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	Molecular Mechanisms in Pentanucleotide Repeat Diseases. Cells, 2022, 11, 205.	1.8	14
2	Lipoplexes and polyplexes as nucleic acids delivery nanosystems: The current state and future considerations. Expert Opinion on Drug Delivery, 2022, 19, 577-594.	2.4	4
3	Joining European Scientific Forces to Face Pandemics. Trends in Microbiology, 2021, 29, 92-97.	3.5	5
4	Antisense Transcription across Nucleotide Repeat Expansions in Neurodegenerative and Neuromuscular Diseases: Progress and Mysteries. Genes, 2020, 11, 1418.	1.0	11
5	Mutational mechanism for DAB1 (ATTTC) _n insertion in SCA37: ATTTT repeat lengthening and nucleotide substitution. Human Mutation, 2019, 40, 404-412.	1.1	21
6	A repeat-primed PCR assay for pentanucleotide repeat alleles in spinocerebellar ataxia type 37. Journal of Human Genetics, 2018, 63, 981-987.	1.1	7
7	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
8	Characterization of polymeric nanoparticles for intravenous delivery: Focus on stability. Colloids and Surfaces B: Biointerfaces, 2017, 150, 326-333.	2.5	20
9	Recent Advances in Nucleic Acid-Based Delivery: From Bench to Clinical Trials in Genetic Diseases. Journal of Biomedical Nanotechnology, 2016, 12, 841-862.	0.5	19
10	Unstable repeat expansions in neurodegenerative diseases: nucleocytoplasmic transport emerges on the scene. Neurobiology of Aging, 2016, 39, 174-183.	1.5	32
11	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.	2.4	19
12	The Prevalence of Familial Hemiplegic Migraine With Cerebellar Ataxia and Spinocerebellar Ataxia Type 6 in <scp>P</scp>ortugal. Headache, 2014, 54, 911-915.	1.8	4
13	Hereditary Ataxia and Spastic Paraplegia in Portugal. JAMA Neurology, 2013, 70, 746.	4.5	106
14	Cerebellar Ataxia, Hemiplegic Migraine, and Related Phenotypes Due to a CACNA1A Missense Mutation. JAMA Neurology, 2013, 70, 235.	4.5	27
15	Autosomal Dominant Spastic Paraplegias. JAMA Neurology, 2013, 70, 481.	4.5	48
16	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
17	â€˜Costa da Morteâ€™ ataxia is spinocerebellar ataxia 36: clinical and genetic characterization. Brain, 2012, 135, 1423-1435.	3.7	78
18	Loss of junctophilinâ€3 contributes to huntington diseaseâ€like 2 pathogenesis. Annals of Neurology, 2012, 71, 245-257.	2.8	71

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19	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.	1.4	6
20	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.	1.1	30
21	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
22	Ancestral Origin of the ATTCT Repeat Expansion in Spinocerebellar Ataxia Type 10 (SCA10). PLoS ONE, 2009, 4, e4553.	1.1	40
23	NovelSPG3AandSPG4mutations in dominant spastic paraplegia families. Acta Neurologica Scandinavica, 2009, 119, 113-118.	1.0	16
24	The spatial learning phenotype of heterozygous leaner mice is robust to systematic variation of the housing environment. Comparative Medicine, 2009, 59, 129-38.	0.4	6
25	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.	1.1	24
26	Motor and cognitive deficits in the heterozygous leaner mouse, a Cav2.1 voltage-gated Ca2+ channel mutant. Neurobiology of Aging, 2008, 29, 1733-1743.	1.5	27
27	Depressive Symptoms in Machado-Joseph Disease (SCA3) Patients and Their Relatives. Public Health Genomics, 2007, 10, 19-26.	1.0	46
28	Asian Origin for the Worldwide-Spread Mutational Event in Machado-Joseph Disease. Archives of Neurology, 2007, 64, 1502.	4.9	65
29	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-???.	0.3	22
30	Spinocerebellar ataxias in 114 Brazilian families: clinical and molecular findings. Clinical Genetics, 2006, 70, 173-176.	1.0	24
31	A novel trinucleotide repeat expansion at chromosome 3q26.2 identified by a CAG/CTG repeat expansion detection array. Human Genetics, 2006, 120, 193-200.	1.8	1
32	Reduced penetrance of intermediate size alleles in spinocerebellar ataxia type 10. Neurology, 2006, 66, 1602-1604.	1.5	38
33	FXTAS, SCA10, and SCA17 in American patients with movement disorders. American Journal of Medical Genetics, Part A, 2005, 136A, 87-89.	0.7	36
34	Haplotype diversity and somatic instability in normal and expanded SCA8 alleles. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2005, 139B, 109-114.	1.1	8
35	A novel H101Q mutation causes PKCÎ³ loss in spinocerebellar ataxia type 14. Journal of Human Genetics, 2005, 50, 523-529.	1.1	32
36	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.	1.0	26

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37	Use of fluoxetine for treatment of Machado-Joseph disease: an open-label study. Acta Neurologica Scandinavica, 2003, 107, 207-210.	1.0	32
38	Portuguese families with dentatorubropallidoluysian atrophy (DRPLA) share a common haplotype of Asian origin. European Journal of Human Genetics, 2003, 11, 808-811.	1.4	37
39	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.	1.0	26
40	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
41	Trinucleotide Repeats in 202 Families With Ataxia. Archives of Neurology, 2002, 59, 623.	4.9	158
42	Ancestral Origins of the Machado-Joseph Disease Mutation: A Worldwide Haplotype Study. American Journal of Human Genetics, 2001, 68, 523-528.	2.6	118
43	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
44	Machado-Joseph disease in South Brazil: clinical and molecular characterization of kindreds. Acta Neurologica Scandinavica, 2001, 104, 224-231.	1.0	44
45	Neurologic Findings in Machado-Joseph Disease. Archives of Neurology, 2001, 58, 899.	4.9	154
46	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
47	Analysis of SCA1, DRPLA, MJD, SCA2, and SCA6 CAG repeats in 48 portuguese ataxia families. , 1998, 81, 134-138.		57
48	Fluorodopa and raclopride PET analysis of patients with Machado-Joseph disease. Neurology, 1997, 49, 1133-1136.	1.5	23
49	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.	0.3	38
50	Limits of Clinical Assessment in the Accurate Diagnosis of Machado-Joseph Disease. Archives of Neurology, 1996, 53, 1168-1174.	4.9	29
51	Clinical and molecular characteristics of a Brazilian family with spinocerebellar ataxia type 1. Arquivos De Neuro-Psiquiatria, 1996, 54, 412-418.	0.3	2
52	Linkage disequilibrium analysis in Machado-Joseph disease patients of different ethnic origins. Human Genetics, 1996, 98, 620-624.	1.8	24
53	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
54	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

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55	Genetic Linkage Studies of Machado-Joseph Disease with Chromosome 14q STRPs in 16 Portuguese-Azorean Kindreds. Genomics, 1994, 21, 645-648.	1.3	34
56	Machado-Joseph Disease Is Genetically Different from Holguin Dominant Ataxia (SCA2). Genomics, 1993, 17, 556-559.	1.3	4