Isabel Silveira

List of Publications by Year
in descending order

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Version: 2024-02-01

56 papers	2,345 citations	186265 28 h-index	214800 47 g-index
56 all docs	56 docs citations	56 times ranked	2414 citing authors

#	Article	IF	Citations
1	Trinucleotide Repeats in 202 Families With Ataxia. Archives of Neurology, 2002, 59, 623.	4.5	158
2	Neurologic Findings in Machado-Joseph Disease. Archives of Neurology, 2001, 58, 899.	4.5	154
3	Ancestral Origins of the Machado-Joseph Disease Mutation: A Worldwide Haplotype Study. American Journal of Human Genetics, 2001, 68, 523-528.	6.2	118
4	A Pentanucleotide ATTTC Repeat Insertion in the Non-coding Region of DAB1, Mapping to SCA37, Causes Spinocerebellar Ataxia. American Journal of Human Genetics, 2017, 101, 87-103.	6.2	112
5	Frequency of spinocerebellar ataxia type 1, dentatorubropallidoluysian atrophy, and Machado-Joseph disease mutations in a large group of spinocerebellar ataxia patients. Neurology, 1996, 46, 214-218.	1.1	106
6	Hereditary Ataxia and Spastic Paraplegia in Portugal. JAMA Neurology, 2013, 70, 746.	9.0	106
7	A survey of spinocerebellar ataxia in South Brazil - 66 new cases with Machado-Joseph disease, SCA7, SCA8, or unidentified disease-causing mutations. Journal of Neurology, 2001, 248, 870-876.	3.6	88
8	High Germinal Instability of the (CTG)n at the SCA8 Locus of Both Expanded and Normal Alleles. American Journal of Human Genetics, 2000, 66, 830-840.	6.2	79
9	â€˜Costa da Morteâ€™ ataxia is spinocerebellar ataxia 36: clinical and genetic characterization. Brain, 2012, 135, 1423-1435.	7.6	78
10	Phenotypes of Spinocerebellar Ataxia Type 6 and Familial Hemiplegic Migraine Caused by a Unique CACNA1A Missense Mutation in Patients From a Large Family. Archives of Neurology, 2003, 60, 610.	4.5	77
11	Loss of junctophilinâ€3 contributes to huntington diseaseâ€like 2 pathogenesis. Annals of Neurology, 2012, 71, 245-257.	5.3	71
12	Asian Origin for the Worldwide-Spread Mutational Event in Machado-Joseph Disease. Archives of Neurology, 2007, 64, 1502.	4.5	65
13	Somatic mosaicism in the central nervous system in spinocerebellar ataxia type 1 and machado-joseph disease. Annals of Neurology, 1996, 40, 199-206.	5.3	59
14	Analysis of SCA1, DRPLA, MJD, SCA2, and SCA6 CAG repeats in 48 portuguese ataxia families. American Journal of Medical Genetics Part A, 1998, 81, 134-138.	2.4	57
15	Epidemiology and population genetics of degenerative ataxias. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2012, 103, 227-251.	1.8	57
16	Autosomal dominant cerebellar ataxia: frequency analysis and clinical characterization of 45 families from Portugal. European Journal of Neurology, 2010, 17, 124-128.	3.3	54
17	Autosomal Dominant Spastic Paraplegias. JAMA Neurology, 2013, 70, 481.	9.0	48
18	Depressive Symptoms in Machado-Joseph Disease (SCA3) Patients and Their Relatives. Public Health Genomics, 2007, 10, 19-26.	1.0	46

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19	Machado-Joseph disease in South Brazil: clinical and molecular characterization of kindreds. Acta Neurologica Scandinavica, 2001, 104, 224-231.	2.1	44
20	Ancestral Origin of the ATTCT Repeat Expansion in Spinocerebellar Ataxia Type 10 (SCA10). PLoS ONE, 2009, 4, e4553.	2.5	40
21	Frequency of the different mutations causing spinocerebellar ataxia (SCA1, SCA2, MJD/SCA3 and DRPLA) in a large group of Brazilian patients. Arquivos De Neuro-Psiquiatria, 1997, 55, 519-529.	0.8	38
22	Reduced penetrance of intermediate size alleles in spinocerebellar ataxia type 10. Neurology, 2006, 66, 1602-1604.	1.1	38
23	Portuguese families with dentatorubropallidoluysian atrophy (DRPLA) share a common haplotype of Asian origin. European Journal of Human Genetics, 2003, 11, 808-811.	2.8	37
24	FXTAS, SCA10, and SCA17 in American patients with movement disorders. American Journal of Medical Genetics, Part A, 2005, 136A, 87-89.	1.2	36
25	Genetic Linkage Studies of Machado-Joseph Disease with Chromosome 14q STRPs in 16 Portuguese-Azorean Kindreds. Genomics, 1994, 21, 645-648.	2.9	34
26	Use of fluoxetine for treatment of Machado-Joseph disease: an open-label study. Acta Neurologica Scandinavica, 2003, 107, 207-210.	2.1	32
27	A novel H101Q mutation causes PKCÎ³ loss in spinocerebellar ataxia type 14. Journal of Human Genetics, 2005, 50, 523-529.	2.3	32
28	Unstable repeat expansions in neurodegenerative diseases: nucleocytoplasmic transport emerges on the scene. Neurobiology of Aging, 2016, 39, 174-183.	3.1	32
29	Common origin of pure and interrupted repeat expansions in spinocerebellar ataxia type 2 (SCA2). American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 524-531.	1.7	30
30	Limits of Clinical Assessment in the Accurate Diagnosis of Machado-Joseph Disease. Archives of Neurology, 1996, 53, 1168-1174.	4.5	29
31	Motor and cognitive deficits in the heterozygous leaner mouse, a Cav2.1 voltage-gated Ca2+ channel mutant. Neurobiology of Aging, 2008, 29, 1733-1743.	3.1	27
32	Cerebellar Ataxia, Hemiplegic Migraine, and Related Phenotypes Due to a CACNA1A Missense Mutation. JAMA Neurology, 2013, 70, 235.	9.0	27
33	Searching for modulating effects of SCA2, SCA6 and DRPLA CAG tracts on the Machado-Joseph disease (SCA3) phenotype. Acta Neurologica Scandinavica, 2003, 107, 211-214.	2.1	26
34	A novel R1347Q mutation in the predicted voltage sensor segment of the P/Q-type calcium-channel Î±1A-subunit in a family with progressive cerebellar ataxia and hemiplegic migraine. Clinical Genetics, 2003, 65, 70-72.	2.0	26
35	Linkage disequilibrium analysis in Machado-Joseph disease patients of different ethnic origins. Human Genetics, 1996, 98, 620-624.	3.8	24
36	Spinocerebellar ataxias in 114 Brazilian families: clinical and molecular findings. Clinical Genetics, 2006, 70, 173-176.	2.0	24

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37	Cisâ€acting factors promoting the CAG intergenerational instability in Machadoâ€“Joseph disease. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 439-446.	1.7	24
38	Fluorodopa and raclopride PET analysis of patients with Machado-Joseph disease. Neurology, 1997, 49, 1133-1136.	1.1	23
39	Machado-Joseph Disease Enhances Genetic Fitness: A Comparison Between Affected and Unaffected Women and Between MJD and the General Population. Annals of Human Genetics, 2007, 72, 070807042352001-???.	0.8	22
40	Mutational mechanism for DAB1 (ATTTC) _n insertion in SCA37: ATTTT repeat lengthening and nucleotide substitution. Human Mutation, 2019, 40, 404-412.	2.5	21
41	Characterization of polymeric nanoparticles for intravenous delivery: Focus on stability. Colloids and Surfaces B: Biointerfaces, 2017, 150, 326-333.	5.0	20
42	Recent advances in characterization of nonviral vectors for delivery of nucleic acids: impact on their biological performance. Expert Opinion on Drug Delivery, 2015, 12, 27-39.	5.0	19
43	Recent Advances in Nucleic Acid-Based Delivery: From Bench to Clinical Trials in Genetic Diseases. Journal of Biomedical Nanotechnology, 2016, 12, 841-862.	1.1	19
44	NovelSPG3AandSPG4mutations in dominant spastic paraplegia families. Acta Neurologica Scandinavica, 2009, 119, 113-118.	2.1	16
45	Molecular Mechanisms in Pentanucleotide Repeat Diseases. Cells, 2022, 11, 205.	4.1	14
46	Antisense Transcription across Nucleotide Repeat Expansions in Neurodegenerative and Neuromuscular Diseases: Progress and Mysteries. Genes, 2020, 11, 1418.	2.4	11
47	Haplotype diversity and somatic instability in normal and expanded SCA8 alleles. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2005, 139B, 109-114.	1.7	8
48	A repeat-primed PCR assay for pentanucleotide repeat alleles in spinocerebellar ataxia type 37. Journal of Human Genetics, 2018, 63, 981-987.	2.3	7
49	FXTAS is rare among Portuguese patients with movement disorders: FMR1 premutations may be associated with a wider spectrum of phenotypes. Behavioral and Brain Functions, 2011, 7, 19.	3.3	6
50	The spatial learning phenotype of heterozygous leaner mice is robust to systematic variation of the housing environment. Comparative Medicine, 2009, 59, 129-38.	1.0	6
51	Joining European Scientific Forces to Face Pandemics. Trends in Microbiology, 2021, 29, 92-97.	7.7	5
52	Machado-Joseph Disease Is Genetically Different from Holguin Dominant Ataxia (SCA2). Genomics, 1993, 17, 556-559.	2.9	4
53	The Prevalence of Familial Hemiplegic Migraine With Cerebellar Ataxia and Spinocerebellar Ataxia Type 6 in <scp>P</scp>ortugal. Headache, 2014, 54, 911-915.	3.9	4
54	Lipoplexes and polyplexes as nucleic acids delivery nanosystems: The current state and future considerations. Expert Opinion on Drug Delivery, 2022, 19, 577-594.	5.0	4

#	Article	IF	Citations
55	Clinical and molecular characteristics of a Brazilian family with spinocerebellar ataxia type 1. Arquivos De Neuro-Psiquiatria, 1996, 54, 412-418.	0.8	2
56	A novel trinucleotide repeat expansion at chromosome 3q26.2 identified by a CAG/CTG repeat expansion detection array. Human Genetics, 2006, 120, 193-200.	3.8	1