Massimo Pandolfo

List of Publications by Citations

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	Regulation of mitochondrial iron accumulation by Yfh1p, a putative homolog of frataxin. *Science*, **1997**, 276, 1709-12	33.3	842
196	HLA-A*3101 and carbamazepine-induced hypersensitivity reactions in Europeans. *New England Journal of Medicine*, **2011**, 364, 1134-43	59.2	689
195	Common genetic variants influence human subcortical brain structures. *Nature*, **2015**, 520, 224-9	50.4	601
194	The ENIGMA Consortium: large-scale collaborative analyses of neuroimaging and genetic data. *Brain Imaging and Behavior*, **2014**, 8, 153-82	4.1	539
193	Identification of common variants associated with human hippocampal and intracranial volumes. *Nature Genetics*, **2012**, 44, 552-61	36.3	498
192	Siponimod versus placebo in secondary progressive multiple sclerosis (EXPAND): a double-blind, randomised, phase 3 study. *Lancet, The*, **2018**, 391, 1263-1273	40	422
191	Senataxin, the ortholog of a yeast RNA helicase, is mutant in ataxia-ocular apraxia 2. *Nature Genetics*, **2004**, 36, 225-7	36.3	385
190	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, 951-62	24.1	359
189	Friedreich's ataxia: point mutations and clinical presentation of compound heterozygotes. *Annals of Neurology*, **1999**, 45, 200-6	9.4	327
188	Friedreich ataxia: the clinical picture. *Journal of Neurology*, **2009**, 256 Suppl 1, 3-8	5.5	317
187	Sticky DNA: self-association properties of long GAA.TTC repeats in R.R.Y triplex structures from Friedreich's ataxia. *Molecular Cell*, **1999**, 3, 465-75	17.6	271
186	Inhibitory effects of expanded GAA.TTC triplet repeats from intron I of the Friedreich ataxia gene on transcription and replication in vivo. *Journal of Biological Chemistry*, **1998**, 273, 14588-95	5.4	239
185	Mutations in DEPDC5 cause familial focal epilepsy with variable foci. *Nature Genetics*, **2013**, 45, 546-51	36.3	238
184	Rare deletions at 16p13.11 predispose to a diverse spectrum of sporadic epilepsy syndromes. *American Journal of Human Genetics*, **2010**, 86, 707-18	11	206
183	Phenotypic variability in Friedreich ataxia: role of the associated GAA triplet repeat expansion. *Annals of Neurology*, **1997**, 41, 675-82	9.4	200
182	The pathogenesis of Friedreich ataxia and the structure and function of frataxin. *Journal of Neurology*, **2009**, 256 Suppl 1, 9-17	5.5	193
181	Diagnosis and treatment of Friedreich ataxia: a European perspective. *Nature Reviews Neurology*, **2009**, 5, 222-34	15	192

180	HDAC inhibitors correct frataxin deficiency in a Friedreich ataxia mouse model. *PLoS ONE*, **2008**, 3, e1958	3.7	174
179	The gene for paroxysmal non-kinesigenic dyskinesia encodes an enzyme in a stress response pathway. *Human Molecular Genetics*, **2004**, 13, 3161-70	5.6	164
178	Friedreich ataxia. *Archives of Neurology*, **2008**, 65, 1296-303		159
177	Multicentre search for genetic susceptibility loci in sporadic epilepsy syndrome and seizure types: a case-control study. *Lancet Neurology, The*, **2007**, 6, 970-80	24.1	152
176	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	24.1	150
175	The neurological presentation of ceruloplasmin gene mutations. *European Neurology*, **2008**, 60, 200-5	2.1	150
174	Sticky DNA, a self-associated complex formed at long GAA*TTC repeats in intron 1 of the frataxin gene, inhibits transcription. *Journal of Biological Chemistry*, **2001**, 276, 27171-7	5.4	149
173	Novel genetic loci underlying human intracranial volume identified through genome-wide association. *Nature Neuroscience*, **2016**, 19, 1569-1582	25.5	147
172	Epilepsy, hippocampal sclerosis and febrile seizures linked by common genetic variation around SCN1A. *Brain*, **2013**, 136, 3140-50	11.2	144
171	Long-term disease progression in spinocerebellar ataxia types 1, 2, 3, and 6: a longitudinal cohort study. *Lancet Neurology, The*, **2015**, 14, 1101-8	24.1	139
170	Homozygosity mapping of Hallervorden-Spatz syndrome to chromosome 20p12.3-p13. *Nature Genetics*, **1996**, 14, 479-81	36.3	139
169	Visualization, quantification and correlation of brain atrophy with clinical symptoms in spinocerebellar ataxia types 1, 3 and 6. *NeuroImage*, **2010**, 49, 158-68	7.9	133
168	Frataxin knockin mouse. *FEBS Letters*, **2002**, 512, 291-7	3.8	128
167	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, 5184-9	11.5	119
166	Common genetic variation and susceptibility to partial epilepsies: a genome-wide association study. *Brain*, **2010**, 133, 2136-47	11.2	115
165	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, 174-82	24.1	113
164	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	4.1	111
163	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	3.7	111

162	Modulation of the age at onset in spinocerebellar ataxia by CAG tracts in various genes. *Brain*, **2014**, 137, 2444-55	11.2	108
161	Aminopyridines correct early dysfunction and delay neurodegeneration in a mouse model of spinocerebellar ataxia type 1. *Journal of Neuroscience*, **2011**, 31, 11795-807	6.6	104
160	Epigenetic therapy for Friedreich ataxia. *Annals of Neurology*, **2014**, 76, 489-508	9.4	101
159	PGC-1alpha down-regulation affects the antioxidant response in Friedreich's ataxia. *PLoS ONE*, **2010**, 5, e10025	3.7	100
158	Functional genomic analysis of frataxin deficiency reveals tissue-specific alterations and identifies the PPARgamma pathway as a therapeutic target in Friedreich's ataxia. *Human Molecular Genetics*, **2009**, 18, 2452-61	5.6	94
157	Genotype-specific patterns of atrophy progression are more sensitive than clinical decline in SCA1, SCA3 and SCA6. *Brain*, **2013**, 136, 905-17	11.2	92
156	Molecular pathogenesis of Friedreich ataxia. *Archives of Neurology*, **1999**, 56, 1201-8		90
155	Deferiprone in Friedreich ataxia: a 6-month randomized controlled trial. *Annals of Neurology*, **2014**, 76, 509-21	9.4	83
154	Friedreich ataxia. *Seminars in Pediatric Neurology*, **2003**, 10, 163-72	2.9	83
153	Spinocerebellar ataxia type 8: molecular genetic comparisons and haplotype analysis of 37 families with ataxia. *American Journal of Human Genetics*, **2004**, 75, 3-16	11	81
152	Genetic architecture of subcortical brain structures in 38,851 individuals. *Nature Genetics*, **2019**, 51, 1624-1636	30.9	81
151	Progression characteristics of the European Friedreich's Ataxia Consortium for Translational Studies (EFACTS): a 2 year cohort study. *Lancet Neurology, The*, **2016**, 15, 1346-1354	24.1	78
150	Characteristics and outcomes of patients with multiple cervical artery dissection. *Stroke*, **2014**, 45, 37-41	6.7	78
149	Mapping of a gene determining familial partial epilepsy with variable foci to chromosome 22q11-q12. *American Journal of Human Genetics*, **1999**, 65, 1698-710	11	78
148	Frataxin fracas. *Nature Genetics*, **1997**, 15, 337-8	36.3	73
147	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. *Journal of Biological Chemistry*, **2001**, 276, 27178-87	5.4	66
146	Spinocerebellar ataxia types 1, 2, 3 and 6: the clinical spectrum of ataxia and morphometric brainstem and cerebellar findings. *Cerebellum*, **2012**, 11, 155-66	4.3	65
145	Early-onset ataxia with cardiomyopathy and retained tendon reflexes maps to the Friedreich's ataxia locus on chromosome 9q. *Annals of Neurology*, **1995**, 37, 359-62	9.4	65

144	Frataxin deficiency enhances apoptosis in cells differentiating into neuroectoderm. *Human Molecular Genetics*, **2001**, 10, 1935-44	5.6	63
143	Manganese superoxide dismutase induction by iron is impaired in Friedreich ataxia cells. *FEBS Letters*, **2001**, 509, 101-5	3.8	63
142	Deferiprone for the treatment of Friedreich's ataxia. *Journal of Neurochemistry*, **2013**, 126 Suppl 1, 142-66		62
141	Grafting neural precursor cells promotes functional recovery in an SCA1 mouse model. *Journal of Neuroscience*, **2009**, 29, 13126-35	6.6	62
140	Decreased brain protein levels of cytochrome oxidase subunits in Alzheimer's disease and in hereditary spinocerebellar ataxia disorders: a nonspecific change?. *Journal of Neurochemistry*, **1999**, 72, 700-7	6	62
139	Frataxin shows developmentally regulated tissue-specific expression in the mouse embryo. *Neurobiology of Disease*, **1997**, 4, 103-13	7.5	61
138	Central role and mechanisms of β-cell dysfunction and death in friedreich ataxia-associated diabetes. *Annals of Neurology*, **2012**, 72, 971-82	9.4	60
137	Consensus clinical management guidelines for Friedreich ataxia. *Orphanet Journal of Rare Diseases*, **2014**, 9, 184	4.2	56
136	Iron metabolism and mitochondrial abnormalities in Friedreich ataxia. *Blood Cells, Molecules, and Diseases*, **2002**, 29, 536-47; discussion 548-52	2.1	56
135	Knock-out of the cyaY gene in Escherichia coli does not affect cellular iron content and sensitivity to oxidants. *FEBS Letters*, **1999**, 456, 13-6	3.8	56
134	Mapping of the congenital generalized hypertrichosis locus to chromosome Xq24-q27.1. *Nature Genetics*, **1995**, 10, 202-7	36.3	55
133	Proprotein convertase subtilisin/kexin type 9 (PCSK9) gene is a risk factor of large-vessel atherosclerosis stroke. *PLoS ONE*, **2007**, 2, e1043	3.7	53
132	Down-regulation of the dopamine receptor D2 in mice lacking ataxin 1. *Human Molecular Genetics*, **2007**, 16, 2122-34	5.6	53
131	Falls in spinocerebellar ataxias: Results of the EuroSCA Fall Study. *Cerebellum*, **2010**, 9, 232-9	4.3	52
130	Mammalian single-stranded DNA binding proteins and heterogeneous nuclear RNA proteins have common antigenic determinants. *Nucleic Acids Research*, **1985**, 13, 337-46	20.1	52
129	Molecular genetics and pathogenesis of Friedreich ataxia. *Neuromuscular Disorders*, **1998**, 8, 409-15	2.9	51
128	A gene expression phenotype in lymphocytes from Friedreich ataxia patients. *Annals of Neurology*, **2011**, 70, 790-804	9.4	49
127	Drug Insight: antioxidant therapy in inherited ataxias. *Nature Clinical Practice Neurology*, **2008**, 4, 86-96		48

126	Familial partial epilepsy with variable foci: clinical features and linkage to chromosome 22q12. *Epilepsia*, **2004**, 45, 1054-60	6.4	48
125	Unveiling a common mechanism of apoptosis in □cells and neurons in Friedreich's ataxia. *Human Molecular Genetics*, **2015**, 24, 2274-86	5.6	47
124	Gene expression profiling in frataxin deficient mice: microarray evidence for significant expression changes without detectable neurodegeneration. *Neurobiology of Disease*, **2006**, 22, 302-11	7.5	47
123	Survival in patients with spinocerebellar ataxia types 1, 2, 3, and 6 (EUROSCA): a longitudinal cohort study. *Lancet Neurology, The*, **2018**, 17, 327-334	24.1	46
122	A 22-Year Follow-up Study of Long-term Cardiac Outcome and Predictors of Survival in Friedreich Ataxia. *JAMA Neurology*, **2015**, 72, 1334-41	17.2	45
121	Single stranded DNA binding proteins derive from hnRNP proteins by proteolysis in mammalian cells. *Nucleic Acids Research*, **1985**, 13, 6577-90	20.1	44
120	Brain glyceraldehyde-3-phosphate dehydrogenase activity in human trinucleotide repeat disorders. *Archives of Neurology*, **1998**, 55, 1299-304		43
119	Increasing frataxin gene expression with histone deacetylase inhibitors as a therapeutic approach for Friedreich's ataxia. *Journal of Neurochemistry*, **2013**, 126 Suppl 1, 147-54	6	42
118	The S18Y polymorphism in the UCHL1 gene is a genetic modifier in Huntington's disease. *Neurogenetics*, **2006**, 7, 27-30	3	42
117	The molecular basis of Friedreich ataxia. *Advances in Experimental Medicine and Biology*, **2002**, 516, 99-11[illegible]6		42
116	Friedreich's ataxia: clinical aspects and pathogenesis. *Seminars in Neurology*, **1999**, 19, 311-21	3.2	40
115	Delayed-onset Friedreich's ataxia revisited. *Movement Disorders*, **2016**, 31, 62-9	7	40
114	A new clinical and molecular form of Unverricht-Lundborg disease localized by homozygosity mapping. *Brain*, **2005**, 128, 652-8	11.2	39
113	Variant on 9p21 strongly associates with coronary heart disease, but lacks association with common stroke. *European Journal of Human Genetics*, **2009**, 17, 1287-93	5.3	38
112	Multifaceted role of galectin-3 on human glioblastoma cell motility. *Biochemical and Biophysical Research Communications*, **2004**, 325, 1393-8	3.4	37
111	Hemicerebellectomy blocks the enhancement of cortical motor output associated with repetitive somatosensory stimulation in the rat. *Journal of Physiology*, **2005**, 567, 293-300	3.9	37
110	Genetic analysis of candidate genes modifying the age-at-onset in Huntington's disease. *Human Genetics*, **2006**, 120, 285-92	6.3	36
109	Genome-wide mapping for clinically relevant predictors of lamotrigine- and phenytoin-induced hypersensitivity reactions. *Pharmacogenomics*, **2012**, 13, 399-405	2.6	34

108	Eight novel mutations in SPG4 in a large sample of patients with hereditary spastic paraplegia. *Archives of Neurology*, **2006**, 63, 750-5		34
107	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, 4847-4855	5.6	33
106	Associations of apolipoprotein E gene with ischemic stroke and intracranial atherosclerosis. *European Journal of Human Genetics*, **2008**, 16, 955-60	5.3	31
105	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	4.1	30
104	Genetics of epilepsy. *Seminars in Neurology*, **2011**, 31, 506-18	3.2	29
103	Variants of the KCNMB3 regulatory subunit of maxi BK channels affect channel inactivation. *Physiological Genomics*, **2003**, 15, 191-8	3.6	29
102	Molecular basis of Friedreich ataxia. *Movement Disorders*, **2001**, 16, 815-21	7	29
101	MME mutation in dominant spinocerebellar ataxia with neuropathy (SCA43). *Neurology: Genetics*, **2016**, 2, e94	3.8	28
100	Pharmacological treatments for Friedreich ataxia. *The Cochrane Library*, **2016**, CD007791	5.2	28
99	Blood-brain barrier promotes differentiation of human fetal neural precursor cells. *Stem Cells*, **2009** , 27, 838-46	5.8	28
98	Normal left ventricular ejection fraction and mass but subclinical myocardial dysfunction in patients with Friedreich's ataxia. *European Heart Journal Cardiovascular Imaging*, **2012**, 13, 346-52	4.1	28
97	Frataxin deficiency and mitochondrial dysfunction. *Mitochondrion*, **2002**, 2, 87-93	4.9	28
96	Large-scale purification of hnRNP proteins from HeLa cells by affinity chromatography on ssDNA-cellulose. *FEBS Journal*, **1987**, 162, 213-20		28
95	Friedreich ataxia: Detection of GAA repeat expansions and frataxin point mutations. *Methods in Molecular Medicine*, **2006**, 126, 197-216		26
94	Cerebellar and afferent ataxias. *CONTINUUM Lifelong Learning in Neurology*, **2013**, 19, 1312-43	3	24
93	The gene for spinal cerebellar ataxia 1 (SCA1) is flanked by two closely linked highly polymorphic microsatellite loci. *Human Molecular Genetics*, **1993**, 2, 1383-7	5.6	24
92	Antioxidants and other pharmacological treatments for Friedreich ataxia. *Cochrane Database of Systematic Reviews*, **2012**, CD007791		23
91	Prenatal diagnosis of Friedreich ataxia. *Prenatal Diagnosis*, **1998**, 18, 831-833	3.2	23

90	Frataxin overexpressing mice. *FEBS Letters*, **2004**, 572, 281-8	3.8	23
89	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. *Cerebellum*, **2005**, 4, 230-8	4.3	23
88	Exenatide induces frataxin expression and improves mitochondrial function in Friedreich ataxia. *JCI Insight*, **2020**, 5,	9.9	23
87	Friedreich ataxia: new pathways. *Journal of Child Neurology*, **2012**, 27, 1204-11	2.5	22
86	A recessive ataxia diagnosis algorithm for the next generation sequencing era. *Annals of Neurology*, **2017**, 82, 892-899	9.4	20
85	Development of histone deacetylase inhibitors as therapeutics for neurological disease. *Future Neurology*, **2009**, 4, 775-784	1.5	20
84	Evidence for genetically determined degeneration of proprioceptive tracts in Friedreich ataxia. *Neurology*, **2019**, 93, e116-e124	6.5	20
83	Mutations in TNK2 in severe autosomal recessive infantile onset epilepsy. *Annals of Neurology*, **2013**, 74, 496-501	9.4	18
82	The myelin basic protein gene is not a major susceptibility locus for multiple sclerosis in Italian patients. *Journal of Neurology*, **1994**, 241, 615-9	5.5	17
81	Cerebellar cognitive disorder parallels cerebellar motor symptoms in Friedreich ataxia. *Annals of Clinical and Translational Neurology*, **2020**, 7, 1050-1054	5.3	16
80	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, 458-63	6.3	16
79	Molecular genetics of the hereditary ataxias. *Advances in Genetics*, **1998**, 38, 31-68	3.3	16
78	A missense mutation (W155R) in an American patient with Friedreich Ataxia. *Human Mutation*, **1999** , 13, 506-506	4.7	16
77	Long-term evolution of patient-reported outcome measures in spinocerebellar ataxias. *Journal of Neurology*, **2018**, 265, 2040-2051	5.5	16
76	Friedreich ataxia. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2012**, 103, 275-943		15
75	Study of the adenosinergic system in the brain of HPRT knockout mouse (Lesch-Nyhan disease). *Clinica Chimica Acta*, **2006**, 373, 104-7	6.2	15
74	Heteroduplexes may confuse the interpretation of PCR-based molecular tests for the Friedreich ataxia GAA triplet repeat. *Human Mutation*, **1999**, 13, 328-30	4.7	15
73	Personality and Neuropsychological Profiles in Friedreich Ataxia. *Cerebellum*, **2018**, 17, 204-212	4.3	14

72	A new locus for familial temporal lobe epilepsy on chromosome 3q. *Epilepsy Research*, **2013**, 106, 338-443		14
71	Body Mass Index Decline Is Related to Spinocerebellar Ataxia Disease Progression. *Movement Disorders Clinical Practice*, **2017**, 4, 689-697	2.2	14
70	Common data elements for clinical research in Friedreich's ataxia. *Movement Disorders*, **2013**, 28, 190-5	7	14
69	Primary proprioceptive neurons from human induced pluripotent stem cells: a cell model for afferent ataxias. *Scientific Reports*, **2020**, 10, 7752	4.9	14
68	Automated functional upper limb evaluation of patients with Friedreich ataxia using serious games rehabilitation exercises. *Journal of NeuroEngineering and Rehabilitation*, **2018**, 15, 87	5.3	14
67	Urinary, bowel and sexual symptoms in a cohort of patients with Friedreich's ataxia. *Orphanet Journal of Rare Diseases*, **2017**, 12, 158	4.2	13
66	150 years of Friedreich ataxia: from its discovery to therapy. *Journal of Neurochemistry*, **2013**, 126 Suppl 1, 1-3	6	13
65	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, 346-9	3.3	13
64	Suggestive evidence for a new locus for epilepsy with heterogeneous phenotypes on chromosome 17q. *Epilepsy Research*, **2010**, 88, 65-75	3	13
63	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, 3425-37	5.6	12
62	Interaction between repetitive stimulation of the sciatic nerve and functional ablation of cerebellar nucleus interpositus in the rat. *Cerebellum*, **2004**, 3, 21-6	4.3	12
61	Striking intrafamilial phenotypic variability and spastic paraplegia in the presence of similar homozygous expansions of the FRDA1 gene. *Movement Disorders*, **2004**, 19, 1424-31	7	12
60	Myorhythmia associated with Hodgkin's lymphoma. *Journal of Neurology*, **2003**, 250, 1382-4	5.5	12
59	A rapid method to isolate (GT)n repeats from yeast artificial chromosomes. *Nucleic Acids Research*, **1992**, 20, 1154	20.1	12
58	Evidence of linkage between susceptibility to multiple sclerosis and HLA-class II loci in Italian multiplex families. *European Journal of Human Genetics*, **1995**, 3, 303-11	5.3	12
57	Friedreich's Ataxia **2006**, 277-296		12
56	Central Nervous System Therapeutic Targets in Friedreich Ataxia. *Human Gene Therapy*, **2020**, 31, 1226-1236	4.8	12
55	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	24.1	12

54	Age of onset determines intrinsic functional brain architecture in Friedreich ataxia. *Annals of Clinical and Translational Neurology*, **2020**, 7, 94-104	5.3	11
53	Friedreich and dominant ataxias: quantitative differences in cerebellar dysfunction measurements. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 559-565	5.5	11
52	Pediatric epilepsy genetics. *Current Opinion in Neurology*, **2013**, 26, 137-45	7.1	11
51	Molecular basis of hypoxanthine-guanine phosphoribosyltransferase deficiency in Italian Lesch-Nyhan patients: identification of nine novel mutations. *Journal of Inherited Metabolic Disease*, **2004**, 27, 767-73	5.4	11
50	Altered neocortical tactile but preserved auditory early change detection responses in Friedreich ataxia. *Clinical Neurophysiology*, **2019**, 130, 1299-1310	4.3	10
49	Neurologic outcomes in Friedreich ataxia: Study of a single-site cohort. *Neurology: Genetics*, **2020**, 6, e415	3.8	10
48	An anonymous DNA probe (LAMP 92) detects a Pvu II polymorphism on human chromosome 9 [D9S29]. *Nucleic Acids Research*, **1988**, 16, 7213	20.1	10
47	Mapping of Friedreich's ataxia locus by identification of recombination events in patients homozygous by descent. *European Journal of Human Genetics*, **1994**, 2, 291-9	5.3	9
46	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. *Neuropharmacology*, **2017**, 123, 34-45	5.5	8
45	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	3.2	8
44	Effects of levetiracetam on the production of nitric oxide--an in vivo study. *Journal of Neurology*, **2005**, 252, 727-30	5.5	8
43	Localization of the human gene for carnitine palmitoyltransferase to 1p13-p11 by nonradioactive in situ hybridization. *Genomics*, **1992**, 13, 1372-4	4.3	8
42	Onset features and time to diagnosis in Friedreich's Ataxia. *Orphanet Journal of Rare Diseases*, **2020**, 15, 198	4.2	8
41	Standardized Assessment of Hereditary Ataxia Patients in Clinical Studies. *Movement Disorders Clinical Practice*, **2016**, 3, 230-240	2.2	7
40	Adult neural precursor cells form connexin-dependent networks that improve their survival. *NeuroReport*, **2015**, 26, 928-36	1.7	7
39	Bilateral high-frequency synchronous discharges: a new form of tremor in humans. *Archives of Neurology*, **2003**, 60, 416-22		7
38	Neuropathology of the inherited ataxias **2001**, 387-406		7
37	Prediction of Survival With Long-Term Disease Progression in Most Common Spinocerebellar Ataxia. *Movement Disorders*, **2019**, 34, 1220-1227	7	6

36	Transient CNS deficits and migrainous auras in individuals without a history of headache. *Headache*, **2014**, 54, 493-9	4.2	6
35	Test-retest reliability of the Friedreich's ataxia rating scale. *Annals of Clinical and Translational Neurology*, **2020**, 7, 1708-1712	5.3	6
34	HLA genotype as a marker of multiple sclerosis prognosis: A pilot study. *Journal of the Neurological Sciences*, **2017**, 375, 348-354	3.2	5
33	Neurodegeneration With Brain Iron Accumulation (NBIA) Syndromes Presenting With Late-Onset Craniocervical Dystonia: An Illustrative Case Series?. *Movement Disorders Clinical Practice*, **2017**, 4, 254-257	2.2	5
32	Building bridges between neuroscientific evidence and policy. *Lancet Neurology, The*, **2015**, 14, 242-5	24.1	5
31	Treatment of Friedreich's ataxia. *Expert Opinion on Orphan Drugs*, **2013**, 1, 221-234	1.1	5
30	Iron metabolism in mice with partial frataxin deficiency. *Cerebellum*, **2003**, 2, 146-53	4.3	5
29	Friedreich's ataxia associated with mitochondrial myopathy: clinicopathologic report. *Journal of Child Neurology*, **2002**, 17, 453-6	2.5	5
28	Cerebellar ataxia, neuropathy, hearing loss, and intellectual disability due to AIFM1 mutation. *Neurology: Genetics*, **2020**, 6, e420	3.8	4
27	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	6.3	4
26	Friedreich's Ataxia and Related DNA Loss-of-Function Disorders **2007**, 277-294		3
25	A TaqI RFLP in the locus D9S29 on human chromosome 9. *Nucleic Acids Research*, **1989**, 17, 10514	20.1	3
24	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	4.2	3
23	Hand Dexterity and Pyramidal Dysfunction in Friedreich Ataxia, A Finger Tapping Study. *Movement Disorders Clinical Practice*, **2021**, 8, 85-91	2.2	3
22	Age of onset modulates resting-state brain network dynamics in Friedreich Ataxia. *Human Brain Mapping*, **2021**, 42, 5334-5344	5.9	3
21	Cardiomyopathy in Friedreich's ataxia. *Acta Neurologica Belgica*, **2011**, 111, 183-7	1.5	3
20	Real-time PCR and linkage studies to identify carriers presenting HPRT deleted gene. *Molecular Medicine*, **2006**, 12, 246-51	6.2	2
19	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, 775-7	5.8	2

18	Evidence of a genetic marker associated with early onset in Friedreich's ataxia. *Journal of Neurology* , **1993**, 240, 254-6	5.5	2
17	A Pvu II RFLP in the human ADH3 gene. *Nucleic Acids Research*, **1988**, 16, 11857	20.1	2
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