

# Francesca Cavalcanti

## List of Publications by Year in descending order

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37  
papers

4,315  
citations

331259

21  
h-index

344852

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. <i>Pediatric Neurology</i> , 2022, , .	1.0	2
2	Interruptions of the FXN GAA Repeat Tract Delay the Age at Onset of Friedreichâ€™s Ataxia in a Location Dependent Manner. <i>International Journal of Molecular Sciences</i> , 2021, 22, 7507.	1.8	10
3	The Mitochondrial Dysfunction Hypothesis in Autism Spectrum Disorders: Current Status and Future Perspectives. <i>International Journal of Molecular Sciences</i> , 2020, 21, 5785.	1.8	29
4	Clinical features and genetic analysis of two siblings with startle disease in an Italian family: a case report. <i>BMC Medical Genetics</i> , 2019, 20, 40.	2.1	4
5	Expanding the global prevalence of spinocerebellar ataxia type 42. <i>Neurology: Genetics</i> , 2018, 4, e232.	0.9	14
6	Large Interruptions of GAA Repeat Expansion Mutations in Friedreich Ataxia Are Very Rare. <i>Frontiers in Cellular Neuroscience</i> , 2018, 12, 443.	1.8	20
7	<i>NeuroArray</i>: A Customized aCGH for the Analysis of Copy Number Variations in Neurological Disorders. <i>Current Genomics</i> , 2018, 19, 431-443.	0.7	3
8	A customized high-resolution array-comparative genomic hybridization to explore copy number variations in Parkinsonâ€™s disease. <i>Neurogenetics</i> , 2016, 17, 233-244.	0.7	10
9	Splicing: is there an alternative contribution to Parkinsonâ€™s disease?. <i>Neurogenetics</i> , 2015, 16, 245-263.	0.7	54
10	Sacsin-Related Spastic Ataxia Caused by a Novel Missense Mutation p.Arg272His in a Patient from Sicily, Southern Italy. <i>Cerebellum</i> , 2013, 12, 589-592.	1.4	3
11	Mutations in PRRT2 result in familial infantile seizures with heterogeneous phenotypes including febrile convulsions and probable SUDEP. <i>Epilepsy Research</i> , 2013, 104, 280-284.	0.8	29
12	Ataxin-1 and ataxin-2 intermediate-length PolyQ expansions in amyotrophic lateral sclerosis. <i>Neurology</i> , 2012, 79, 2315-2320.	1.5	70
13	An Axon Regeneration Signature in a Charcot-Marie-Tooth Disease Type 2 Patient. <i>Journal of Neurogenetics</i> , 2009, 23, 324-328.	0.6	4
14	Vitamin E deficiency due to chylomicron retention disease in Marinesco-Sjögren syndrome. <i>Annals of Neurology</i> , 2000, 47, 260-264.	2.8	40
15	Accuracy of clinical diagnostic criteria for Friedreich's ataxia. <i>Movement Disorders</i> , 2000, 15, 1255-1258.	2.2	39
16	Atypical Friedreich ataxia phenotype associated with a novel missense mutation in the <i>X25 gene</i>. <i>Neurology</i> , 2000, 54, 496-496.	1.5	26
17	Relation between trinucleotide GAA repeat length and sensory neuropathy in Friedreich's ataxia. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 1999, 66, 93-96.	0.9	59
18	Why do some Friedreich's ataxia patients retain tendon reflexes?. <i>Journal of Neurology</i> , 1999, 246, 353-357.	1.8	36

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19	Friedreich's ataxia: Point mutations and clinical presentation of compound heterozygotes. <i>Annals of Neurology</i> , 1999, 45, 200-206.	2.8	371
20	Determinants of onset age in Friedreich's ataxia. <i>Journal of Neurology</i> , 1998, 245, 166-168.	1.8	35
21	A New Locus for Autosomal Recessive Hereditary Spastic Paraplegia Maps to Chromosome 16q24.3. <i>American Journal of Human Genetics</i> , 1998, 63, 135-139.	2.6	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. <i>Human Molecular Genetics</i> , 1998, 7, 1901-1906.	1.4	75
23	The Friedreich ataxia GAA triplet repeat: premutation and normal alleles. <i>Human Molecular Genetics</i> , 1997, 6, 1261-1266.	1.4	188
24	Broadened Friedreich's ataxia phenotype after gene cloning. <i>Neurology</i> , 1997, 49, 1617-1620.	1.5	55
25	1-29-08 Linkage study in an Italian family with autosomal recessive spastic paraplegia. <i>Journal of the Neurological Sciences</i> , 1997, 150, S41.	0.3	0
26	Frataxin fracas. <i>Nature Genetics</i> , 1997, 15, 337-338.	9.4	78
27	Friedreich's Ataxia: Autosomal Recessive Disease Caused by an Intronic GAA Triplet Repeat Expansion. <i>Science</i> , 1996, 271, 1423-1427.	6.0	2,642
28	Childhood Onset of Friedreich Ataxia: A Clinical and Genetic Study of 36 Cases. <i>Neuropediatrics</i> , 1996, 27, 3-7.	0.3	36
29	Has spinocerebellar ataxia type 2 a distinct phenotype? Genetic and clinical study of an Italian family. <i>Neurology</i> , 1995, 45, 793-796.	1.5	46
30	Linkage disequilibrium between FD1-D9S202 haplotypes and the Friedreich's ataxia locus in a central-southern Italian population.. <i>Journal of Medical Genetics</i> , 1994, 31, 133-135.	1.5	6
31	Late onset Friedreich's disease: clinical features and mapping of mutation to the FRDA locus.. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 1994, 57, 977-979.	0.9	99
32	Isolation of a New Gene in the Friedreich Ataxia Candidate Region on Human Chromosome 9 by cDNA Direct Selection. <i>Biochemical Medicine and Metabolic Biology</i> , 1994, 52, 115-119.	0.7	7
33	Evidence of a genetic marker associated with early onset in Friedreich's ataxia. <i>Journal of Neurology</i> , 1993, 240, 254-256.	1.8	2
34	A dinucleotide repeat polymorphism (D9S202) in the Friedreich's ataxia region on chromosome 9q13-q21.1. <i>Human Molecular Genetics</i> , 1993, 2, 822-822.	1.4	12
35	Linkage Disequilibrium Analysis of Friedreich's Ataxia in 140 Caucasian Families: Positioning of the Disease Locus and Evaluation of Allelic Heterogeneity. <i>European Journal of Human Genetics</i> , 1993, 1, 133-143.	1.4	13
36	Intrafamilial phenotype variation in Friedreich's disease: possible exceptions to diagnostic criteria. <i>Journal of Neurology</i> , 1991, 238, 147-150.	1.8	17

#	ARTICLE	IF	CITATIONS
37	Clinical and genetic heterogeneity in early onset cerebellar ataxia with retained tendon reflexes.. Journal of Neurology, Neurosurgery and Psychiatry, 1990, 53, 667-670.	0.9	34