Maria Joo Prata

List of Publications by Year in Descending Order

106 papers	2,088 citations	24 h-index	43 g-index
108 ext. papers	2,313 ext. citations	3.2 avg, IF	4.17 L-index

#	Paper	IF	Citations
106	Splicing Modulation as a Promising Therapeutic Strategy for Lysosomal Storage Disorders: The Mucopolysaccharidoses Example. *Life*, **2022**, 12, 608	3	
105	Evolutionary dynamics of the human pseudoautosomal regions. *PLoS Genetics*, **2021**, 17, e1009532	6	0
104	Compensatory epistasis explored by molecular dynamics simulations. *Human Genetics*, **2021**, 140, 1329-1342	3.9	1
103	Common polymorphic OTC variants can act as genetic modifiers of enzymatic activity. *Human Mutation*, **2021**, 42, 978-989	4.7	2
102	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	4.9	0
101	Molecular Characterization of a Novel Splicing Mutation underlying Mucopolysaccharidosis (MPS) type VI-Indirect Proof of Principle on Its Pathogenicity. *Diagnostics*, **2020**, 10,	3.8	2
100	GBA3: a polymorphic pseudogene in humans that experienced repeated gene loss during mammalian evolution. *Scientific Reports*, **2020**, 10, 11565	4.9	0
99	Development of an Antisense Oligonucleotide-Mediated Exon Skipping Therapeutic Strategy for Mucolipidosis II: Validation at RNA Level. *Human Gene Therapy*, **2020**, 31, 775-783	4.8	4
98	European Roma groups show complex West Eurasian admixture footprints and a common South Asian genetic origin. *PLoS Genetics*, **2019**, 15, e1008417	6	15
97	Unusual Globin Haplotype Distribution in Newborns from Bengo, Angola. *Hemoglobin*, **2019**, 43, 149-154	0.6	3
96	Essential genetic findings in neurodevelopmental disorders. *Human Genomics*, **2019**, 13, 31	6.8	17
95	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	1.7	8
94	Y chromosome diversity in a linguistic isolate (Mirandese, NE Portugal). *American Journal of Human Biology*, **2016**, 28, 671-80	2.7	2
93	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	1.6	4
92	Mosaic maternal ancestry in the Great Lakes region of East Africa. *Human Genetics*, **2015**, 134, 1013-27	6.3	11
91	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	0.5	
90	Exploring the relationship between lifestyles, diets and genetic adaptations in humans. *BMC Genetics*, **2015**, 16, 55	2.6	11

89	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, 2712-21	6.9	10
88	Portuguese mitochondrial DNA genetic diversity-An update and a phylogenetic revision. *Forensic Science International: Genetics*, **2015**, 15, 27-32	4.3	10
87	Mirandese language and genetic differentiation in Iberia: a study using X chromosome markers. *Annals of Human Biology*, **2015**, 42, 20-5	1.7	9
86	Molecular and computational analyses of genes involved in mannose 6-phosphate independent trafficking. *Clinical Genetics*, **2015**, 88, 190-4	4	3
85	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, 810-7	1.2	5
84	Pharmacogenetic Polymorphisms in a Portuguese Gypsy Population. *Current Pharmacogenomics and Personalized Medicine*, **2015**, 13, 36-40	0.4	1
83	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, 245-51	5.3	13
82	Distinctive patterns of evolution of the β-globin gene (HBD) in primates. *PLoS ONE*, **2015**, 10, e0123365	3.7	4
81	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, 368-76	4.7	21
80	Prenatal skeletal dysplasia phenotype in severe MLII alpha/beta with novel GNPTAB mutation. *Gene* , **2014**, 542, 266-8	3.8	10
79	Therapeutic strategies based on modified U1 snRNAs and chaperones for Sanfilippo C splicing mutations. *Orphanet Journal of Rare Diseases*, **2014**, 9, 180	4.2	31
78	Assessing the potential application of X-chromosomal haploblocks in population genetics and forensic studies. *Forensic Science International: Genetics Supplement Series*, **2013**, 4, e9-e10	0.5	1
77	Linguistic isolates in Portugal: insights from the mitochondrial DNA pattern. *Forensic Science International: Genetics*, **2013**, 7, 618-623	4.3	10
76	Sortilin and the risk of cardiovascular disease. *Revista Portuguesa De Cardiologia*, **2013**, 32, 793-9	1	4
75	Molecular characterization of maple syrup urine disease patients from Tunisia. *Gene*, **2013**, 517, 116-9	3.8	9
74	Evolutionary constraints in the β-globin cluster: the signature of purifying selection at the β-globin (HBD) locus and its role in developmental gene regulation. *Genome Biology and Evolution*, **2013**, 5, 559-71	3.9	13
73	Lysosomal multienzymatic complex-related diseases: a genetic study among Portuguese patients. *Clinical Genetics*, **2012**, 81, 379-93	4	10
72	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	3.1	39

71	A shortcut to the lysosome: the mannose-6-phosphate-independent pathway. *Molecular Genetics and Metabolism*, **2012**, 107, 257-66	3.7	67
70	Mannose-6-phosphate pathway: a review on its role in lysosomal function and dysfunction. *Molecular Genetics and Metabolism*, **2012**, 105, 542-50	3.7	139
69	Mucolipidosis type II [illegible] with a homozygous missense mutation in the GNPTAB gene. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1225-8	2.5	8
68	Refining the genetic portrait of Portuguese Roma through X-chromosomal markers. *American Journal of Physical Anthropology*, **2012**, 148, 389-94	2.5	6
67	Alu-Alu Recombination Underlying the First Large Genomic Deletion in GlcNAc-Phosphotransferase Alpha/Beta (GNPTAB) Gene in a MLII Alpha/Beta Patient. *JIMD Reports*, **2012**, 4, 117-24	1.9	4
66	Study of 25 X-chromosome Single Nucleotide Polymorphisms in African and Asian populations. *Forensic Science International: Genetics Supplement Series*, **2011**, 3, e139-e140	0.5	1
65	Study of 25 X-chromosome SNPs in the Portuguese. *Forensic Science International: Genetics*, **2011**, 5, 336-43	4	7
64	Origin and spread of a common deletion causing mucolipidosis type II: insights from patterns of haplotypic diversity. *Clinical Genetics*, **2011**, 80, 273-80	4	13
63	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	0.5	4
62	Reconstructing the Indian origin and dispersal of the European Roma: a maternal genetic perspective. *PLoS ONE*, **2011**, 6, e15988	3.7	53
61	High prevalence of the GSTM3*A/B polymorphism in sub-Sarahan African populations. *Brazilian Journal of Medical and Biological Research*, **2010**, 43, 677-80	2.8	3
60	Incidence of maple syrup urine disease in Portugal. *Molecular Genetics and Metabolism*, **2010**, 100, 385-7	3.7	13
59	VKORC1 polymorphisms in Brazilians: comparison with the Portuguese and Portuguese-speaking Africans and pharmacogenetic implications. *Pharmacogenomics*, **2010**, 11, 1257-67	2.6	21
58	Genetic profiling of the Azores Islands (Portugal): data from 10 X-chromosome STRs. *American Journal of Human Biology*, **2010**, 22, 221-3	2.7	4
57	Genetic characterization of uniparental lineages in populations from Southwest Iberia with past malaria endemicity. *American Journal of Human Biology*, **2010**, 22, 588-95	2.7	6
56	A genetic historical sketch of European Gypsies: The perspective from autosomal markers. *American Journal of Physical Anthropology*, **2010**, 141, 507-14	2.5	14
55	Patterns of pharmacogenetic diversity in African populations: role of ancient and recent history. *Pharmacogenomics*, **2009**, 10, 1413-22	2.6	8
54	Polymorphisms in one-carbon metabolism pathway genes and risk for bladder cancer in a Tunisian population. *Cancer Genetics and Cytogenetics*, **2009**, 195, 43-53		20

53	Molecular analysis of the GNPTAB and GNPTG genes in 13 patients with mucolipidosis type II or type III - identification of eight novel mutations. *Clinical Genetics*, **2009**, 76, 76-84	4	41
52	In search of the pre- and post-neolithic genetic substrates in Iberia: evidence from Y-chromosome in Pyrenean populations. *Annals of Human Genetics*, **2009**, 73, 42-53	2.2	23
51	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	2.2	11
50	The Karimojong from Uganda: genetic characterization using an X-STR decaplex system. *Forensic Science International: Genetics*, **2009**, 3, e127-8	4.3	20
49	Association between STRs from the X chromosome in a sample of Portuguese Gypsies. *Forensic Science International: Genetics Supplement Series*, **2009**, 2, 391-393	0.5	3
48	A perspective on the history of the Iberian gypsies provided by phylogeographic analysis of Y-chromosome lineages. *Annals of Human Genetics*, **2008**, 72, 215-27	2.2	39
47	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, 251-6	4	11
46	Molecular characterization of Portuguese patients with mucopolysaccharidosis IIIC: two novel mutations in the HGSNAT gene. *Clinical Genetics*, **2008**, 74, 194-5	4	12
45	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, 148-56	3.7	24
44	Refining the analysis of Y-chromosomal diversity in Alentejo (Portugal). *Forensic Science International: Genetics Supplement Series*, **2008**, 1, 208-209	0.5	1
43	Y-chromosomal STR haplotypes in a Gypsy population from Portugal. *Forensic Science International: Genetics Supplement Series*, **2008**, 1, 212-213	0.5	1
42	The African contribution to the present-day population of the Azores Islands (Portugal): analysis of the Y chromosome haplogroup E. *American Journal of Human Biology*, **2007**, 19, 854-60	2.7	5
41	Population data for Y-chromosome haplotypes defined by 17 STRs (AmpFlSTR YFiler) in Portugal. *Forensic Science International*, **2007**, 171, 250-5	2.6	35
40	Dissecting the genetic history of São Tomé e Príncipe: a new window from Y-chromosome biallelic markers. *Annals of Human Genetics*, **2007**, 71, 77-85	2.2	4
39	Methylenetetrahydrofolate reductase and methionine synthase polymorphisms and risk of bladder cancer in a Tunisian population. *Cancer Genetics and Cytogenetics*, **2007**, 176, 48-53		24
38	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, 703-6	2.8	26
37	Pharmacogenetically relevant polymorphisms in Portugal. *Pharmacogenomics*, **2007**, 8, 703-12	2.6	31
36	Analysis of 10 X-STRs in three African populations. *Forensic Science International: Genetics*, **2007**, 1, 208-14	4.3	26

35	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		1
34	Genetic analysis of autosomal and Y-specific STRs in the Karimojong population from Uganda. *International Congress Series*, **2006**, 1288, 213-215		5
33	High-resolution analysis of Y-biallelic markers in three populations from São Tomé e Príncipe. *International Congress Series*, **2006**, 1288, 28-30		
32	Outcome in acute lymphoblastic leukemia: Influence of thiopurine methyltransferase genetic polymorphisms. *International Congress Series*, **2006**, 1288, 789-791		
31	Frequency of the thiopurine S-methyltransferase alleles in the ancient genetic population isolate of Sardinia. *Journal of Clinical Pharmacy and Therapeutics*, **2006**, 31, 283-7	2.2	13
30	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, 743-54	5.4	28
29	The MTHFR C677T and A1298C polymorphisms and susceptibility to childhood acute lymphoblastic leukemia in Portugal. *Journal of Pediatric Hematology/Oncology*, **2005**, 27, 425-9	1.2	43
28	Peopling of the Azores Islands (Portugal): data from the Y chromosome. *Human Biology*, **2005**, 77, 189-99	1.2	4
27	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	2.2	4
26	Pattern of mtDNA variation in three populations from São Tomé e Príncipe. *Annals of Human Genetics*, **2004**, 68, 40-54	2.2	29
25	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, 313-23	2.2	21
24	GATA C4 allele 17 as a marker for sub-Saharan origin of Y-chromosome lineages. *International Congress Series*, **2004**, 1261, 281-283		
23	Insights from pattern of mtDNA variation into the genetic history of São Tomé e Príncipe. *International Congress Series*, **2004**, 1261, 377-379		1
22	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	3.1	74
21	Phylogeny of the mtDNA haplogroup U6. Analysis of the sequences observed in North Africa and Iberia. *International Congress Series*, **2003**, 1239, 491-493		2
20	An evaluation of the proportion of identical Y-STR haplotypes due to recurrent mutation. *International Congress Series*, **2003**, 1239, 57-60		2
19	Evolution of a VNTR located within the promoter region of the thiopurine methyltransferase gene: inferences from population and sequence data. *Human Genetics*, **2002**, 111, 172-8	6.3	7
18	Mitochondrial portrait of the Cabo Verde archipelago: the Senegambian outpost of Atlantic slave trade. *Annals of Human Genetics*, **2002**, 66, 49-60	2.2	55

17	Mismatch distribution analysis of Y-STR haplotypes as a tool for the evaluation of identity-by-state proportions and significance of matches--the European picture. *Forensic Science International*, **2002**, 130, 147-55	2.6	11
16	The GSTM1 and GSTT1 genetic polymorphisms and susceptibility to acute lymphoblastic leukemia in children from north Portugal. *Leukemia*, **2002**, 16, 1565-7	10.7	30
15	Bantu and European Y-lineages in Sub-Saharan Africa. *Annals of Human Genetics*, **2002**, 66, 369-78	2.2	13
14	STR data from S. Tomé Príncipe (Gulf of Guinea, West Africa). *Forensic Science International*, **2001**, 116, 53-4	2.6	6
13	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, 165-74	6.1	58
12	STR data for the AmpFlSTR profiler plus loci from Macau (China). *Forensic Science International*, **2001**, 123, 74-5	2.6	9
11	Phylogeography of the human mitochondrial haplogroup L3e: a snapshot of African prehistory and Atlantic slave trade. *Annals of Human Genetics*, **2001**, 65, 549-63	2.2	25
10	Evidence for population sub-structuring in São Tomé Príncipe as inferred from Y-chromosome STR analysis. *Annals of Human Genetics*, **2001**, 65, 271-83	2.2	1
9	Screening of thiopurine S-methyltransferase mutations by horizontal conformation-sensitive gel electrophoresis. *Human Mutation*, **2000**, 15, 246-53	4.7	22
8	Diversity of mtDNA lineages in Portugal: not a genetic edge of European variation. *Annals of Human Genetics*, **2000**, 64, 491-506	2.2	103
7	Allelic affinities between the F13A common gene products inferred by the analysis of an (AAAG)n STR polymorphism within the 5Suntranslated region. *Human Heredity*, **2000**, 50, 189-93	1.1	1
6	Y-chromosomal diversity in Europe is clinal and influenced primarily by geography, rather than by language. *American Journal of Human Genetics*, **2000**, 67, 1526-43	11	471
5	Detection of additional structural variation at the FES/FPS system and population data from S. Tomé Príncipe and North Portugal. *International Journal of Legal Medicine*, **1999**, 112, 204-6	3.1	1
4	Failed PCR amplifications of MBP-STR alleles due to polymorphism in the primer annealing region. *International Journal of Legal Medicine*, **1996**, 108, 313-5	3.1	25
3	The STR system hTPO: population and segregation data. *International Journal of Legal Medicine*, **1995**, 108, 167-9	3.1	10
2	Genetic polymorphism of human peptidase C, PEPC (E.C.3.4.1.1): formal genetic and population data. *Human Genetics*, **1989**, 83, 197-8	6.3	2
1	Hb F Levels in β-Thalassemia Carriers and Normal Individuals: Known and Unknown Quantitative Trait Loci in the β-Globin Gene Cluster. *Hemoglobin*,1-8	0.6	