Maria do Carmo Costa

List of Publications by Year
in descending order

35 papers	1,599 citations	331259 21 h-index	360668 35 g-index
42 all docs	42 docs citations	42 times ranked	1496 citing authors

#	Article	IF	Citations
1	Toward understanding Machadoâ€“Joseph disease. Progress in Neurobiology, 2012, 97, 239-257.	2.8	228
2	Early Changes in Cerebellar Physiology Accompany Motor Dysfunction in the Polyglutamine Disease Spinocerebellar Ataxia Type 3. Journal of Neuroscience, 2011, 31, 13002-13014.	1.7	190
3	Improvement in the Molecular Diagnosis of Machado-Joseph Disease. Archives of Neurology, 2001, 58, 1821.	4.9	121
4	Toward RNAi Therapy for the Polyglutamine Disease Machadoâ€“Joseph Disease. Molecular Therapy, 2013, 21, 1898-1908.	3.7	102
5	Silencing Mutant ATXN3 Expression Resolves Molecular Phenotypes in SCA3 Transgenic Mice. Molecular Therapy, 2013, 21, 1909-1918.	3.7	100
6	Towards a Structural Understanding of the Fibrillization Pathway in Machado-Joseph's Disease: Trapping Early Oligomers of Non-expanded Ataxin-3. Journal of Molecular Biology, 2005, 353, 642-654.	2.0	68
7	Functional genomics and biochemical characterization of the C. elegans orthologue of the Machadoâ€Joseph disease protein ataxinâ€3. FASEB Journal, 2007, 21, 1126-1136.	0.2	62
8	Motor uncoordination and neuropathology in a transgenic mouse model of Machadoâ€“Joseph disease lacking intranuclear inclusions and ataxin-3 cleavage products. Neurobiology of Disease, 2010, 40, 163-176.	2.1	62
9	Nonsense mutation in TITF1 in a Portuguese family with benign hereditary chorea. Neurogenetics, 2005, 6, 209-215.	0.7	59
10	Population Genetics of Wild-Type CAG Repeats in the Machado-Joseph Disease Gene in Portugal. Human Heredity, 2005, 60, 156-163.	0.4	43
11	Absence of ataxin-3 leads to cytoskeletal disorganization and increased cell death. Biochimica Et Biophysica Acta - Molecular Cell Research, 2010, 1803, 1154-1163.	1.9	42
12	A knockin mouse model of spinocerebellar ataxia type 3 exhibits prominent aggregate pathology and aberrant splicing of the disease gene transcript. Human Molecular Genetics, 2015, 24, 1211-1224.	1.4	41
13	Interaction of the polyglutamine protein ataxin-3 with Rad23 regulates toxicity in Drosophila models of Spinocerebellar Ataxia Type 3. Human Molecular Genetics, 2017, 26, 1419-1431.	1.4	40
14	Increased transcript diversity: novel splicing variants of Machadoâ€“Joseph Disease gene (ATXN3). Neurogenetics, 2010, 11, 193-202.	0.7	37
15	Unbiased screen identifies aripiprazole as a modulator of abundance of the polyglutamine disease protein, ataxin-3. Brain, 2016, 139, 2891-2908.	3.7	37
16	Antisense oligonucleotide therapy rescues aggresome formation in a novel spinocerebellar ataxia type 3 human embryonic stem cell line. Stem Cell Research, 2019, 39, 101504.	0.3	35
17	The CAG repeat at the Huntington disease gene in the Portuguese population: insights into its dynamics and to the origin of the mutation. Journal of Human Genetics, 2006, 51, 189-195.	1.1	29
18	Genotypes at the APOE and SCA2 loci do not predict the course of multiple sclerosis in patients of Portuguese origin. Multiple Sclerosis Journal, 2004, 10, 153-157.	1.4	27

#	Article	IF	Citations
19	Genomic structure, promoter activity, and developmental expression of the mouse homologue of the Machadoâ€“Joseph disease (MJD) geneâ˜†. Genomics, 2004, 84, 361-373.	1.3	26
20	Exclusion of mutations in the PRNP, JPH3, TBP, ATN1, CREBBP, POU3F2 and FTL genes as a cause of disease in Portuguese patients with a Huntington-like phenotype. Journal of Human Genetics, 2006, 51, 645-651.	1.1	26
21	Dominant negative effect of polyglutamine expansion perturbs normal function of ataxin-3 in neuronal cells. Human Molecular Genetics, 2015, 24, 100-117.	1.4	26
22	Ataxin-3 Plays a Role in Mouse Myogenic Differentiation through Regulation of Integrin Subunit Levels. PLoS ONE, 2010, 5, e11728.	1.1	25
23	Differential recruitment of UBQLN2 to nuclear inclusions in the polyglutamine diseases HD and SCA3. Neurobiology of Disease, 2015, 82, 281-288.	2.1	24
24	Citalopram Reduces Aggregation of ATXN3 in a YAC Transgenic Mouse Model of Machado-Joseph Disease. Molecular Neurobiology, 2019, 56, 3690-3701.	1.9	24
25	The Deubiquitinating Enzyme Ataxin-3 Regulates Ciliogenesis and Phagocytosis in the Retina. Cell Reports, 2020, 33, 108360.	2.9	23
26	In Vivo Molecular Signatures of Cerebellar Pathology in Spinocerebellar Ataxia Type 3. Movement Disorders, 2020, 35, 1774-1786.	2.2	23
27	Molecular diagnosis of Huntington disease in Portugal: implications for genetic counselling and clinical practice. European Journal of Human Genetics, 2003, 11, 872-878.	1.4	18
28	Druggable genome screen identifies new regulators of the abundance and toxicity of ATXN3, the Spinocerebellar Ataxia type 3 disease protein. Neurobiology of Disease, 2020, 137, 104697.	2.1	12
29	Ataxin-3 Links NOD2 and TLR2 Mediated Innate Immune Sensing and Metabolism in Myeloid Cells. Frontiers in Immunology, 2019, 10, 1495.	2.2	11
30	Recent therapeutic prospects for Machadoâ€“Joseph disease. Current Opinion in Neurology, 2020, 33, 519-526.	1.8	11
31	Identification of three novel polymorphisms in the MJD1 gene and study of their frequency in the Portuguese population. Journal of Human Genetics, 2002, 47, 205-207.	1.1	5
32	Selection of Reference Genes for Normalization of Gene Expression Data in Blood of Machado-Joseph Disease/Spinocerebellar Ataxia Type 3 (MJD/SCA3) Subjects. Journal of Molecular Neuroscience, 2019, 69, 450-455.	1.1	5
33	New hope for therapy in neurodegenerative diseases. Cell Research, 2013, 23, 1159-1160.	5.7	4
34	Altered retinal structure and function in Spinocerebellar ataxia type 3. Neurobiology of Disease, 2022, 170, 105774.	2.1	4
35	The Deubiquitinating Enzyme Ataxin-3 Regulates Ciliogenesis and Phagocytosis in the Retina. SSRN Electronic Journal, 0, , .	0.4	0