Margherita Baldassarri

List of Publications by Year
in descending order

32 papers	990 citations	516710 16 h-index	477307 29 g-index
42 all docs	42 docs citations	42 times ranked	2522 citing authors

#	Article	IF	Citations
1	Common, low-frequency, rare, and ultra-rare coding variants contribute to COVID-19 severity. Human Genetics, 2022, 141, 147-173.	3.8	22
2	The polymorphism L412F in TLR3 inhibits autophagy and is a marker of severe COVID-19 in males. Autophagy, 2022, 18, 1662-1672.	9.1	25
3	The 2019 and 2021 International Workshops on Alport Syndrome. European Journal of Human Genetics, 2022, 30, 507-516.	2.8	12
4	Rare variants in Toll-like receptor 7 results in functional impairment and downregulation of cytokine-mediated signaling in COVID-19 patients. Genes and Immunity, 2022, 23, 51-56.	4.1	41
5	Carriers of ADAMTS13 Rare Variants Are at High Risk of Life-Threatening COVID-19. Viruses, 2022, 14, 1185.	3.3	1
6	Pathogen-sugar interactions revealed by universal saturation transfer analysis. Science, 2022, 377, .	12.6	24
7	MET somatic activating mutations are responsible for lymphovenous malformation and can be identified using cell-free DNA next generation sequencing liquid biopsy. Journal of Vascular Surgery: Venous and Lymphatic Disorders, 2021, 9, 740-744.	1.6	7
8	Employing a systematic approach to biobanking and analyzing clinical and genetic data for advancing COVID-19 research. European Journal of Human Genetics, 2021, 29, 745-759.	2.8	35
9	Epilepsy in Nicolaidesâ€“Baraitser Syndrome: Review of Literature and Report of 25 Patients Focusing on Treatment Aspects. Neuropediatrics, 2021, 52, 109-122.	0.6	2
10	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males. EBioMedicine, 2021, 65, 103246.	6.1	52
11	Association of Toll-like receptor 7 variants with life-threatening COVID-19 disease in males: findings from a nested case-control study. ELife, 2021, 10, .	6.0	145
12	Protective Role of a TMPRSS2 Variant on Severe COVID-19 Outcome in Young Males and Elderly Women. Genes, 2021, 12, 596.	2.4	39
13	In response to the letter to the editor by Soha Ghanian etÂal. re our publication â€œShorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European malesâ€• EBioMedicine, 2021, 68, 103426.	6.1	0
14	Severe COVID-19 in Hospitalized Carriers of Single CFTR Pathogenic Variants. Journal of Personalized Medicine, 2021, 11, 558.	2.5	16
15	C9orf72 Intermediate Repeats Confer Genetic Risk for Severe COVID-19 Pneumonia Independently of Age. International Journal of Molecular Sciences, 2021, 22, 6991.	4.1	12
16	Exome Sequencing in 200 Intellectual Disability/Autistic Patients: New Candidates and Atypical Presentations. Brain Sciences, 2021, 11, 936.	2.3	17
17	SELP Asp603Asn and severe thrombosis in COVID-19 males. Journal of Hematology and Oncology, 2021, 14, 123.	17.0	11
18	13q Deletion Syndrome Involving RB1: Characterization of a New Minimal Critical Region for Psychomotor Delay. Genes, 2021, 12, 1318.	2.4	2

#	Article	IF	Citations
19	Identification of a Novel Pathogenic Variant in the NAGLU Gene in a Child with Neurodevelopmental Delay. Journal of Autism and Developmental Disorders, 2021, , 1.	2.7	0
20	Exome sequencing in BRCA1-2 candidate familias: the contribution of other cancer susceptibility genes. Frontiers in Oncology, 2021, 11, 649435.	2.8	2
21	New Candidates for Autism/Intellectual Disability Identified by Whole-Exome Sequencing. International Journal of Molecular Sciences, 2021, 22, 13439.	4.1	23
22	ACE2 gene variants may underlie interindividual variability and susceptibility to COVID-19 in the Italian population. European Journal of Human Genetics, 2020, 28, 1602-1614.	2.8	208
23	A novel mutation in LMX1B gene in a newborn with nailâ€patella syndrome: Clinical and dermoscopic findings. Pediatric Dermatology, 2020, 37, 1205-1206.	0.9	2
24	Clinical and molecular characterization of COVID-19 hospitalized patients. PLoS ONE, 2020, 15, e0242534.	2.5	25
25	Usefulness and Limitations of Comprehensive Characterization of mRNA Splicing Profiles in the Definition of the Clinical Relevance of BRCA1/2 Variants of Uncertain Significance. Cancers, 2019, 11, 295.	3.7	24
26	Urine-derived podocytes-lineage cells: A promising tool for precision medicine in Alport Syndrome. Human Mutation, 2018, 39, 302-314.	2.5	16
27	Omic Approach in Non-smoker Female with Lung Squamous Cell Carcinoma Pinpoints to Germline Susceptibility and Personalized Medicine. Cancer Research and Treatment, 2018, 50, 356-365.	3.0	20
28	Microduplication of the ARID1A gene causes intellectual disability with recognizable syndromic features. Genetics in Medicine, 2017, 19, 701-710.	2.4	13
29	Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes are a common well recognizable cause of intellectual disability. Brain and Development, 2015, 37, 527-536.	1.1	32
30	FANCMc.5791C>T nonsense mutation (rs144567652) induces exon skipping, affects DNA repair activity and is a familial breast cancer risk factor. Human Molecular Genetics, 2015, 24, 5345-5355.	2.9	91
31	Oligogenic germline mutations identified in early non-smokers lung adenocarcinoma patients. Lung Cancer, 2014, 85, 168-174.	2.0	30
32	Shorter Androgen Receptor PolyQ Alleles Protect Against Life-Threatening COVID-19 Disease in Males. SSRN Electronic Journal, 0, , .	0.4	2