

Massimo Pandolfo

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

197
papers

13,601
citations

60
h-index

113
g-index

215
ext. papers

15,446
ext. citations

8.8
avg, IF

6.06
L-index

#	Paper	IF	Citations
197	Recessive cerebellar and afferent ataxias - clinical challenges and future directions.. <i>Nature Reviews Neurology</i> , 2022 ,	15	1
196	DNA methylation in Friedreich ataxia silences expression of frataxin isoform E.. <i>Scientific Reports</i> , 2022 , 12, 5031	4.9	1
195	Research priorities for rare neurological diseases: a representative view of patient representatives and healthcare professionals from the European Reference Network for Rare Neurological Diseases. <i>Orphanet Journal of Rare Diseases</i> , 2021 , 16, 135	4.2	3
194	Progression characteristics of the European Friedreich's Ataxia Consortium for Translational Studies (EFACTS): a 4-year cohort study. <i>Lancet Neurology, The</i> , 2021 , 20, 362-372	24.1	12
193	Hand Dexterity and Pyramidal Dysfunction in Friedreich Ataxia, A Finger Tapping Study. <i>Movement Disorders Clinical Practice</i> , 2021 , 8, 85-91	2.2	3
192	Age of onset modulates resting-state brain network dynamics in Friedreich Ataxia. <i>Human Brain Mapping</i> , 2021 , 42, 5334-5344	5.9	3
191	Cerebellar ataxia, neuropathy, hearing loss, and intellectual disability due to AIFM1 mutation. <i>Neurology: Genetics</i> , 2020 , 6, e420	3.8	4
190	Cerebellar cognitive disorder parallels cerebellar motor symptoms in Friedreich ataxia. <i>Annals of Clinical and Translational Neurology</i> , 2020 , 7, 1050-1054	5.3	16
189	Friedreich ataxia 2020 , 99-112		
188	Age of onset determines intrinsic functional brain architecture in Friedreich ataxia. <i>Annals of Clinical and Translational Neurology</i> , 2020 , 7, 94-104	5.3	11
187	Neurologic outcomes in Friedreich ataxia: Study of a single-site cohort. <i>Neurology: Genetics</i> , 2020 , 6, e415	5.8	10
186	Exenatide induces frataxin expression and improves mitochondrial function in Friedreich ataxia. <i>JCI Insight</i> , 2020 , 5,	9.9	23
185	Primary proprioceptive neurons from human induced pluripotent stem cells: a cell model for afferent ataxias. <i>Scientific Reports</i> , 2020 , 10, 7752	4.9	14
184	Central Nervous System Therapeutic Targets in Friedreich Ataxia. <i>Human Gene Therapy</i> , 2020 , 31, 1226-1236	12.86	12
183	Onset features and time to diagnosis in Friedreich's Ataxia. <i>Orphanet Journal of Rare Diseases</i> , 2020 , 15, 198	4.2	8
182	Test-retest reliability of the Friedreich's ataxia rating scale. <i>Annals of Clinical and Translational Neurology</i> , 2020 , 7, 1708-1712	5.3	6
181	Altered neocortical tactile but preserved auditory early change detection responses in Friedreich ataxia. <i>Clinical Neurophysiology</i> , 2019 , 130, 1299-1310	4.3	10

180	Prediction of Survival With Long-Term Disease Progression in Most Common Spinocerebellar Ataxia. <i>Movement Disorders</i> , 2019 , 34, 1220-1227	7	6
179	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). <i>Neurological Research and Practice</i> , 2019 , 1, 33	3.2	8
178	Evidence for genetically determined degeneration of proprioceptive tracts in Friedreich ataxia. <i>Neurology</i> , 2019 , 93, e116-e124	6.5	20
177	Genetic architecture of subcortical brain structures in 38,851 individuals. <i>Nature Genetics</i> , 2019 , 51, 1624-1636	5.6	81
176	Friedreich and dominant ataxias: quantitative differences in cerebellar dysfunction measurements. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018 , 89, 559-565	5.5	11
175	Siponimod versus placebo in secondary progressive multiple sclerosis (EXPAND): a double-blind, randomised, phase 3 study. <i>Lancet, The</i> , 2018 , 391, 1263-1273	4.0	422
174	Survival in patients with spinocerebellar ataxia types 1, 2, 3, and 6 (EUROSCA): a longitudinal cohort study. <i>Lancet Neurology, The</i> , 2018 , 17, 327-334	24.1	46
173	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. <i>Lancet Neurology, The</i> , 2018 , 17, 405-415	24.1	150
172	Personality and Neuropsychological Profiles in Friedreich Ataxia. <i>Cerebellum</i> , 2018 , 17, 204-212	4.3	14
171	Automated functional upper limb evaluation of patients with Friedreich ataxia using serious games rehabilitation exercises. <i>Journal of NeuroEngineering and Rehabilitation</i> , 2018 , 15, 87	5.3	14
170	Low Prevalence Estimates of Late-Onset Glycogen Storage Disease Type II in French-Speaking Belgium are not Due to Missed Diagnoses. <i>Journal of Neuromuscular Diseases</i> , 2018 , 5, 471-480	5	1
169	Long-term evolution of patient-reported outcome measures in spinocerebellar ataxias. <i>Journal of Neurology</i> , 2018 , 265, 2040-2051	5.5	16
168	Erythropoietin and small molecule agonists of the tissue-protective erythropoietin receptor increase FXN expression in neuronal cells in vitro and in Fxn-deficient KIKO mice in vivo. <i>Neuropharmacology</i> , 2017 , 123, 34-45	5.5	8
167	HLA genotype as a marker of multiple sclerosis prognosis: A pilot study. <i>Journal of the Neurological Sciences</i> , 2017 , 375, 348-354	3.2	5
166	A recessive ataxia diagnosis algorithm for the next generation sequencing era. <i>Annals of Neurology</i> , 2017 , 82, 892-899	9.4	20
165	Urinary, bowel and sexual symptoms in a cohort of patients with Friedreich's ataxia. <i>Orphanet Journal of Rare Diseases</i> , 2017 , 12, 158	4.2	13
164	Body Mass Index Decline Is Related to Spinocerebellar Ataxia Disease Progression. <i>Movement Disorders Clinical Practice</i> , 2017 , 4, 689-697	2.2	14
163	Neurodegeneration With Brain Iron Accumulation (NBIA) Syndromes Presenting With Late-Onset Craniocervical Dystonia: An Illustrative Case Series?. <i>Movement Disorders Clinical Practice</i> , 2017 , 4, 254-257	2.2	5

162	MME mutation in dominant spinocerebellar ataxia with neuropathy (SCA43). <i>Neurology: Genetics</i> , 2016 , 2, e94	3.8	28
161	Pharmacological treatments for Friedreich ataxia. <i>The Cochrane Library</i> , 2016 , CD007791	5.2	28
160	Progression characteristics of the European Friedreich's Ataxia Consortium for Translational Studies (EFACTS): a 2 year cohort study. <i>Lancet Neurology, The</i> , 2016 , 15, 1346-1354	24.1	78
159	Standardized Assessment of Hereditary Ataxia Patients in Clinical Studies. <i>Movement Disorders Clinical Practice</i> , 2016 , 3, 230-240	2.2	7
158	Friedreich ataxia-induced pluripotent stem cell-derived neurons show a cellular phenotype that is corrected by a benzamide HDAC inhibitor. <i>Human Molecular Genetics</i> , 2016 , 25, 4847-4855	5.6	33
157	Novel genetic loci underlying human intracranial volume identified through genome-wide association. <i>Nature Neuroscience</i> , 2016 , 19, 1569-1582	25.5	147
156	Delayed-onset Friedreich's ataxia revisited. <i>Movement Disorders</i> , 2016 , 31, 62-9	7	40
155	Common genetic variants influence human subcortical brain structures. <i>Nature</i> , 2015 , 520, 224-9	50.4	601
154	Friedreich Ataxia 2015 , 833-843		1
153	Animal Models of Friedreich Ataxia 2015 , 1017-1024		1
152	Friedreich Ataxia 2015 , 984-1002		
151	A 22-Year Follow-up Study of Long-term Cardiac Outcome and Predictors of Survival in Friedreich Ataxia. <i>JAMA Neurology</i> , 2015 , 72, 1334-41	17.2	45
150	Long-term disease progression in spinocerebellar ataxia types 1, 2, 3, and 6: a longitudinal cohort study. <i>Lancet Neurology, The</i> , 2015 , 14, 1101-8	24.1	139
149	URINARY, BOWEL AND SEXUAL FUNCTION IN PATIENTS WITH FRIEDREICH'S ATAXIA. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2015 , 86, e4.167-e4	5.5	
148	Adult neural precursor cells form connexin-dependent networks that improve their survival. <i>NeuroReport</i> , 2015 , 26, 928-36	1.7	7
147	Unveiling a common mechanism of apoptosis in E-cells and neurons in Friedreich's ataxia. <i>Human Molecular Genetics</i> , 2015 , 24, 2274-86	5.6	47
146	Building bridges between neuroscientific evidence and policy. <i>Lancet Neurology, The</i> , 2015 , 14, 242-5	24.1	5
145	Biological and clinical characteristics of the European Friedreich's Ataxia Consortium for Translational Studies (EFACTS) cohort: a cross-sectional analysis of baseline data. <i>Lancet Neurology, The</i> , 2015 , 14, 174-82	24.1	113

144	Transient CNS deficits and migrainous auras in individuals without a history of headache. <i>Headache</i> , 2014 , 54, 493-9	4.2	6
143	The ENIGMA Consortium: large-scale collaborative analyses of neuroimaging and genetic data. <i>Brain Imaging and Behavior</i> , 2014 , 8, 153-82	4.1	539
142	Epigenetic therapy for Friedreich ataxia. <i>Annals of Neurology</i> , 2014 , 76, 489-508	9.4	101
141	Modulation of the age at onset in spinocerebellar ataxia by CAG tracts in various genes. <i>Brain</i> , 2014 , 137, 2444-55	11.2	108
140	Deferiprone in Friedreich ataxia: a 6-month randomized controlled trial. <i>Annals of Neurology</i> , 2014 , 76, 509-21	9.4	83
139	Characteristics and outcomes of patients with multiple cervical artery dissection. <i>Stroke</i> , 2014 , 45, 37-416.7		78
138	Consensus clinical management guidelines for Friedreich ataxia. <i>Orphanet Journal of Rare Diseases</i> , 2014 , 9, 184	4.2	56
137	A new locus for familial temporal lobe epilepsy on chromosome 3q. <i>Epilepsy Research</i> , 2013 , 106, 338-443		14
136	Mutations in DEPDC5 cause familial focal epilepsy with variable foci. <i>Nature Genetics</i> , 2013 , 45, 546-51	36.3	238
135	150 years of Friedreich ataxia: from its discovery to therapy. <i>Journal of Neurochemistry</i> , 2013 , 126 Suppl 1, 1-3	6	13
134	Genotype-specific patterns of atrophy progression are more sensitive than clinical decline in SCA1, SCA3 and SCA6. <i>Brain</i> , 2013 , 136, 905-17	11.2	92
133	Epilepsy, hippocampal sclerosis and febrile seizures linked by common genetic variation around SCN1A. <i>Brain</i> , 2013 , 136, 3140-50	11.2	144
132	A novel function of Ataxin-1 in the modulation of PP2A activity is dysregulated in the spinocerebellar ataxia type 1. <i>Human Molecular Genetics</i> , 2013 , 22, 3425-37	5.6	12
131	Cerebellar and afferent ataxias. <i>CONTINUUM Lifelong Learning in Neurology</i> , 2013 , 19, 1312-43	3	24
130	Pediatric epilepsy genetics. <i>Current Opinion in Neurology</i> , 2013 , 26, 137-45	7.1	11
129	Neurons and cardiomyocytes derived from induced pluripotent stem cells as a model for mitochondrial defects in Friedreich's ataxia. <i>DMM Disease Models and Mechanisms</i> , 2013 , 6, 608-21	4.1	111
128	Treatment of Friedreich's ataxia. <i>Expert Opinion on Orphan Drugs</i> , 2013 , 1, 221-234	1.1	5
127	Deferiprone for the treatment of Friedreich's ataxia. <i>Journal of Neurochemistry</i> , 2013 , 126 Suppl 1, 142-66		62

126	Mutations in TNK2 in severe autosomal recessive infantile onset epilepsy. <i>Annals of Neurology</i> , 2013 , 74, 496-501	9.4	18
125	Increasing frataxin gene expression with histone deacetylase inhibitors as a therapeutic approach for Friedreich's ataxia. <i>Journal of Neurochemistry</i> , 2013 , 126 Suppl 1, 147-54	6	42
124	Common data elements for clinical research in Friedreich's ataxia. <i>Movement Disorders</i> , 2013 , 28, 190-5	7	14
123	Friedreich ataxia. <i>Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn</i> , 2012 , 103, 275-94	3	15
122	Central role and mechanisms of cell dysfunction and death in friedreich ataxia-associated diabetes. <i>Annals of Neurology</i> , 2012 , 72, 971-82	9.4	60
121	Identification of common variants associated with human hippocampal and intracranial volumes. <i>Nature Genetics</i> , 2012 , 44, 552-61	36.3	498
120	Antioxidants and other pharmacological treatments for Friedreich ataxia. <i>Cochrane Database of Systematic Reviews</i> , 2012 , CD007791		23
119	Genetic risk factors for ischaemic stroke and its subtypes (the METASTROKE collaboration): a meta-analysis of genome-wide association studies. <i>Lancet Neurology</i> , 2012 , 11, 951-62	24.1	359
118	Spinocerebellar ataxia types 1, 2, 3 and 6: the clinical spectrum of ataxia and morphometric brainstem and cerebellar findings. <i>Cerebellum</i> , 2012 , 11, 155-66	4.3	65
117	Clinical data and characterization of the liver conditional mouse model exclude neoplasia as a non-neurological manifestation associated with Friedreich's ataxia. <i>DMM Disease Models and Mechanisms</i> , 2012 , 5, 860-9	4.1	30
116	Genome-wide mapping for clinically relevant predictors of lamotrigine- and phenytoin-induced hypersensitivity reactions. <i>Pharmacogenomics</i> , 2012 , 13, 399-405	2.6	34
115	Friedreich ataxia: new pathways. <i>Journal of Child Neurology</i> , 2012 , 27, 1204-11	2.5	22
114	Normal left ventricular ejection fraction and mass but subclinical myocardial dysfunction in patients with Friedreich's ataxia. <i>European Heart Journal Cardiovascular Imaging</i> , 2012 , 13, 346-52	4.1	28
113	HLA-A*3101 and carbamazepine-induced hypersensitivity reactions in Europeans. <i>New England Journal of Medicine</i> , 2011 , 364, 1134-43	59.2	689
112	Two novel homozygous SACS mutations in unrelated patients including the first reported case of paternal UPD as an etiologic cause of ARSACS. <i>Journal of Molecular Neuroscience</i> , 2011 , 43, 346-9	3.3	13
111	A gene expression phenotype in lymphocytes from Friedreich ataxia patients. <i>Annals of Neurology</i> , 2011 , 70, 790-804	9.4	49
110	Genetics of epilepsy. <i>Seminars in Neurology</i> , 2011 , 31, 506-18	3.2	29
109	Aminopyridines correct early dysfunction and delay neurodegeneration in a mouse model of spinocerebellar ataxia type 1. <i>Journal of Neuroscience</i> , 2011 , 31, 11795-807	6.6	104

108	Cardiomyopathy in Friedreich's ataxia. <i>Acta Neurologica Belgica</i> , 2011 , 111, 183-7	1.5	3
107	Two new pimelic diphenylamide HDAC inhibitors induce sustained frataxin upregulation in cells from Friedreich's ataxia patients and in a mouse model. <i>PLoS ONE</i> , 2010 , 5, e8825	3.7	111
106	Communication via gap junctions underlies early functional and beneficial interactions between grafted neural stem cells and the host. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2010 , 107, 5184-9	11.5	119
105	Common genetic variation and susceptibility to partial epilepsies: a genome-wide association study. <i>Brain</i> , 2010 , 133, 2136-47	11.2	115
104	Visualization, quantification and correlation of brain atrophy with clinical symptoms in spinocerebellar ataxia types 1, 3 and 6. <i>NeuroImage</i> , 2010 , 49, 158-68	7.9	133
103	Rare deletions at 16p13.11 predispose to a diverse spectrum of sporadic epilepsy syndromes. <i>American Journal of Human Genetics</i> , 2010 , 86, 707-18	11	206
102	Falls in spinocerebellar ataxias: Results of the EuroSCA Fall Study. <i>Cerebellum</i> , 2010 , 9, 232-9	4.3	52
101	Suggestive evidence for a new locus for epilepsy with heterogeneous phenotypes on chromosome 17q. <i>Epilepsy Research</i> , 2010 , 88, 65-75	3	13
100	PGC-1alpha down-regulation affects the antioxidant response in Friedreich's ataxia. <i>PLoS ONE</i> , 2010 , 5, e10025	3.7	100
99	Grafting neural precursor cells promotes functional recovery in an SCA1 mouse model. <i>Journal of Neuroscience</i> , 2009 , 29, 13126-35	6.6	62
98	Development of histone deacetylase inhibitors as therapeutics for neurological disease. <i>Future Neurology</i> , 2009 , 4, 775-784	1.5	20
97	Functional genomic analysis of frataxin deficiency reveals tissue-specific alterations and identifies the PPARgamma pathway as a therapeutic target in Friedreich's ataxia. <i>Human Molecular Genetics</i> , 2009 , 18, 2452-61	5.6	94
96	Friedreich ataxia: the clinical picture. <i>Journal of Neurology</i> , 2009 , 256 Suppl 1, 3-8	5.5	317
95	The pathogenesis of Friedreich ataxia and the structure and function of frataxin. <i>Journal of Neurology</i> , 2009 , 256 Suppl 1, 9-17	5.5	193
94	Blood-brain barrier promotes differentiation of human fetal neural precursor cells. <i>Stem Cells</i> , 2009 , 27, 838-46	5.8	28
93	Variant on 9p21 strongly associates with coronary heart disease, but lacks association with common stroke. <i>European Journal of Human Genetics</i> , 2009 , 17, 1287-93	5.3	38
92	Diagnosis and treatment of Friedreich ataxia: a European perspective. <i>Nature Reviews Neurology</i> , 2009 , 5, 222-34	15	192
91	Associations of apolipoprotein E gene with ischemic stroke and intracranial atherosclerosis. <i>European Journal of Human Genetics</i> , 2008 , 16, 955-60	5.3	31

90	Drug Insight: antioxidant therapy in inherited ataxias. <i>Nature Clinical Practice Neurology</i> , 2008 , 4, 86-96		48
89	The neurological presentation of ceruloplasmin gene mutations. <i>European Neurology</i> , 2008 , 60, 200-5	2.1	150
88	Friedreich ataxia. <i>Archives of Neurology</i> , 2008 , 65, 1296-303		159
87	HDAC inhibitors correct frataxin deficiency in a Friedreich ataxia mouse model. <i>PLoS ONE</i> , 2008 , 3, e1958	3.7	174
86	Author response to Reply to Drug Insight: antioxidant therapy in inherited ataxias <i>Nature Clinical Practice Neurology</i> , 2008 , 4, E2-E2		
85	Proprotein convertase subtilisin/kexin type 9 (PCSK9) gene is a risk factor of large-vessel atherosclerosis stroke. <i>PLoS ONE</i> , 2007 , 2, e1043	3.7	53
84	Friedreich's Ataxia and Related DNA Loss-of-Function Disorders 2007 , 277-294		3
83	Multicentre search for genetic susceptibility loci in sporadic epilepsy syndrome and seizure types: a case-control study. <i>Lancet Neurology</i> , 2007 , 6, 970-80	24.1	152
82	Down-regulation of the dopamine receptor D2 in mice lacking ataxin 1. <i>Human Molecular Genetics</i> , 2007 , 16, 2122-34	5.6	53
81	Chapter 7 Friedreich Ataxia. <i>Blue Books of Neurology</i> , 2007 , 31, 186-201		
80	Gene expression profiling in frataxin deficient mice: microarray evidence for significant expression changes without detectable neurodegeneration. <i>Neurobiology of Disease</i> , 2006 , 22, 302-11	7.5	47
79	Friedreich ataxia: Detection of GAA repeat expansions and frataxin point mutations. <i>Methods in Molecular Medicine</i> , 2006 , 126, 197-216		26
78	Eight novel mutations in SPG4 in a large sample of patients with hereditary spastic paraplegia. <i>Archives of Neurology</i> , 2006 , 63, 750-5		34
77	Study of the adenosinergic system in the brain of HPRT knockout mouse (Lesch-Nyhan disease). <i>Clinica Chimica Acta</i> , 2006 , 373, 104-7	6.2	15
76	Real-time PCR and linkage studies to identify carriers presenting HPRT deleted gene. <i>Molecular Medicine</i> , 2006 , 12, 246-51	6.2	2
75	The S18Y polymorphism in the UCHL1 gene is a genetic modifier in Huntington's disease. <i>Neurogenetics</i> , 2006 , 7, 27-30	3	42
74	Genetic analysis of candidate genes modifying the age-at-onset in Huntington's disease. <i>Human Genetics</i> , 2006 , 120, 285-92	6.3	36
73	Friedreich's Ataxia 2006 , 277-296		12

72	A new clinical and molecular form of Unverricht-Lundborg disease localized by homozygosity mapping. <i>Brain</i> , 2005 , 128, 652-8	11.2	39
71	Depression of extra-cellular GABA and increase of NMDA-induced nitric oxide following acute intra-nuclear administration of alcohol in the cerebellar nuclei of the rat. <i>Cerebellum</i> , 2005 , 4, 230-8	4.3	23
70	Hemicerebellectomy blocks the enhancement of cortical motor output associated with repetitive somatosensory stimulation in the rat. <i>Journal of Physiology</i> , 2005 , 567, 293-300	3.9	37
69	Effects of levetiracetam on the production of nitric oxide--an in vivo study. <i>Journal of Neurology</i> , 2005 , 252, 727-30	5.5	8
68	The gene for paroxysmal non-kinesigenic dyskinesia encodes an enzyme in a stress response pathway. <i>Human Molecular Genetics</i> , 2004 , 13, 3161-70	5.6	164
67	Familial partial epilepsy with variable foci: clinical features and linkage to chromosome 22q12. <i>Epilepsia</i> , 2004 , 45, 1054-60	6.4	48
66	Senataxin, the ortholog of a yeast RNA helicase, is mutant in ataxia-ocular apraxia 2. <i>Nature Genetics</i> , 2004 , 36, 225-7	36.3	385
65	Interaction between repetitive stimulation of the sciatic nerve and functional ablation of cerebellar nucleus interpositus in the rat. <i>Cerebellum</i> , 2004 , 3, 21-6	4.3	12
64	Molecular basis of hypoxanthine-guanine phosphoribosyltransferase deficiency in Italian Lesch-Nyhan patients: identification of nine novel mutations. <i>Journal of Inherited Metabolic Disease</i> , 2004 , 27, 767-73	5.4	11
63	New clues on the origin of the Friedreich ataxia expanded alleles from the analysis of new polymorphisms closely linked to the mutation. <i>Human Genetics</i> , 2004 , 114, 458-63	6.3	16
62	Striking intrafamilial phenotypic variability and spastic paraplegia in the presence of similar homozygous expansions of the FRDA1 gene. <i>Movement Disorders</i> , 2004 , 19, 1424-31	7	12
61	Recurrent transient ischemic attacks in a patient with intrapulmonary arteriovenous shunting detected after closure of a patent foramen ovale. <i>Journal of the American Society of Echocardiography</i> , 2004 , 17, 775-7	5.8	2
60	Frataxin overexpressing mice. <i>FEBS Letters</i> , 2004 , 572, 281-8	3.8	23
59	Multifaceted role of galectin-3 on human glioblastoma cell motility. <i>Biochemical and Biophysical Research Communications</i> , 2004 , 325, 1393-8	3.4	37
58	Spinocerebellar ataxia type 8: molecular genetic comparisons and haplotype analysis of 37 families with ataxia. <i>American Journal of Human Genetics</i> , 2004 , 75, 3-16	11	81
57	Variants of the KCNMB3 regulatory subunit of maxi BK channels affect channel inactivation. <i>Physiological Genomics</i> , 2003 , 15, 191-8	3.6	29
56	Bilateral high-frequency synchronous discharges: a new form of tremor in humans. <i>Archives of Neurology</i> , 2003 , 60, 416-22		7
55	Iron metabolism in mice with partial frataxin deficiency. <i>Cerebellum</i> , 2003 , 2, 146-53	4.3	5

54	Myorhythmia associated with Hodgkin's lymphoma. <i>Journal of Neurology</i> , 2003 , 250, 1382-4	5.5	12
53	Friedreich ataxia. <i>Seminars in Pediatric Neurology</i> , 2003 , 10, 163-72	2.9	83
52	Friedreich Ataxia 2003 , 165-178		
51	The Hereditary Ataxias. <i>Cerebellum</i> , 2002 , 1, 143-158	4.3	
50	Friedreich's ataxia associated with mitochondrial myopathy: clinicopathologic report. <i>Journal of Child Neurology</i> , 2002 , 17, 453-6	2.5	5
49	Iron metabolism and mitochondrial abnormalities in Friedreich ataxia. <i>Blood Cells, Molecules, and Diseases</i> , 2002 , 29, 536-47; discussion 548-52	2.1	56
48	Frataxin knockin mouse. <i>FEBS Letters</i> , 2002 , 512, 291-7	3.8	128
47	Frataxin deficiency and mitochondrial dysfunction. <i>Mitochondrion</i> , 2002 , 2, 87-93	4.9	28
46	The molecular basis of Friedreich ataxia. <i>Advances in Experimental Medicine and Biology</i> , 2002 , 516, 99-1186		42
45	A symposium of the Society for Experimental Neuropathology under the auspices of the American Neurological Association. <i>Cerebellum</i> , 2002 , 1, 143-158	4.3	
44	Molecular basis of Friedreich ataxia. <i>Movement Disorders</i> , 2001 , 16, 815-21	7	29
43	Frataxin deficiency enhances apoptosis in cells differentiating into neuroectoderm. <i>Human Molecular Genetics</i> , 2001 , 10, 1935-44	5.6	63
42	Sticky DNA, a self-associated complex formed at long GAA*TTC repeats in intron 1 of the frataxin gene, inhibits transcription. <i>Journal of Biological Chemistry</i> , 2001 , 276, 27171-7	5.4	149
41	GGA*TCC-interrupted triplets in long GAA*TTC repeats inhibit the formation of triplex and sticky DNA structures, alleviate transcription inhibition, and reduce genetic instabilities. <i>Journal of Biological Chemistry</i> , 2001 , 276, 27178-87	5.4	66
40	Manganese superoxide dismutase induction by iron is impaired in Friedreich ataxia cells. <i>FEBS Letters</i> , 2001 , 509, 101-5	3.8	63
39	Neuropathology of the inherited ataxias 2001 , 387-406		7
38	Friedreich's ataxia: clinical aspects and pathogenesis. <i>Seminars in Neurology</i> , 1999 , 19, 311-21	3.2	40
37	Decreased brain protein levels of cytochrome oxidase subunits in Alzheimer's disease and in hereditary spinocerebellar ataxia disorders: a nonspecific change?. <i>Journal of Neurochemistry</i> , 1999 , 72, 700-7	6	62

36	Friedreich's ataxia: point mutations and clinical presentation of compound heterozygotes. <i>Annals of Neurology</i> , 1999 , 45, 200-6	9.4	327
35	Heteroduplexes may confuse the interpretation of PCR-based molecular tests for the Friedreich ataxia GAA triplet repeat. <i>Human Mutation</i> , 1999 , 13, 328-30	4.7	15
34	A missense mutation (W155R) in an American patient with Friedreich Ataxia. <i>Human Mutation</i> , 1999 , 13, 506-506	4.7	16
33	Mapping of a gene determining familial partial epilepsy with variable foci to chromosome 22q11-q12. <i>American Journal of Human Genetics</i> , 1999 , 65, 1698-710	11	78
32	Sticky DNA: self-association properties of long GAA.TTC repeats in R.R.Y triplex structures from Friedreich's ataxia. <i>Molecular Cell</i> , 1999 , 3, 465-75	17.6	271
31	Knock-out of the <i>cyaY</i> gene in <i>Escherichia coli</i> does not affect cellular iron content and sensitivity to oxidants. <i>FEBS Letters</i> , 1999 , 456, 13-6	3.8	56
30	Molecular pathogenesis of Friedreich ataxia. <i>Archives of Neurology</i> , 1999 , 56, 1201-8		90
29	Friedreich's ataxia: Point mutations and clinical presentation of compound heterozygotes 1999 , 45, 200		2
28	Prenatal diagnosis of Friedreich ataxia. <i>Prenatal Diagnosis</i> , 1998 , 18, 831-833	3.2	23
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23	Regulation of mitochondrial iron accumulation by Yfh1p, a putative homolog of frataxin. <i>Science</i> , 1997 , 276, 1709-12	33.3	842
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12	Evidence of a genetic marker associated with early onset in Friedreich's ataxia. <i>Journal of Neurology</i> , 1993 , 240, 254-6	5.5	2
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9	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. <i>Human Genetics</i> , 1991 , 86, 525-8	6.3	1
8	Regional localization by in situ hybridization of a human chromosome 9 marker tightly linked to the Friedreich's ataxia locus. <i>Human Genetics</i> , 1990 , 85, 125-6	6.3	4
7	A TaqI RFLP in the locus D9S29 on human chromosome 9. <i>Nucleic Acids Research</i> , 1989 , 17, 10514	20.1	3
6	An anonymous DNA probe (LAMP 92) detects a Pvu II polymorphism on human chromosome 9 [D9S29]. <i>Nucleic Acids Research</i> , 1988 , 16, 7213	20.1	10
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3	Mammalian single-stranded DNA binding proteins and heterogeneous nuclear RNA proteins have common antigenic determinants. <i>Nucleic Acids Research</i> , 1985 , 13, 337-46	20.1	52
2	Single stranded DNA binding proteins derive from hnRNP proteins by proteolysis in mammalian cells. <i>Nucleic Acids Research</i> , 1985 , 13, 6577-90	20.1	44
1	Oxidative stress and loss of Fe-S proteins in Friedreich ataxia induced pluripotent stem cell-derived PSNs can be reversed by restoring FXN expression with a benzamide HDAC inhibitor		1

