

Mafalda Raposo

List of Publications by Year in descending order

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Version: 2024-02-01

26
papers

543
citations

840776

11
h-index

677142

22
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28
all docs

28
docs citations

28
times ranked

865
citing authors

#	ARTICLE	IF	CITATIONS
1	Genetic Variation in ATXN3 (Ataxin-3) 3'UTR: Insights into the Downstream Regulatory Elements of the Causative Gene of Machado-Joseph Disease/Spinocerebellar Ataxia Type 3. <i>Cerebellum</i> , 2023, 22, 37-45.	2.5	4
2	Novel Machado-Joseph disease-modifying genes and pathways identified by whole-exome sequencing. <i>Neurobiology of Disease</i> , 2022, 162, 105578.	4.4	3
3	Polyglutamine-Expanded Ataxin-3: A Target Engagement Marker for Spinocerebellar Ataxia Type 3 in Peripheral Blood. <i>Movement Disorders</i> , 2021, 36, 2675-2681.	3.9	22
4	The repeat variant in MSH3 is not a genetic modifier for spinocerebellar ataxia type 3 and Friedreich's ataxia. <i>Brain</i> , 2020, 143, e25-e25.	7.6	3
5	Genome-wide association study identifies genetic factors that modify age at onset in Machado-Joseph disease. <i>Aging</i> , 2020, 12, 4742-4756.	3.1	10
6	Selection of Reference Genes for Normalization of Gene Expression Data in Blood of Machado-Joseph Disease/Spinocerebellar Ataxia Type 3 (MJD/SCA3) Subjects. <i>Journal of Molecular Neuroscience</i> , 2019, 69, 450-455.	2.3	5
7	Accumulation of Mitochondrial DNA Common Deletion Since The Preataxic Stage of Machado-Joseph Disease. <i>Molecular Neurobiology</i> , 2019, 56, 119-124.	4.0	24
8	Towards the Identification of Molecular Biomarkers of Spinocerebellar Ataxia Type 3 (SCA3)/Machado-Joseph Disease (MJD). <i>Advances in Experimental Medicine and Biology</i> , 2018, 1049, 309-319.	1.6	12
9	Promoter Variation and Expression Levels of Inflammatory Genes IL1A, IL1B, IL6 and TNF in Blood of Spinocerebellar Ataxia Type 3 (SCA3) Patients. <i>NeuroMolecular Medicine</i> , 2017, 19, 41-45.	3.4	21
10	Promoter Variant Alters Expression of the Autophagic BECN1 Gene: Implications for Clinical Manifestations of Machado-Joseph Disease. <i>Cerebellum</i> , 2017, 16, 957-963.	2.5	15
11	B48...DNA repair pathways as a common genetic mechanism modulating the age at onset in polyglutamine diseases. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, A26.1-A26.	1.9	0
12	Triplet Repeat Primed PCR (TP-PCR) in Molecular Diagnostic Testing for Spinocerebellar Ataxia Type 3 (SCA3). <i>Molecular Diagnosis and Therapy</i> , 2016, 20, 617-622.	3.8	10
13	DNA repair pathways underlie a common genetic mechanism modulating onset in polyglutamine diseases. <i>Annals of Neurology</i> , 2016, 79, 983-990.	5.3	183
14	Verification of Inter-laboratorial Genotyping Consistency in the Molecular Diagnosis of Polyglutamine Spinocerebellar Ataxias. <i>Journal of Molecular Neuroscience</i> , 2016, 58, 83-87.	2.3	3
15	Replicating studies of genetic modifiers in spinocerebellar ataxia type 3: can homogeneous cohorts aid?. <i>Brain</i> , 2015, 138, e398-e398.	7.6	26
16	Novel candidate blood-based transcriptional biomarkers of machado-joseph disease. <i>Movement Disorders</i> , 2015, 30, 968-975.	3.9	28
17	Familial hypercholesterolemia: Molecular characterization of possible cases from the Azores Islands (Portugal). <i>Meta Gene</i> , 2014, 2, 638-645.	0.6	3
18	Nystagmus as an early ocular alteration in Machado-Joseph disease (MJD/SCA3). <i>BMC Neurology</i> , 2014, 14, 17.	1.8	24

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19	Transcript Diversity of Machado-Joseph Disease Gene (ATXN3) Is Not Directly Determined by SNPs in Exonic or Flanking Intronic Regions. <i>Journal of Molecular Neuroscience</i> , 2013, 49, 539-543.	2.3	3
20	Patterns of Mitochondrial DNA Damage in Blood and Brain Tissues of a Transgenic Mouse Model of Machado-Joseph Disease. <i>Neurodegenerative Diseases</i> , 2013, 11, 206-214.	1.4	55
21	Psychological Well-Being and Family Satisfaction Levels Five Years After Being Confirmed as a Carrier of the Machado-Joseph Disease Mutation. <i>Genetic Testing and Molecular Biomarkers</i> , 2012, 16, 1363-1368.	0.7	12
22	Sequence Analysis of 5' Regulatory Regions of the Machado-Joseph Disease Gene (ATXN3). <i>Cerebellum</i> , 2012, 11, 1045-1050.	2.5	7
23	Cross-sectional study of risk factors for atherosclerosis in the Azorean population. <i>Annals of Human Biology</i> , 2011, 38, 354-359.	1.0	3
24	Parkinsonian phenotype in Machado-Joseph disease (MJD/SCA3): a two-case report. <i>BMC Neurology</i> , 2011, 11, 131.	1.8	30
25	The <i>APOE</i> ϵ 2 Allele Increases the Risk of Earlier Age at Onset in Machado-Joseph Disease. <i>Archives of Neurology</i> , 2011, 68, 1580.	4.5	33
26	Polymorphism in cardiovascular diseases (CVD) susceptibility loci in the azores islands (Portugal). <i>Open Journal of Genetics</i> , 2011, 01, 48-53.	0.1	3