Luisa Azevedo

List of Publications by Year
in descending order

48 papers	1,285 citations	471509 17 h-index	395702 33 g-index
51 all docs	51 docs citations	51 times ranked	2424 citing authors

#	Article	IF	Citations
1	The Human Gene Mutation Database (HGMDÂ®): optimizing its use in a clinical diagnostic or research setting. Human Genetics, 2020, 139, 1197-1207.	3.8	353
2	An X-Linked Haplotype of Neandertal Origin Is Present Among All Non-African Populations. Molecular Biology and Evolution, 2011, 28, 1957-1962.	8.9	87
3	Frequency and Pattern of Heteroplasmy in the Complete Human Mitochondrial Genome. PLoS ONE, 2013, 8, e74636.	2.5	69
4	Gains, Losses and Changes of Function after Gene Duplication: Study of the Metallothionein Family. PLoS ONE, 2011, 6, e18487.	2.5	67
5	Novel L2HGDH mutations in 21 patients with L-2-hydroxyglutaric aciduria of Portuguese origin. Human Mutation, 2005, 26, 395-396.	2.5	47
6	Epistatic interactions: how strong in disease and evolution?. Trends in Genetics, 2006, 22, 581-585.	6.7	45
7	Novel TTC19 mutation in a family with severe psychiatric manifestations and complex III deficiency. Neurogenetics, 2013, 14, 153-160.	1.4	42
8	Essential genetic findings in neurodevelopmental disorders. Human Genomics, 2019, 13, 31.	2.9	41
9	Ancestral Origin of the ATTCT Repeat Expansion in Spinocerebellar Ataxia Type 10 (SCA10). PLoS ONE, 2009, 4, e4553.	2.5	40
10	Trans-species polymorphism in humans and the great apes is generally maintained by balancing selection that modulates the host immune response. Human Genomics, 2015, 9, 21.	2.9	39
11	Epistatic interactions modulate the evolution of mammalian mitochondrial respiratory complex components. BMC Genomics, 2009, 10, 266.	2.8	33
12	The Yeast Saccharomyces cerevisiae as a Model for Understanding RAS Proteins and their Role in Human Tumorigenesis. Cells, 2018, 7, 14.	4.1	33
13	Discussion on common data analysis strategies used in MSâ€based proteomics. Proteomics, 2011, 11, 604-619.	2.2	31
14	Molecular mechanisms underlying large genomic deletions in ornithine transcarbamylase (OTC) gene. Clinical Genetics, 2009, 75, 457-464.	2.0	24
15	Improving the in silico assessment of pathogenicity for compensated variants. European Journal of Human Genetics, 2017, 25, 2-7.	2.8	24
16	NAMPT and NAPRT1: novel polymorphisms and distribution of variants between normal tissues and tumor samples. Scientific Reports, 2014, 4, 6311.	3.3	21
17	3-Methylcrotonyl-CoA carboxylase deficiency: Mutational spectrum derived from comprehensive newborn screening. Gene, 2016, 594, 203-210.	2.2	20
18	Characterization of human NLZ1/ZNF703 identifies conserved domains essential for proper subcellular localization and transcriptional repression. Journal of Cellular Biochemistry, 2013, 114, 120-133.	2.6	18

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19	Major influence of repetitive elements on disease-associated copy number variants (CNVs). Human Genomics, 2016, 10, 30.	2.9	18
20	Relative frequency of known causes of multiple mtDNA deletions: Two novel POLG mutations. Neuromuscular Disorders, 2011, 21, 483-488.	0.6	16
21	Trimethylaminuria (fish odor syndrome): Genotype characterization among Portuguese patients. Gene, 2013, 527, 366-370.	2.2	16
22	In vitro demonstration of intra-locus compensation using the ornithine transcarbamylase protein as model. Human Molecular Genetics, 2007, 16, 2209-2214.	2.9	15
23	SLC35A2-CDG: Novel variant and review. Molecular Genetics and Metabolism Reports, 2021, 26, 100717.	1.1	15
24	Congenital Disorder of Glycosylation Type Ia: Searching for the Origin of Common Mutations inPMM2. Annals of Human Genetics, 2007, 71, 348-353.	0.8	13
25	New polymorphic sites within ornithine transcarbamylase gene: population genetics studies and implications for diagnosis. Molecular Genetics and Metabolism, 2003, 78, 152-157.	1.1	12
26	Evolutionary History and Functional Diversification of Phosphomannomutase Genes. Journal of Molecular Evolution, 2010, 71, 119-127.	1.8	11
27	Evaluation of InnoQuantÂ® HY and InnoTyperÂ® 21 kits in the DNA analysis of rootless hair samples. Forensic Science International: Genetics, 2019, 39, 61-65.	3.1	10
28	Congenital Disorders of Glycosylation in Portugalâ€"Two Decades of Experience. Journal of Pediatrics, 2021, 231, 148-156.	1.8	9
29	Consequences of primer binding-sites polymorphisms on genotyping practice. Open Journal of Genetics, 2011, 01, 15-17.	0.1	9
30	Ornithine transcarbamylase deficiency: a novel splice site mutation in a family with meiotic recombination and a new useful SNP for diagnosis. Molecular Genetics and Metabolism, 2002, 76, 68-70.	1.1	8
31	Mutational Spectrum and Linkage Disequilibrium Patterns at the Ornithine Transcarbamylase Gene (OTC). Annals of Human Genetics, 2006, 70, 797-801.	0.8	8
32	Comparative analyses of the Conserved Oligomeric Golgi (COG) complex in vertebrates. BMC Evolutionary Biology, 2010, 10, 212.	3.2	8
33	The Evolutionary Portrait of Metazoan NAD Salvage. PLoS ONE, 2013, 8, e64674.	2.5	8
34	Characterization of the Human Ornithine Transcarbamylase 3â€² Untranslated Regulatory Region. DNA and Cell Biology, 2012, 31, 427-433.	1.9	7
35	Human carbamoyl phosphate synthetase I (CPSI): Insights on the structural role of the unknown function domains. Biochemical and Biophysical Research Communications, 2012, 421, 409-412.	2.1	7
36	Internal validation of two new retrotransposons-based kits (InnoQuant Â® HY and InnoTyper Â® 21) at a forensic lab. Forensic Science International, 2018, 283, 1-8.	2.2	7

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37	The Role of Recombination in the Origin and Evolution of Alu Subfamilies. PLoS ONE, 2013, 8, e64884.	2.5	7
38	Genetic Variability of the Functional Domains of Chromodomains Helicase DNA-Binding (CHD) Proteins. Genes, 2021, 12, 1827.	2.4	7
39	Identification of novel L2HGDH gene mutations and update of the pathological spectrum. Journal of Human Genetics, 2010, 55, 55-58.	2.3	6
40	Compensatory epistasis explored by molecular dynamics simulations. Human Genetics, 2021, 140, 1329-1342.	3.8	6
41	Common polymorphic OTC variants can act as genetic modifiers of enzymatic activity. Human Mutation, 2021, 42, 978-989.	2.5	6
42	Transcriptional regulation of the human mitochondrial peptide deformylase (PDF). Biochemical and Biophysical Research Communications, 2012, 421, 825-831.	2.1	5
43	Evidence for mutational cis-acting factors affecting mutagenesis in the ornithine transcarbamylase gene. Human Mutation, 2004, 24, 273-273.	2.5	4
44	Successful COG8 and PDF overlap is mediated by alterations in splicing and polyadenylation signals. Human Genetics, 2012, 131, 265-274.	3.8	4
45	Identification of maternal uniparental isodisomy of chromosome 10 in a patient with mitochondrial DNA depletion syndrome. Molecular Genetics and Metabolism, 2013, 110, 493-494.	1.1	3
46	Haplotype study of microsatellites flanking the t(15;17) breakpoint in acute promyelocytic leukemia patients from North Portugal. Leukemia, 2002, 16, 1353-1357.	7.2	2
47	The mitochondrial genome of the pinewood nematode (Bursaphelenchus xylophilus) lineage introduced in Europe. Mitochondrial DNA, 2014, 25, 420-421.	0.6	2
48	GBA3: a polymorphic pseudogene in humans that experienced repeated gene loss during mammalian evolution. Scientific Reports, 2020, 10, 11565.	3.3	2