Grainne Gorman

List of Publications by Year
in descending order

110 papers	6,707 citations	81900 39 h-index	69250 77 g-index
112 all docs	112 docs citations	112 times ranked	7851 citing authors

#	Article	IF	Citations
1	Forecasting stroke-like episodes and outcomes in mitochondrial disease. Brain, 2022, 145, 542-554.	7.6	25
2	Natural History of Leigh Syndrome: A Study of Disease Burden and Progression. Annals of Neurology, 2022, 91, 117-130.	5.3	17
3	The application of Raman spectroscopy to the diagnosis of mitochondrial muscle disease: A preliminary comparison between fibre optic probe and microscope formats. Journal of Raman Spectroscopy, 2022, 53, 172-181.	2.5	5
4	Pathogenic SLC25A26 variants impair SAH transport activity causing mitochondrial disease. Human Molecular Genetics, 2022, 31, 2049-2062.	2.9	3
5	COVID-19â€“Related Outcomes in Primary Mitochondrial Diseases. Neurology, 2022, 98, 576-582.	1.1	7
6	Circulating small RNA signatures differentiate accurately the subtypes of muscular dystrophies: small-RNA next-generation sequencing analytics and functional insights. RNA Biology, 2022, 19, 507-518.	3.1	1
7	Rapid identification of human muscle disease with fibre optic Raman spectroscopy. Analyst, The, 2022, 147, 2533-2540.	3.5	9
8	RRM1 variants cause a mitochondrial DNA maintenance disorder via impaired de novo nucleotide synthesis. Journal of Clinical Investigation, 2022, 132, .	8.2	6
9	Neuromuscular Junction Abnormalities in Mitochondrial Disease. Neurology: Clinical Practice, 2021, 11, 97-104.	1.6	10
10	Comment on â€œA severe linezolidâ€induced rhabdomyolysis and lactic acidosis in Leigh syndromeâ€. Journal of Inherited Metabolic Disease, 2021, 44, 6-7.	3.6	2
11	POLRMT mutations impair mitochondrial transcription causing neurological disease. Nature Communications, 2021, 12, 1135.	12.8	21
12	Risk of cardiac manifestations in adult mitochondrial disease caused by nuclear genetic defects. Open Heart, 2021, 8, e001510.	2.3	3
13	Mitochondrial disease in adults: recent advances and future promise. Lancet Neurology, The, 2021, 20, 573-584.	10.2	96
14	miR-223-3p and miR-24-3p as novel serum-based biomarkers for myotonic dystrophy type 1. Molecular Therapy - Methods and Clinical Development, 2021, 23, 169-183.	4.1	6
15	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€” Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880.	27.0	352
16	Systematic review of cognitive deficits in adult mitochondrial disease. European Journal of Neurology, 2020, 27, 3-17.	3.3	17
17	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.	2.0	7
18	Activities of daily living in myotonic dystrophy type 1. Acta Neurologica Scandinavica, 2020, 141, 380-387.	2.1	7

#	Article	IF	Citations
19	Lewy body pathology is more prevalent in older individuals with mitochondrial disease than controls. Acta Neuropathologica, 2020, 139, 219-221.	7.7	11
20	Ultrasensitive deletion detection links mitochondrial DNA replication, disease, and aging. Genome Biology, 2020, 21, 248.	8.8	48
21	Mitochondrial Diseases: Hope for the Future. Cell, 2020, 181, 168-188.	28.9	243
22	Change over time in ability to perform activities of daily living in myotonic dystrophy type 1. Journal of Neurology, 2020, 267, 3235-3242.	3.6	3
23	Safety of drug use in patients with a primary mitochondrial disease: An international Delphiâ€based consensus. Journal of Inherited Metabolic Disease, 2020, 43, 800-818.	3.6	42
24	A study protocol for quantifying patient preferences in neuromuscular disorders: a case study of the IMI PREFER Project. Wellcome Open Research, 2020, 5, 253.	1.8	4
25	Instability of the mitochondrial alanyl-tRNA synthetase underlies fatal infantile-onset cardiomyopathy. Human Molecular Genetics, 2019, 28, 258-268.	2.9	19
26	Analyzing walking speeds with ankle and wrist worn accelerometers in a cohort with myotonic dystrophy. Disability and Rehabilitation, 2019, 41, 2972-2978.	1.8	13
27	Cognitive deficits in adult m.3243A>Gâ€and m.8344A>Gâ€related mitochondrial disease: importance of correcting for baseline intellectual ability. Annals of Clinical and Translational Neurology, 2019, 6, 826-836.	3.7	10
28	Pathogenic variants in MTâ€ATP6: A United Kingdomâ€“based mitochondrial disease cohort study. Annals of Neurology, 2019, 86, 310-315.	5.3	33
29	Mitochondrial Donation â€” Which Women Could Benefit?. New England Journal of Medicine, 2019, 380, 1971-1972.	27.0	25
30	Height as a Clinical Biomarker of Disease Burden in Adult Mitochondrial Disease. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 2057-2066.	3.6	19
31	Leigh syndrome caused by mutations in <scp>MTFMT</scp> is associated with a better prognosis. Annals of Clinical and Translational Neurology, 2019, 6, 515-524.	3.7	17
32	Disease burden of myotonic dystrophy type 1. Journal of Neurology, 2019, 266, 998-1006.	3.6	21
33	Genetic determinants of disease severity in the myotonic dystrophy type 1 OPTIMISTIC cohort. Neurology, 2019, 93, e995-e1009.	1.1	71
34	Consensus-based statements for the management of mitochondrial stroke-like episodes. Wellcome Open Research, 2019, 4, 201.	1.8	66
35	MT-ND5 Mutation Exhibits Highly Variable Neurological Manifestations at Low Mutant Load. EBioMedicine, 2018, 30, 86-93.	6.1	47
36	Pathological mechanisms underlying single largeâ€scale mitochondrial <scp>DNA</scp> deletions. Annals of Neurology, 2018, 83, 115-130.	5.3	42

#	Article	IF	Citations
37	Topoisomerase 3Î± Is Required for Decatenation and Segregation of Human mtDNA. Molecular Cell, 2018, 69, 9-23.e6.	9.7	102
38	Multifocal demyelinating motor neuropathy and hamartoma syndrome associated with a de novo PTEN mutation. Neurology, 2018, 90, e1842-e1848.	1.1	4
39	Phenotypic heterogeneity in m.3243A>G mitochondrial disease: The role of nuclear factors. Annals of Clinical and Translational Neurology, 2018, 5, 333-345.	3.7	102
40	Scientific and Ethical Issues in Mitochondrial Donation. New Bioethics, 2018, 24, 57-73.	1.1	25
41	Retrospective natural history of thymidine kinase 2 deficiency. Journal of Medical Genetics, 2018, 55, 515-521.	3.2	73
42	Sideroblastic anemia with myopathy secondary to novel, pathogenic missense variants in the YARS2 gene. Haematologica, 2018, 103, e564-e566.	3.5	5
43	Mitochondrial donation: from test tube to clinic. Lancet, The, 2018, 392, 1191-1192.	13.7	30
44	Skeletal muscle mitochondrial oxidative phosphorylation function in idiopathic pulmonary arterial hypertension: in vivo and in vitro study. Pulmonary Circulation, 2018, 8, 1-5.	1.7	10
45	Mutations in TOP3A Cause a Bloom Syndrome-like Disorder. American Journal of Human Genetics, 2018, 103, 221-231.	6.2	65
46	mt <scp>DNA</scp> heteroplasmy level and copy number indicate disease burden in m.3243A>G mitochondrial disease. EMBO Molecular Medicine, 2018, 10, .	6.9	199
47	Cognitive behavioural therapy with optional graded exercise therapy in patients with severe fatigue with myotonic dystrophy type 1: a multicentre, single-blind, randomised trial. Lancet Neurology, The, 2018, 17, 671-680.	10.2	95
48	Review: Central nervous system involvement in mitochondrial disease. Neuropathology and Applied Neurobiology, 2017, 43, 102-118.	3.2	42
49	Clinical Features, Molecular Heterogeneity, and Prognostic Implications in YARS2-Related Mitochondrial Myopathy. JAMA Neurology, 2017, 74, 686.	9.0	41
50	Novel reproductive technologies to prevent mitochondrial disease. Human Reproduction Update, 2017, 23, 501-519.	10.8	59
51	Decreased male reproductive success in association with mitochondrial dysfunction. European Journal of Human Genetics, 2017, 25, 1162-1164.	2.8	18
52	International Workshop:. Neuromuscular Disorders, 2017, 27, 1126-1137.	0.6	58
53	Pathophysiology of exercise intolerance in chronic diseases: the role of diminished cardiac performance in mitochondrial and heart failure patients. Open Heart, 2017, 4, e000632.	2.3	19
54	Measuring Habitual Physical Activity inÂNeuromuscular Disorders: A Systematic Review. Journal of Neuromuscular Diseases, 2017, 4, 25-52.	2.6	28

#	Article	IF	Citations
55	Opening One's Eyes to Mosaicism in Progressive External Ophthalmoplegia. Neurology: Genetics, 2017, 3, e202.	1.9	1
56	Clinical, Genetic, and Radiological Features of Extrapyramidal Movement Disorders in Mitochondrial Disease. JAMA Neurology, 2016, 73, 668.	9.0	69
57	Nutritional interventions in primary mitochondrial disorders: Developing an evidence base. Molecular Genetics and Metabolism, 2016, 119, 187-206.	1.1	41
58	Pathogenic mtDNA mutations causing mitochondrial myopathy: The need for muscle biopsy. Neurology: Genetics, 2016, 2, e82.	1.9	24
59	Reply. Annals of Neurology, 2016, 80, 314-314.	5.3	0
60	The Spectrum of Mitochondrial Ultrastructural Defects in Mitochondrial Myopathy. Scientific Reports, 2016, 6, 30610.	3.3	165
61	Pseudoâ€obstruction, stroke, and mitochondrial dysfunction: A lethal combination. Annals of Neurology, 2016, 80, 686-692.	5.3	40
62	Mitochondrial diseases. Nature Reviews Disease Primers, 2016, 2, 16080.	30.5	1,001
63	Solid organ transplantation in primary mitochondrial disease: Proceed with caution. Molecular Genetics and Metabolism, 2016, 118, 178-184.	1.1	55
64	Both mitochondrial DNA and mitonuclear gene mutations cause hearing loss through cochlear dysfunction. Brain, 2016, 139, e33-e33.	7.6	15
65	Sudden adult death syndrome in m.3243A>G-related mitochondrial disease: an unrecognized clinical entity in young, asymptomatic adults. European Heart Journal, 2016, 37, 2552-2559.	2.2	53
66	Extensive respiratory chain defects in inhibitory interneurones in patients with mitochondrial disease. Neuropathology and Applied Neurobiology, 2016, 42, 180-193.	3.2	43
67	Epilepsy in adults with mitochondrial disease: A cohort study. Annals of Neurology, 2015, 78, 949-957.	5.3	62
68	Preliminary Evaluation of Clinician Rated Outcome Measures in Mitochondrial Disease. Journal of Neuromuscular Diseases, 2015, 2, 151-155.	2.6	8
69	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.	2.6	22
70	Prevalence of nuclear and mitochondrial <scp>DNA</scp> mutations related to adult mitochondrial disease. Annals of Neurology, 2015, 77, 753-759.	5.3	706
71	Clonal Expansion of Secondary Mitochondrial DNA Deletions Associated With Spinocerebellar Ataxia Type 28. JAMA Neurology, 2015, 72, 106.	9.0	41
72	Adultâ€onset myoclonus ataxia associated with the mitochondrial m.8993<scp>T</scp>><scp>C</scp> â€œ<scp>NARP</scp>â€•mutation. Movement Disorders, 2015, 30, 1432-1433.	3.9	3

#	Article	IF	Citations
73	Mitochondrial Donation â€" How Many Women Could Benefit?. New England Journal of Medicine, 2015, 372, 885-887.	27.0	87
74	Novel MTND1 mutations cause isolated exercise intolerance, complex I deficiency and increased assembly factor expression. Clinical Science, 2015, 128, 895-904.	4.3	21
75	Perceived fatigue is highly prevalent and debilitating in patients with mitochondrial disease. Neuromuscular Disorders, 2015, 25, 563-566.	0.6	67
76	A CLINICAL AUDIT OF ACUTE MANAGEMENT OF STROKE-LIKE EPISODES FROM A NATIONAL MITOCHONDRIAL DISEASE CENTRE. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, e4.161-e4.	1.9	0
77	The diagnosis of posterior reversible encephalopathy syndrome. Lancet Neurology, The, 2015, 14, 1073.	10.2	5
78	The urinary proteome and metabonome differ from normal in adults with mitochondrial disease. Kidney International, 2015, 87, 610-622.	5.2	41
79	Adult-onset Mendelian PEO Associated with Mitochondrial Disease. Journal of Neuromuscular Diseases, 2014, 1, 119-133.	2.6	19
80	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.	7.4	304
81	Distal weakness with respiratory insufficiency caused by the m.8344A>G â€œMERRFâ€•mutation. Neuromuscular Disorders, 2014, 24, 533-536.	0.6	26
82	Orthostatic intolerance is common in chronic disease â€" A clinical cohort study. International Journal of Cardiology, 2014, 174, 861-863.	1.7	11
83	Discrete gait characteristics are associated with m.3243A>G and m.8344A>G variants of mitochondrial disease and its pathological consequences. Journal of Neurology, 2014, 261, 73-82.	3.6	11
84	Disease progression in patients with single, large-scale mitochondrial DNA deletions. Brain, 2014, 137, 323-334.	7.6	103
85	Mutations in the SPG7 gene cause chronic progressive external ophthalmoplegia through disordered mitochondrial DNA maintenance. Brain, 2014, 137, 1323-1336.	7.6	151
86	Adult-onset Mendelian PEO Associated with Mitochondrial Disease. Journal of Neuromuscular Diseases, 2014, 1, 119-133.	2.6	9
87	RRM2B-Related Mitochondrial Disease. , 2013, , 171-182.		3
88	Initial development and validation of a mitochondrial disease quality of life scale. Neuromuscular Disorders, 2013, 23, 324-329.	0.6	11
89	Defining cardiac adaptations and safety of endurance training in patients with m.3243A>G-related mitochondrial disease. International Journal of Cardiology, 2013, 168, 3599-3608.	1.7	43
90	Concentric hypertrophic remodelling and subendocardial dysfunction in mitochondrial DNA point mutation carriersâ€. European Heart Journal Cardiovascular Imaging, 2013, 14, 650-658.	1.2	30

#	Article	IF	Citations
91	Late-onset respiratory failure due to TK2 mutations causing multiple mtDNA deletions. Neurology, 2013, 81, 2051-2053.	1.1	23
92	Mitochondrial DNA deletions in muscle satellite cells: implications for therapies. Human Molecular Genetics, 2013, 22, 4739-4747.	2.9	33
93	Childhood presentation of â€œadultâ€•polyglucosan body disease. Annals of Neurology, 2013, 73, 317-318.	5.3	2
94	ANO5Gene Analysis in a Large Cohort of Patients with Anoctaminopathy: Confirmation of Male Prevalence and High Occurrence of the Common Exon 5 Gene Mutation. Human Mutation, 2013, 34, 1111-1118.	2.5	64
95	Extraocular Muscle Atrophy and Central Nervous System Involvement in Chronic Progressive External Ophthalmoplegia. PLoS ONE, 2013, 8, e75048.	2.5	27
96	Adult-onset cerebellar ataxia due to mutations inCABC1/ADCK3. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 174-178.	1.9	99
97	Adults with RRM2B-related mitochondrial disease have distinct clinical and molecular characteristics. Brain, 2012, 135, 3392-3403.	7.6	70
98	Diagnostic investigations of patients with chronic progressive external ophthalmoplegia. British Journal of Ophthalmology, 2012, 96, 1536.2-1536.	3.9	6
99	Cardiomyopathy is common in patients with the mitochondrial DNA m.3243A>G mutation and correlates with mutation load. Neuromuscular Disorders, 2012, 22, 592-596.	0.6	34
100	Mitochondrial DNA abnormalities in ophthalmological disease. Saudi Journal of Ophthalmology, 2011, 25, 395-404.	0.3	9
101	Habitual Physical Activity in Mitochondrial Disease. PLoS ONE, 2011, 6, e22294.	2.5	37
102	The phenotypic spectrum of neutral lipid storage myopathy due to mutations in the PNPLA2 gene. Journal of Neurology, 2011, 258, 1987-1997.	3.6	87
103	RRM2B mutations are frequent in familial PEO with multiple mtDNA deletions. Neurology, 2011, 76, 2032-2034.	1.1	59
104	The clinical, histochemical, and molecular spectrum of PEO1 (Twinkle)-linked adPEO. Neurology, 2010, 74, 1619-1626.	1.1	84
105	Older mothers are not at risk of having grandchildren with sporadic mtDNA deletions. Genetics in Medicine, 2010, 12, 313-314.	2.4	3
106	Multi-system neurological disease is common in patients with OPA1 mutations. Brain, 2010, 133, 771-786.	7.6	385
107	Clinical Reasoning: An unusual case of papilledema after orthotopic liver transplantation. Neurology, 2009, 73, e25-9.	1.1	0
108	Vertigo and vestibular abnormalities in spinocerebellar ataxia type 6. Journal of Neurology, 2009, 256, 78-82.	3.6	34

#	Article	IF	Citations
109	Generating hand dysaesthesiae: the "GHD phenomenon" - straight to the diagnosis. BMJ Case Reports, 2009, 2009, bcr0220091544-bcr0220091544.	0.5	0
110	How Can We Treat Mitochondrial Encephalomyopathies? Approaches to Therapy. Neurotherapeutics, 2008, 5, 558-568.	4.4	33