Maria JoÃ£o Prata

List of Publications by Year
in descending order

108 papers	2,564 citations	218592 26 h-index	223716 46 g-index
108 all docs	108 docs citations	108 times ranked	3189 citing authors

#	Article	IF	Citations
1	Splicing Modulation as a Promising Therapeutic Strategy for Lysosomal Storage Disorders: The Mucopolysaccharidoses Example. Life, 2022, 12, 608.	1.1	1
2	Evolutionary dynamics of the human pseudoautosomal regions. PLoS Genetics, 2021, 17, e1009532.	1.5	16
3	Compensatory epistasis explored by molecular dynamics simulations. Human Genetics, 2021, 140, 1329-1342.	1.8	6
4	Common polymorphic OTC variants can act as genetic modifiers of enzymatic activity. Human Mutation, 2021, 42, 978-989.	1.1	6
5	Insights into the Middle Eastern paternal genetic pool in Tunisia: high prevalence of T-M70 haplogroup in an Arab population. Scientific Reports, 2021, 11, 15728.	1.6	1
6	Genetic Variability of the Functional Domains of Chromodomains Helicase DNA-Binding (CHD) Proteins. Genes, 2021, 12, 1827.	1.0	7
7	GBA3: a polymorphic pseudogene in humans that experienced repeated gene loss during mammalian evolution. Scientific Reports, 2020, 10, 11565.	1.6	2
8	Lysosomal Storage Disease-Associated Neuropathy: Targeting Stable Nucleic Acid Lipid Particle (SNALP)-Formulated siRNAs to the Brain as a Therapeutic Approach. International Journal of Molecular Sciences, 2020, 21, 5732.	1.8	5
9	Molecular Characterization of a Novel Splicing Mutation Underlying Mucopolysaccharidosis (MPS) Type VIâ€"Indirect Proof of Principle on Its Pathogenicity. Diagnostics, 2020, 10, 58.	1.3	4
10	Development of an Antisense Oligonucleotide-Mediated Exon Skipping Therapeutic Strategy for Mucolipidosis II: Validation at RNA Level. Human Gene Therapy, 2020, 31, 775-783.	1.4	10
11	Unusual Î²-Globin Haplotype Distribution in Newborns from Bengo, Angola. Hemoglobin, 2019, 43, 149-154.	0.4	5
12	Essential genetic findings in neurodevelopmental disorders. Human Genomics, 2019, 13, 31.	1.4	41
13	European Roma groups show complex West Eurasian admixture footprints and a common South Asian genetic origin. PLoS Genetics, 2019, 15, e1008417.	1.5	28
14	The Orientalisation of North Africa: New hints from the study of autosomal STRs in an Arab population. Annals of Human Biology, 2017, 44, 180-190.	0.4	12
15	Y chromosome diversity in a linguistic isolate (Mirandese, NE Portugal). American Journal of Human Biology, 2016, 28, 671-680.	0.8	2
16	Solving a case of allelic dropout in the GNPTAB gene: implications in the molecular diagnosis of mucolipidosis type III alpha/beta. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 1225-1228.	0.4	5
17	Data in support of a functional analysis of splicing mutations in the IDS gene and the use of antisense oligonucleotides to exploit an alternative therapy for MPS II. Data in Brief, 2015, 5, 810-817.	0.5	6
18	Pharmacogenetic Polymorphisms in a Portuguese Gypsy Population. Current Pharmacogenomics and Personalized Medicine, 2015, 13, 36-40.	0.2	2

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19	The peopling of Greenland: further insights from the analysis of genetic diversity using autosomal and X-chromosomal markers. European Journal of Human Genetics, 2015, 23, 245-251.	1.4	15
20	Mosaic maternal ancestry in the Great Lakes region of East Africa. Human Genetics, 2015, 134, 1013-1027.	1.8	18
21	Enriching the knowledge on East Asia populations: Characterization of male lineages from Macau and Shanghai. Forensic Science International: Genetics Supplement Series, 2015, 5, e322-e324.	0.1	0
22	Exploring the relationship between lifestyles, diets and genetic adaptations in humans. BMC Genetics, 2015, 16, 55.	2.7	15
23	Functional analysis of splicing mutations in the IDS gene and the use of antisense oligonucleotides to exploit an alternative therapy for MPS II. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2015, 1852, 2712-2721.	1.8	13
24	Portuguese mitochondrial DNA genetic diversityâ€"An update and a phylogenetic revision. Forensic Science International: Genetics, 2015, 15, 27-32.	1.6	10
25	Mirandese language and genetic differentiation in Iberia: a study using X chromosome markers. Annals of Human Biology, 2015, 42, 20-25.	0.4	10
26	Molecular and computational analyses of genes involved in mannose 6â€phosphate independent trafficking. Clinical Genetics, 2015, 88, 190-194.	1.0	4
27	Distinctive Patterns of Evolution of the Î´-Globin Gene (HBD) in Primates. PLoS ONE, 2015, 10, e0123365.	1.1	7
28	Therapeutic strategies based on modified U1 snRNAs and chaperones for Sanfilippo C splicing mutations. Orphanet Journal of Rare Diseases, 2014, 9, 180.	1.2	42
29	Mucolipidosis II-Related Mutations Inhibit the Exit from the Endoplasmic Reticulum and Proteolytic Cleavage of GlcNAc-1-Phosphotransferase Precursor Protein (GNPTAB). Human Mutation, 2014, 35, 368-376.	1.1	21
30	Prenatal skeletal dysplasia phenotype in severe MLII alpha/beta with novel GNPTAB mutation. Gene, 2014, 542, 266-268.	1.0	10
31	Assessing the potential application of X-chromosomal haploblocks in population genetics and forensic studies. Forensic Science International: Genetics Supplement Series, 2013, 4, e9-e10.	0.1	1
32	Linguistic isolates in Portugal: Insights from the mitochondrial DNA pattern. Forensic Science International: Genetics, 2013, 7, 618-623.	1.6	10
33	Sortilina e risco de doenÃ§a cardiovascular. Revista Portuguesa De Cardiologia, 2013, 32, 793-799.	0.2	8
34	Molecular characterization of maple syrup urine disease patients from Tunisia. Gene, 2013, 517, 116-119.	1.0	13
35	Evolutionary Constraints in the Î²-Globin Cluster: The Signature of Purifying Selection at the Î´-Globin (HBD) Locus and Its Role in Developmental Gene Regulation. Genome Biology and Evolution, 2013, 5, 559-571.	1.1	18
36	A shortcut to the lysosome: The mannose-6-phosphate-independent pathway. Molecular Genetics and Metabolism, 2012, 107, 257-266.	0.5	93

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37	Mannose-6-phosphate pathway: A review on its role in lysosomal function and dysfunction. Molecular Genetics and Metabolism, 2012, 105, 542-550.	0.5	188
38	Mucolipidosis type II Î±/Î² with a homozygous missense mutation in the GNPTAB gene. American Journal of Medical Genetics, Part A, 2012, 158A, 1225-1228.	0.7	10
39	Refining the genetic portrait of Portuguese Roma through Xâ€chromosomal markers. American Journal of Physical Anthropology, 2012, 148, 389-394.	2.1	9
40	Lysosomal multienzymatic complexâ€related diseases: a genetic study among Portuguese patients. Clinical Genetics, 2012, 81, 379-393.	1.0	14
41	A method for the analysis of 32 X chromosome insertion deletion polymorphisms in a single PCR. International Journal of Legal Medicine, 2012, 126, 97-105.	1.2	45
42	Genetic characterization of Somali and Iraqi populations using a set of 33 X-chromosome Indels. Forensic Science International: Genetics Supplement Series, 2011, 3, e137-e138.	0.1	4
43	Study of 25 X-chromosome Single Nucleotide Polymorphisms in African and Asian populations. Forensic Science International: Genetics Supplement Series, 2011, 3, e139-e140.	0.1	1
44	Study of 25 X-chromosome SNPs in the Portuguese. Forensic Science International: Genetics, 2011, 5, 336-338.	1.6	9
45	Origin and spread of a common deletion causing mucolipidosis type II: insights from patterns of haplotypic diversity. Clinical Genetics, 2011, 80, 273-280.	1.0	15
46	Alu-Alu Recombination Underlying the First Large Genomic Deletion in GlcNAc-Phosphotransferase Alpha/Beta (GNPTAB) Gene in a MLII Alpha/Beta Patient. JIMD Reports, 2011, 4, 117-124.	0.7	5
47	Reconstructing the Indian Origin and Dispersal of the European Roma: A Maternal Genetic Perspective. PLoS ONE, 2011, 6, e15988.	1.1	61
48	Genetic profiling of the Azores Islands (Portugal): Data from 10 Xâ€chromosome STRs. American Journal of Human Biology, 2010, 22, 221-223.	0.8	5
49	Genetic characterization of uniparental lineages in populations from Southwest Iberia with past malaria endemicity. American Journal of Human Biology, 2010, 22, 588-595.	0.8	6
50	A genetic historical sketch of European Gypsies: The perspective from autosomal markers. American Journal of Physical Anthropology, 2010, 141, 507-514.	2.1	19
51	High prevalence of the GSTM3*A/B polymorphism in sub-Sarahan African populations. Brazilian Journal of Medical and Biological Research, 2010, 43, 677-680.	0.7	3
52	Incidence of maple syrup urine disease in Portugal. Molecular Genetics and Metabolism, 2010, 100, 385-387.	0.5	18
53	VKORC1 polymorphisms in Brazilians: comparison with the Portuguese and Portuguese-speaking Africans and pharmacogenetic implications. Pharmacogenomics, 2010, 11, 1257-1267.	0.6	23
54	Patterns of pharmacogenetic diversity in African populations: role of ancient and recent history. Pharmacogenomics, 2009, 10, 1413-1422.	0.6	9

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55	Polymorphisms in one-carbon metabolism pathway genes and risk for bladder cancer in a Tunisian population. Cancer Genetics and Cytogenetics, 2009, 195, 43-53.	1.0	26
56	Molecular analysis of the GNPTAB and GNPTG genes in 13 patients with mucolipidosis type II or type III â€“ identification of eight novel mutations. Clinical Genetics, 2009, 76, 76-84.	1.0	42
57	In search of the Preâ€•and Postâ€Neolithic Genetic Substrates in Iberia: Evidence from Yâ€Chromosome in Pyrenean Populations. Annals of Human Genetics, 2009, 73, 42-53.	0.3	26
58	Revisiting MSUD in Portuguese Gypsies: Evidence for a Founder Mutation and for a Mutational Hotspot within the BCKDHA Gene. Annals of Human Genetics, 2009, 73, 298-303.	0.3	12
59	The Karimojong from Uganda: Genetic characterization using an X-STR decaplex system. Forensic Science International: Genetics, 2009, 3, e127-e128.	1.6	21
60	Association between STRs from the X chromosome in a sample of Portuguese Gypsies. Forensic Science International: Genetics Supplement Series, 2009, 2, 391-393.	0.1	3
61	A Perspective on the History of the Iberian Gypsies Provided by Phylogeographic Analysis of Yâ€Chromosome Lineages. Annals of Human Genetics, 2008, 72, 215-227.	0.3	45
62	Molecular analysis of mucopolysaccharidosis type IIIB in Portugal: evidence of a single origin for a common mutation (R234C) in the Iberian Peninsula. Clinical Genetics, 2008, 73, 251-256.	1.0	14
63	Molecular characterization of Portuguese patients with mucopolysaccharidosis IIIC: two novel mutations in the HGSNAT gene. Clinical Genetics, 2008, 74, 194-195.	1.0	15
64	Molecular and structural analyses of maple syrup urine disease and identification of a founder mutation in a Portuguese Gypsy community. Molecular Genetics and Metabolism, 2008, 94, 148-156.	0.5	27
65	Refining the analysis of Y-chromosomal diversity in Alentejo (Portugal). Forensic Science International: Genetics Supplement Series, 2008, 1, 208-209.	0.1	1
66	Y-chromosomal STR haplotypes in a Gypsy population from Portugal. Forensic Science International: Genetics Supplement Series, 2008, 1, 212-213.	0.1	1
67	Pharmacogenetically relevant polymorphisms in Portugal. Pharmacogenomics, 2007, 8, 703-712.	0.6	34
68	Analysis of 10 X-STRs in three African populations. Forensic Science International: Genetics, 2007, 1, 208-211.	1.6	27
69	The African contribution to the presentâ€day population of the Azores Islands (Portugal): Analysis of the Y chromosome haplogroup E. American Journal of Human Biology, 2007, 19, 854-860.	0.8	6
70	Population data for Y-chromosome haplotypes defined by 17 STRs (AmpFlSTR YFiler) in Portugal. Forensic Science International, 2007, 171, 250-255.	1.3	36
71	Dissecting the Genetic History of SÃ£o TomÃ© e PrÃncipe: A New Window from Y-Chromosome Biallelic Markers. Annals of Human Genetics, 2007, 71, 77-85.	0.3	9
72	Methylenetetrahydrofolate reductase and methionine synthase polymorphisms and risk of bladder cancer in a Tunisian population. Cancer Genetics and Cytogenetics, 2007, 176, 48-53.	1.0	30

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73	Do the distribution patterns of polymorphisms at the thiopurine S-methyltransferase locus in sub-Saharan populations need revision? Hints from Cabinda and Mozambique. European Journal of Clinical Pharmacology, 2007, 63, 703-706.	0.8	35
74	Y-STR polymorphisms from Basque-speaking region of Cinco Villas (Navarra) in the context of the Pyrenean genetic landscape. International Congress Series, 2006, 1288, 198-200.	0.2	1
75	Genetic analysis of autosomal and Y-specific STRs in the Karimojong population from Uganda. International Congress Series, 2006, 1288, 213-215.	0.2	5
76	High-resolution analysis of Y-biallelic markers in three populations from SÃ£o TomÃ© e PrÃncipe. International Congress Series, 2006, 1288, 28-30.	0.2	0
77	Outcome in acute lymphoblastic leukemia: Influence of thiopurine methyltransferase genetic polymorphisms. International Congress Series, 2006, 1288, 789-791.	0.2	1
78	Frequency of the thiopurine S-methyltransferase alleles in the ancient genetic population isolate of Sardinia. Journal of Clinical Pharmacy and Therapeutics, 2006, 31, 283-287.	0.7	16
79	Molecular characterization of Portuguese patients with mucopolysaccharidosis type II shows evidence that the IDS gene is prone to splicing mutations. Journal of Inherited Metabolic Disease, 2006, 29, 743-754.	1.7	34
80	The MTHFR C677T and A1298C Polymorphisms and Susceptibility to Childhood Acute Lymphoblastic Leukemia in Portugal. Journal of Pediatric Hematology/Oncology, 2005, 27, 425-429.	0.3	45
81	Peopling of the Azore Islands (Portugal): Data from the Y Chromosome. Human Biology, 2005, 77, 189-199.	0.4	5
82	Analysis of Y-chromosome variability and its comparison with mtDNA variability reveals different demographic histories between islands in the Azores Archipelago (Portugal). Annals of Human Genetics, 2005, 69, 135-44.	0.3	10
83	Pattern of mtDNA Variation in Three Populations from Sao Tome e Principe. Annals of Human Genetics, 2004, 68, 40-54.	0.3	35
84	Tracing the Origin of the Most Common Thiopurine Methyltransferase (TPMT) Variants: Preliminary Data from the Patterns of Haplotypic Association with Two CA Repeats. Annals of Human Genetics, 2004, 68, 313-323.	0.3	22
85	GATA C4 allele 17 as a marker for sub-Saharan origin of Y-chromosome lineages. International Congress Series, 2004, 1261, 281-283.	0.2	0
86	Insights from pattern of mtDNA variation into the genetic history of SÃ£o TomÃ© e PrÄ±ìncipe. International Congress Series, 2004, 1261, 377-379.	0.2	1
87	A Recent Shift from Polygyny to Monogamy in Humans Is Suggested by the Analysis of Worldwide Y-Chromosome Diversity. Journal of Molecular Evolution, 2003, 57, 85-97.	0.8	90
88	Phylogeny of the mtDNA haplogroup U6. Analysis of the sequences observed in North Africa and Iberia. International Congress Series, 2003, 1239, 491-493.	0.2	2
89	An evaluation of the proportion of identical Y-STR haplotypes due to recurrent mutation. International Congress Series, 2003, 1239, 57-60.	0.2	2
90	Evolution of a VNTR located within the promoter region of the thiopurine methyltransferase gene: inferences from population and sequence data. Human Genetics, 2002, 111, 172-178.	1.8	11

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91	Mitochondrial portrait of the Cabo Verde archipelago: the Senegambian outpost of Atlantic slave trade. Annals of Human Genetics, 2002, 66, 49-60.	0.3	61
92	Mismatch distribution analysis of Y-STR haplotypes as a tool for the evaluation of identity-by-state proportions and significance of matchesâ€"the European picture. Forensic Science International, 2002, 130, 147-155.	1.3	14
93	The GSTM1 and GSTT1 genetic polymorphisms and susceptibility to acute lymphoblastic leukemia in children from north Portugal. Leukemia, 2002, 16, 1565-1567.	3.3	34
94	Bantu and European Y-lineages in Sub-Saharan Africa. Annals of Human Genetics, 2002, 66, 369-78.	0.3	26
95	STR data from S. TomÃ© e PrÄ±ncipe (Gulf of Guinea, West Africa). Forensic Science International, 2001, 116, 53-54.	1.3	8
96	Influence of the variable number of tandem repeats located in the promoter region of the thiopurine methyltransferase gene on enzymatic activity. Clinical Pharmacology and Therapeutics, 2001, 70, 165-174.	2.3	69
97	STR data for the AmpFlSTR profiler plus loci from Macau (China). Forensic Science International, 2001, 123, 74-75.	1.3	10
98	Phylogeography of the human mitochondrial haplogroup L3e: a snapshot of African prehistory and Atlantic slave trade. Annals of Human Genetics, 2001, 65, 549-63.	0.3	33
99	Evidence for population sub-structuring in SÃ£o TomÃ© e PrÃncipe as inferred from Y-chromosome STR analysis. Annals of Human Genetics, 2001, 65, 271-83.	0.3	7
100	Screening of thiopurine Sâ€methyltransferase mutations by horizontal conformationâ€sensitive gel electrophoresis. Human Mutation, 2000, 15, 246-253.	1.1	28
101	Diversity of mtDNA lineages in Portugal: not a genetic edge of European variation. Annals of Human Genetics, 2000, 64, 491-506.	0.3	110
102	Allelic Affinities between the F13A Common Gene Products Inferred by the Analysis of an (AAAG)n STR Polymorphism within the 5â€² Untranslated Region. Human Heredity, 2000, 50, 189-193.	0.4	1
103	Y-Chromosomal Diversity in Europe Is Clinal and Influenced Primarily by Geography, Rather than by Language. American Journal of Human Genetics, 2000, 67, 1526-1543.	2.6	519
104	Detection of additional structural variation at the FES/FPS system and population data from S. TomÃ© e PrÃncipe and North Portugal. International Journal of Legal Medicine, 1999, 112, 204-206.	1.2	3
105	Failed PCR amplifications of MBP-STR alleles due to polymorphism in the primer annealing region. International Journal of Legal Medicine, 1996, 108, 313-315.	1.2	26
106	The STR system hTPO: Population and segregation data. International Journal of Legal Medicine, 1995, 108, 167-169.	1.2	11
107	Genetic polymorphism of human peptidase C, PEPC (E.C.3.4.1.1): formal genetic and population data. Human Genetics, 1989, 83, 197-198.	1.8	3
108	Hb F Levels in Î²-Thalassemia Carriers and Normal Individuals: Known and Unknown Quantitative Trait Loci in the Î²-Globin Gene Cluster. Hemoglobin, 0, , 1-8.	0.4	0