## Cristina Santos

## List of Publications by Citations

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66
papers

1,016
citations

h-index

20
g-index

3.82
ext. papers

ext. citations

20
g-index

3.82
L-index

#	Paper	IF	Citations
66	Understanding differences between phylogenetic and pedigree-derived mtDNA mutation rate: a model using families from the Azores Islands (Portugal). <i>Molecular Biology and Evolution</i> , <b>2005</b> , 22, 149	o- <sup>8</sup> 505	74
65	A novel mutation in JARID1C gene associated with mental retardation. <i>European Journal of Human Genetics</i> , <b>2006</b> , 14, 583-6	5.3	60
64	Frequency and pattern of heteroplasmy in the complete human mitochondrial genome. <i>PLoS ONE</i> , <b>2013</b> , 8, e74636	3.7	58
63	Human mitochondrial DNA complete amplification and sequencing: a new validated primer set that prevents nuclear DNA sequences of mitochondrial origin co-amplification. <i>Electrophoresis</i> , <b>2009</b> , 30, 15	8 <del>7</del> :93	55
62	Mitochondrial DNA damage patterns and aging: revising the evidences for humans and mice <b>2013</b> , 4, 337-50		49
61	Genetic structure and origin of peopling in the Azores islands (Portugal): the view from mtDNA. <i>Annals of Human Genetics</i> , <b>2003</b> , 67, 433-56	2.2	44
60	Nuclear insertions of mitochondrial origin: Database updating and usefulness in cancer studies. <i>Mitochondrion</i> , <b>2011</b> , 11, 946-53	4.9	38
59	Frequency and pattern of heteroplasmy in the control region of human mitochondrial DNA. <i>Journal of Molecular Evolution</i> , <b>2008</b> , 67, 191-200	3.1	36
58	Population genetics of wild-type CAG repeats in the Machado-Joseph disease gene in Portugal. <i>Human Heredity</i> , <b>2005</b> , 60, 156-63	1.1	33
57	Tracing the origin of the east-west population admixture in the Altai region (Central Asia). <i>PLoS ONE</i> , <b>2012</b> , 7, e48904	3.7	31
56	The APOE I allele increases the risk of earlier age at onset in Machado-Joseph disease. <i>Archives of Neurology</i> , <b>2011</b> , 68, 1580-3		31
55	The decrease of Esynuclein in cortical brain areas defines a molecular subgroup of dementia with Lewy bodies. <i>Brain</i> , <b>2010</b> , 133, 3724-33	11.2	30
54	GBA Mutations Are Associated With Earlier Onset and Male Sex in Dementia With Lewy Bodies. <i>Movement Disorders</i> , <b>2016</b> , 31, 1066-70	7	26
53	Mitochondrial DNA mutations in cancer: a review. Current Topics in Medicinal Chemistry, 2008, 8, 1351-6	563	26
52	Validated primer set that prevents nuclear DNA sequences of mitochondrial origin co-amplification: a revision based on the New Human Genome Reference Sequence (GRCh37). <i>Electrophoresis</i> , <b>2011</b> , 32, 782-3	3.6	25
51	Increased transcript diversity: novel splicing variants of Machado-Joseph disease gene (ATXN3). <i>Neurogenetics</i> , <b>2010</b> , 11, 193-202	3	24
50	Parkinsonian phenotype in Machado-Joseph disease (MJD/SCA3): a two-case report. <i>BMC</i> Neurology, <b>2011</b> , 11, 131	3.1	23

## (2019-2004)

49	Determination of human caucasian mitochondrial DNA haplogroups by means of a hierarchical approach. <i>Human Biology</i> , <b>2004</b> , 76, 431-53	1.2	21	
48	Analysis of segregation patterns in Machado-Joseph disease pedigrees. <i>Journal of Human Genetics</i> , <b>2008</b> , 53, 920-923	4.3	20	
47	Mitochondrial DNA patterns in the Iberian Northern plateau: population dynamics and substructure of the Zamora province. <i>American Journal of Physical Anthropology</i> , <b>2010</b> , 142, 531-9	2.5	19	
46	Mutation patterns of mtDNA: empirical inferences for the coding region. <i>BMC Evolutionary Biology</i> , <b>2008</b> , 8, 167	3	19	
45	Segregation distortion of wild-type alleles at the Machado-Joseph disease locus: a study in normal families from the Azores islands (Portugal). <i>Journal of Human Genetics</i> , <b>2008</b> , 53, 333-339	4.3	19	
44	Sensitivity of mitochondrial DNA heteroplasmy detection using Next Generation Sequencing. <i>Mitochondrion</i> , <b>2020</b> , 50, 88-93	4.9	18	
43	Differential mtDNA damage patterns in a transgenic mouse model of Machado-Joseph disease (MJD/SCA3). <i>Journal of Molecular Neuroscience</i> , <b>2015</b> , 55, 449-53	3.3	15	
42	The role of control region mitochondrial DNA mutations in cardiovascular disease: stroke and myocardial infarction. <i>Scientific Reports</i> , <b>2020</b> , 10, 2766	4.9	15	
41	Accumulation of Mitochondrial DNA Common Deletion Since The Preataxic Stage of Machado-Joseph Disease. <i>Molecular Neurobiology</i> , <b>2019</b> , 56, 119-124	6.2	14	
40	The (CAG)n tract of Machado-Joseph Disease gene (ATXN3): a comparison between DNA and mRNA in patients and controls. <i>European Journal of Human Genetics</i> , <b>2010</b> , 18, 621-3	5.3	13	
39	Evaluation of two methods for computational HLA haplotypes inference using a real dataset. <i>BMC Bioinformatics</i> , <b>2008</b> , 9, 68	3.6	13	
38	The presence of nuclear families in prehistoric collective burials revisited: the bronze age burial of Montanissell Cave (Spain) in the light of aDNA. <i>American Journal of Physical Anthropology</i> , <b>2011</b> , 146, 406-13	2.5	12	
37	Short-term psychological impact of predictive testing for Machado-Joseph disease: depression and anxiety levels in individuals at risk from the Azores (Portugal). <i>Public Health Genomics</i> , <b>2004</b> , 7, 196-201		12	
36	Y-chromosome variation in South Iberia: insights into the North African contribution. <i>American Journal of Human Biology</i> , <b>2009</b> , 21, 407-9	2.7	11	
35	Linguistic isolates in Portugal: insights from the mitochondrial DNA pattern. <i>Forensic Science International: Genetics</i> , <b>2013</b> , 7, 618-623	4.3	10	
34	Mitochondrial DNA and Y-chromosome structure at the Mediterranean and Atlantic falldes of the Iberian Peninsula. <i>American Journal of Human Biology</i> , <b>2014</b> , 26, 130-41	2.7	9	
33	Mitochondrial DNA patterns in the Macaronesia islands: Variation within and among archipelagos. <i>American Journal of Physical Anthropology</i> , <b>2010</b> , 141, 610-9	2.5	8	
32	Mitochondrial DNA haplogroups and age at onset of Machado-Joseph disease/spinocerebellar ataxia type 3: a study in patients from multiple populations. <i>European Journal of Neurology</i> , <b>2019</b> , 26, 506-512	6	7	

31	Sequence analysis of 5Tregulatory regions of the Machado-Joseph disease gene (ATXN3). <i>Cerebellum</i> , <b>2012</b> , 11, 1045-50	4.3	6
30	The African contribution to the present-day population of the Azores Islands (Portugal): analysis of the Y chromosome haplogroup E. <i>American Journal of Human Biology</i> , <b>2007</b> , 19, 854-60	2.7	5
29	Perimortem fracture pattern in ribs by blunt force trauma. <i>International Journal of Legal Medicine</i> , <b>2018</b> , 132, 1205-1213	3.1	4
28	Genetic profiling of the Azores Islands (Portugal): data from 10 X-chromosome STRs. <i>American Journal of Human Biology</i> , <b>2010</b> , 22, 221-3	2.7	4
27	Biodemographic and Genetic Structure of Zamora Province (Spain): Insights from Surname Analysis. <i>Human Ecology</i> , <b>2010</b> , 38, 831-839	2	4
26	Testing hierarchical levels of population sub-structuring: the Azores islands (Portugal) as a case study. <i>Journal of Biosocial Science</i> , <b>2008</b> , 40, 607-21	1.6	4
25	Genetic structure of Flores island (Azores, Portugal) in the 19th century and in the present day: evidence from surname analysis. <i>Human Biology</i> , <b>2005</b> , 77, 317-41	1.2	4
24	Analysis of Y-chromosome variability and its comparison with mtDNA variability reveals different demographic histories between islands in the Azores Archipelago (Portugal). <i>Annals of Human Genetics</i> , <b>2005</b> , 69, 135-44	2.2	4
23	INDEL Length and Haplotypes in the Esynuclein Gene: A Key to Differentiate Dementia with Lewy Bodies?. <i>Journal of Alzheimerts Disease</i> , <b>2018</b> , 65, 207-219	4.3	3
22	Primer effect in the detection of mitochondrial DNA point heteroplasmy by automated sequencing. <i>Mitochondrial DNA</i> , <b>2013</b> , 24, 303-11		3
21	Cross-sectional study of risk factors for atherosclerosis in the Azorean population. <i>Annals of Human Biology</i> , <b>2011</b> , 38, 354-9	1.7	3
20	Polymorphism of the APOE locus in the Azores Islands (Portugal). <i>Human Biology</i> , <b>2006</b> , 78, 509-12	1.2	3
19	New applanation tonometer for myopic patients after laser refractive surgery. <i>Scientific Reports</i> , <b>2020</b> , 10, 7053	4.9	3
18	Genetic identification of Spanish civil war victims. The state of the art in Catalonia (Northeastern Spain). <i>Forensic Science International: Genetics Supplement Series</i> , <b>2019</b> , 7, 419-421	0.5	3
17	Role of mitochondrial DNA variants in the development of fragile X-associated tremor/ataxia syndrome. <i>Mitochondrion</i> , <b>2020</b> , 52, 157-162	4.9	2
16	Verification of Inter-laboratorial Genotyping Consistency in the Molecular Diagnosis of Polyglutamine Spinocerebellar Ataxias. <i>Journal of Molecular Neuroscience</i> , <b>2016</b> , 58, 83-7	3.3	2
15	Human DNA extraction from highly degraded skeletal remains: How to find a suitable method?. <i>Electrophoresis</i> , <b>2020</b> , 41, 2149	3.6	2
14	Sex selection in late Iberian infant burials: Integrating evidence from morphological and genetic data. <i>American Journal of Human Biology</i> , <b>2019</b> , 31, e23204	2.7	2

## LIST OF PUBLICATIONS

Series, **2006**, 1288, 88-90

13	Involvement of mitochondrial haplogroups in myocardial infarction and stroke: A case-control study in Castile and Leon (Spain) population. <i>Mitochondrion</i> , <b>2019</b> , 44, 1-6	4.9	2	
12	Genetic structure of the Azores Islands: a study using 15 autosomal short tandem repeat loci. <i>Collegium Antropologicum</i> , <b>2009</b> , 33, 991-9	0.1	2	
11	Glucocerebrosidase regulators SCARB2 and TFEB are up-regulated in Lewy body disease brain. <i>Neuroscience Letters</i> , <b>2019</b> , 706, 164-168	3.3	1	
10	Y-chromosome analysis in a Northwest Iberian population: unraveling the impact of Northern African lineages. <i>American Journal of Human Biology</i> , <b>2014</b> , 26, 740-6	2.7	1	
9	Molecular polymorphism of the ABO blood group: a study in Poland, Spain, and Andorra. <i>American Journal of Human Biology</i> , <b>2014</b> , 26, 556-8	2.7	1	
8	Peopling, demographic history and genetic structure of the Azores Islands: Integrating data from mtDNA and Y-chromosome. <i>International Congress Series</i> , <b>2006</b> , 1288, 85-87		1	
7	Reappraising the human mitochondrial DNA recombination dogma		1	
6	The role of control region mitochondrial DNA mutations in cardiovascular disease: stroke and myocardial infarction		1	
5	Assessing DNA recovery from highly degraded skeletal remains by using silica-based extraction methods. <i>Forensic Science International: Genetics Supplement Series</i> , <b>2019</b> , 7, 810-812	0.5	1	
4	Challenges in using both eyes of the same patient when evaluating the results after implantation of intrastromal corneal ring segments. <i>Acta Ophthalmologica</i> , <b>2021</b> , 99, e319-e323	3.7	1	
3	Mitochondrial DNA haplogroups J and T increase the risk of glioma. <i>Mitochondrion</i> , <b>2021</b> , 58, 95-101	4.9	О	
2	Intraocular pressure after myopic laser refractive surgery measured with a new Goldmann convex prism: correlations with GAT and ORA <i>BMC Ophthalmology</i> , <b>2022</b> , 22, 79	2.3	Ο	
1	Diversity of maternal and paternal lineages in the geographic extremes of the Azores (Santa Maria and Flores Islands): Insights from mtDNA, Y-chromosome and surname data. <i>International Congress</i>			