

Isabel Silveira

List of Publications by Year in descending order

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56
papers

2,345
citations

185998

28
h-index

223531

46
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.	4.9	158
2	Neurologic Findings in Machado-Joseph Disease. Archives of Neurology, 2001, 58, 899.	4.9	154
3	Ancestral Origins of the Machado-Joseph Disease Mutation: A Worldwide Haplotype Study. American Journal of Human Genetics, 2001, 68, 523-528.	2.6	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.	2.6	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.	1.5	106
6	Hereditary Ataxia and Spastic Paraplegia in Portugal. JAMA Neurology, 2013, 70, 746.	4.5	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.	1.8	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.	2.6	79
9	â€œCosta da Morteâ€™ ataxia is spinocerebellar ataxia 36: clinical and genetic characterization. Brain, 2012, 135, 1423-1435.	3.7	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.	4.9	77
11	Loss of junctophilinâ€³ contributes to huntington diseaseâ€like 2 pathogenesis. Annals of Neurology, 2012, 71, 245-257.	2.8	71
12	Asian Origin for the Worldwide-Spread Mutational Event in Machado-Joseph Disease. Archives of Neurology, 2007, 64, 1502.	4.9	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.	2.8	59
14	Analysis of SCA1, DRPLA, MJD, SCA2, and SCA6 CAG repeats in 48 portuguese ataxia families. , 1998, 81, 134-138.		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.	1.0	57
16	Autosomal dominant cerebellar ataxia: frequency analysis and clinical characterization of 45 families from Portugal. European Journal of Neurology, 2010, 17, 124-128.	1.7	54
17	Autosomal Dominant Spastic Paraplegias. JAMA Neurology, 2013, 70, 481.	4.5	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. <i>Acta Neurologica Scandinavica</i> , 2001, 104, 224-231.	1.0	44
20	Ancestral Origin of the ATTCT Repeat Expansion in Spinocerebellar Ataxia Type 10 (SCA10). <i>PLoS ONE</i> , 2009, 4, e4553.	1.1	40
21	Frequency of the different mutations causing spinocerebellar ataxia (SCA1, SCA2, MJD/SCA3 and DRPLA) in a large group of Brazilian patients. <i>Arquivos De Neuro-Psiquiatria</i> , 1997, 55, 519-529.	0.3	38
22	Reduced penetrance of intermediate size alleles in spinocerebellar ataxia type 10. <i>Neurology</i> , 2006, 66, 1602-1604.	1.5	38
23	Portuguese families with dentatorubropallidoluysian atrophy (DRPLA) share a common haplotype of Asian origin. <i>European Journal of Human Genetics</i> , 2003, 11, 808-811.	1.4	37
24	FXTAS, SCA10, and SCA17 in American patients with movement disorders. <i>American Journal of Medical Genetics, Part A</i> , 2005, 136A, 87-89.	0.7	36
25	Genetic Linkage Studies of Machado-Joseph Disease with Chromosome 14q STRPs in 16 Portuguese-Azorean Kindreds. <i>Genomics</i> , 1994, 21, 645-648.	1.3	34
26	Use of fluoxetine for treatment of Machado-Joseph disease: an open-label study. <i>Acta Neurologica Scandinavica</i> , 2003, 107, 207-210.	1.0	32
27	A novel H101Q mutation causes PKC δ loss in spinocerebellar ataxia type 14. <i>Journal of Human Genetics</i> , 2005, 50, 523-529.	1.1	32
28	Unstable repeat expansions in neurodegenerative diseases: nucleocytoplasmic transport emerges on the scene. <i>Neurobiology of Aging</i> , 2016, 39, 174-183.	1.5	32
29	Common origin of pure and interrupted repeat expansions in spinocerebellar ataxia type 2 (SCA2). <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2010, 153B, 524-531.	1.1	30
30	Limits of Clinical Assessment in the Accurate Diagnosis of Machado-Joseph Disease. <i>Archives of Neurology</i> , 1996, 53, 1168-1174.	4.9	29
31	Motor and cognitive deficits in the heterozygous leaner mouse, a Cav2.1 voltage-gated Ca ²⁺ channel mutant. <i>Neurobiology of Aging</i> , 2008, 29, 1733-1743.	1.5	27
32	Cerebellar Ataxia, Hemiplegic Migraine, and Related Phenotypes Due to a CACNA1A Missense Mutation. <i>JAMA Neurology</i> , 2013, 70, 235.	4.5	27
33	Searching for modulating effects of SCA2, SCA6 and DRPLA CAG tracts on the Machado-Joseph disease (SCA3) phenotype. <i>Acta Neurologica Scandinavica</i> , 2003, 107, 211-214.	1.0	26
34	A novel R1347Q mutation in the predicted voltage sensor segment of the P/Q-type calcium-channel α 1A-subunit in a family with progressive cerebellar ataxia and hemiplegic migraine. <i>Clinical Genetics</i> , 2003, 65, 70-72.	1.0	26
35	Linkage disequilibrium analysis in Machado-Joseph disease patients of different ethnic origins. <i>Human Genetics</i> , 1996, 98, 620-624.	1.8	24
36	Spinocerebellar ataxias in 114 Brazilian families: clinical and molecular findings. <i>Clinical Genetics</i> , 2006, 70, 173-176.	1.0	24

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37	<i>Cis</i> -acting factors promoting the CAG intergenerational instability in Machado-Joseph disease. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2008, 147B, 439-446.	1.1	24
38	Fluorodopa and raclopride PET analysis of patients with Machado-Joseph disease. <i>Neurology</i> , 1997, 49, 1133-1136.	1.5	23
39	Machado-Joseph Disease Enhances Genetic Fitness: A Comparison Between Affected and Unaffected Women and Between MJD and the General Population. <i>Annals of Human Genetics</i> , 2007, 72, 070807042352001-???	0.3	22
40	Mutational mechanism for <i>DAB1</i> (ATTC) _n insertion in SCA37: ATTTT repeat lengthening and nucleotide substitution. <i>Human Mutation</i> , 2019, 40, 404-412.	1.1	21
41	Characterization of polymeric nanoparticles for intravenous delivery: Focus on stability. <i>Colloids and Surfaces B: Biointerfaces</i> , 2017, 150, 326-333.	2.5	20
42	Recent advances in characterization of nonviral vectors for delivery of nucleic acids: impact on their biological performance. <i>Expert Opinion on Drug Delivery</i> , 2015, 12, 27-39.	2.4	19
43	Recent Advances in Nucleic Acid-Based Delivery: From Bench to Clinical Trials in Genetic Diseases. <i>Journal of Biomedical Nanotechnology</i> , 2016, 12, 841-862.	0.5	19
44	Novel <i>SPG3A</i> and <i>SPG4</i> mutations in dominant spastic paraplegia families. <i>Acta Neurologica Scandinavica</i> , 2009, 119, 113-118.	1.0	16
45	Molecular Mechanisms in Pentanucleotide Repeat Diseases. <i>Cells</i> , 2022, 11, 205.	1.8	14
46	Antisense Transcription across Nucleotide Repeat Expansions in Neurodegenerative and Neuromuscular Diseases: Progress and Mysteries. <i>Genes</i> , 2020, 11, 1418.	1.0	11
47	Haplotype diversity and somatic instability in normal and expanded SCA8 alleles. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2005, 139B, 109-114.	1.1	8
48	A repeat-primed PCR assay for pentanucleotide repeat alleles in spinocerebellar ataxia type 37. <i>Journal of Human Genetics</i> , 2018, 63, 981-987.	1.1	7
49	FXTAS is rare among Portuguese patients with movement disorders: FMR1 premutations may be associated with a wider spectrum of phenotypes. <i>Behavioral and Brain Functions</i> , 2011, 7, 19.	1.4	6
50	The spatial learning phenotype of heterozygous leaner mice is robust to systematic variation of the housing environment. <i>Comparative Medicine</i> , 2009, 59, 129-38.	0.4	6
51	Joining European Scientific Forces to Face Pandemics. <i>Trends in Microbiology</i> , 2021, 29, 92-97.	3.5	5
52	Machado-Joseph Disease Is Genetically Different from Holguin Dominant Ataxia (SCA2). <i>Genomics</i> , 1993, 17, 556-559.	1.3	4
53	The Prevalence of Familial Hemiplegic Migraine With Cerebellar Ataxia and Spinocerebellar Ataxia Type 6 in Portugal. <i>Headache</i> , 2014, 54, 911-915.	1.8	4
54	Lipoplexes and polyplexes as nucleic acids delivery nanosystems: The current state and future considerations. <i>Expert Opinion on Drug Delivery</i> , 2022, 19, 577-594.	2.4	4

#	ARTICLE	IF	CITATIONS
55	Clinical and molecular characteristics of a Brazilian family with spinocerebellar ataxia type 1. <i>Arquivos De Neuro-Psiquiatria</i> , 1996, 54, 412-418.	0.3	2
56	A novel trinucleotide repeat expansion at chromosome 3q26.2 identified by a CAG/CTG repeat expansion detection array. <i>Human Genetics</i> , 2006, 120, 193-200.	1.8	1