

Maria Monticelli

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

11
papers

237
citations

8
h-index

15
g-index

18
ext. papers

360
ext. citations

5.3
avg, IF

3.33
L-index

#	Paper	IF	Citations
11	Liver involvement in congenital disorders of glycosylation (CDG). A systematic review of the literature. <i>Journal of Inherited Metabolic Disease</i> , 2017 , 40, 195-207	5.4	59
10	Immunological aspects of congenital disorders of glycosylation (CDG): a review. <i>Journal of Inherited Metabolic Disease</i> , 2016 , 39, 765-780	5.4	37
9	Looking for protein stabilizing drugs with thermal shift assay. <i>Drug Testing and Analysis</i> , 2015 , 7, 831-4	3.5	34
8	Pharmacological Chaperones: A Therapeutic Approach for Diseases Caused by Destabilizing Missense Mutations. <i>International Journal of Molecular Sciences</i> , 2020 , 21,	6.3	23
7	The Analysis of Variants in the General Population Reveals That Is Extremely Tolerant to Missense Mutations and That Diagnosis of PMM2-CDG Can Benefit from the Identification of Modifiers. <i>International Journal of Molecular Sciences</i> , 2018 , 19,	6.3	19
6	Loss-of-function maternal-effect mutations of PADI6 are associated with familial and sporadic Beckwith-Wiedemann syndrome with multi-locus imprinting disturbance. <i>Clinical Epigenetics</i> , 2020 , 12, 139	7.7	19
5	β-Glucose-1,6-Bisphosphate Stabilizes Pathological Phosphomannomutase2 Mutants In Vitro and Represents a Lead Compound to Develop Pharmacological Chaperones for the Most Common Disorder of Glycosylation, PMM2-CDG. <i>International Journal of Molecular Sciences</i> , 2019 , 20,	6.3	17
4	Protective Role of a Variant on Severe COVID-19 Outcome in Young Males and Elderly Women. <i>Genes</i> , 2021 , 12,	4.2	14
3	Why does SARS-CoV-2 hit in different ways? Host genetic factors can influence the acquisition or the course of COVID-19. <i>European Journal of Medical Genetics</i> , 2021 , 64, 104227	2.6	8
2	E-Learning for Rare Diseases: An Example Using Fabry Disease. <i>International Journal of Molecular Sciences</i> , 2017 , 18,	6.3	2
1	Bioinformatics tools for marine biotechnology: a practical tutorial with a metagenomic approach. <i>BMC Bioinformatics</i> , 2020 , 21, 348	3.6	0