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	Regulation of Mitochondrial Iron Accumulation by Yfh1p, a Putative Homolog of Frataxin. Science, 1997, 276, 1709-1712.	12.6	942
2	HLA-A*3101 and Carbamazepine-Induced Hypersensitivity Reactions in Europeans. New England Journal of Medicine, 2011, 364, 1134-1143.	27.0	815
3	Common genetic variants influence human subcortical brain structures. Nature, 2015, 520, 224-229.	27.8	772
4	The ENIGMA Consortium: large-scale collaborative analyses of neuroimaging and genetic data. Brain Imaging and Behavior, 2014, 8, 153-182.	2.1	696
5	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
6	Identification of common variants associated with human hippocampal and intracranial volumes. Nature Genetics, 2012, 44, 552-561.	21.4	594
7	Senataxin, the ortholog of a yeast RNA helicase, is mutant in ataxia-ocular apraxia 2. Nature Genetics, 2004, 36, 225-227.	21.4	454
8	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
9	Friedreich ataxia: The clinical picture. Journal of Neurology, 2009, 256, 3-8.	3.6	380
10	Friedreich's ataxia: Point mutations and clinical presentation of compound heterozygotes. Annals of Neurology, 1999, 45, 200-206.	5.3	371
11	Sticky DNA. Molecular Cell, 1999, 3, 465-475.	9.7	305
12	Mutations in DEPDC5 cause familial focal epilepsy with variable foci. Nature Genetics, 2013, 45, 546-551.	21.4	301
13	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
14	Phenotypic variability in friedreich ataxia: Role of the associated GAA triplet repeat expansion. Annals of Neurology, 1997, 41, 675-682.	5.3	249
15	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
16	Diagnosis and treatment of Friedreich ataxia: a European perspective. Nature Reviews Neurology, 2009, 5, 222-234.	10.1	231
17	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
18	The pathogenesis of Friedreich ataxia and the structure and function of frataxin. Journal of Neurology, 2009, 256, 9-17.	3.6	220

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19	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
20	Novel genetic loci underlying human intracranial volume identified through genome-wide association. Nature Neuroscience, 2016, 19, 1569-1582.	14.8	213
21	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
22	Friedreich Ataxia. Archives of Neurology, 2008, 65, 1296-303.	4.5	194
23	HDAC Inhibitors Correct Frataxin Deficiency in a Friedreich Ataxia Mouse Model. PLoS ONE, 2008, 3, e1958.	2.5	193
24	Genetic architecture of subcortical brain structures in 38,851 individuals. Nature Genetics, 2019, 51, 1624-1636.	21.4	192
25	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
26	The Neurological Presentation of Ceruloplasmin Gene Mutations. European Neurology, 2008, 60, 200-205.	1.4	171
27	Epilepsy, hippocampal sclerosis and febrile seizures linked by common genetic variation around SCN1A. Brain, 2013, 136, 3140-3150.	7.6	168
28	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
29	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
30	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
31	Homozygosity mapping of Hallervordenâ€“Spatz syndrome to chromosome 20p12.3â€“p13. Nature Genetics, 1996, 14, 479-481.	21.4	158
32	Frataxin knockin mouse. FEBS Letters, 2002, 512, 291-297.	2.8	155
33	Modulation of the age at onset in spinocerebellar ataxia by CAG tracts in various genes. Brain, 2014, 137, 2444-2455.	7.6	144
34	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
35	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
36	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

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37	Common genetic variation and susceptibility to partial epilepsies: a genome-wide association study. Brain, 2010, 133, 2136-2147.	7.6	132
38	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
39	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
40	Epigenetic therapy for <scp>F</scp>riedreich ataxia. Annals of Neurology, 2014, 76, 489-508.	5.3	128
41	PGC-1alpha Down-Regulation Affects the Antioxidant Response in Friedreich's Ataxia. PLoS ONE, 2010, 5, e10025.	2.5	118
42	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
43	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
44	Deferiprone in <scp>F</scp>riedreich ataxia: A 6â€Month randomized controlled trial. Annals of Neurology, 2014, 76, 509-521.	5.3	109
45	Molecular Pathogenesis of Friedreich Ataxia. Archives of Neurology, 1999, 56, 1201.	4.5	103
46	Characteristics and Outcomes of Patients With Multiple Cervical Artery Dissection. Stroke, 2014, 45, 37-41.	2.0	96
47	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
48	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
49	Friedreich ataxia. Seminars in Pediatric Neurology, 2003, 10, 163-172.	2.0	87
50	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
51	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
52	Frataxin fracas. Nature Genetics, 1997, 15, 337-338.	21.4	78
53	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
54	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

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55	Consensus clinical management guidelines for Friedreich ataxia. Orphanet Journal of Rare Diseases, 2014, 9, 184.	2.7	76
56	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
57	Manganese superoxide dismutase induction by iron is impaired in Friedreich ataxia cells. FEBS Letters, 2001, 509, 101-105.	2.8	71
58	Frataxin deficiency enhances apoptosis in cells differentiating into neuroectoderm. Human Molecular Genetics, 2001, 10, 1935-1944.	2.9	71
59	Familial Partial Epilepsy with Variable Foci: Clinical Features and Linkage to Chromosome 22q12. Epilepsia, 2004, 45, 1054-1060.	5.1	71
60	Deferiprone for the treatment of Friedreich's ataxia. Journal of Neurochemistry, 2013, 126, 142-146.	3.9	71
61	Grafting Neural Precursor Cells Promotes Functional Recovery in an SCA1 Mouse Model. Journal of Neuroscience, 2009, 29, 13126-13135.	3.6	70
62	Frataxin Shows Developmentally Regulated Tissue-Specific Expression in the Mouse Embryo. Neurobiology of Disease, 1997, 4, 103-113.	4.4	69
63	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
64	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
65	Mapping of the congenital generalized hypertrichosis locus to chromosome Xq24â€“q27.1. Nature Genetics, 1995, 10, 202-207.	21.4	68
66	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
67	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
68	Iron Metabolism and Mitochondrial Abnormalities in Friedreich Ataxia. Blood Cells, Molecules, and Diseases, 2002, 29, 536-547.	1.4	61
69	Down-regulation of the dopamine receptor D2 in mice lacking ataxin 1. Human Molecular Genetics, 2007, 16, 2122-2134.	2.9	61
70	Falls in Spinocerebellar Ataxias: Results of the EuroSCA Fall Study. Cerebellum, 2010, 9, 232-239.	2.5	59
71	A gene expression phenotype in lymphocytes from friedreich ataxia patients. Annals of Neurology, 2011, 70, 790-804.	5.3	58
72	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

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73	Molecular genetics and pathogenesis of Friedreich ataxia. Neuromuscular Disorders, 1998, 8, 409-415.	0.6	57
74	Delayedâ€onset Friedreich's ataxia revisited. Movement Disorders, 2016, 31, 62-69.	3.9	54
75	Drug Insight: antioxidant therapy in inherited ataxias. Nature Clinical Practice Neurology, 2008, 4, 86-96.	2.5	53
76	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
77	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
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	Brain Glyceraldehyde-3-Phosphate Dehydrogenase Activity in Human Trinucleotide Repeat Disorders. Archives of Neurology, 1998, 55, 1299.	4.5	50
80	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
81	The S18Y polymorphism in the UCHL1 gene is a genetic modifier in Huntingtonâ€™s disease. Neurogenetics, 2006, 7, 27-30.	1.4	47
82	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
83	Single stranded DNA binding proteins derive from hnRNP proteins by proteolysis in mammalian cells. Nucleic Acids Research, 1985, 13, 6577-6590.	14.5	45
84	A new clinical and molecular form of Unverricht-Lundborg disease localized by homozygosity mapping. Brain, 2005, 128, 652-658.	7.6	45
85	The Molecular Basis of Friedreich Ataxia. Advances in Experimental Medicine and Biology, 2002, 516, 99-118.	1.6	45
86	Pharmacological treatments for Friedreich ataxia. The Cochrane Library, 2016, 2016, CD007791.	2.8	44
87	Friedreich's Ataxia: Clinical Aspects and Pathogenesis. Seminars in Neurology, 1999, 19, 311-321.	1.4	43
88	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
89	MME mutation in dominant spinocerebellar ataxia with neuropathy (SCA43). Neurology: Genetics, 2016, 2, e94.	1.9	41
90	Multifaceted role of galectin-3 on human glioblastoma cell motility. Biochemical and Biophysical Research Communications, 2004, 325, 1393-1398.	2.1	40

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91	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
92	Genetic analysis of candidate genes modifying the age-at-onset in Huntingtonâ€™s disease. Human Genetics, 2006, 120, 285-292.	3.8	39
93	Eight Novel Mutations in SPG4 in a Large Sample of Patients With Hereditary Spastic Paraplegia. Archives of Neurology, 2006, 63, 750.	4.5	39
94	Associations of apolipoprotein E gene with ischemic stroke and intracranial atherosclerosis. European Journal of Human Genetics, 2008, 16, 955-960.	2.8	39
95	Genetics of Epilepsy. Seminars in Neurology, 2011, 31, 506-518.	1.4	39
96	Cerebellar and Afferent Ataxias. CONTINUUM Lifelong Learning in Neurology, 2013, 19, 1312-1343.	0.8	39
97	Exenatide induces frataxin expression and improves mitochondrial function in Friedreich ataxia. JCI Insight, 2020, 5, .	5.0	39
98	Molecular basis of Friedreich ataxia. Movement Disorders, 2001, 16, 815-821.	3.9	38
99	Genome-wide mapping for clinically relevant predictors of lamotrigine- and phenytoin-induced hypersensitivity reactions. Pharmacogenomics, 2012, 13, 399-405.	1.3	38
100	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
101	Variants of the KCNMB3 regulatory subunit of maxi BK channels affect channel inactivation. Physiological Genomics, 2003, 15, 191-198.	2.3	34
102	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
103	Long-term evolution of patient-reported outcome measures in spinocerebellar ataxias. Journal of Neurology, 2018, 265, 2040-2051.	3.6	34
104	Cerebellar cognitive disorder parallels cerebellar motor symptoms in Friedreich ataxia. Annals of Clinical and Translational Neurology, 2020, 7, 1050-1054.	3.7	32
105	Large-scale purification of hnRNP proteins from HeLa cells by affinity chromatography on ssDNA-cellulose. FEBS Journal, 1987, 162, 213-220.	0.2	31
106	Frataxin deficiency and mitochondrial dysfunction. Mitochondrion, 2002, 2, 87-93.	3.4	31
107	Blood-Brain Barrier Promotes Differentiation of Human Fetal Neural Precursor Cells. Stem Cells, 2009, 27, 838-846.	3.2	31
108	Evidence for genetically determined degeneration of proprioceptive tracts in Friedreich ataxia. Neurology, 2019, 93, e116-e124.	1.1	30

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109	Frataxin overexpressing mice. FEBS Letters, 2004, 572, 281-288.	2.8	27
110	Friedreich Ataxia: Detection of GAA Repeat Expansions and Frataxin Point Mutations. , 2006, 126, 197-216.		27
111	A recessive ataxia diagnosis algorithm for the next generation sequencing era. Annals of Neurology, 2017, 82, 892-899.	5.3	27
112	Onset features and time to diagnosis in Friedreichâ€™s Ataxia. Orphanet Journal of Rare Diseases, 2020, 15, 198.	2.7	27
113	Neurologic outcomes in Friedreich ataxia. Neurology: Genetics, 2020, 6, e415.	1.9	27
114	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
115	Prenatal diagnosis of Friedreich ataxia. Prenatal Diagnosis, 1998, 18, 831-833.	2.3	26
116	Antioxidants and other pharmacological treatments for Friedreich ataxia. , 2012, , CD007791.		26
117	Central Nervous System Therapeutic Targets in Friedreich Ataxia. Human Gene Therapy, 2020, 31, 1226-1236.	2.7	26
118	Development of histone deacetylase inhibitors as therapeutics for neurological disease. Future Neurology, 2009, 4, 775-784.	0.5	25
119	Friedreich Ataxia. Journal of Child Neurology, 2012, 27, 1204-1211.	1.4	25
120	Friedreich ataxia. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2012, 103, 275-294.	1.8	25
121	Body Mass Index Decline Is Related to Spinocerebellar Ataxia Disease Progression. Movement Disorders Clinical Practice, 2017, 4, 689-697.	1.5	25
122	Primary proprioceptive neurons from human induced pluripotent stem cells: a cell model for afferent ataxias. Scientific Reports, 2020, 10, 7752.	3.3	24
123	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
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	Mutations in TNK2 in severe autosomal recessive infantile onset epilepsy. Annals of Neurology, 2013, 74, 496-501.	5.3	22
126	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

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127	Personality and Neuropsychological Profiles in Friedreich Ataxia. Cerebellum, 2018, 17, 204-212.	2.5	21
128	Age of onset determines intrinsic functional brain architecture in Friedreich ataxia. Annals of Clinical and Translational Neurology, 2020, 7, 94-104.	3.7	21
129	2 Molecular Genetics of the Hereditary Ataxias. Advances in Genetics, 1998, 38, 31-68.	1.8	19
130	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
131	Localization of the human gene for carnitine palmitoyltransferase to 1p13â€“p11 by nonradioactive in situ hybridization. Genomics, 1992, 13, 1372-1374.	2.9	18
132	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
133	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
134	A rapid method to isolate (GT)n repeats from yeast artificial chromosomes. Nucleic Acids Research, 1992, 20, 1154-1154.	14.5	17
135	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
136	A missense mutation (W155R) in an American patient with Friedreich Ataxia. Human Mutation, 1999, 13, 506-506.	2.5	17
137	A new locus for familial temporal lobe epilepsy on chromosome 3q. Epilepsy Research, 2013, 106, 338-344.	1.6	17
138	150Âyears of Friedreich Ataxia: from its discovery to therapy. Journal of Neurochemistry, 2013, 126, 1-3.	3.9	17
139	Pediatric epilepsy genetics. Current Opinion in Neurology, 2013, 26, 137-145.	3.6	17
140	Suggestive evidence for a new locus for epilepsy with heterogeneous phenotypes on chromosome 17q. Epilepsy Research, 2010, 88, 65-75.	1.6	16
141	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
142	Friedreich and dominant ataxias: quantitative differences in cerebellar dysfunction measurements. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 559-565.	1.9	16
143	Friedreich's Ataxia. , 2006, , 277-296.		16
144	Myorhythmia associated with Hodgkin?s lymphoma. Journal of Neurology, 2003, 250, 1382-1384.	3.6	15

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145	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
146	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
147	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
148	Common data elements for clinical research in Friedreich's ataxia. Movement Disorders, 2013, 28, 190-195.	3.9	14
149	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
150	Prediction of Survival With Longâ€Term Disease Progression in Most Common Spinocerebellar Ataxia. Movement Disorders, 2019, 34, 1220-1227.	3.9	14
151	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
152	Standardized Assessment of Hereditary Ataxia Patients in Clinical Studies. Movement Disorders Clinical Practice, 2016, 3, 230-240.	1.5	13
153	Altered neocortical tactile but preserved auditory early change detection responses in Friedreich ataxia. Clinical Neurophysiology, 2019, 130, 1299-1310.	1.5	13
154	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
155	Adult neural precursor cells form connexin-dependent networks that improve their survival. NeuroReport, 2015, 26, 928-936.	1.2	12
156	Testâ€“retest reliability of the Friedreichâ€™s ataxia rating scale. Annals of Clinical and Translational Neurology, 2020, 7, 1708-1712.	3.7	12
157	Recessive cerebellar and afferent ataxias â€” clinical challenges and future directions. Nature Reviews Neurology, 2022, 18, 257-272.	10.1	12
158	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
159	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
160	Cerebellar ataxia, neuropathy, hearing loss, and intellectual disability due to AIFM1 mutation. Neurology: Genetics, 2020, 6, e420.	1.9	10
161	Neuropathology of the inherited ataxias. , 2001, , 387-406.		10
162	Friedreich's Ataxia Associated With Mitochondrial Myopathy: Clinicopathologic Report. Journal of Child Neurology, 2002, 17, 453-456.	1.4	8

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163	Bilateral High-Frequency Synchronous Discharges. Archives of Neurology, 2003, 60, 416.	4.5	8
164	Effects of levetiracetam on the production of nitric oxide. Journal of Neurology, 2005, 252, 727-730.	3.6	8
165	Introduction. Journal of Neurology, 2009, 256, 1-2.	3.6	8
166	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
167	HLA genotype as a marker of multiple sclerosis prognosis: A pilot study. Journal of the Neurological Sciences, 2017, 375, 348-354.	0.6	8
168	Age of onset modulates restingâ€state brain network dynamics in Friedreich Ataxia. Human Brain Mapping, 2021, 42, 5334-5344.	3.6	8
169	Treatment of Friedreich's ataxia. Expert Opinion on Orphan Drugs, 2013, 1, 221-234.	0.8	7
170	Transient <scp>CNS</scp> Deficits and Migrainous Auras in Individuals Without a History of Headache. Headache, 2014, 54, 493-499.	3.9	7
171	DNA methylation in Friedreich ataxia silences expression of frataxin isoform E. Scientific Reports, 2022, 12, 5031.	3.3	7
172	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
173	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
174	Iron metabolism in mice with partial frataxin deficiency. Cerebellum, 2003, 2, 146-153.	2.5	5
175	Building bridges between neuroscientific evidence and policy. Lancet Neurology, The, 2015, 14, 242-245.	10.2	5
176	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
177	A TaqI RFLP in the locus D9S29 on human chromosome 9. Nucleic Acids Research, 1989, 17, 10514-10514.	14.5	4
178	Hand Dexterity and Pyramidal Dysfunction in Friedreich Ataxia, A Finger Tapping Study. Movement Disorders Clinical Practice, 2021, 8, 85-91.	1.5	4
179	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
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