Francesca Cavalcanti

List of Publications by Year
in descending order

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37 papers	4,315 citations	331670 21 h-index	345221 36 g-index
37 all docs	37 docs citations	37 times ranked	3731 citing authors

#	Article	IF	Citations
1	Hereditary hyperekplexia: a new family and a systematic review of GLRA1 gene-related phenotypes. Pediatric Neurology, 2022, , .	2.1	2
2	Interruptions of the FXN GAA Repeat Tract Delay the Age at Onset of Friedreichâ€™s Ataxia in a Location Dependent Manner. International Journal of Molecular Sciences, 2021, 22, 7507.	4.1	10
3	The Mitochondrial Dysfunction Hypothesis in Autism Spectrum Disorders: Current Status and Future Perspectives. International Journal of Molecular Sciences, 2020, 21, 5785.	4.1	29
4	Clinical features and genetic analysis of two siblings with startle disease in an Italian family: a case report. BMC Medical Genetics, 2019, 20, 40.	2.1	4
5	Expanding the global prevalence of spinocerebellar ataxia type 42. Neurology: Genetics, 2018, 4, e232.	1.9	14
6	Large Interruptions of GAA Repeat Expansion Mutations in Friedreich Ataxia Are Very Rare. Frontiers in Cellular Neuroscience, 2018, 12, 443.	3.7	20
7	NeuroArray: A Customized aCGH for the Analysis of Copy Number Variations in Neurological Disorders. Current Genomics, 2018, 19, 431-443.	1.6	3
8	A customized high-resolution array-comparative genomic hybridization to explore copy number variations in Parkinsonâ€™s disease. Neurogenetics, 2016, 17, 233-244.	1.4	10
9	Splicing: is there an alternative contribution to Parkinsonâ€™s disease?. Neurogenetics, 2015, 16, 245-263.	1.4	54
10	Sacsin-Related Spastic Ataxia Caused by a Novel Missense Mutation p.Arg272His in a Patient from Sicily, Southern Italy. Cerebellum, 2013, 12, 589-592.	2.5	3
11	Mutations in PRRT2 result in familial infantile seizures with heterogeneous phenotypes including febrile convulsions and probable SUDEP. Epilepsy Research, 2013, 104, 280-284.	1.6	29
12	Ataxin-1 and ataxin-2 intermediate-length PolyQ expansions in amyotrophic lateral sclerosis. Neurology, 2012, 79, 2315-2320.	1.1	70
13	An Axon Regeneration Signature in a Charcot-Marie-Tooth Disease Type 2 Patient. Journal of Neurogenetics, 2009, 23, 324-328.	1.4	4
14	Vitamin E deficiency due to chylomicron retention disease in Marinesco-Sjï¿½gren syndrome. Annals of Neurology, 2000, 47, 260-264.	5.3	40
15	Accuracy of clinical diagnostic criteria for Friedreich's ataxia. Movement Disorders, 2000, 15, 1255-1258.	3.9	39
16	Atypical Friedreich ataxia phenotype associated with a novel missense mutation in the X25 gene. Neurology, 2000, 54, 496-496.	1.1	26
17	Relation between trinucleotide GAA repeat length and sensory neuropathy in Friedreich's ataxia. Journal of Neurology, Neurosurgery and Psychiatry, 1999, 66, 93-96.	1.9	59
18	Why do some Friedreich's ataxia patients retain tendon reflexes?. Journal of Neurology, 1999, 246, 353-357.	3.6	36

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19	Friedreich's ataxia: Point mutations and clinical presentation of compound heterozygotes. Annals of Neurology, 1999, 45, 200-206.	5.3	371
20	Determinants of onset age in Friedreichâ€™s ataxia. Journal of Neurology, 1998, 245, 166-168.	3.6	35
21	A New Locus for Autosomal Recessive Hereditary Spastic Paraplegia Maps to Chromosome 16q24.3. American Journal of Human Genetics, 1998, 63, 135-139.	6.2	147
22	Parental gender, age at birth and expansion length influence GAA repeat intergenerational instability in the X25 gene: pedigree studies and analysis of sperm from patients with Friedreich's Ataxia. Human Molecular Genetics, 1998, 7, 1901-1906.	2.9	75
23	The Friedreich ataxia GAA triplet repeat: premutation and normal alleles. Human Molecular Genetics, 1997, 6, 1261-1266.	2.9	188
24	Broadened Friedreich's ataxia phenotype after gene cloning. Neurology, 1997, 49, 1617-1620.	1.1	55
25	1-29-08 Linkage study in an Italian family with autosomal recessive spastic paraplegia. Journal of the Neurological Sciences, 1997, 150, S41.	0.6	0
26	Frataxin fracas. Nature Genetics, 1997, 15, 337-338.	21.4	78
27	Friedreich's Ataxia: Autosomal Recessive Disease Caused by an Intronic GAA Triplet Repeat Expansion. Science, 1996, 271, 1423-1427.	12.6	2,642
28	Childhood Onset of Friedreich Ataxia: A Clinical and Genetic Study of 36 Cases. Neuropediatrics, 1996, 27, 3-7.	0.6	36
29	Has spinocerebellar ataxia type 2 a distinct phenotype? Genetic and clinical study of an Italian family. Neurology, 1995, 45, 793-796.	1.1	46
30	Linkage disequilibrium between FD1-D9S202 haplotypes and the Friedreich's ataxia locus in a central-southern Italian population.. Journal of Medical Genetics, 1994, 31, 133-135.	3.2	6
31	Late onset Friedreich's disease: clinical features and mapping of mutation to the FRDA locus.. Journal of Neurology, Neurosurgery and Psychiatry, 1994, 57, 977-979.	1.9	99
32	Isolation of a New Gene in the Friedreich Ataxia Candidate Region on Human Chromosome 9 by cDNA Direct Selection. Biochemical Medicine and Metabolic Biology, 1994, 52, 115-119.	0.7	7
33	Evidence of a genetic marker associated with early onset in Friedreich's ataxia. Journal of Neurology, 1993, 240, 254-256.	3.6	2
34	A dinucleotide repeat polymorphism (D9S202) in the Friedreich's ataxia region on chromosome 9q13-q21.1. Human Molecular Genetics, 1993, 2, 822-822.	2.9	12
35	Linkage Disequilibrium Analysis of Friedreich's Ataxia in 140 Caucasian Families: Positioning of the Disease Locus and Evaluation of Allelic Heterogeneity. European Journal of Human Genetics, 1993, 1, 133-143.	2.8	13
36	Intrafamilial phenotype variation in Friedreich's disease: possible exceptions to diagnostic criteria. Journal of Neurology, 1991, 238, 147-150.	3.6	17

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37	Clinical and genetic heterogeneity in early onset cerebellar ataxia with retained tendon reflexes.. Journal of Neurology, Neurosurgery and Psychiatry, 1990, 53, 667-670.	1.9	34