

Francesco Musacchia

List of Publications by Year in descending order

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Version: 2024-02-01

29
papers

727
citations

840776

11
h-index

580821

25
g-index

36
all docs

36
docs citations

36
times ranked

1874
citing authors

#	ARTICLE	IF	CITATIONS
1	CantÃ1 syndrome: Report of a patient with a novel variant in <scp> <i>KCNJ8</i> </scp> and revision of literature. American Journal of Medical Genetics, Part A, 2022, 188, 1661-1666.	1.2	5
2	Therapeutic homology-independent targeted integration in retina and liver. Nature Communications, 2022, 13, 1963.	12.8	14
3	Identification of LINE retrotransposons and long non-coding RNAs expressed in the octopus brain. BMC Biology, 2022, 20, 116.	3.8	6
4	Milder presentation of TELO2-related syndrome in two sisters homozygous for the p.Arg609His pathogenic variant. European Journal of Medical Genetics, 2021, 64, 104116.	1.3	5
5	Linked-Read Whole Genome Sequencing Solves a Double DMD Gene Rearrangement. Genes, 2021, 12, 133.	2.4	8
6	Exome reanalysis and proteomic profiling identified TRIP4 as a novel cause of cerebellar hypoplasia and spinal muscular atrophy (PCH1). European Journal of Human Genetics, 2021, 29, 1348-1353.	2.8	10
7	VarGenius-HZD Allows Accurate Detection of Rare Homozygous or Hemizygous Deletions in Targeted Sequencing Leveraging Breadth of Coverage. Genes, 2021, 12, 1979.	2.4	4
8	Clinical and Genetic Analysis of a European Cohort with Pericentral Retinitis Pigmentosa. International Journal of Molecular Sciences, 2020, 21, 86.	4.1	25
9	Parkinsonism, Intellectual Disability, and Catatonia in a Young Male With <i>MECP2</i> Variant. Movement Disorders Clinical Practice, 2020, 7, 118-119.	1.5	7
10	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
11	Sinus pericranii, skull defects, and structural brain anomalies in TRAF7 â€related disorder. Birth Defects Research, 2020, 112, 1085-1092.	1.5	5
12	Congenital posterior cervical spine malformation due to biallelic c.240â€4T>G <i>RIPPLY2</i> variant: A discrete entity. American Journal of Medical Genetics, Part A, 2020, 182, 1466-1472.	1.2	5
13	A draft genome sequence of the elusive giant squid, Architeuthis dux. GigaScience, 2020, 9, .	6.4	37
14	Intraspecific Diversity in the Cold Stress Response of Transposable Elements in the Diatom Leptocylindrus aporus. Genes, 2020, 11, 9.	2.4	16
15	Spectrum of Disease Severity in Patients With X-Linked Retinitis Pigmentosa Due to <i>RPGR</i> Mutations. , 2020, 61, 36.		17
16	Consolidating the Role of TDP2 Mutations in Recessive Spinocerebellar Ataxia Associated with Pediatric Onset Drug Resistant Epilepsy and Intellectual Disability (SCAR23). Cerebellum, 2019, 18, 972-975.	2.5	12
17	O.27Ultra-exome: a new tool to solve the unsolved NMD. Neuromuscular Disorders, 2019, 29, S123-S124.	0.6	0
18	Expanding the clinical and molecular spectrum of <i>PRMT7</i> mutations: 3 additional patients and review. Clinical Genetics, 2018, 93, 675-681.	2.0	28

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19	Novel CNS malformations and skeletal anomalies in a patient with Beaulieu-Boycott-Innes syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2835-2840.	1.2	9
20	VarGenius executes cohort-level DNA-seq variant calling and annotation and allows to manage the resulting data through a PostgreSQL database. BMC Bioinformatics, 2018, 19, 477.	2.6	23
21	Genomewide transcriptional reprogramming in the seagrass <i>Cymodocea nodosa</i> under experimental ocean acidification. Molecular Ecology, 2017, 26, 4241-4259.	3.9	27
22	Marine diatoms change their gene expression profile when exposed to microscale turbulence under nutrient replete conditions. Scientific Reports, 2017, 7, 3826.	3.3	27
23	De novo assembly of a transcriptome from the eggs and early embryos of <i>Astropecten aranciacus</i> . PLoS ONE, 2017, 12, e0184090.	2.5	9
24	Transcriptome sequencing of three <i>Pseudo-nitzschia</i> species reveals comparable gene sets and the presence of Nitric Oxide Synthase genes in diatoms. Scientific Reports, 2015, 5, 12329.	3.3	58
25	De novo assembly and sex-specific transcriptome profiling in the sand fly <i>Phlebotomus perniciosus</i> (Diptera, Phlebotominae), a major Old World vector of <i>Leishmania infantum</i> . BMC Genomics, 2015, 16, 847.	2.8	23
26	Annocript: a flexible pipeline for the annotation of transcriptomes able to identify putative long noncoding RNAs. Bioinformatics, 2015, 31, 2199-2201.	4.1	94
27	Solving Biclustering with a GRASP-Like Metaheuristic: Two Case-Studies on Gene Expression Analysis. Lecture Notes in Computer Science, 2012, , 253-267.	1.3	1
28	CONTENT-BASED IMAGE RETRIEVAL BY A FUZZY SCALE-SPACE APPROACH. International Journal of Pattern Recognition and Artificial Intelligence, 2006, 20, 849-867.	1.2	6
29	A Fuzzy Scale-Space Approach to Feature-Based Image Representation and Retrieval. Lecture Notes in Computer Science, 2005, , 377-385.	1.3	5