

# Isabel Marques

## List of Publications by Year in descending order

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Version: 2024-02-01

15  
papers

291  
citations

1478505

6  
h-index

1199594

12  
g-index

15  
all docs

15  
docs citations

15  
times ranked

493  
citing authors

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