Mafalda Raposo

List of Publications by Year
in descending order

26	543	840776 11	677142 22
papers	citations	h-index	g-index

28	28	28	865
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	DNA repair pathways underlie a common genetic mechanism modulating onset in polyglutamine diseases. Annals of Neurology, 2016, 79, 983-990.	5.3	183
2	Patterns of Mitochondrial DNA Damage in Blood and Brain Tissues of a Transgenic Mouse Model of Machado-Joseph Disease. Neurodegenerative Diseases, 2013, 11, 206-214.	1.4	55
3	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
4	Parkinsonian phenotype in Machado-Joseph disease (MJD/SCA3): a two-case report. BMC Neurology, 2011, 11, 131.	1.8	30
5	Novel candidate bloodâ€based transcriptional biomarkers of machadoâ€joseph disease. Movement Disorders, 2015, 30, 968-975.	3.9	28
6	Replicating studies of genetic modifiers in spinocerebellar ataxia type 3: can homogeneous cohorts aid?. Brain, 2015, 138, e398-e398.	7.6	26
7	Nystagmus as an early ocular alteration in Machado-Joseph disease (MJD/SCA3). BMC Neurology, 2014, 14, 17.	1.8	24
8	Accumulation of Mitochondrial DNA Common Deletion Since The Preataxic Stage of Machado-Joseph Disease. Molecular Neurobiology, 2019, 56, 119-124.	4.0	24
9	Polyglutamineâ€Expanded Ataxinâ€3: A Target Engagement Marker for Spinocerebellar Ataxia Type 3 in Peripheral Blood. Movement Disorders, 2021, 36, 2675-2681.	3.9	22
10	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
11	Promoter Variant Alters Expression of the Autophagic BECN1 Gene: Implications for Clinical Manifestations of Machado-Joseph Disease. Cerebellum, 2017, 16, 957-963.	2.5	15
12	Psychological Well-Being and Family Satisfaction Levels Five Years After Being Confirmed as a Carrier of the Machado-Joseph Disease Mutation. Genetic Testing and Molecular Biomarkers, 2012, 16, 1363-1368.	0.7	12
13	Towards the Identification of Molecular Biomarkers of Spinocerebellar Ataxia Type 3 (SCA3)/Machado-Joseph Disease (MJD). Advances in Experimental Medicine and Biology, 2018, 1049, 309-319.	1.6	12
14	Triplet Repeat Primed PCR (TP-PCR) in Molecular Diagnostic Testing for Spinocerebellar Ataxia Type 3 (SCA3). Molecular Diagnosis and Therapy, 2016, 20, 617-622.	3.8	10
15	Genome-wide association study identifies genetic factors that modify age at onset in Machado-Joseph disease. Aging, 2020, 12, 4742-4756.	3.1	10
16	Sequence Analysis of 5â€² Regulatory Regions of the Machadoâ€“Joseph Disease Gene (ATXN3). Cerebellum, 2012, 11, 1045-1050.	2.5	7
17	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.	2.3	5
18	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

#	Article	IF	Citations
19	Cross-sectional study of risk factors for atherosclerosis in the Azorean population. Annals of Human Biology, 2011, 38, 354-359.	1.0	3
20	Transcript Diversity of Machadoâ€“Joseph Disease Gene (ATXN3) Is Not Directly Determined by SNPs in Exonic or Flanking Intronic Regions. Journal of Molecular Neuroscience, 2013, 49, 539-543.	2.3	3
21	Familial hypercholesterolemia: Molecular characterization of possible cases from the Azores Islands (Portugal). Meta Gene, 2014, 2, 638-645.	0.6	3
22	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
23	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
24	Polymorphism in cardiovascular diseases (CVD) susceptibility loci in the azores islands (Portugal). Open Journal of Genetics, 2011, 01, 48-53.	0.1	3
25	Novel Machado-Joseph disease-modifying genes and pathways identified by whole-exome sequencing. Neurobiology of Disease, 2022, 162, 105578.	4.4	3
26	B48â€…DNA repair pathways as a common genetic mechanism modulating the age at onset in polyglutamine diseases. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, A26.1-A26.	1.9	0