Margherita Baldassarri

List of Publications by Year
in descending order

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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	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
2	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
3	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
4	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males. EBioMedicine, 2021, 65, 103246.	6.1	52
5	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
6	Protective Role of a TMPRSS2 Variant on Severe COVID-19 Outcome in Young Males and Elderly Women. Genes, 2021, 12, 596.	2.4	39
7	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
8	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
9	Oligogenic germline mutations identified in early non-smokers lung adenocarcinoma patients. Lung Cancer, 2014, 85, 168-174.	2.0	30
10	Clinical and molecular characterization of COVID-19 hospitalized patients. PLoS ONE, 2020, 15, e0242534.	2.5	25
11	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
12	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
13	Pathogen-sugar interactions revealed by universal saturation transfer analysis. Science, 2022, 377, .	12.6	24
14	New Candidates for Autism/Intellectual Disability Identified by Whole-Exome Sequencing. International Journal of Molecular Sciences, 2021, 22, 13439.	4.1	23
15	Common, low-frequency, rare, and ultra-rare coding variants contribute to COVID-19 severity. Human Genetics, 2022, 141, 147-173.	3.8	22
16	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
17	Exome Sequencing in 200 Intellectual Disability/Autistic Patients: New Candidates and Atypical Presentations. Brain Sciences, 2021, 11, 936.	2.3	17
18	Urine-derived podocytes-lineage cells: A promising tool for precision medicine in Alport Syndrome. Human Mutation, 2018, 39, 302-314.	2.5	16

#	Article	IF	Citations
19	Severe COVID-19 in Hospitalized Carriers of Single CFTR Pathogenic Variants. Journal of Personalized Medicine, 2021, 11, 558.	2.5	16
20	Microduplication of the ARID1A gene causes intellectual disability with recognizable syndromic features. Genetics in Medicine, 2017, 19, 701-710.	2.4	13
21	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
22	The 2019 and 2021 International Workshops on Alport Syndrome. European Journal of Human Genetics, 2022, 30, 507-516.	2.8	12
23	SELP Asp603Asn and severe thrombosis in COVID-19 males. Journal of Hematology and Oncology, 2021, 14, 123.	17.0	11
24	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
25	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
26	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
27	13q Deletion Syndrome Involving RB1: Characterization of a New Minimal Critical Region for Psychomotor Delay. Genes, 2021, 12, 1318.	2.4	2
28	Shorter Androgen Receptor PolyQ Alleles Protect Against Life-Threatening COVID-19 Disease in Males. SSRN Electronic Journal, 0, , .	0.4	2
29	Exome sequencing in BRCA1-2 candidate familias: the contribution of other cancer susceptibility genes. Frontiers in Oncology, 2021, 11, 649435.	2.8	2
30	Carriers of ADAMTS13 Rare Variants Are at High Risk of Life-Threatening COVID-19. Viruses, 2022, 14, 1185.	3.3	1
31	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
32	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