SÃ©rgio D J Pena

List of Publications by Year
in descending order

63 papers	5,157 citations	147801 31 h-index	133252 59 g-index
63 all docs	63 docs citations	63 times ranked	5228 citing authors

#	Article	IF	Citations
1	Peeking into the mysterious world of Trypanosoma cruzi and Chagas disease. Memorias Do Instituto Oswaldo Cruz, 2022, 117, e210193chgsa.	1.6	0
2	Structural analysis of new compound heterozygous variants in PEPD gene identified in a patient with Prolidase Deficiency diagnosed by exome sequencing. Genetics and Molecular Biology, 2021, 44, e20200393.	1.3	2
3	Genetic admixture in Brazil. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 928-938.	1.6	45
4	â€œExome sequencing identifies a novel homozygous variant in NDRG4 in a family with infantile myofibromatosis (Linhares etÂal., 2014)â€•turns out to be EBV+ leiomyomatosis caused by CARMIL2 mutations. European Journal of Medical Genetics, 2018, 61, 106.	1.3	4
5	Nephrotic syndrome and adrenal insufficiency caused by a variant in SGPL1. CKJ: Clinical Kidney Journal, 2018, 11, 462-467.	2.9	32
6	Familial STAG2 germline mutation defines a new human cohesinopathy. Npj Genomic Medicine, 2017, 2, 7.	3.8	56
7	Mendel,MD: A user-friendly open-source web tool for analyzing WES and WGS in the diagnosis of patients with Mendelian disorders. PLoS Computational Biology, 2017, 13, e1005520.	3.2	10
8	New native South American Y chromosome lineages. Journal of Human Genetics, 2016, 61, 593-603.	2.3	28
9	Association of Genetic Variants with Self-Assessed Color Categories in Brazilians. PLoS ONE, 2014, 9, e83926.	2.5	38
10	Exome sequencing identifies a novel homozygous variant in NDRG4 in a family with infantile myofibromatosis. European Journal of Medical Genetics, 2014, 57, 643-648.	1.3	17
11	Two ancient human genomes reveal Polynesian ancestry among the indigenous Botocudos of Brazil. Current Biology, 2014, 24, R1035-R1037.	3.9	73
12	Global Pharmacogenomics: Distribution of CYP3A5 Polymorphisms and Phenotypes in the Brazilian Population. PLoS ONE, 2014, 9, e83472.	2.5	34
13	Distribution of CYP2D6 Alleles and Phenotypes in the Brazilian Population. PLoS ONE, 2014, 9, e110691.	2.5	49
14	Tracing the genomic ancestry of Peruvians reveals a major legacy of pre-Columbian ancestors. Journal of Human Genetics, 2013, 58, 627-634.	2.3	58
15	Application of the F_{ST} statistics to explore pharmacogenomic diversity in the Brazilian population. Pharmacogenomics, 2012, 13, 771-777.	1.3	22
16	Automated Genotyping of a Highly Informative Panel of 40 Short Insertion-Deletion Polymorphisms Resolved in Polyacrylamide Gels for Forensic Identification and Kinship Analysis. Transfusion Medicine and Hemotherapy, 2012, 39, 211-216.	1.6	15
17	Influence of Genomic Ancestry on the Distribution of SLCO1B1, SLCO1B3 and ABCB1 Gene Polymorphisms among Brazilians. Basic and Clinical Pharmacology and Toxicology, 2012, 110, 460-468.	2.5	31
18	The Genomic Ancestry of Individuals from Different Geographical Regions of Brazil Is More Uniform Than Expected. PLoS ONE, 2011, 6, e17063.	2.5	489

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19	Extensive admixture in Brazilian sickle cell patients: implications for the mapping of genetic modifiers. Blood, 2011, 118, 4493-4495.	1.4	20
20	Estimating the degree of identity by descent in consanguineous couples. Human Mutation, 2011, 32, 1350-1358.	2.5	9
21	The Biological Nonexistence versus the Social Existence of Human Races: Can Science Instruct the Social Ethos?. , 2011, , 69-99.		14
22	VKORC1 polymorphisms in Brazilians: comparison with the Portuguese and Portuguese-speaking Africans and pharmacogenetic implications. Pharmacogenomics, 2010, 11, 1257-1267.	1.3	23
23	Color, Race, and Genomic Ancestry in Brazil. Current Anthropology, 2009, 50, 787-819.	1.6	111
24	Trypanosoma cruzi: ancestral genomes and population structure. Memorias Do Instituto Oswaldo Cruz, 2009, 104, 108-114.	1.6	23
25	The Phylogeography of African Brazilians. Human Heredity, 2008, 65, 23-32.	0.8	62
26	Self-reported skin color, genomic ancestry and the distribution of GST polymorphisms. Pharmacogenetics and Genomics, 2007, 17, 765-771.	1.5	63
27	Impact of population admixture on the distribution of theCYP3A5*3polymorphism. Pharmacogenomics, 2007, 8, 1299-1306.	1.3	44
28	A Worldwide Phylogeography for the Human X Chromosome. PLoS ONE, 2007, 2, e557.	2.5	15
29	The Genetic Structure of Human Populations Studied Through Short Insertion-Deletion Polymorphisms. Annals of Human Genetics, 2006, 70, 658-665.	0.8	111
30	Phylogeography of Haplotypes of Five Microsatellites Located in a Low-recombination Region of the X Chromosome: Studies Worldwide and in Brazilian Populations. Genetica, 2006, 126, 243-250.	1.1	6
31	Y Chromosome Diversity in Brazilians: Switching Perspectives from Slow to Fast Evolving Markers. Genetica, 2006, 126, 251-260.	1.1	18
32	Multiplex Protocol Suitable for Screening for MECP2 Mutations in Girls with Mental Retardation. Clinical Chemistry, 2006, 52, 539-540.	3.2	3
33	Ancestral Genomes, Sex, and the Population Structure of Trypanosoma cruzi. PLoS Pathogens, 2006, 2, e24.	4.7	225
34	Color and Genomic Ancestry in Brazilians: A Study with Forensic Microsatellites. Human Heredity, 2006, 62, 190-195.	0.8	144
35	Detection of in a white Brazilian subject. Clinical Pharmacology and Therapeutics, 2005, 77, 587-588.	4.7	11
36	Trypanosoma cruzi: genetic structure of populations and relevance of genetic variability to the pathogenesis of chagas disease. Memorias Do Instituto Oswaldo Cruz, 2004, 99, 1-12.	1.6	241

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37	The mutational spectrum of human autosomal tetranucleotide microsatellites. Human Mutation, 2003, 21, 71-79.	2.5	56
38	Color and genomic ancestry in Brazilians. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 177-182.	7.1	796
39	Single-Nucleotide Polymorphisms of the Trypanosoma cruzi MSH2 Gene Support the Existence of Three Phylogenetic Lineages Presenting Differences in Mismatch-Repair Efficiency. Genetics, 2003, 164, 117-126.	2.9	40
40	Chagas disease: role of parasite genetic variation in pathogenesis. Expert Reviews in Molecular Medicine, 2002, 4, 1-16.	3.9	48
41	The Phylogeography of Brazilian Y-Chromosome Lineages. American Journal of Human Genetics, 2001, 68, 281-286.	6.2	309
42	Genetic Differentiation in South Amerindians Is Related to Environmental and Cultural Diversity: Evidence from the Y Chromosome. American Journal of Human Genetics, 2001, 68, 1485-1496.	6.2	179
43	Microsatellite instability in tumors as a model to study the process of microsatellite mutations. Human Molecular Genetics, 2000, 9, 347-352.	2.9	31
44	Genetic Characterization of Trypanosoma cruzi Directly from Tissues of Patients with Chronic Chagas Disease. American Journal of Pathology, 2000, 156, 1805-1809.	3.8	222
45	Molecular characterization and population study of an X chromosome homolog of the Y-linked microsatellite DYS391. Gene, 2000, 247, 233-240.	2.2	9
46	The Ancestry of Brazilian mtDNA Lineages. American Journal of Human Genetics, 2000, 67, 444-461.	6.2	549
47	Diagnosis of the fragile X syndrome in males using methylation-specific PCR of the FMRI locus. Genetics and Molecular Biology, 1999, 22, 169-172.	1.3	3
48	Divergent Human Y-Chromosome Microsatellite Evolution Rates. Journal of Molecular Evolution, 1999, 49, 204-214.	1.8	33
49	Differential tissue distribution of diverse clones of Trypanosoma cruzi in infected mice. Molecular and Biochemical Parasitology, 1999, 100, 163-172.	1.1	168
50	Fully mutated and gray-zoneFRAXA alleles in Brazilian mentally retarded boys. , 1999, 84, 198-201.		27
51	The Central Siberian Origin for Native American Y Chromosomes. American Journal of Human Genetics, 1999, 64, 619-628.	6.2	184
52	PCR-based DNA Profiling of Human Y Chromosomes. , 1999, , 133-152.		4
53	Origin of the Amerindians. Science, 1999, 283, 2017b-2017.	12.6	3
54	Polarity of mutations in tumor-associated microsatellite instability. Human Genetics, 1998, 102, 231-235.	3.8	15

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55	Molecular Cytogenetics: : PCR-based diagnosis of human trisomies using computer-assisted laser densitometry. Genetics and Molecular Biology, 1998, 21, 317-322.	1.3	7
56	Simultaneous detection of size and sequence polymorphisms in the transcribed trinucleotide repeat D2S196E (EST00493). Human Genetics, 1997, 99, 796-800.	3.8	12
57	Geographic differences in the allele frequencies of the human Y-linked tetranucleotide polymorphism DYS19. Human Genetics, 1996, 97, 309-313.	3.8	64
58	A PCR-based test suitable for screening for fragile X syndrome among mentally retarded males. Human Genetics, 1996, 97, 808-812.	3.8	56
59	A PCR-based test suitable for screening for fragile X syndrome among mentally retarded males. Human Genetics, 1996, 97, 808-812.	3.8	9
60	A major founder Yâ€“chromosome haplotype in Amerindians. Nature Genetics, 1995, 11, 15-16.	21.4	86
61	PCR haplotypes for the human Y chromosome based on alphoid satellite DNA variants and heteroduplex analysis. Gene, 1995, 165, 191-198.	2.2	55
62	CAT repeat polymorphism in a human expressed sequence tag (EST00444) (D1 3S308). Human Molecular Genetics, 1993, 2, 1748-1748.	2.9	8
63	DNA bioprints: Simple nonisotopic DNA fingerprints with biotinylated probes. Electrophoresis, 1991, 12, 146-152.	2.4	38