Massimo Pandolfo

List of Publications by Year
in descending order

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202 papers	16,948 citations	17440 63 h-index	16650 123 g-index
215 all docs	215 docs citations	215 times ranked	19110 citing authors

#	Article	IF	Citations
1	Recessive cerebellar and afferent ataxias â€" clinical challenges and future directions. Nature Reviews Neurology, 2022, 18, 257-272.	10.1	12
2	DNA methylation in Friedreich ataxia silences expression of frataxin isoform E. Scientific Reports, 2022, 12, 5031.	3.3	7
3	Hand Dexterity and Pyramidal Dysfunction in Friedreich Ataxia, A Finger Tapping Study. Movement Disorders Clinical Practice, 2021, 8, 85-91.	1.5	4
4	Neurology: Genetics Year in Review. Neurology: Genetics, 2021, 7, e556.	1.9	0
5	Research priorities for rare neurological diseases: a representative view of patient representatives and healthcare professionals from the European Reference Network for Rare Neurological Diseases. Orphanet Journal of Rare Diseases, 2021, 16, 135.	2.7	5
6	Progression characteristics of the European Friedreich's Ataxia Consortium for Translational Studies (EFACTS): a 4-year cohort study. Lancet Neurology, The, 2021, 20, 362-372.	10.2	53
7	Age of onset modulates restingâ€state brain network dynamics in Friedreich Ataxia. Human Brain Mapping, 2021, 42, 5334-5344.	3.6	8
8	Primary proprioceptive neurons from human induced pluripotent stem cells: a cell model for afferent ataxias. Scientific Reports, 2020, 10, 7752.	3.3	24
9	Central Nervous System Therapeutic Targets in Friedreich Ataxia. Human Gene Therapy, 2020, 31, 1226-1236.	2.7	26
10	Onset features and time to diagnosis in Friedreichâ€™s Ataxia. Orphanet Journal of Rare Diseases, 2020, 15, 198.	2.7	27
11	Testâ€"retest reliability of the Friedreichâ€™s ataxia rating scale. Annals of Clinical and Translational Neurology, 2020, 7, 1708-1712.	3.7	12
12	Cerebellar ataxia, neuropathy, hearing loss, and intellectual disability due to AIFM1 mutation. Neurology: Genetics, 2020, 6, e420.	1.9	10
13	Cerebellar cognitive disorder parallels cerebellar motor symptoms in Friedreich ataxia. Annals of Clinical and Translational Neurology, 2020, 7, 1050-1054.	3.7	32
14	Friedreich ataxia. , 2020, , 99-112.		0
15	Age of onset determines intrinsic functional brain architecture in Friedreich ataxia. Annals of Clinical and Translational Neurology, 2020, 7, 94-104.	3.7	21
16	Neurologic outcomes in Friedreich ataxia. Neurology: Genetics, 2020, 6, e415.	1.9	27
17	Exenatide induces frataxin expression and improves mitochondrial function in Friedreich ataxia. JCI Insight, 2020, 5, .	5.0	39
18	Protocol of a randomized, double-blind, placebo-controlled, parallel-group, multicentre study of the efficacy and safety of nicotinamide in patients with Friedreich ataxia (NICOFA). Neurological Research and Practice, 2019, 1, 33.	2.0	14

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19	Altered neocortical tactile but preserved auditory early change detection responses in Friedreich ataxia. Clinical Neurophysiology, 2019, 130, 1299-1310.	1.5	13
20	Prediction of Survival With Longâ€Term Disease Progression in Most Common Spinocerebellar Ataxia. Movement Disorders, 2019, 34, 1220-1227.	3.9	14
21	Evidence for genetically determined degeneration of proprioceptive tracts in Friedreich ataxia. Neurology, 2019, 93, e116-e124.	1.1	30
22	Rating scales for rare neurological diseases. Neurology: Genetics, 2019, 5, e380.	1.9	2
23	Genetic architecture of subcortical brain structures in 38,851 individuals. Nature Genetics, 2019, 51, 1624-1636.	21.4	192
24	Friedreich and dominant ataxias: quantitative differences in cerebellar dysfunction measurements. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 559-565.	1.9	16
25	2017 Year in Review and Message from the Editors to Our Reviewers. Neurology: Genetics, 2018, 4, e221.	1.9	0
26	Siponimod versus placebo in secondary progressive multiple sclerosis (EXPAND): a double-blind, randomised, phase 3 study. Lancet, The, 2018, 391, 1263-1273.	13.7	684
27	Survival in patients with spinocerebellar ataxia types 1, 2, 3, and 6 (EUROSCA): a longitudinal cohort study. Lancet Neurology, The, 2018, 17, 327-334.	10.2	69
28	Effect of natalizumab on disease progression in secondary progressive multiple sclerosis (ASCEND): a phase 3, randomised, double-blind, placebo-controlled trial with an open-label extension. Lancet Neurology, The, 2018, 17, 405-415.	10.2	238
29	Personality and Neuropsychological Profiles in Friedreich Ataxia. Cerebellum, 2018, 17, 204-212.	2.5	21
30	Automated functional upper limb evaluation of patients with Friedreich ataxia using serious games rehabilitation exercises. Journal of NeuroEngineering and Rehabilitation, 2018, 15, 87.	4.6	22
31	Low Prevalence Estimates of Late-Onset Glycogen Storage Disease Type II in French-Speaking Belgium are not Due to Missed Diagnoses. Journal of Neuromuscular Diseases, 2018, 5, 471-480.	2.6	1
32	Long-term evolution of patient-reported outcome measures in spinocerebellar ataxias. Journal of Neurology, 2018, 265, 2040-2051.	3.6	34
33	2016 in Review and Message from the Editors to our Reviewers. Neurology: Genetics, 2017, 3, e132.	1.9	0
34	Erythropoietin and small molecule agonists of the tissue-protective erythropoietin receptor increase FXN expression in neuronal cells inÂvitro and in Fxn-deficient KIKO mice inÂvivo. Neuropharmacology, 2017, 123, 34-45.	4.1	8
35	HLA genotype as a marker of multiple sclerosis prognosis: A pilot study. Journal of the Neurological Sciences, 2017, 375, 348-354.	0.6	8
36	A recessive ataxia diagnosis algorithm for the next generation sequencing era. Annals of Neurology, 2017, 82, 892-899.	5.3	27

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37	Body Mass Index Decline Is Related to Spinocerebellar Ataxia Disease Progression. Movement Disorders Clinical Practice, 2017, 4, 689-697.	1.5	25
38	Neurodegeneration With Brain Iron Accumulation (NBIA) Syndromes Presenting With Late-Onset Craniocervical Dystonia: An Illustrative Case Seriesâ€Ž. Movement Disorders Clinical Practice, 2017, 4, 254-257.	1.5	6
39	Urinary, bowel and sexual symptoms in a cohort of patients with Friedreichâ€™s ataxia. Orphanet Journal of Rare Diseases, 2017, 12, 158.	2.7	15
40	This variant alters protein function, but is it pathogenic?. Neurology: Genetics, 2017, 3, e173.	1.9	3
41	Friedreich ataxia-induced pluripotent stem cell-derived neurons show a cellular phenotype that is corrected by a benzamide HDAC inhibitor. Human Molecular Genetics, 2016, 25, ddw308.	2.9	46
42	Novel genetic loci underlying human intracranial volume identified through genome-wide association. Nature Neuroscience, 2016, 19, 1569-1582.	14.8	213
43	Delayedâ€onset Friedreich's ataxia revisited. Movement Disorders, 2016, 31, 62-69.	3.9	54
44	MME mutation in dominant spinocerebellar ataxia with neuropathy (SCA43). Neurology: Genetics, 2016, 2, e94.	1.9	41
45	Pharmacological treatments for Friedreich ataxia. The Cochrane Library, 2016, 2016, CD007791.	2.8	44
46	Progression characteristics of the European Friedreichâ€™s Ataxia Consortium for Translational Studies (EFACTS): a 2 year cohort study. Lancet Neurology, The, 2016, 15, 1346-1354.	10.2	117
47	Standardized Assessment of Hereditary Ataxia Patients in Clinical Studies. Movement Disorders Clinical Practice, 2016, 3, 230-240.	1.5	13
48	URINARY, BOWEL AND SEXUAL FUNCTION IN PATIENTS WITH FRIEDREICH'S ATAXIA. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, e4.167-e4.	1.9	0
49	Adult neural precursor cells form connexin-dependent networks that improve their survival. NeuroReport, 2015, 26, 928-936.	1.2	12
50	Unveiling a common mechanism of apoptosis in Î²-cells and neurons in Friedreich's ataxia. Human Molecular Genetics, 2015, 24, 2274-2286.	2.9	58
51	Building bridges between neuroscientific evidence and policy. Lancet Neurology, The, 2015, 14, 242-245.	10.2	5
52	Biological and clinical characteristics of the European Friedreich's Ataxia Consortium for Translational Studies (EFACTS) cohort: a cross-sectional analysis of baseline data. Lancet Neurology, The, 2015, 14, 174-182.	10.2	159
53	Common genetic variants influence human subcortical brain structures. Nature, 2015, 520, 224-229.	27.8	772
54	Friedreich Ataxia. , 2015, , 833-843.		1

#	Article	IF	Citations
55	Animal Models of Friedreich Ataxia. , 2015, , 1017-1024.		1
56	Friedreich Ataxia. , 2015, , 984-1002.		0
57	A 22-Year Follow-up Study of Long-term Cardiac Outcome and Predictors of Survival in Friedreich Ataxia. JAMA Neurology, 2015, 72, 1334.	9.0	69
58	Long-term disease progression in spinocerebellar ataxia types 1, 2, 3, and 6: a longitudinal cohort study. Lancet Neurology, The, 2015, 14, 1101-1108.	10.2	213
59	Consensus clinical management guidelines for Friedreich ataxia. Orphanet Journal of Rare Diseases, 2014, 9, 184.	2.7	76
60	Transient <scp>CNS</scp> Deficits and Migrainous Auras in Individuals Without a History of Headache. Headache, 2014, 54, 493-499.	3.9	7
61	The ENIGMA Consortium: large-scale collaborative analyses of neuroimaging and genetic data. Brain Imaging and Behavior, 2014, 8, 153-182.	2.1	696
62	Epigenetic therapy for <scp>F</scp>riedreich ataxia. Annals of Neurology, 2014, 76, 489-508.	5.3	128
63	Modulation of the age at onset in spinocerebellar ataxia by CAG tracts in various genes. Brain, 2014, 137, 2444-2455.	7.6	144
64	Deferiprone in <scp>F</scp>riedreich ataxia: A 6â€Month randomized controlled trial. Annals of Neurology, 2014, 76, 509-521.	5.3	109
65	Characteristics and Outcomes of Patients With Multiple Cervical Artery Dissection. Stroke, 2014, 45, 37-41.	2.0	96
66	A new locus for familial temporal lobe epilepsy on chromosome 3q. Epilepsy Research, 2013, 106, 338-344.	1.6	17
67	Mutations in DEPDC5 cause familial focal epilepsy with variable foci. Nature Genetics, 2013, 45, 546-551.	21.4	301
68	150Âyears of Friedreich Ataxia: from its discovery to therapy. Journal of Neurochemistry, 2013, 126, 1-3.	3.9	17
69	Genotype-specific patterns of atrophy progression are more sensitive than clinical decline in SCA1, SCA3 and SCA6. Brain, 2013, 136, 905-917.	7.6	128
70	Epilepsy, hippocampal sclerosis and febrile seizures linked by common genetic variation around SCN1A. Brain, 2013, 136, 3140-3150.	7.6	168
71	A novel function of Ataxin-1 in the modulation of PP2A activity is dysregulated in the spinocerebellar ataxia type 1. Human Molecular Genetics, 2013, 22, 3425-3437.	2.9	15
72	Cerebellar and Afferent Ataxias. CONTINUUM Lifelong Learning in Neurology, 2013, 19, 1312-1343.	0.8	39

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73	Pediatric epilepsy genetics. Current Opinion in Neurology, 2013, 26, 137-145.	3.6	17
74	Neurons and cardiomyocytes derived from induced pluripotent stem cells as a model for mitochondrial defects in Friedreich's ataxia. DMM Disease Models and Mechanisms, 2013, 6, 608-21.	2.4	142
75	Treatment of Friedreich's ataxia. Expert Opinion on Orphan Drugs, 2013, 1, 221-234.	0.8	7
76	Deferiprone for the treatment of Friedreich's ataxia. Journal of Neurochemistry, 2013, 126, 142-146.	3.9	71
77	Mutations in TNK2 in severe autosomal recessive infantile onset epilepsy. Annals of Neurology, 2013, 74, 496-501.	5.3	22
78	Increasing frataxin gene expression with histone deacetylase inhibitors as a therapeutic approach for Friedreich's ataxia. Journal of Neurochemistry, 2013, 126, 147-154.	3.9	51
79	Common data elements for clinical research in Friedreich's ataxia. Movement Disorders, 2013, 28, 190-195.	3.9	14
80	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
81	Genome-wide mapping for clinically relevant predictors of lamotrigine- and phenytoin-induced hypersensitivity reactions. Pharmacogenomics, 2012, 13, 399-405.	1.3	38
82	Friedreich Ataxia. Journal of Child Neurology, 2012, 27, 1204-1211.	1.4	25
83	Normal left ventricular ejection fraction and mass but subclinical myocardial dysfunction in patients with Friedreich's ataxia. European Heart Journal Cardiovascular Imaging, 2012, 13, 346-352.	1.2	35
84	Friedreich ataxia. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2012, 103, 275-294.	1.8	25
85	Central role and mechanisms of Î²â€cell dysfunction and death in friedreich ataxiaâ€“associated diabetes. Annals of Neurology, 2012, 72, 971-982.	5.3	84
86	Identification of common variants associated with human hippocampal and intracranial volumes. Nature Genetics, 2012, 44, 552-561.	21.4	594
87	Antioxidants and other pharmacological treatments for Friedreich ataxia. , 2012, , CD007791.		26
88	Genetic risk factors for ischaemic stroke and its subtypes (the METASTROKE Collaboration): a meta-analysis of genome-wide association studies. Lancet Neurology, The, 2012, 11, 951-962.	10.2	445
89	Spinocerebellar Ataxia Types 1, 2, 3 and 6: the Clinical Spectrum of Ataxia and Morphometric Brainstem and Cerebellar Findings. Cerebellum, 2012, 11, 155-166.	2.5	74
90	HLA-A*3101 and Carbamazepine-Induced Hypersensitivity Reactions in Europeans. New England Journal of Medicine, 2011, 364, 1134-1143.	27.0	815

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91	Two Novel Homozygous SACS Mutations in Unrelated Patients Including the First Reported Case of Paternal UPD as an Etiologic Cause of ARSACS. Journal of Molecular Neuroscience, 2011, 43, 346-349.	2.3	16
92	A gene expression phenotype in lymphocytes from friedreich ataxia patients. Annals of Neurology, 2011, 70, 790-804.	5.3	58
93	Genetics of Epilepsy. Seminars in Neurology, 2011, 31, 506-518.	1.4	39
94	Aminopyridines Correct Early Dysfunction and Delay Neurodegeneration in a Mouse Model of Spinocerebellar Ataxia Type 1. Journal of Neuroscience, 2011, 31, 11795-11807.	3.6	137
95	Cardiomyopathy in Friedreich's ataxia. Acta Neurologica Belgica, 2011, 111, 183-7.	1.1	3
96	Rare Deletions at 16p13.11 Predispose to a Diverse Spectrum of Sporadic Epilepsy Syndromes. American Journal of Human Genetics, 2010, 86, 707-718.	6.2	231
97	Falls in Spinocerebellar Ataxias: Results of the EuroSCA Fall Study. Cerebellum, 2010, 9, 232-239.	2.5	59
98	Suggestive evidence for a new locus for epilepsy with heterogeneous phenotypes on chromosome 17q. Epilepsy Research, 2010, 88, 65-75.	1.6	16
99	Two New Pimelic Diphenylamide HDAC Inhibitors Induce Sustained Frataxin Upregulation in Cells from Friedreich's Ataxia Patients and in a Mouse Model. PLoS ONE, 2010, 5, e8825.	2.5	129
100	Communication via gap junctions underlies early functional and beneficial interactions between grafted neural stem cells and the host. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 5184-5189.	7.1	133
101	Common genetic variation and susceptibility to partial epilepsies: a genome-wide association study. Brain, 2010, 133, 2136-2147.	7.6	132
102	Visualization, quantification and correlation of brain atrophy with clinical symptoms in spinocerebellar ataxia types 1, 3 and 6. NeuroImage, 2010, 49, 158-168.	4.2	162
103	PGC-1alpha Down-Regulation Affects the Antioxidant Response in Friedreich's Ataxia. PLoS ONE, 2010, 5, e10025.	2.5	118
104	Grafting Neural Precursor Cells Promotes Functional Recovery in an SCA1 Mouse Model. Journal of Neuroscience, 2009, 29, 13126-13135.	3.6	70
105	Development of histone deacetylase inhibitors as therapeutics for neurological disease. Future Neurology, 2009, 4, 775-784.	0.5	25
106	Functional genomic analysis of frataxin deficiency reveals tissue-specific alterations and identifies the PPARÎ³ pathway as a therapeutic target in Friedreichâ€™s ataxia. Human Molecular Genetics, 2009, 18, 2452-2461.	2.9	109
107	Introduction. Journal of Neurology, 2009, 256, 1-2.	3.6	8
108	Friedreich ataxia: The clinical picture. Journal of Neurology, 2009, 256, 3-8.	3.6	380

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109	The pathogenesis of Friedreich ataxia and the structure and function of frataxin. Journal of Neurology, 2009, 256, 9-17.	3.6	220
110	Blood-Brain Barrier Promotes Differentiation of Human Fetal Neural Precursor Cells. Stem Cells, 2009, 27, 838-846.	3.2	31
111	Variant on 9p21 strongly associates with coronary heart disease, but lacks association with common stroke. European Journal of Human Genetics, 2009, 17, 1287-1293.	2.8	42
112	Diagnosis and treatment of Friedreich ataxia: a European perspective. Nature Reviews Neurology, 2009, 5, 222-234.	10.1	231
113	Associations of apolipoprotein E gene with ischemic stroke and intracranial atherosclerosis. European Journal of Human Genetics, 2008, 16, 955-960.	2.8	39
114	Drug Insight: antioxidant therapy in inherited ataxias. Nature Clinical Practice Neurology, 2008, 4, 86-96.	2.5	53
115	The Neurological Presentation of Ceruloplasmin Gene Mutations. European Neurology, 2008, 60, 200-205.	1.4	171
116	Friedreich Ataxia. Archives of Neurology, 2008, 65, 1296-303.	4.5	194
117	HDAC Inhibitors Correct Frataxin Deficiency in a Friedreich Ataxia Mouse Model. PLoS ONE, 2008, 3, e1958.	2.5	193
118	Author response to â€œReply to Drug Insight: antioxidant therapy in inherited ataxiasâ€• Nature Clinical Practice Neurology, 2008, 4, E2-E2.	2.5	0
119	Down-regulation of the dopamine receptor D2 in mice lacking ataxin 1. Human Molecular Genetics, 2007, 16, 2122-2134.	2.9	61
120	Chapter 7 Friedreich Ataxia. Blue Books of Neurology, 2007, 31, 186-201.	0.1	0
121	Proprotein Convertase Subtilisin/Kexin Type 9 (PCSK9) Gene Is a Risk Factor of Large-Vessel Atherosclerosis Stroke. PLoS ONE, 2007, 2, e1043.	2.5	67
122	Friedreich's Ataxia and Related DNA Loss-of-Function Disorders. , 2007, , 277-294.		3
123	Multicentre search for genetic susceptibility loci in sporadic epilepsy syndrome and seizure types: a case-control study. Lancet Neurology, The, 2007, 6, 970-980.	10.2	175
124	Study of the adenosinergic system in the brain of HPRT knockout mouse (Leschâ€“Nyhan disease). Clinica Chimica Acta, 2006, 373, 104-107.	1.1	23
125	Real-Time PCR and Linkage Studies to Identify Carriers Presenting HPRT Deleted Gene. Molecular Medicine, 2006, 12, 246-251.	4.4	2
126	The S18Y polymorphism in the UCHL1 gene is a genetic modifier in Huntingtonâ€™s disease. Neurogenetics, 2006, 7, 27-30.	1.4	47

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127	Genetic analysis of candidate genes modifying the age-at-onset in Huntingtonâ€™s disease. Human Genetics, 2006, 120, 285-292.	3.8	39
128	Gene expression profiling in frataxin deficient mice: Microarray evidence for significant expression changes without detectable neurodegeneration. Neurobiology of Disease, 2006, 22, 302-311.	4.4	50
129	Friedreich Ataxia: Detection of GAA Repeat Expansions and Frataxin Point Mutations. , 2006, 126, 197-216.		27
130	Eight Novel Mutations in SPG4 in a Large Sample of Patients With Hereditary Spastic Paraplegia. Archives of Neurology, 2006, 63, 750.	4.5	39
131	Friedreich's Ataxia. , 2006, , 277-296.		16
132	Depression of extraâ€cellular GABA and increase of NMDAâ€induced nitric oxide following acute intraâ€nuclear administration of alcohol in the cerebellar nuclei of the rat. Cerebellum, 2005, 4, 230-238.	2.5	23
133	Hemicerebellectomy blocks the enhancement of cortical motor output associated with repetitive somatosensory stimulation in the rat. Journal of Physiology, 2005, 567, 293-300.	2.9	39
134	Effects of levetiracetam on the production of nitric oxide. Journal of Neurology, 2005, 252, 727-730.	3.6	8
135	A new clinical and molecular form of Unverricht-Lundborg disease localized by homozygosity mapping. Brain, 2005, 128, 652-658.	7.6	45
136	The gene for paroxysmal non-kinesigenic dyskinesia encodes an enzyme in a stress response pathway. Human Molecular Genetics, 2004, 13, 3161-3170.	2.9	196
137	Familial Partial Epilepsy with Variable Foci: Clinical Features and Linkage to Chromosome 22q12. Epilepsia, 2004, 45, 1054-1060.	5.1	71
138	Senataxin, the ortholog of a yeast RNA helicase, is mutant in ataxia-ocular apraxia 2. Nature Genetics, 2004, 36, 225-227.	21.4	454
139	Interaction between repetitive stimulation of the sciatic nerve and functional ablation of cerebellar nucleus interpositus in the rat. Cerebellum, 2004, 3, 21-26.	2.5	13
140	Molecular basis of hypoxanthine-guanine phosphoribosyltransferase deficiency in Italian Lesch-Nyhan patients: Identification of nine novel mutations. Journal of Inherited Metabolic Disease, 2004, 27, 767-773.	3.6	12
141	New clues on the origin of the Friedreich ataxia expanded alleles from the analysis of new polymorphisms closely linked to the mutation. Human Genetics, 2004, 114, 458-463.	3.8	19
142	Striking intrafamilial phenotypic variability and spastic paraplegia in the presence of similar homozygous expansions of theFRDA1gene. Movement Disorders, 2004, 19, 1424-1431.	3.9	14
143	Recurrent transient ischemic attacks in a patient with intrapulmonary arteriovenous shunting detected after closure of a patent foramen ovale. Journal of the American Society of Echocardiography, 2004, 17, 775-777.	2.8	3
144	Frataxin overexpressing mice. FEBS Letters, 2004, 572, 281-288.	2.8	27

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145	Multifaceted role of galectin-3 on human glioblastoma cell motility. Biochemical and Biophysical Research Communications, 2004, 325, 1393-1398.	2.1	40
146	Spinocerebellar Ataxia Type 8: Molecular Genetic Comparisonsand Haplotype Analysis of 37 Families with Ataxia. American Journal of Human Genetics, 2004, 75, 3-16.	6.2	88
147	Iron metabolism in mice with partial frataxin deficiency. Cerebellum, 2003, 2, 146-153.	2.5	5
148	Myorhythmia associated with Hodgkin?s lymphoma. Journal of Neurology, 2003, 250, 1382-1384.	3.6	15
149	Friedreich ataxia. Seminars in Pediatric Neurology, 2003, 10, 163-172.	2.0	87
150	Variants of the KCNMB3 regulatory subunit of maxi BK channels affect channel inactivation. Physiological Genomics, 2003, 15, 191-198.	2.3	34
151	Bilateral High-Frequency Synchronous Discharges. Archives of Neurology, 2003, 60, 416.	4.5	8
152	Friedreich Ataxia. , 2003, , 165-178.		1
153	Friedreich's Ataxia Associated With Mitochondrial Myopathy: Clinicopathologic Report. Journal of Child Neurology, 2002, 17, 453-456.	1.4	8
154	Iron Metabolism and Mitochondrial Abnormalities in Friedreich Ataxia. Blood Cells, Molecules, and Diseases, 2002, 29, 536-547.	1.4	61
155	Frataxin knockin mouse. FEBS Letters, 2002, 512, 291-297.	2.8	155
156	Frataxin deficiency and mitochondrial dysfunction. Mitochondrion, 2002, 2, 87-93.	3.4	31
157	The Hereditary Ataxias. Cerebellum, 2002, 1, 143-158.	2.5	0
158	The Molecular Basis of Friedreich Ataxia. Advances in Experimental Medicine and Biology, 2002, 516, 99-118.	1.6	45
159	A symposium of the Society for Experimental Neuropathology under the auspices of the American Neurological Association. Cerebellum, 2002, 1, 143-158.	2.5	0
160	Manganese superoxide dismutase induction by iron is impaired in Friedreich ataxia cells. FEBS Letters, 2001, 509, 101-105.	2.8	71
161	Molecular basis of Friedreich ataxia. Movement Disorders, 2001, 16, 815-821.	3.9	38
162	Frataxin deficiency enhances apoptosis in cells differentiating into neuroectoderm. Human Molecular Genetics, 2001, 10, 1935-1944.	2.9	71

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163	Sticky DNA, a Self-associated Complex Formed at Long GAAÂ·TTC Repeats in Intron 1 of the Frataxin Gene, Inhibits Transcription. Journal of Biological Chemistry, 2001, 276, 27171-27177.	3.4	165
164	GGAÂ·TCC-interrupted Triplets in Long GAAÂ·TTC Repeats Inhibit the Formation of Triplex and Sticky DNA Structures, Alleviate Transcription Inhibition, and Reduce Genetic Instabilities. Journal of Biological Chemistry, 2001, 276, 27178-27187.	3.4	78
165	Neuropathology of the inherited ataxias. , 2001, , 387-406.		10
166	Friedreich's Ataxia: Clinical Aspects and Pathogenesis. Seminars in Neurology, 1999, 19, 311-321.	1.4	43
167	Decreased Brain Protein Levels of Cytochrome Oxidase Subunits in Alzheimer's Disease and in Hereditary Spinocerebella Ataxia Disorders. Journal of Neurochemistry, 1999, 72, 700-707.	3.9	76
168	Friedreich's ataxia: Point mutations and clinical presentation of compound heterozygotes. Annals of Neurology, 1999, 45, 200-206.	5.3	371
169	Heteroduplexes may confuse the interpretation of PCR-based molecular tests for the Friedreich ataxia GAA triplet repeat. Human Mutation, 1999, 13, 328-330.	2.5	17
170	A missense mutation (W155R) in an American patient with Friedreich Ataxia. Human Mutation, 1999, 13, 506-506.	2.5	17
171	Mapping of a Gene Determining Familial Partial Epilepsy with Variable Foci to Chromosome 22q11-q12. American Journal of Human Genetics, 1999, 65, 1698-1710.	6.2	89
172	Sticky DNA. Molecular Cell, 1999, 3, 465-475.	9.7	305
173	Knock-out of the cyaY gene in Escherichia coli does not affect cellular iron content and sensitivity to oxidants. FEBS Letters, 1999, 456, 13-16.	2.8	65
174	Molecular Pathogenesis of Friedreich Ataxia. Archives of Neurology, 1999, 56, 1201.	4.5	103
175	Friedreich's ataxia: Point mutations and clinical presentation of compound heterozygotes. , 1999, 45, 200.		2
176	Prenatal diagnosis of Friedreich ataxia. Prenatal Diagnosis, 1998, 18, 831-833.	2.3	26
177	Molecular genetics and pathogenesis of Friedreich ataxia. Neuromuscular Disorders, 1998, 8, 409-415.	0.6	57
178	Inhibitory Effects of Expanded GAAÂ·TTC Triplet Repeats from Intron I of the Friedreich Ataxia Gene on Transcription and Replicationin Vivo. Journal of Biological Chemistry, 1998, 273, 14588-14595.	3.4	288
179	Brain Glyceraldehyde-3-Phosphate Dehydrogenase Activity in Human Trinucleotide Repeat Disorders. Archives of Neurology, 1998, 55, 1299.	4.5	50
180	2 Molecular Genetics of the Hereditary Ataxias. Advances in Genetics, 1998, 38, 31-68.	1.8	19

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181	Regulation of Mitochondrial Iron Accumulation by Yfh1p, a Putative Homolog of Frataxin. Science, 1997, 276, 1709-1712.	12.6	942
182	Frataxin Shows Developmentally Regulated Tissue-Specific Expression in the Mouse Embryo. Neurobiology of Disease, 1997, 4, 103-113.	4.4	69
183	Frataxin fracas. Nature Genetics, 1997, 15, 337-338.	21.4	78
184	Phenotypic variability in friedreich ataxia: Role of the associated GAA triplet repeat expansion. Annals of Neurology, 1997, 41, 675-682.	5.3	249
185	Homozygosity mapping of Hallervordenâ€“Spatz syndrome to chromosome 20p12.3â€“p13. Nature Genetics, 1996, 14, 479-481.	21.4	158
186	Early-onset ataxia with cardiomyopathy and retained tendon reflexes maps to the friedreich's ataxia locus on chromosome 9q. Annals of Neurology, 1995, 37, 359-362.	5.3	83
187	Mapping of the congenital generalized hypertrichosis locus to chromosome Xq24â€“q27.1. Nature Genetics, 1995, 10, 202-207.	21.4	68
188	Evidence of Linkage between Susceptibility to Multiple Sclerosis and HLA-Class II Loci in Italian Multiplex Families. European Journal of Human Genetics, 1995, 3, 303-311.	2.8	18
189	The myelin basic protein gene is not a major susceptibility locus for multiple sclerosis in Italian patients. Journal of Neurology, 1994, 241, 615-619.	3.6	18
190	Mapping of Friedreich's Ataxia Locus by Identification of Recombination Events in Patients Homozygous by Descent. European Journal of Human Genetics, 1994, 2, 291-299.	2.8	11
191	Evidence of a genetic marker associated with early onset in Friedreich's ataxia. Journal of Neurology, 1993, 240, 254-256.	3.6	2
192	The gene for spinal cerebellar ataxia 1 (SCA1) is flanked by two closely linked highly polymorphic microsatellite loci. Human Molecular Genetics, 1993, 2, 1383-1387.	2.9	26
193	A rapid method to isolate (GT)n repeats from yeast artificial chromosomes. Nucleic Acids Research, 1992, 20, 1154-1154.	14.5	17
194	Localization of the human gene for carnitine palmitoyltransferase to 1p13â€“p11 by nonradioactive in situ hybridization. Genomics, 1992, 13, 1372-1374.	2.9	18
195	Localization of DNA probes tightly linked to the Friedreich's ataxia locus by in situ hybridization in a case of pericentric inversion of chromosome 9. Human Genetics, 1991, 86, 525-528.	3.8	1
196	Regional localization by in situ hybridization of a human chromosome 9 marker tightly linked to the Friedreich's ataxia locus. Human Genetics, 1990, 85, 125-6.	3.8	6
197	A TaqI RFLP in the locus D9S29 on human chromosome 9. Nucleic Acids Research, 1989, 17, 10514-10514.	14.5	4
198	An anonymous DNA probe (LAMP 92) detects a Pvu II polymorphism on human chromosome 9 [D9S29]. Nucleic Acids Research, 1988, 16, 7213-7213.	14.5	10

#	Article	IF	Citations
199	A Pvu II RFLP in the human ADH3 gene. Nucleic Acids Research, 1988, 16, 11857-11857.	14.5	2
200	Large-scale purification of hnRNP proteins from HeLa cells by affinity chromatography on ssDNA-cellulose. FEBS Journal, 1987, 162, 213-220.	0.2	31
201	Mammalian single-stranded DNA binding proteins and heterogeneous nuclear RNA proteins have common antigenic determinants. Nucleic Acids Research, 1985, 13, 337-346.	14.5	52
202	Single stranded DNA binding proteins derive from hnRNP proteins by proteolysis in mammalian cells. Nucleic Acids Research, 1985, 13, 6577-6590.	14.5	45