Jorge Amigo

List of Publications by Year in descending order

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47 papers	1,476 citations	331670 21 h-index	330143 37 g-index
48 all docs	48 docs citations	48 times ranked	2732 citing authors

#	Article	IF	Citations
1	Genome-wide identification of hypoxia-inducible factor binding sites and target genes by a probabilistic model integrating transcription-profiling data and in silico binding site prediction. Nucleic Acids Research, 2010, 38, 2332-2345.	14.5	179
2	BigBWA: approaching the Burrowsâ€“Wheeler aligner to Big Data technologies. Bioinformatics, 2015, 31, 4003-4005.	4.1	107
3	Analysis of global variability in 15 established and 5 new European Standard Set (ESS) STRs using the CEPH human genome diversity panel. Forensic Science International: Genetics, 2011, 5, 155-169.	3.1	103
4	SPSmart: adapting population based SNP genotype databases for fast and comprehensive web access. BMC Bioinformatics, 2008, 9, 428.	2.6	95
5	SparkBWA: Speeding Up the Alignment of High-Throughput DNA Sequencing Data. PLoS ONE, 2016, 11, e0155461.	2.5	72
6	Building a custom large-scale panel of novel microhaplotypes for forensic identification using MiSeq and Ion S5 massively parallel sequencing systems. Forensic Science International: Genetics, 2020, 45, 102213.	3.1	70
7	The impact of modern migrations on present-day multi-ethnic Argentina as recorded on the mitochondrial DNA genome. BMC Genetics, 2011, 12, 77.	2.7	63
8	SNP analysis to results (SNPator): a web-based environment oriented to statistical genomics analyses upon SNP data. Bioinformatics, 2008, 24, 1643-1644.	4.1	61
9	The SNPforID browser: an online tool for query and display of frequency data from the SNPforID project. International Journal of Legal Medicine, 2008, 122, 435-440.	2.2	47
10	Genetic Analysis of Arrhythmogenic Diseases in the Era of NGS: The Complexity of Clinical Decision-Making in Brugada Syndrome. PLoS ONE, 2015, 10, e0133037.	2.5	46
11	Tetra-allelic SNPs: Informative forensic markers compiled from public whole-genome sequence data. Forensic Science International: Genetics, 2015, 19, 100-106.	3.1	44
12	A novel stop mutation in the vascular endothelial growth factor-C gene (VEGFC) results in Milroy-like disease. Journal of Medical Genetics, 2014, 51, 475-478.	3.2	43
13	pop.STRâ€”An online population frequency browser for established and new forensic STRs. Forensic Science International: Genetics Supplement Series, 2009, 2, 361-362.	0.3	38
14	ENGINES: exploring single nucleotide variation in entire human genomes. BMC Bioinformatics, 2011, 12, 105.	2.6	34
15	A compilation of tri-allelic SNPs from 1000 Genomes and use of the most polymorphic loci for a large-scale human identification panel. Forensic Science International: Genetics, 2020, 46, 102232.	3.1	34
16	CSVS, a crowdsourcing database of the Spanish population genetic variability. Nucleic Acids Research, 2021, 49, D1130-D1137.	14.5	34
17	Completion of a worldwide reference panel of samples for an ancestry informative Indel assay. Forensic Science International: Genetics, 2015, 17, 75-80.	3.1	30
18	Whole Exome Sequencing reveals new candidate genes in host genomic susceptibility to Respiratory Syncytial Virus Disease. Scientific Reports, 2017, 7, 15888.	3.3	29

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19	Next generation sequencing challenges in the analysis of cardiac sudden death due to arrhythmogenic disorders. Electrophoresis, 2014, 35, 3111-3116.	2.4	24
20	Comprehensive molecular testing in patients with high functioning autism spectrum disorder. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2016, 784-785, 46-52.	1.0	24
21	A Genome-Wide Study of Modern-Day Tuscans: Revisiting Herodotus's Theory on the Origin of the Etruscans. PLoS ONE, 2014, 9, e105920.	2.5	23
22	D5S2500 is an ambiguously characterized STR: Identification and description of forensic microsatellites in the genomics age.. Forensic Science International: Genetics, 2016, 23, 19-24.	3.1	21
23	Adaptive selection of an incretin gene in Eurasian populations. Genome Research, 2011, 21, 21-32.	5.5	19
24	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.	1.1	19
25	Global population variability in Qiagen Investigator HDplex STRs. Forensic Science International: Genetics, 2014, 8, 36-43.	3.1	19
26	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.	3.1	18
27	Longitudinal analysis on parasite diversity in honeybee colonies: new taxa, high frequency of mixed infections and seasonal patterns of variation. Scientific Reports, 2020, 10, 10454.	3.3	18
28	Viability of in-house datamarting approaches for population genetics analysis of SNP genotypes. BMC Bioinformatics, 2009, 10, S5.	2.6	17
29	Broadening the Applicability of a Custom Multi-Platform Panel of Microhaplotypes: Bio-Geographical Ancestry Inference and Expanded Reference Data. Frontiers in Genetics, 2020, 11, 581041.	2.3	17
30	Mitogenomes from The 1000 Genome Project Reveal New Near Eastern Features in Present-Day Tuscans. PLoS ONE, 2015, 10, e0119242.	2.5	15
31	A Reduced Number of mtSNPs Saturates Mitochondrial DNA Haplotype Diversity of Worldwide Population Groups. PLoS ONE, 2010, 5, e10218.	2.5	13
32	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.	2.2	13
33	A Generalized Model to Estimate the Statistical Power in Mitochondrial Disease Studies Involving 2Ã—k Tables. PLoS ONE, 2013, 8, e73567.	2.5	11
34	tagFinder: A Novel Tag Analysis Methodology That Enables Detection of Molecules from DNA-Encoded Chemical Libraries. SLAS Discovery, 2018, 23, 397-404.	2.7	10
35	Whole Exome Sequencing Identifies New Host Genomic Susceptibility Factors in Empyema Caused by Streptococcus pneumoniae in Children: A Pilot Study. Genes, 2018, 9, 240.	2.4	9
36	Early Colorectal Cancers Provide New Evidence for a Lynch Syndrome-to-CMMRD Phenotypic Continuum. Cancers, 2019, 11, 1081.	3.7	8

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37	An efficient screening method for simultaneous detection of recurrent copy number variants associated with psychiatric disorders. Clinica Chimica Acta, 2015, 445, 34-40.	1.1	7
38	Evaluating the Calling Performance of a Rare Disease NGS Panel for Single Nucleotide and Copy Number Variants. Molecular Diagnosis and Therapy, 2017, 21, 303-313.	3.8	7
39	Trio-based exome sequencing reveals a high rate of the de novo variants in intellectual disability. European Journal of Human Genetics, 2022, 30, 938-945.	2.8	7
40	Targeted resequencing of regulatory regions at schizophrenia risk loci: Role of rare functional variants at chromatin repressive states. Schizophrenia Research, 2016, 174, 10-16.	2.0	6
41	Identification of putative second genetic hits in schizophrenia carriers of high-risk copy number variants and resequencing in additional samples. European Archives of Psychiatry and Clinical Neuroscience, 2018, 268, 585-592.	3.2	6
42	Normalization of DNA encoded library affinity selection results driven by high throughput sequencing and HPLC purification. Bioorganic and Medicinal Chemistry, 2021, 40, 116178.	3.0	6
43	Call for participation in the neurogenetics consortium within the Human Variome Project. Neurogenetics, 2011, 12, 169-173.	1.4	5
44	Targeted NGS meets expert clinical characterization: Efficient diagnosis of spastic paraplegia type 11. Applied & Translational Genomics, 2015, 5, 33-36.	2.1	3
45	GDF: Dealing with High-throughput Genotyping Multiplatform Data for Medical and Population Genetic Applications. Journal of Proteomics and Bioinformatics, 2012, 05, .	0.4	2
46	Viability of in-house datamarting approaches for population genetics analysis of snp genotypes. , 2008, , .		0
47	Identification of genes carrying rare variants of moderate to large effect in schizophrenia: A replication study. Schizophrenia Research, 2018, 197, 577-578.	2.0	0