Maria Brion

List of Publications by Year in Descending Order

97 papers	2,876 citations	26 h-index	52 g-index
99 ext. papers	3,220 ext. citations	3.8 avg, IF	4.24 L-index

#	Paper	IF	Citations
97	Searching for genetic modulators of the phenotypic heterogeneity in Brugada syndrome.. *PLoS ONE*, **2022**, 17, e0263469	3.7	
96	Association of Genetic Variants With Outcomes in Patients With Nonischemic Dilated Cardiomyopathy. *Journal of the American College of Cardiology*, **2021**, 78, 1682-1699	15.1	5
95	Estudio molecular del s□ndrome de plaqueta pegajosa mediante secuenciaci□ de exoma. *Revista Facultad De Medicina*, **2021**, 69, e76806	0.4	
94	Association of variants in MYH7, MYBPC3 and TNNT2 with sudden cardiac death-related risk factors in Brazilian patients with hypertrophic cardiomyopathy. *Forensic Science International: Genetics*, **2021**, 52, 102478	4.3	4
93	COVID-19 and treatment guided by biochemical and molecular diagnostic tests to reduce myocardial damage and cardiotoxicity. *Revista Espanola De Cardiologia (English Ed)*, **2020**, 73, 691-693	0.7	
92	Polygenic risk score as a key factor in cardiovascular clinical prediction models. *Revista Espanola De Cardiologia (English Ed)*, **2020**, 73, 608-610	0.7	
91	Genetic variants of uncertain significance: How to match scientific rigour and standard of proof in sudden cardiac death?. *Legal Medicine*, **2020**, 45, 101712	1.9	12
90	La puntuaci□ de riesgo poligᅵnico como factor clave en los modelos de predicci□ clᅵnica cardiovascular. *Revista Espanola De Cardiologia*, **2020**, 73, 608-610	1.5	
89	Una nueva delecci□ de calsequestrina 2 que causa taquicardia ventricular polimᅵrfica catecolaminᅵrgica y muerte s□bita cardiaca. *Revista Espanola De Cardiologia*, **2019**, 72, 681-683	1.5	1
88	Sudden infant death as the most severe phenotype caused by genetic modulation in a family with atrial fibrillation. *Forensic Science International: Genetics*, **2019**, 43, 102159	4.3	0
87	A Novel Calsequestrin 2 Deletion Causing Catecholaminergic Polymorphic Ventricular Tachycardia and Sudden Cardiac Death. *Revista Espanola De Cardiologia (English Ed)*, **2019**, 72, 681-683	0.7	1
86	Research update for articles published in EJCI in 2016. *European Journal of Clinical Investigation*, **2018**, 48, e13016	4.6	
85	Innovative Strategies in Heart Failure: Present and Future. *Archives of Medical Research*, **2018**, 49, 558-56	7.6	2
84	Sudden unexpected death in the young - Value of massive parallel sequencing in postmortem genetic analyses. *Forensic Science International*, **2018**, 293, 70-76	2.6	13
83	Postmortem genetic testing should be recommended in sudden cardiac death cases due to thoracic aortic dissection. *International Journal of Legal Medicine*, **2017**, 131, 1211-1219	3.1	8
82	Relationship between fibrillin-1 genotype and severity of cardiovascular involvement in Marfan syndrome. *Heart*, **2017**, 103, 1795-1799	5.1	49
81	The genetic component of bicuspid aortic valve and aortic dilation. An exome-wide association study. *Journal of Molecular and Cellular Cardiology*, **2017**, 102, 3-9	5.8	8

80	ABCC3 Polymorphisms and mRNA Expression Influence the Concentration of a Carboxylic Acid Metabolite in Patients on Clopidogrel and Aspirin Therapy. *Basic and Clinical Pharmacology and Toxicology*, **2017**, 120, 466-474	3.1	2
79	Medico-legal perspectives on sudden cardiac death in young athletes. *International Journal of Legal Medicine*, **2017**, 131, 393-409	3.1	15
78	The relationship between surname frequency and Y chromosome variation in Spain. *European Journal of Human Genetics*, **2016**, 24, 120-8	5.3	20
77	PRKG1 and genetic diagnosis of early-onset thoracic aortic disease. *European Journal of Clinical Investigation*, **2016**, 46, 787-94	4.6	13
76	Prospective and Retrospective Diagnosis of Barth Syndrome Aided by Next-Generation Sequencing. *American Journal of Clinical Pathology*, **2016**, 145, 507-13	1.9	7
75	Resequencing and association analysis of coding regions at twenty candidate genes suggest a role for rare risk variation at AKAP9 and protective variation at NRXN1 in schizophrenia susceptibility. *Journal of Psychiatric Research*, **2015**, 66-67, 38-44	5.2	12
74	Massive parallel sequencing applied to the molecular autopsy in sudden cardiac death in the young. *Forensic Science International: Genetics*, **2015**, 18, 160-70	4.3	25
73	Broad-based molecular autopsy: a potential tool to investigate the involvement of subtle cardiac conditions in sudden unexpected death in infancy and early childhood. *Archives of Disease in Childhood*, **2015**, 100, 952-6	2.2	22
72	Predicting proliferative vitreoretinopathy: temporal and external validation of models based on genetic and clinical variables. *British Journal of Ophthalmology*, **2015**, 99, 41-8	5.5	16
71	Genetic Analysis of Arrhythmogenic Diseases in the Era of NGS: The Complexity of Clinical Decision-Making in Brugada Syndrome. *PLoS ONE*, **2015**, 10, e0133037	3.7	32
70	Association between SNPs of Metalloproteinases and Prostaglandin F2 Receptor Genes and Latanoprost Response in Open-Angle Glaucoma. *Ophthalmology*, **2015**, 122, 1040-8.e4	7.3	19
69	Y-chromosomal DNA analysis in French male lineages. *Forensic Science International: Genetics*, **2014**, 9, 162-8	4.3	12
68	Whole exome sequencing for the identification of a new mutation in TGFB2 involved in a familial case of non-syndromic aortic disease. *Clinica Chimica Acta*, **2014**, 437, 88-92	6.2	14
67	Next generation sequencing challenges in the analysis of cardiac sudden death due to arrhythmogenic disorders. *Electrophoresis*, **2014**, 35, 3111-6	3.6	17
66	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. *Nature Genetics*, **2014**, 46, 826-36	36.3	199
65	Involvement of LCA5 in Leber congenital amaurosis and retinitis pigmentosa in the Spanish population. *Ophthalmology*, **2014**, 121, 399-407	7.3	17
64	Pharmacogenomics of anti-platelet therapy focused on peripheral blood cells of coronary arterial disease patients. *Clinica Chimica Acta*, **2013**, 425, 9-17	6.2	5
63	Genetic variants in genes related to lipid metabolism and atherosclerosis, dyslipidemia and atorvastatin response. *Clinica Chimica Acta*, **2013**, 417, 8-11	6.2	23

62	Novel genes detected by transcriptional profiling from whole-blood cells in patients with early onset of acute coronary syndrome. *Clinica Chimica Acta*, **2013**, 421, 184-90	6.2	29
61	A genetic case-control study confirms the implication of SMAD7 and TNF locus in the development of proliferative vitreoretinopathy **2013**, 54, 1665-78		33
60	Increased clopidogrel response is associated with ABCC3 expression: a pilot study. *Clinica Chimica Acta*, **2012**, 413, 417-21	6.2	13
59	Sarcomeric gene mutations in sudden infant death syndrome (SIDS). *Forensic Science International*, **2012**, 219, 278-81	2.6	42
58	Genetic association study of age-related macular degeneration in the Spanish population. *Acta Ophthalmologica*, **2011**, 89, e12-22	3.7	31
57	Prevalence of HCM and long QT syndrome mutations in young sudden cardiac death-related cases. *International Journal of Legal Medicine*, **2011**, 125, 565-72	3.1	22
56	Identification of a novel mutation in the human PDE6A gene in autosomal recessive retinitis pigmentosa: homology with the nmf28/nmf28 mice model. *Clinical Genetics*, **2010**, 78, 495-8	4	19
55	A strong genetic association between the tumor necrosis factor locus and proliferative vitreoretinopathy: the retina 4 project. *Ophthalmology*, **2010**, 117, 2417-2423.e1-2	7.3	44
54	Y-chromosome lineages in native South American population. *Forensic Science International: Genetics*, **2010**, 4, 187-93	4.3	20
53	Genetic variants near TIMP3 and high-density lipoprotein-associated loci influence susceptibility to age-related macular degeneration. *Proceedings of the National Academy of Sciences of the United States of America*, **2010**, 107, 7401-6	11.5	417
52	New technologies in the genetic approach to sudden cardiac death in the young. *Forensic Science International*, **2010**, 203, 15-24	2.6	17
51	A new approach to long QT syndrome mutation detection by Sequenom MassARRAY system. *Electrophoresis*, **2010**, 31, 1648-55	3.6	12
50	Ancestry analysis in the 11-M Madrid bomb attack investigation. *PLoS ONE*, **2009**, 4, e6583	3.7	95
49	Development of predictive models of proliferative vitreoretinopathy based on genetic variables: the Retina 4 project **2009**, 50, 2384-90		29
48	Moors and Saracens in Europe: estimating the medieval North African male legacy in southern Europe. *European Journal of Human Genetics*, **2009**, 17, 848-52	5.3	34
47	The genetic male component of two South-Western Colombian populations. *Forensic Science International: Genetics*, **2009**, 3, e59-61	4.3	8
46	Phylogeography of French male lineages. *Forensic Science International: Genetics Supplement Series*, **2009**, 2, 439-441	0.5	8
45	Involvement of hypertrophic cardiomyopathy genes in sudden infant death syndrome (SIDS). *Forensic Science International: Genetics Supplement Series*, **2009**, 2, 495-496	0.5	12

44	Sequenom MassArray application in the long QT syndrome mutation detection. *Forensic Science International: Genetics Supplement Series*, **2009**, 2, 497-498	0.5	
43	RAS gene polymorphisms, classical risk factors and the advent of coronary artery disease in the Portuguese population. *BMC Cardiovascular Disorders*, **2008**, 8, 15	2.3	34
42	Methylenetetrahydrofolate reductase gene, homocysteine and coronary artery disease: the A1298C polymorphism does matter. Inferences from a case study (Madeira, Portugal). *Thrombosis Research*, **2008**, 122, 648-56	8.2	18
41	Forensic validation and implementation of Y-chromosome SNP multiplexes. *Forensic Science International: Genetics Supplement Series*, **2008**, 1, 181-183	0.5	1
40	Large scale analysis of HCM mutations in sudden cardiac death. *Forensic Science International: Genetics Supplement Series*, **2008**, 1, 549-550	0.5	3
39	Chapter 30 The human Y chromosome male-specific polymorphisms and forensic genetics. *Handbook of Analytical Separations*, **2008**, 969-1000	0.7	1
38	Novel splice donor site mutation in MERTK gene associated with retinitis pigmentosa. *British Journal of Ophthalmology*, **2008**, 92, 1419-23	5.5	25
37	Molecular genetics of sudden cardiac death. *Forensic Science International*, **2008**, 182, 1-12	2.6	66
36	The genetic legacy of religious diversity and intolerance: paternal lineages of Christians, Jews, and Muslims in the Iberian Peninsula. *American Journal of Human Genetics*, **2008**, 83, 725-36	11	151
35	Novel high-throughput SNP genotyping cosegregation analysis for genetic diagnosis of autosomal recessive retinitis pigmentosa and Leber congenital amaurosis. *Human Mutation*, **2007**, 28, 511-6	4.7	27
34	The genetic male legacy from El Salvador. *Forensic Science International*, **2007**, 171, 198-203	2.6	13
33	Forensic validation of the SNPforID 52-plex assay. *Forensic Science International: Genetics*, **2007**, 1, 186-90	4.3	66
32	Phylogenetic evidence for multiple independent duplication events at the DYS19 locus. *Forensic Science International: Genetics*, **2007**, 1, 287-90	4.3	7
31	Analysis of artificially degraded DNA using STRs and SNPs--results of a collaborative European (EDNAP) exercise. *Forensic Science International*, **2006**, 164, 33-44	2.6	112
30	The presence of antibodies to oxidative modified proteins in serum from polycystic ovary syndrome patients. *Clinical and Experimental Immunology*, **2006**, 144, 217-22	6.2	77
29	Autosomal microsatellite data from Northwestern Colombia. *Forensic Science International*, **2006**, 160, 217-20	2.6	5
28	Y chromosome SNP analysis using the single-base extension: a hierarchical multiplex design. *Methods in Molecular Biology*, **2005**, 297, 229-42	1.4	6
27	A collaborative study of the EDNAP group regarding Y-chromosome binary polymorphism analysis. *Forensic Science International*, **2005**, 153, 103-8	2.6	10

26	Y-chromosome haplotype analysis in Antioquia (Colombia). *Forensic Science International*, **2005**, 151, 85-91	2.6	9
25	SNPs in forensic genetics: a review on SNP typing methodologies. *Forensic Science International*, **2005**, 154, 181-94	2.6	299
24	Introduction of an single nucleodite polymorphism-based "Major Y-chromosome haplogroup typing kit" suitable for predicting the geographical origin of male lineages. *Electrophoresis*, **2005**, 26, 4411-20	3.6	70
23	Hierarchical analysis of 30 Y-chromosome SNPs in European populations. *International Journal of Legal Medicine*, **2005**, 119, 10-5	3.1	88
22	Duplications of the Y-chromosome specific loci P25 and 92R7 and forensic implications. *Forensic Science International*, **2004**, 140, 241-50	2.6	22
21	Nineteen autosomal microsatellite data from Antioquia (Colombia). *Forensic Science International*, **2004**, 143, 69-71	2.6	5
20	Y-chromosome STR-haplotype typing in El Salvador. *Forensic Science International*, **2004**, 142, 45-9	2.6	10
19	Micro-geographical differentiation in Northern Iberia revealed by Y-chromosomal DNA analysis. *Gene*, **2004**, 329, 17-25	3.8	36
18	Selecting single nucleotide polymorphisms for forensic applications. *International Congress Series*, **2004**, 1261, 18-20		7
17	SNP genotyping with single base extension-tag microarrays. *International Congress Series*, **2004**, 1261, 331-333		9
16	GATA C4 allele 17 as a marker for sub-Saharan origin of Y-chromosome lineages. *International Congress Series*, **2004**, 1261, 281-283		
15	29 Y-chromosome SNP analysis in European populations. *International Congress Series*, **2004**, 1261, 73-75		1
14	Insights into Iberian population origins through the construction of highly informative Y-chromosome haplotypes using biallelic markers, STRs, and the MSY1 minisatellite. *American Journal of Physical Anthropology*, **2003**, 122, 147-61	2.5	25
13	Population data of Galicia (NW Spain) on the new Y-STRs DYS437, DYS438, DYS439, GATA A10, GATA A7.1, GATA A7.2, GATA C4 and GATA H4. *Forensic Science International*, **2003**, 131, 220-4	2.6	6
12	Study of eight novel Y-chromosome STRs in a sample from Valencia (East of Spain): analysis of gene and haplotypes frequencies. *International Congress Series*, **2003**, 1239, 431-434		
11	Microgeographic patterns of highly informative Y-chromosome haplotypes (using biallelic markers and STRs) in Galicia (NW Spain): forensic and anthropological implications. *International Congress Series*, **2003**, 1239, 61-66		
10	Typing Y-chromosome single nucleotide polymorphisms with DNA microarray technology. *International Congress Series*, **2003**, 1239, 21-25		4
9	New method to measure minisatellite variant repeat variation in population genetic studies. *American Journal of Human Biology*, **2002**, 14, 421-8	2.7	8

8	Point mutations in the flanking regions of the Y-chromosome specific STRs DYS391, DYS437 and DYS438. *International Journal of Legal Medicine*, **2002**, 116, 322-6	3.1	10
7	Genetic data on eight STRs (D5S818, D7S820, F13B, LPL, TH01, TPOX, VWA31, CSF1PO) from a Colombian population. *Forensic Science International*, **2002**, 129, 216-8	2.6	7
6	Y-chromosome variation in a Norwegian population sample. *Forensic Science International*, **2001**, 117, 163-73	2.6	29
5	The use of the LightCycler for the detection of Y chromosome SNPs. *Forensic Science International*, **2001**, 118, 163-8	2.6	23
4	Chapter 20C The human Y-chromosome. Male-specific polymorphisms and forensic genetics. *Handbook of Analytical Separations*, **2000**, 2, 721-735	0.7	1
3	Distribution of Y-chromosome STR defined haplotypes in Iberia. *Forensic Science International*, **2000** , 110, 117-26	2.6	42
2	Y chromosome specific polymorphisms in forensic analysis. *Legal Medicine*, **1999**, 1, 55-60	1.9	14
1	Robustness of the Y STRs DYS19, DYS389 I and II, DYS390 and DYS393: optimization of a PCR pentaplex. *Forensic Science International*, **1999**, 106, 163-72	2.6	69