microvascular integrity in the subthalamic nucleus in Parkinson's disease. Neurobiology of Disease, 2015, 74, 392-405.	4.4	86
13	mtDNA mutations and common neurodegenerative disorders. Trends in Genetics, 2005, 21, 583-586.	6.7	78
14	Mitochondrial DNA disease: new options for prevention. Human Molecular Genetics, 2011, 20, R168-R174.	2.9	72
15	Perceived fatigue is highly prevalent and debilitating in patients with mitochondrial disease. Neuromuscular Disorders, 2015, 25, 563-566.	0.6	67
16	Comparison of Mitochondrial Mutation Spectra in Ageing Human Colonic Epithelium and Disease: Absence of Evidence for Purifying Selection in Somatic Mitochondrial DNA Point Mutations. PLoS Genetics, 2012, 8, e1003082.	3.5	61
17	Mitochondrial DNA clonality in the dock: can surveillance swing the case?. Trends in Genetics, 2006, 22, 603-607.	6.7	56
18	Mitochondrial Abnormality Associates with Type-Specific Neuronal Loss and Cell Morphology Changes in the Pedunculopontine Nucleus in Parkinson Disease. American Journal of Pathology, 2013, 183, 1826-1840.	3.8	53

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19	Pathogenic mitochondrial tRNA mutations - Which mutations are inherited and why?. Human Mutation, 2009, 30, E984-E992.	2.5	49
20	An animal model mimicking pedunculopontine nucleus cholinergic degeneration in Parkinsonâ€™s disease. Brain Structure and Function, 2015, 220, 479-500.	2.3	49
21	Mitochondrial DNA changes in pedunculopontine cholinergic neurons in Parkinson disease. Annals of Neurology, 2017, 82, 1016-1021.	5.3	45
22	African Haplogroup L mtDNA Sequences Show Violations of Clock-like Evolution. Molecular Biology and Evolution, 2004, 21, 1843-1854.	8.9	43
23	Relationship Between Mitochondria and Î±-Synuclein. Archives of Neurology, 2012, 69, 385.	4.5	43
24	Pharmacogenetic stimulation of cholinergic pedunculopontine neurons reverses motor deficits in a rat model of Parkinsonâ€™s disease. Molecular Neurodegeneration, 2015, 10, 47.	10.8	41
25	Habitual Physical Activity in Mitochondrial Disease. PLoS ONE, 2011, 6, e22294.	2.5	37
26	The role of the mitochondrial ribosome in human disease: searching for mutations in 12S mitochondrial rRNA with high disruptive potential. Human Molecular Genetics, 2014, 23, 949-967.	2.9	35
27	Relative Rates of Evolution in the Coding and Control Regions of African mtDNAs. Molecular Biology and Evolution, 2007, 24, 2213-2221.	8.9	33
28	Characterization of mtDNA variation in a cohort of South African paediatric patients with mitochondrial disease. European Journal of Human Genetics, 2012, 20, 650-656.	2.8	30
29	Mitochondrial DNA as a Risk Factor for False Positives in Case-Control Association Studies. Journal of Genetics and Genomics, 2015, 42, 169-172.	3.9	30
30	The presence of highly disruptive 16S rRNA mutations in clinical samples indicates a wider role for mutations of the mitochondrial ribosome in human disease. Mitochondrion, 2015, 25, 17-27.	3.4	29
31	A proposed consensus panel of organisms for determining evolutionary conservation of mt-tRNA point mutations. Mitochondrion, 2012, 12, 533-538.	3.4	28
32	The low abundance of clonally expanded mitochondrial DNA point mutations in aged substantia nigra neurons. Aging Cell, 2009, 8, 496-498.	6.7	26
33	Aggregation, impaired degradation and immunization targeting of amyloid-beta dimers in Alzheimerâ€™s disease: a stochastic modelling approach. Molecular Neurodegeneration, 2012, 7, 32.	10.8	25
34	Design and evaluation of bi-functional iron chelators for protection of dopaminergic neurons from toxicants. Archives of Toxicology, 2020, 94, 3105-3123.	4.2	24
35	The effect of myalgic encephalomyelitis/chronic fatigue syndrome (ME/CFS) severity on cellular bioenergetic function. PLoS ONE, 2020, 15, e0231136.	2.5	24
36	Mitochondrial complex activity in permeabilised cells of chronic fatigue syndrome patients using two cell types. PeerJ, 2019, 7, e6500.	2.0	24

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37	MtDNA population variation in Myalgic encephalomyelitis/Chronic fatigue syndrome in two populations: a study of mildly deleterious variants. Scientific Reports, 2019, 9, 2914.	3.3	21
38	The length of cytochrome c oxidase-negative segments in muscle fibres in patients with mtDNA myopathy. Neuromuscular Disorders, 2002, 12, 858-864.	0.6	20
39	Using MutPred derived mtDNA load scores to evaluate mtDNA variation in hypertension and diabetes in a two-population cohort: The SABPA study. Journal of Genetics and Genomics, 2017, 44, 139-149.	3.9	20
40	The unresolved role of mitochondrial DNA in Parkinson's disease: An overview of published studies, their limitations, and future prospects. Neurochemistry International, 2019, 129, 104495.	3.8	19
41	MutPred mutational load analysis shows mildly deleterious mitochondrial DNA variants are not more prevalent in Alzheimer's patients, but may be under-represented in healthy older individuals. Mitochondrion, 2017, 34, 141-146.	3.4	18
42	Mitochondrial DNA sequence context in the penetrance of mitochondrial t-RNA mutations: A study across multiple lineages with diagnostic implications. PLoS ONE, 2017, 12, e0187862.	2.5	18
43	DREADD Activation of Pedunculopontine Cholinergic Neurons Reverses Motor Deficits and Restores Striatal Dopamine Signaling in Parkinsonian Rats. Neurotherapeutics, 2020, 17, 1120-1141.	4.4	18
44	Does mitochondrial DNA predispose to neuromyelitis optica (Devic's disease)?. Brain, 2008, 131, e93-e93.	7.6	17
45	Pedunculopontine cell loss and protein aggregation direct microglia activation in parkinsonian rats. Brain Structure and Function, 2016, 221, 2319-2341.	2.3	17
46	Associating Mitochondrial DNA Variation with Complex Traits. American Journal of Human Genetics, 2007, 80, 378-382.	6.2	16
47	A new approach to find biomarkers in chronic fatigue syndrome/myalgic encephalomyelitis (CFS/ME) by single-cell Raman micro-spectroscopy. Analyst, The, 2019, 144, 913-920.	3.5	16
48	Clinically proven mtDNA mutations are not common in those with chronic fatigue syndrome. BMC Medical Genetics, 2017, 18, 29.	2.1	15
49	The aetiology of cardiovascular disease: a role for mitochondrial DNA?. Cardiovascular Journal of Africa, 2018, 29, 122-132.	0.4	14
50	Mitochondrial DNA haplogroups and amyotrophic lateral sclerosis. Neurogenetics, 2007, 8, 65-67.	1.4	13
51	New mtDNA Association Model, MutPred Variant Load, Suggests Individuals With Multiple Mildly Deleterious mtDNA Variants Are More Likely to Suffer From Atherosclerosis. Frontiers in Genetics, 2018, 9, 702.	2.3	12
52	Panel-Based Nuclear and Mitochondrial Next-Generation Sequencing Outcomes of an Ethnically Diverse Pediatric Patient Cohort with Mitochondrial Disease. Journal of Molecular Diagnostics, 2019, 21, 503-513.	2.8	12
53	Testing the adaptive selection of human mtDNA haplogroups: an experimental bioenergetics approach. Biochemical Journal, 2007, 404, e3-5.	3.7	10
54	Haplogroup Context is Less Important in the Penetrance of Mitochondrial DNA Complex I Mutations Compared to mt-tRNA Mutations. Journal of Molecular Evolution, 2018, 86, 395-403.	1.8	10

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55	Mitochondrial DNA mutations induce mitochondrial biogenesis and increase the tumorigenic potential of Hodgkin and Reedâ€“Sternberg cells. Carcinogenesis, 2020, 41, 1735-1745.	2.8	10
56	Heterologous Inferential Analysis (HIA) as a Method to Understand the Role of Mitochondrial rRNA Mutations in Pathogenesis. Methods in Molecular Biology, 2015, 1264, 369-383.	0.9	10
57	Assessing cellular energy dysfunction in CFS/ME using a commercially available laboratory test. Scientific Reports, 2019, 9, 11464.	3.3	9
58	Toward a mtDNA locus-specific mutation database using the LOVD platform. Human Mutation, 2012, 33, 1352-1358.	2.5	8
59	Altered motor, anxiety-related and attentional task performance at baseline associate with multiple gene copies of the vesicular acetylcholine transporter and related protein overexpression in ChAT::Cre+ rats. Brain Structure and Function, 2019, 224, 3095-3116.	2.3	8
60	Substrate utilisation of cultured skeletal muscle cells in patients with CFS. Scientific Reports, 2020, 10, 18232.	3.3	8
61	Novel 1-hydroxypyridin-2-one metal chelators prevent and rescue ubiquitin proteasomal-related neuronal injury in an in vitro model of Parkinsonâ€™s disease. Archives of Toxicology, 2020, 94, 813-831.	4.2	8
62	Mitochondrial DNA variation in Parkinsonâ€™s disease: Analysis of â€œout-of-placeâ€•population variants as a risk factor. Frontiers in Aging Neuroscience, 0, 14, .	3.4	7
63	Raising Doubts about the Pathogenicity of Mitochondrial DNA Mutation m.3308T>C in Left Ventricular Hypertraveculation/Noncompaction. Cardiology, 2012, 122, 113-115.	1.4	6
64	Temporal-Spatial Profiling of Pedunculopontine Galanin-Cholinergic Neurons in the Lactacystin Rat Model of Parkinsonâ€™s Disease. Neurotoxicity Research, 2018, 34, 16-31.	2.7	6
65	The role of mitochondria in ME/CFS: a perspective. Fatigue: Biomedicine, Health and Behavior, 2019, 7, 52-58.	1.9	6
66	Call for participation in the neurogenetics consortium within the Human Variome Project. Neurogenetics, 2011, 12, 169-173.	1.4	5
67	What can a comparative genomics approach tell us about the pathogenicity of mtDNA mutations in human populations?. Evolutionary Applications, 2019, 12, 1912-1930.	3.1	5
68	A broad comparative genomics approach to understanding the pathogenicity of Complex I mutations. Scientific Reports, 2021, 11, 19578.	3.3	5
69	Could we offer mitochondrial donation or similar assisted reproductive technology to South African patients with mitochondrial DNA disease?. South African Medical Journal, 2016, 106, 234.	0.6	4
70	Heterologous Inferential Analysis (HIA) and Other Emerging Concepts: In Understanding Mitochondrial Variation In Pathogenesis: There is no More Low-Hanging Fruit. Methods in Molecular Biology, 2021, 2277, 203-245.	0.9	4
71	Older mothers are not at risk of having grandchildren with sporadic mtDNA deletions. Genetics in Medicine, 2010, 12, 313-314.	2.4	3
72	Implementing a new variant load model to investigate the role of mtDNA in oxidative stress and inflammation in a bi-ethnic cohort: the SABPA study. Mitochondrial DNA Part A: DNA Mapping, Sequencing, and Analysis, 2019, 30, 440-447.	0.7	3

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73	Evolutionary dissection of mtDNA hg H: a susceptibility factor for hypertrophic cardiomyopathy. Mitochondrial DNA Part A: DNA Mapping, Sequencing, and Analysis, 2020, 31, 238-244.	0.7	3
74	Investigation of the correlation between mildly deleterious mtDNA Variations and the clinical progression of multiple sclerosis. Multiple Sclerosis and Related Disorders, 2021, 53, 103055.	2.0	3
75	Mitochondrial DNA copy number is not associated with fatigue status in Primary SjÃ¶grenâ€™s Syndrome. Fatigue: Biomedicine, Health and Behavior, 2018, 6, 123-131.	1.9	2
76	Single-Cell Approaches for Studying the Role of Mitochondrial DNA in Neurodegenerative Disease. Methods in Molecular Biology, 2021, 2277, 299-329.	0.9	1
77	Common mtDNA Polymorphisms and Neurodegenerative Disorders. , 2012, , 63-78.		0