Conceicao Bettencourt

List of Publications by Year
in descending order

55 papers	2,134 citations	279778 23 h-index	243610 44 g-index
57 all docs	57 docs citations	57 times ranked	4100 citing authors

#	Article	IF	Citations
1	Individual dopaminergic neurons show raised iron levels in Parkinson disease. Neurology, 2007, 68, 1820-1825.	1.1	237
2	DNA repair pathways underlie a common genetic mechanism modulating onset in polyglutamine diseases. Annals of Neurology, 2016, 79, 983-990.	5.3	183
3	Genetic and phenotypic characterization of complex hereditary spastic paraplegia. Brain, 2016, 139, 1904-1918.	7.6	170
4	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
5	Machado-Joseph Disease: from first descriptions to new perspectives. Orphanet Journal of Rare Diseases, 2011, 6, 35.	2.7	132
6	A Missense Mutation in KCTD17 Causes Autosomal Dominant Myoclonus-Dystonia. American Journal of Human Genetics, 2015, 96, 938-947.	6.2	109
7	Truncating and Missense Mutations in IGHMBP2 Cause Charcot-Marie Tooth Disease Type 2. American Journal of Human Genetics, 2014, 95, 590-601.	6.2	75
8	Dominant Mutations in GRM1 Cause Spinocerebellar Ataxia Type 44. American Journal of Human Genetics, 2017, 101, 451-458.	6.2	62
9	Insights From Cerebellar Transcriptomic Analysis Into the Pathogenesis of Ataxia. JAMA Neurology, 2014, 71, 831.	9.0	60
10	A homozygous lossâ€ofâ€function mutation in PDE2A associated to earlyâ€onset hereditary chorea. Movement Disorders, 2018, 33, 482-488.	3.9	52
11	Coenzyme Q10 Levels Are Decreased in the Cerebellum of Multiple-System Atrophy Patients. PLoS ONE, 2016, 11, e0149557.	2.5	48
12	Brain iron accumulation affects myelin-related molecular systems implicated in a rare neurogenetic disease family with neuropsychiatric features. Molecular Psychiatry, 2016, 21, 1599-1607.	7.9	45
13	A loss-of-function homozygous mutation in DDX59 implicates a conserved DEAD-box RNA helicase in nervous system development and function. Human Mutation, 2018, 39, 187-192.	2.5	44
14	White matter DNA methylation profiling reveals deregulation of HIP1, LMAN2, MOBP, and other loci in multiple system atrophy. Acta Neuropathologica, 2020, 139, 135-156.	7.7	42
15	Rare variants in SQSTM1 and VCP genes and risk of sporadic inclusion body myositis. Neurobiology of Aging, 2016, 47, 218.e1-218.e9.	3.1	40
16	Genetic and clinical characteristics ofNEFL-related Charcot-Marie-Tooth disease. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 575-585.	1.9	34
17	The <emph type="ital">APOE</emph> Îµ2 Allele Increases the Risk of Earlier Age at Onset in Machado-Joseph Disease. Archives of Neurology, 2011, 68, 1580.	4.5	33
18	Clinical and Neuropathological Features of Spastic Ataxia in a Spanish Family with Novel Compound Heterozygous Mutations in STUB1. Cerebellum, 2015, 14, 378-381.	2.5	33

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19	A 30-unit hexanucleotide repeat expansion in C9orf72 induces pathological lesions with dipeptide-repeat proteins and RNA foci, but not TDP-43 inclusions and clinical disease. Acta Neuropathologica, 2015, 130, 599-601.	7.7	31
20	Analysis of segregation patterns in Machadoâ€“Joseph disease pedigrees. Journal of Human Genetics, 2008, 53, 920-923.	2.3	28
21	Sporadic inclusion body myositis: the genetic contributions to the pathogenesis. Orphanet Journal of Rare Diseases, 2014, 9, 88.	2.7	28
22	Novel candidate bloodâ€based transcriptional biomarkers of machadoâ€joseph disease. Movement Disorders, 2015, 30, 968-975.	3.9	28
23	Trehalose Improves Human Fibroblast Deficits in a New CHIP-Mutation Related Ataxia. PLoS ONE, 2014, 9, e106931.	2.5	28
24	Replicating studies of genetic modifiers in spinocerebellar ataxia type 3: can homogeneous cohorts aid?. Brain, 2015, 138, e398-e398.	7.6	26
25	Pure Cerebellar Ataxia with Homozygous Mutations in the PNPLA6 Gene. Cerebellum, 2017, 16, 262-267.	2.5	26
26	Segregation distortion of wild-type alleles at the Machado-Joseph disease locus: a study in normal families from the Azores islands (Portugal). Journal of Human Genetics, 2008, 53, 333-339.	2.3	25
27	Heterogeneity in clinical features and disease severity in ataxia-associated SYNE1 mutations. Journal of Neurology, 2016, 263, 1503-1510.	3.6	24
28	Gene co-expression networks shed light into diseases of brain iron accumulation. Neurobiology of Disease, 2016, 87, 59-68.	4.4	24
29	Accumulation of Mitochondrial DNA Common Deletion Since The Preataxic Stage of Machado-Joseph Disease. Molecular Neurobiology, 2019, 56, 119-124.	4.0	24
30	Identification of multiple system atrophy mimicking Parkinsonâ€™s disease or progressive supranuclear palsy. Brain, 2021, 144, 1138-1151.	7.6	24
31	The (CAG)n tract of Machadoâ€“Joseph Disease gene (ATXN3): a comparison between DNA and mRNA in patients and controls. European Journal of Human Genetics, 2010, 18, 621-623.	2.8	21
32	Expanding the Phenotype and Genetic Defects Associated with the <scp>GOSR</scp>2 Gene. Movement Disorders Clinical Practice, 2015, 2, 271-273.	1.5	21
33	Promoter Variation and Expression Levels of Inflammatory Genes IL1A, IL1B, IL6 and TNF in Blood of Spinocerebellar Ataxia Type 3 (SCA3) Patients. NeuroMolecular Medicine, 2017, 19, 41-45.	3.4	21
34	Transcriptional profiling of multiple system atrophy cerebellar tissue highlights differences between the parkinsonian and cerebellar sub-types of the disease. Acta Neuropathologica Communications, 2020, 8, 76.	5.2	20
35	Transcriptomic analysis of frontotemporal lobar degeneration with TDP-43 pathology reveals cellular alterations across multiple brain regions. Acta Neuropathologica, 2022, 143, 383-401.	7.7	20
36	Exome sequencing expands the mutational spectrum of SPG8 in a family with spasticity responsive to l-DOPA treatment. Journal of Neurology, 2013, 260, 2414-2416.	3.6	18

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37	Genotype-phenotype correlations and expansion of the molecular spectrum of AP4M1-related hereditary spastic paraplegia. Orphanet Journal of Rare Diseases, 2017, 12, 172.	2.7	17
38	The effects of an intronic polymorphism in TOMM40 and APOE genotypes in sporadic inclusion body myositis. Neurobiology of Aging, 2015, 36, 1766.e1-1766.e3.	3.1	16
39	Neurodegenerative movement disorders: An epigenetics perspective and promise for the future. Neuropathology and Applied Neurobiology, 2021, 47, 897-909.	3.2	16
40	Revisiting genotype-phenotype overlap in neurogenetics: Triplet-repeat expansions mimicking spastic paraplegias. Human Mutation, 2012, 33, 1315-1323.	2.5	15
41	Multiple system atrophy: genetic risks and alpha-synuclein mutations. F1000Research, 2017, 6, 2072.	1.6	14
42	MOBP and HIP1 in multiple system atrophy: New Î±â€synuclein partners in glial cytoplasmic inclusions implicated in the disease pathogenesis. Neuropathology and Applied Neurobiology, 2021, 47, 640-652.	3.2	11
43	Paroxysmal Movement Disorder and Epilepsy Caused by a De Novo Truncating Mutation in KAT6A. Journal of Pediatric Genetics, 2018, 07, 114-116.	0.7	10
44	Pathological relationships involving iron and myelin may constitute a shared mechanism linking various rare and common brain diseases. Rare Diseases (Austin, Tex), 2016, 4, e1198458.	1.8	7
45	Sequencing analysis of the SCA6 CAG expansion excludes an influence of repeat interruptions on disease onset. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 1226-1227.	1.9	7
46	In silico comparative analysis of LRRK2 interactomes from brain, kidney and lung. Brain Research, 2021, 1765, 147503.	2.2	6
47	Epigenomics and transcriptomics analyses of multiple system atrophy brain tissue supports a role for inflammatory processes in disease pathogenesis. Acta Neuropathologica Communications, 2020, 8, 71.	5.2	5
48	Novel single base-pair deletion in exon 1 of XK gene leading to McLeod syndrome with chorea, muscle wasting, peripheral neuropathy, acanthocytosis and haemolysis. Journal of the Neurological Sciences, 2014, 339, 220-222.	0.6	4
49	Genetic Variation in ATXN3 (Ataxin-3) 3â€²UTR: Insights into the Downstream Regulatory Elements of the Causative Gene of Machado-Joseph Disease/Spinocerebellar Ataxia Type 3. Cerebellum, 2023, 22, 37-45.	2.5	4
50	Verification of Inter-laboratorial Genotyping Consistency in the Molecular Diagnosis of Polyglutamine Spinocerebellar Ataxias. Journal of Molecular Neuroscience, 2016, 58, 83-87.	2.3	3
51	The repeat variant in MSH3 is not a genetic modifier for spinocerebellar ataxia type 3 and Friedreichâ€™s ataxia. Brain, 2020, 143, e25-e25.	7.6	3
52	A novel frameshift deletion in autosomal recessive SBF1-related syndromic neuropathy with necklace fibres. Journal of Neurology, 2020, 267, 2705-2712.	3.6	3
53	Novel Machado-Joseph disease-modifying genes and pathways identified by whole-exome sequencing. Neurobiology of Disease, 2022, 162, 105578.	4.4	3
54	Peopling, demographic history and genetic structure of the Azores Islands: Integrating data from mtDNA and Y-chromosome. International Congress Series, 2006, 1288, 85-87.	0.2	1

#	Article	IF	Citations
55	Uniparental isodisomy of chromosome 2 causing MRPL44-related multisystem mitochondrial disease. Molecular Biology Reports, 2021, 48, 2093-2104.	2.3	1