Isabel Marques

List of Publications by Year
in descending order

15	291	1478505 6	1199594 12
papers	citations	h-index	g-index

15	15	15	493
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Supramolecular Organization of the Respiratory Chain in Neurospora crassa Mitochondria. Eukaryotic Cell, 2007, 6, 2391-2405.	3.4	88
2	New findings of Neurospora in Europe and comparisons of diversity in temperate climates on continental scales. Mycologia, 2006, 98, 550-559.	1.9	64
3	Composition of complex I from Neurospora crassa and disruption of two â€œaccessoryâ€•subunits. Biochimica Et Biophysica Acta - Bioenergetics, 2005, 1707, 211-220.	1.0	49
4	The 9.8 kDa Subunit of Complex I, Related to Bacterial Na+-translocating NADH Dehydrogenases, is Required for Enzyme Assembly and Function in Neurospora crassa. Journal of Molecular Biology, 2003, 329, 283-290.	4.2	21
5	Unraveling the pathogenesis of ARX polyalanine tract variants using a clinical and molecular interfacing approach. Molecular Genetics & Genomic Medicine, 2015, 3, 203-214.	1.2	21
6	Contraction of fully expanded FMR1 alleles to the normal range: predisposing haplotype or rare events?. Journal of Human Genetics, 2017, 62, 269-275.	2.3	18
7	Role of the Conserved Cysteine Residues of the 11.5 kDa Subunit in Complex I Catalytic Properties. Journal of Biochemistry, 2007, 141, 489-493.	1.7	6
8	FXTAS is rare among Portuguese patients with movement disorders: FMR1 premutations may be associated with a wider spectrum of phenotypes. Behavioral and Brain Functions, 2011, 7, 19.	3.3	6
9	Development and validation of a multiplex-PCR assay for X-linked intellectual disability. BMC Medical Genetics, 2013, 14, 80.	2.1	6
10	Classical fragile-X phenotype in a female infant disclosed by comprehensive genomic studies. BMC Medical Genetics, 2018, 19, 74.	2.1	6
11	Two Novel Pathogenic MID1 Variants and Genotype-Phenotype Correlation Reanalysis in X-Linked Opitz G/BBB Syndrome. Molecular Syndromology, 2018, 9, 45-51.	0.8	3
12	Development and Validation of a Mathematical Model to Predict the Complexity of FMR1 Allele Combinations. Frontiers in Genetics, 2020, 11, 557147.	2.3	2
13	Development and validation in 500 female samples of a TP-PCR assay to identify AFF2 GCC expansions. Scientific Reports, 2021, 11, 14676.	3.3	1
14	Usher syndrome and Nebulinâ€•associated myopathy in a single patient due to variants in MYO7A and NEB. Clinical Case Reports (discontinued), 2020, 8, 2476-2482.	0.5	0
15	Use of the FMR1 Gene Methylation Status to Assess the X-Chromosome Inactivation Pattern: A Stepwise Analysis. Genes, 2022, 13, 419.	2.4	0