David R Lynch

List of Publications by Year
in descending order

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180 papers	15,794 citations	38738 50 h-index	17588 121 g-index
183 all docs	183 docs citations	183 times ranked	10667 citing authors

#	Article	IF	Citations
1	Anti-NMDA-receptor encephalitis: case series and analysis of the effects of antibodies. Lancet Neurology, The, 2008, 7, 1091-1098.	10.2	2,696
2	Paraneoplastic antiâ€“Nâ€methylâ€<scp>D</scp>â€aspartate receptor encephalitis associated with ovarian teratoma. Annals of Neurology, 2007, 61, 25-36.	5.3	2,166
3	Antiâ€“Nâ€methylâ€Dâ€aspartate receptor (NMDAR) encephalitis in children and adolescents. Annals of Neurology, 2009, 66, 11-18.	5.3	969
4	Cellular and Synaptic Mechanisms of Anti-NMDA Receptor Encephalitis. Journal of Neuroscience, 2010, 30, 5866-5875.	3.6	959
5	Antibody titres at diagnosis and during follow-up of anti-NMDA receptor encephalitis: a retrospective study. Lancet Neurology, The, 2014, 13, 167-177.	10.2	758
6	AMPA receptor antibodies in limbic encephalitis alter synaptic receptor location. Annals of Neurology, 2009, 65, 424-434.	5.3	712
7	N-methyl-D-aspartate Receptor Subtypes: Multiple Roles in Excitotoxicity and Neurological Disease. Neuroscientist, 2005, 11, 37-49.	3.5	313
8	ADCK3, an Ancestral Kinase, Is Mutated in a Form of Recessive Ataxia Associated with Coenzyme Q10 Deficiency. American Journal of Human Genetics, 2008, 82, 661-672.	6.2	290
9	Anti-NMDA Receptor Encephalitis Antibody Binding Is Dependent on Amino Acid Identity of a Small Region within the GluN1 Amino Terminal Domain. Journal of Neuroscience, 2012, 32, 11082-11094.	3.6	247
10	Mortality in Friedreich Ataxia. Journal of the Neurological Sciences, 2011, 307, 46-49.	0.6	236
11	A Phase 3, Double-blind, Placebo-Controlled Trial of Idebenone in Friedreich Ataxia. Archives of Neurology, 2010, 67, 941-7.	4.5	187
12	Idiopathic Pulmonary Fibrosis: A Genetic Disease That Involves Mucociliary Dysfunction of the Peripheral Airways. Physiological Reviews, 2016, 96, 1567-1591.	28.8	186
13	Excitotoxicity: Perspectives Based onN-Methyl-d-Aspartate Receptor Subtypes. Journal of Pharmacology and Experimental Therapeutics, 2002, 300, 717-723.	2.5	184
14	Selective alterations in glutamate and GABA receptor subunit mRNA expression in dysplastic neurons and giant cells of cortical tubers. Annals of Neurology, 2001, 49, 67-78.	5.3	158
15	Autosomal dominant â€œOpitzâ€GBBB syndrome due to a 22q11. 2 deletion. American Journal of Medical Genetics Part A, 1995, 59, 103-113.	2.4	152
16	Safety and Efficacy of Omaveloxolone in Friedreich Ataxia (<scp>MOXIe</scp> Study). Annals of Neurology, 2021, 89, 212-225.	5.3	128
17	Mitochondrial dysfunction in the development and progression of neurodegenerative diseases. Archives of Biochemistry and Biophysics, 2021, 702, 108698.	3.0	126
18	Idebenone in Friedreich ataxia cardiomyopathyâ€”results from a 6-month phase III study (IONIA). American Heart Journal, 2011, 161, 639-645.e1.	2.7	121

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19	Progression of Friedreich ataxia: quantitative characterization over 5 years. Annals of Clinical and Translational Neurology, 2016, 3, 684-694.	3.7	117
20	Compound heterozygous FXN mutations and clinical outcome in friedreich ataxia. Annals of Neurology, 2016, 79, 485-495.	5.3	115
21	High prevalence of <scp>NMDA</scp> receptor IgA/IgM antibodies in different dementia types. Annals of Clinical and Translational Neurology, 2014, 1, 822-832.	3.7	114
22	Friedreich Ataxia. Archives of Neurology, 2002, 59, 743-7.	4.5	110
23	Apelin, an endogenous neuronal peptide, protects hippocampal neurons against excitotoxic injury. Journal of Neurochemistry, 2007, 102, 1905-1917.	3.9	110
24	NMDA Receptor Pharmacology: Perspectives from Molecular Biology. Current Drug Targets, 2001, 2, 215-231.	2.1	110
25	Safety, pharmacodynamics, and potential benefit of omaveloxolone in Friedreich ataxia. Annals of Clinical and Translational Neurology, 2019, 6, 15-26.	3.7	105
26	Measuring the rate of progression in Friedreich ataxia: Implications for clinical trial design. Movement Disorders, 2010, 25, 426-432.	3.9	102
27	FXN methylation predicts expression and clinical outcome in Friedreich ataxia. Annals of Neurology, 2012, 71, 487-497.	5.3	101
28	Specific proteolysis of the NR2 subunit at multiple sites by calpain. Journal of Neurochemistry, 2001, 78, 1083-1093.	3.9	100
29	A rapid, noninvasive immunoassay for frataxin: Utility in assessment of Friedreich ataxia. Molecular Genetics and Metabolism, 2010, 101, 238-245.	1.1	91
30	Assessment of neurological efficacy of idebenone in pediatric patients with Friedreich's ataxia: data from a 6-month controlled study followed by a 12-month open-label extension study. Journal of Neurology, 2012, 259, 284-291.	3.6	88
31	Excision of Expanded GAA Repeats Alleviates the Molecular Phenotype of Friedreich's Ataxia. Molecular Therapy, 2015, 23, 1055-1065.	8.2	79
32	Fyn-mediated Phosphorylation of NR2B Tyr-1336 Controls Calpain-mediated NR2B Cleavage in Neurons and Heterologous Systems. Journal of Biological Chemistry, 2007, 282, 20075-20087.	3.4	76
33	Consensus clinical management guidelines for Friedreich ataxia. Orphanet Journal of Rare Diseases, 2014, 9, 184.	2.7	76
34	A0001 in Friedreich ataxia: Biochemical characterization and effects in a clinical trial. Movement Disorders, 2012, 27, 1026-1033.	3.9	75
35	Interactions of Postsynaptic Density-95 and the NMDA Receptor 2 Subunit Control Calpain-Mediated Cleavage of the NMDA Receptor. Journal of Neuroscience, 2004, 24, 11035-11045.	3.6	73
36	Axonal Î±7 nicotinic ACh receptors modulate presynaptic NMDA receptor expression and structural plasticity of glutamatergic presynaptic boutons. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 16661-16666.	7.1	67

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37	Anti-NMDA receptor encephalitis and nonencephalitic HSV-1 infection. Neurology: Neuroimmunology and NeuroInflammation, 2018, 5, e458.	6.0	67
38	Advancements in the pathophysiology of Friedreichâ€™s Ataxia and new prospects for treatments. Molecular Genetics and Metabolism, 2007, 92, 23-35.	1.1	65
39	Pharmacological therapeutics in Friedreich ataxia: the present state. Expert Review of Neurotherapeutics, 2017, 17, 895-907.	2.8	63
40	Antigenic and mechanistic characterization of antiâ€•<scp>AMPA</scp> receptor encephalitis. Annals of Clinical and Translational Neurology, 2014, 1, 180-189.	3.7	62
41	Anti-NMDA Receptor Encephalitis: Clinical Features and Basic Mechanisms. Advances in Pharmacology, 2018, 82, 235-260.	2.0	62
42	Double-blind, randomized and controlled trial of EPI-743 in Friedreich's ataxia. Neurodegenerative Disease Management, 2018, 8, 233-242.	2.2	62
43	Normal serum iron and ferritin concentrations in patients with Friedreich's ataxia. Annals of Neurology, 1998, 44, 132-134.	5.3	61
44	Pharmacological Characterization of Heterodimeric NMDA Receptors Composed of NR 1a and 2B Subunits: Differences with Receptors Formed from NR 1a and 2A. Journal of Neurochemistry, 2002, 64, 1462-1468.	3.9	59
45	NMDA receptor modulation by the neuropeptide apelin: implications for excitotoxic injury. Journal of Neurochemistry, 2011, 118, 1113-1123.	3.9	59
46	A gene expression phenotype in lymphocytes from friedreich ataxia patients. Annals of Neurology, 2011, 70, 790-804.	5.3	58
47	Psychometric properties of the Friedreich Ataxia Rating Scale. Neurology: Genetics, 2019, 5, 371.	1.9	57
48	Near infrared muscle spectroscopy in patients with Friedreich's ataxia. Muscle and Nerve, 2002, 25, 664-673.	2.2	56
49	Validation of Software Gating: A Practical Technology for Respiratory Motion Correction in PET. Radiology, 2016, 281, 239-248.	7.3	56
50	Analysis of the visual system in Friedreich ataxia. Journal of Neurology, 2013, 260, 2362-2369.	3.6	55
51	Cortical synaptic NMDA receptor deficits in Î±7 nicotinic acetylcholine receptor gene deletion models: Implications for neuropsychiatric diseases. Neurobiology of Disease, 2014, 63, 129-140.	4.4	55
52	Cortical parvalbumin GABAergic deficits with Î±7 nicotinic acetylcholine receptor deletion: implications for schizophrenia. Molecular and Cellular Neurosciences, 2014, 61, 163-175.	2.2	55
53	Frataxin levels in peripheral tissue in Friedreich ataxia. Annals of Clinical and Translational Neurology, 2015, 2, 831-842.	3.7	55
54	Early cerebellar deficits in mitochondrial biogenesis and respiratory chain complexes in the KIKO mouse model of Friedreich ataxia. DMM Disease Models and Mechanisms, 2017, 10, 1343-1352.	2.4	55

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55	Expanded GAA repeats impede transcription elongation through theFXNgene and induce transcriptional silencing that is restricted to theFXNlocus. Human Molecular Genetics, 2015, 24, ddv397.	2.9	54
56	Analysis of Echocardiograms in a Large Heterogeneous Cohort of Patients With Friedreich Ataxia. American Journal of Cardiology, 2012, 109, 401-405.	1.6	50
57	Lack of effect of polymorphisms in dopamine metabolism related genes on imaging of TRODAT-1 in striatum of asymptomatic volunteers and patients with Parkinson's disease. Movement Disorders, 2003, 18, 804-812.	3.9	48
58	Friedreich Ataxia Clinical Outcome Measures. Journal of Child Neurology, 2012, 27, 1152-1158.	1.4	48
59	Rating disease progression of Friedreichâ€™s ataxia by the International Cooperative Ataxia Rating Scale: analysis of a 603-patient database. Brain, 2013, 136, 259-268.	7.6	48
60	Somatic instability of the expanded GAA repeats in Friedreichâ€™s ataxia. PLoS ONE, 2017, 12, e0189990.	2.5	48
61	Characterization of Glutamate Binding Sites in Receptors Assembled from Transfected NMDA Receptor Subunits. Journal of Neurochemistry, 1996, 67, 608-616.	3.9	47
62	Increased serum transferrin receptor concentrations in Friedreich ataxia. Annals of Neurology, 2000, 47, 659-661.	5.3	46
63	Performance measures in Friedreich ataxia: Potential utility as clinical outcome tools. Movement Disorders, 2005, 20, 777-782.	3.9	46
64	Health related quality of life measures in Friedreich Ataxia. Journal of the Neurological Sciences, 2008, 272, 123-128.	0.6	46
65	Blood cells from Friedreich ataxia patients harbor frataxin deficiency without a loss of mitochondrial function. Mitochondrion, 2011, 11, 342-350.	3.4	44
66	D-Serine and Serine Racemase Are Associated with PSD-95 and Glutamatergic Synapse Stability. Frontiers in Cellular Neuroscience, 2016, 10, 34.	3.7	43
67	Therapeutic approaches for the treatment of Friedreichâ€™s ataxia. Expert Review of Neurotherapeutics, 2014, 14, 947-955.	2.8	41
68	Predictors of loss of ambulation in Friedreich's ataxia. EClinicalMedicine, 2020, 18, 100213.	7.1	40
69	Opposing Contributions of NR1 and NR2 to Protein Kinase C Modulation of NMDA Receptors. Journal of Neurochemistry, 1998, 71, 1471-1481.	3.9	39
70	Comprehensive analysis of gene expression patterns in Friedreich's ataxia fibroblasts by RNA sequencing reveals altered levels of protein synthesis factors and solute carriers. DMM Disease Models and Mechanisms, 2017, 10, 1353-1369.	2.4	38
71	Muscle oxidative phosphorylation quantitation using creatine chemical exchange saturation transfer (CrCEST) MRI in mitochondrial disorders. JCI Insight, 2016, 1, e88207.	5.0	38
72	Role of excitatory amino acids in developmental epilepsies. Mental Retardation and Developmental Disabilities Research Reviews, 2001, 7, 254-260.	3.6	37

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73	Liquid Chromatography-High Resolution Mass Spectrometry Analysis of Platelet Frataxin as a Protein Biomarker for the Rare Disease Friedreichâ€™s Ataxia. Analytical Chemistry, 2018, 90, 2216-2223.	6.5	37
74	Peripheral blood gene expression reveals an inflammatory transcriptomic signature in Friedreichâ€™s ataxia patients. Human Molecular Genetics, 2018, 27, 2965-2977.	2.9	36
75	miR-886-3p Levels Are Elevated in Friedreich Ataxia. Journal of Neuroscience, 2012, 32, 9369-9373.	3.6	35
76	Characterization of a new N-terminally acetylated extra-mitochondrial isoform of frataxin in human erythrocytes. Scientific Reports, 2018, 8, 17043.	3.3	35
77	Clinical data and characterization of the liver conditional mouse model exclude neoplasia as a non-neurological manifestation associated with Friedreich's ataxia. DMM Disease Models and Mechanisms, 2012, 5, 860-9.	2.4	34
78	Development of Frataxin Gene Expression Measures for the Evaluation of Experimental Treatments in Friedreichâ€™s Ataxia. PLoS ONE, 2013, 8, e63958.	2.5	34
79	New developments in pharmacotherapy for Friedreich ataxia. Expert Opinion on Pharmacotherapy, 2019, 20, 1855-1867.	1.8	34
80	Antioxidant use in Friedreich ataxia. Journal of the Neurological Sciences, 2008, 267, 174-176.	0.6	33
81	Antibodies to dendritic neuronal surface antigens in opsoclonus myoclonus ataxia syndrome. Journal of Neuroimmunology, 2015, 286, 86-92.	2.3	33
82	PPAR gamma agonist leriglitazone improves frataxin-loss impairments in cellular and animal models of Friedreich Ataxia. Neurobiology of Disease, 2021, 148, 105162.	4.4	33
83	Pediatric antiâ€<scp>NMDA</scp> receptor encephalitis is seasonal. Annals of Clinical and Translational Neurology, 2014, 1, 921-925.	3.7	32
84	Randomized, doubleâ€blind, placeboâ€controlled study of interferonâ€Î³ 1b in Friedreich Ataxia. Annals of Clinical and Translational Neurology, 2019, 6, 546-553.	3.7	32
85	Unanswered Questions in Friedreich Ataxia. Journal of Child Neurology, 2012, 27, 1223-1229.	1.4	31
86	Pharmacotherapy for Friedreich Ataxia. CNS Drugs, 2009, 23, 213-223.	5.9	29
87	Methylated and unmethylated epialleles support variegated epigenetic silencing in Friedreich ataxia. Human Molecular Genetics, 2021, 29, 3818-3829.	2.9	29
88	Ovarian failure in ataxia with oculomotor apraxia type 2. American Journal of Medical Genetics, Part A, 2007, 143A, 1775-1777.	1.2	28
89	Health-Related Quality of Life in Children With Friedreich Ataxia. Pediatric Neurology, 2010, 42, 335-337.	2.1	28
90	Role of frataxin protein deficiency and metabolic dysfunction in Friedreich ataxia, an autosomal recessive mitochondrial disease. Neuronal Signaling, 2018, 2, NS20180060.	3.2	28

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91	Contrast Letter Acuity as a Measure of Visual Dysfunction in Patients with Friedreich Ataxia. Journal of Neuro-Ophthalmology, 2002, 22, 270-274.	0.8	26
92	Cross-Sectional Analysis of Electrocardiograms in a Large Heterogeneous Cohort of Friedreich Ataxia Subjects. Journal of Child Neurology, 2012, 27, 1187-1192.	1.4	26
93	Stable isotopes and LCâ€“MS for monitoring metabolic disturbances in Friedreich's ataxia platelets. Bioanalysis, 2015, 7, 1843-1855.	1.5	26
94	Selected missense mutations impair frataxin processing in Friedreich ataxia. Annals of Clinical and Translational Neurology, 2017, 4, 575-584.	3.7	26
95	Central Nervous System Therapeutic Targets in Friedreich Ataxia. Human Gene Therapy, 2020, 31, 1226-1236.	2.7	26
96	Friedreich Ataxia: Multidisciplinary Clinical Care. Journal of Multidisciplinary Healthcare, 2021, Volume 14, 1645-1658.	2.7	26
97	Management and therapy for cardiomyopathy in Friedreichâ€™s ataxia. Expert Review of Cardiovascular Therapy, 2012, 10, 767-777.	1.5	25
98	Longitudinal Strain in Friedreich Ataxia: A Potential Marker for Early Left Ventricular Dysfunction. Echocardiography, 2014, 31, 50-57.	0.9	24
99	Early VGLUT1-specific parallel fiber synaptic deficits and dysregulated cerebellar circuit in the KIKO mouse model of Friedreich ataxia. DMM Disease Models and Mechanisms, 2017, 10, 1529-1538.	2.4	24
100	Modulation of the N-Methyl-d-Aspartate Receptor by Haloperidol: NR2B-Specific Interactions. Journal of Neurochemistry, 2002, 70, 2120-2128.	3.9	23
101	Urinary isoprostanes in Friedreich ataxia: Lack of correlation with disease features. Movement Disorders, 2008, 23, 1920-1922.	3.9	22
102	Natural History of Friedreich Ataxia. Neurology, 2022, 99, .	1.1	21
103	Practical Approaches to Neurogenetic Disease. Journal of Neuro-Ophthalmology, 2002, 22, 297-304.	0.8	20
104	Establishment and Maintenance of Primary Fibroblast Repositories for Rare Diseasesâ€”Friedreich's Ataxia Example. Biopreservation and Biobanking, 2016, 14, 324-329.	1.0	20
105	The current state of biomarker research for Friedreichâ€™s ataxia: a report from the 2018 FARA biomarker meeting. Future Science OA, 2019, 5, FSO398.	1.9	20
106	Human platelets as a platform to monitor metabolic biomarkers using stable isotopes and LCâ€“MS. Bioanalysis, 2013, 5, 3009-3021.	1.5	19
107	Glutamatergic autoencephalitides: an emerging field. Journal of Neural Transmission, 2014, 121, 957-968.	2.8	19
108	Compound heterozygote mutations in SPG7 in a family with adult-onset primary lateral sclerosis. Neurology: Genetics, 2016, 2, e60.	1.9	19

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109	Mitochondrial and metabolic dysfunction in Friedreich ataxia: update on pathophysiological relevance and clinical interventions. Neuronal Signaling, 2021, 5, NS20200093.	3.2	19
110	The attitude of patients with progressive ataxias towards clinical trials. Orphanet Journal of Rare Diseases, 2022, 17, 1.	2.7	19
111	Subjective Improvement in Proprioception in 2 Patients With Atypical Friedreich Ataxia Treated With Varenicline (Chantix). Journal of Clinical Neuromuscular Disease, 2009, 10, 191-193.	0.7	18
112	Health related quality of life in Friedreich Ataxia in a large heterogeneous cohort. Journal of the Neurological Sciences, 2020, 410, 116642.	0.6	18
113	Extra-mitochondrial mouse frataxin and its implications for mouse models of Friedreichâ€™s ataxia. Scientific Reports, 2020, 10, 15788.	3.3	17
114	Friedreich ataxia: clinical features and new developments. Neurodegenerative Disease Management, 2022, 12, 267-283.	2.2	17
115	Clinical measures of dysarthria in Friedreich Ataxia. Movement Disorders, 2010, 25, 108-111.	3.9	16
116	Elevation of serum cardiac troponin I in a cross-sectional cohort of asymptomatic subjects with Friedreich ataxia. International Journal of Cardiology, 2013, 167, 1622-1624.	1.7	16
117	Cross-sectional analysis of glucose metabolism in Friedreich Ataxia. Journal of the Neurological Sciences, 2014, 342, 29-35.	0.6	16
118	Cardiac transplantation in Friedreich Ataxia: Extended follow-up. Journal of the Neurological Sciences, 2017, 375, 471-473.	0.6	16
119	Impact of diabetes in the Friedreich ataxia clinical outcome measures study. Annals of Clinical and Translational Neurology, 2017, 4, 622-631.	3.7	16
120	Longitudinal analysis of contrast acuity in Friedreich ataxia. Neurology: Genetics, 2018, 4, e250.	1.9	15
121	Common data elements for clinical research in Friedreich's ataxia. Movement Disorders, 2013, 28, 190-195.	3.9	14
122	Usefulness of frataxin immunoassays for the diagnosis of Friedreich ataxia. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 994-1002.	1.9	14
123	Effects of genetic severity on glucose homeostasis in Friedreich ataxia. Muscle and Nerve, 2016, 54, 887-894.	2.2	14
124	Picking Up the Pieces: The Roles of Functional Remnants of Calpain-Mediated Proteolysis. Neuron, 2007, 53, 317-319.	8.1	13
125	IFN-Î³ for Friedreich ataxia: present evidence. Neurodegenerative Disease Management, 2015, 5, 497-504.	2.2	13
126	Drp1â€dependent peptide reverse mitochondrial fragmentation, a homeostatic response in Friedreich ataxia. Pharmacology Research and Perspectives, 2021, 9, e00755.	2.4	13

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127	Low apolipoprotein A-I levels in Friedreichâ€™s ataxia and in frataxin-deficient cells: Implications for therapy. PLoS ONE, 2018, 13, e0192779.	2.5	13
128	Pediatric Ataxia: Focus on Chronic Disorders. Seminars in Pediatric Neurology, 2018, 25, 54-64.	2.0	12
129	Testâ€“retest reliability of the Friedreichâ€™s ataxia rating scale. Annals of Clinical and Translational Neurology, 2020, 7, 1708-1712.	3.7	12
130	Omaveloxolone: potential new agent for Friedreich ataxia. Neurodegenerative Disease Management, 2021, 11, 91-98.	2.2	12
131	Functional NMDA receptors are expressed by human pulmonary artery smooth muscle cells. Scientific Reports, 2021, 11, 8205.	3.3	12
132	Pregnancy with Friedreich ataxia: a retrospective review of medical risks and psychosocial implications. American Journal of Obstetrics and Gynecology, 2010, 203, 224.e1-224.e5.	1.3	11
133	GRP75 overexpression rescues frataxin deficiency and mitochondrial phenotypes in Friedreich ataxia cellular models. Human Molecular Genetics, 2019, 28, 1594-1607.	2.9	11
134	Scoliosis in Friedreich's ataxia: longitudinal characterization in a large heterogeneous cohort. Annals of Clinical and Translational Neurology, 2021, 8, 1239-1250.	3.7	11
135	Digital endpoints for selfâ€administered homeâ€based functional assessment in pediatric Friedreichâ€™s ataxia. Annals of Clinical and Translational Neurology, 2021, 8, 1845-1856.	3.7	11
136	High-Throughput Immunoassay for the Biochemical Diagnosis of Friedreich Ataxia in Dried Blood Spots and Whole Blood. Clinical Chemistry, 2013, 59, 1461-1469.	3.2	10
137	Neurofilament light chain as a potential biomarker of disease status in Friedreich ataxia. Journal of Neurology, 2020, 267, 2594-2598.	3.6	10
138	Targeting 3â€² and 5â€² untranslated regions with antisense oligonucleotides to stabilize frataxin mRNA and increase protein expression. Nucleic Acids Research, 2021, 49, 11560-11574.	14.5	10
139	Designing phase II clinical trials in Friedreich ataxia. Expert Opinion on Emerging Drugs, 2021, 26, 415-423.	2.4	10
140	Skin fibroblast metabolomic profiling reveals that lipid dysfunction predicts the severity of Friedreichâ€™s ataxia. Journal of Lipid Research, 2022, 63, 100255.	4.2	10
141	Friedreich Ataxia and nephrotic syndrome: a series of two patients. BMC Neurology, 2016, 16, 3.	1.8	9
142	Clinical trial design for Friedreich ataxia - Where are we now and what do we need?. Expert Opinion on Orphan Drugs, 2018, 6, 219-230.	0.8	9
143	A Comprehensive Transcriptome Analysis Identifies FXN and BDNF as Novel Targets of miRNAs in Friedreichâ€™s Ataxia Patients. Molecular Neurobiology, 2020, 57, 2639-2653.	4.0	9
144	Results of a randomized doubleâ€blind study evaluating luvadaxistat in adults with Friedreich ataxia. Annals of Clinical and Translational Neurology, 2021, 8, 1343-1352.	3.7	9

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145	Malignant oligodendroglioma arising after radiation therapy for lymphoma. Medical and Pediatric Oncology, 1994, 22, 45-52.	1.0	8
146	Openâ€label pilot study of oral methylprednisolone for the treatment of patients with friedreich ataxia. Muscle and Nerve, 2019, 60, 571-575.	2.2	8
147	Neuronal serine racemase associates with Disrupted-In-Schizophrenia-1 and DISC1 agglomerates: Implications for schizophrenia. Neuroscience Letters, 2019, 692, 107-114.	2.1	8
148	Ectopic Burden via Holter Monitors in Friedreich Ataxia. Pediatric Neurology, 2021, 117, 29-33.	2.1	8
149	Comparison of neutrophil to lymphocyte ratio and prognostic nutritional index with other clinical and molecular biomarkers for prediction of glioblastoma multiforme outcome. PLoS ONE, 2021, 16, e0252614.	2.5	8
150	Friedreichâ€™s Ataxia related Diabetes: Epidemiology and management practices. Diabetes Research and Clinical Practice, 2022, 186, 109828.	2.8	8
151	Frataxin deficiency lowers lean mass and triggers the integrated stress response in skeletal muscle. JCI Insight, 2022, 7, .	5.0	8
152	Novel Diagnostic Paradigms for Friedreich Ataxia. Journal of Child Neurology, 2012, 27, 1146-1151.	1.4	7
153	DNA methylation in Friedreich ataxia silences expression of frataxin isoform E. Scientific Reports, 2022, 12, 5031.	3.3	7
154	Simultaneous Quantification of Mitochondrial Mature Frataxin and Extra-Mitochondrial Frataxin Isoform E in Friedreichâ€™s Ataxia Blood. Frontiers in Neuroscience, 2022, 16, 874768.	2.8	7
155	Clinical Monitoring in a Patient With Friedreich Ataxia and Osteogenic Sarcoma. Journal of Child Neurology, 2012, 27, 1159-1163.	1.4	6
156	<scp>F</scp>riedreich Ataxia: New findings, new challenges. Annals of Neurology, 2014, 76, 487-488.	5.3	6
157	Comorbid Medical Conditions in Friedreich Ataxia. Journal of Child Neurology, 2016, 31, 1161-1165.	1.4	6
158	Reverse Phase Protein Array Reveals Correlation of Retinoic Acid Metabolism With Cardiomyopathy in Friedreich's Ataxia. Molecular and Cellular Proteomics, 2021, 20, 100094.	3.8	6
159	Epigenetic Heterogeneity in Friedreich Ataxia Underlies Variable FXN Reactivation. Frontiers in Neuroscience, 2021, 15, 752921.	2.8	6
160	Clinical Evidence for Variegated Silencing in Patients With Friedreich Ataxia. Neurology: Genetics, 2022, 8, .	1.9	6
161	Impact of Mobility Device Use on Quality of Life in Children With Friedreich Ataxia. Journal of Child Neurology, 2018, 33, 397-404.	1.4	5
162	Identification of a novel missense mutation in Friedreich's ataxia â€“<scp>FXN^W</scp>^{168R}. Annals of Clinical and Translational Neurology, 2019, 6, 812-816.	3.7	5

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163	Cardiac Dysfunction Exacerbated by Endocrinopathies in Friedreich Ataxia. Journal of Child Neurology, 2012, 27, 1316-1319.	1.4	4
164	Nicotinamide in Friedreich's ataxia: useful or not?. Lancet, The, 2014, 384, 474-475.	13.7	4
165	Evaluation of antibodies for western blot analysis of frataxin protein isoforms. Journal of Immunological Methods, 2019, 474, 112629.	1.4	4
166	Correlation of Visual Quality of Life With Clinical and Visual Status in Friedreich Ataxia. Journal of Neuro-Ophthalmology, 2020, 40, 213-217.	0.8	4
167	The Role of Serum Levels of Neurofilament Light (NfL) Chain as a Biomarker in Friedreich Ataxia. Frontiers in Neuroscience, 2021, 15, 653241.	2.8	4
168	Cerebellar Pathology in an Inducible Mouse Model of Friedreich Ataxia. Frontiers in Neuroscience, 2022, 16, 819569.	2.8	4
169	Body Mass Index and Height in the Friedreich Ataxia Clinical Outcome Measures Study. Neurology: Genetics, 2021, 7, e638.	1.9	3
170	Friedreich's ataxia: the European consortium. Lancet Neurology, The, 2015, 14, 130-131.	10.2	2
171	Challenges ahead for trials in Friedreichâ€™s ataxia. Lancet Neurology, The, 2016, 15, 1300-1301.	10.2	2
172	Etravirine in Friedreich's ataxia: Lessons from HIV?. Movement Disorders, 2019, 34, 305-306.	3.9	2
173	Increased serum transferrin receptor concentrations in Friedreich ataxia. , 2000, 47, 659.		2
174	In vivo assessment of OXPHOS capacity using 3ÂT CrCEST MRI in Friedreichâ€™s ataxia. Journal of Neurology, 2022, 269, 2527-2538.	3.6	2
175	Efficacy and safety of idebenone in the treatment of Friedreich ataxia: a review of early results and future prospects. Future Neurology, 2008, 3, 375-384.	0.5	1
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