Maria Monticelli

List of Publications by Year in Descending Order

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	Protective Role of a Variant on Severe COVID-19 Outcome in Young Males and Elderly Women. *Genes*, **2021**, 12,	4.2	14
10	Why does SARS-CoV-2 hit in different ways? Host genetic factors can influence the acquisition or the course of COVID-19. *European Journal of Medical Genetics*, **2021**, 64, 104227	2.6	8
9	Pharmacological Chaperones: A Therapeutic Approach for Diseases Caused by Destabilizing Missense Mutations. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	23
8	Bioinformatics tools for marine biotechnology: a practical tutorial with a metagenomic approach. *BMC Bioinformatics*, **2020**, 21, 348	3.6	0
7	Loss-of-function maternal-effect mutations of PADI6 are associated with familial and sporadic Beckwith-Wiedemann syndrome with multi-locus imprinting disturbance. *Clinical Epigenetics*, **2020**, 12, 139	7.7	19
6	Glucose-1,6-Bisphosphate Stabilizes Pathological Phophomannomutase2 Mutants In Vitro and Represents a Lead Compound to Develop Pharmacological Chaperones for the Most Common Disorder of Glycosylation, PMM2-CDG. *International Journal of Molecular Sciences*, **2019**, 20,	6.3	17
5	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. *International Journal of Molecular Sciences*, **2018**, 19,	6.3	19
4	Liver involvement in congenital disorders of glycosylation (CDG). A systematic review of the literature. *Journal of Inherited Metabolic Disease*, **2017**, 40, 195-207	5.4	59
3	E-Learning for Rare Diseases: An Example Using Fabry Disease. *International Journal of Molecular Sciences*, **2017**, 18,	6.3	2
2	Immunological aspects of congenital disorders of glycosylation (CDG): a review. *Journal of Inherited Metabolic Disease*, **2016**, 39, 765-780	5.4	37
1	Looking for protein stabilizing drugs with thermal shift assay. *Drug Testing and Analysis*, **2015**, 7, 831-4	3.5	34