Gabriella Maria Squeo

List of Publications by Year
in descending order

19	424	759233 12	794594 19
papers	citations	h-index	g-index

21	21	21	922
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Clinical heterogeneity of Kabuki syndrome in a cohort of Italian patients and review of the literature. European Journal of Pediatrics, 2022, 181, 171-187.	2.7	13
2	DNA methylation episignature testing improves molecular diagnosis of Mendelian chromatinopathies. Genetics in Medicine, 2022, 24, 51-60.	2.4	24
3	Common, low-frequency, rare, and ultra-rare coding variants contribute to COVID-19 severity. Human Genetics, 2022, 141, 147-173.	3.8	22
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	Identification of a DNA Methylation Episignature in the 22q11.2 Deletion Syndrome. International Journal of Molecular Sciences, 2021, 22, 8611.	4.1	15
6	TRIM8 interacts with KIF11 and KIFC1 and controls bipolar spindle formation and chromosomal stability. Cancer Letters, 2020, 473, 98-106.	7.2	16
7	Loss of Function of the Gene Encoding the Histone Methyltransferase KMT2D Leads to Deregulation of Mitochondrial Respiration. Cells, 2020, 9, 1685.	4.1	10
8	Clinical Genetics Can Solve the Pitfalls of Genome-Wide Investigations: Lesson from Mismapping a Loss-of-Function Variant in KANSL1. Genes, 2020, 11, 1177.	2.4	3
9	DNA Methylation in the Diagnosis of Monogenic Diseases. Genes, 2020, 11, 355.	2.4	28
10	Customised next-generation sequencing multigene panel to screen a large cohort of individuals with chromatin-related disorder. Journal of Medical Genetics, 2020, 57, 760-768.	3.2	15
11	A restricted spectrum of missense KMT2D variants cause a multiple malformations disorder distinct fromKabuki syndrome. Genetics in Medicine, 2020, 22, 867-877.	2.4	41
12	A small 7q11.23 microduplication involving <scp>GTF2I</scp> in a family with intellectual disability. Clinical Genetics, 2020, 97, 940-942.	2.0	4
13	Generation of the induced human pluripotent stem cell lines CSSi009-A from a patient with a GNB5 pathogenic variant, and CSSi010-A from a CRISPR/Cas9 engineered GNB5 knock-out human cell line. Stem Cell Research, 2019, 40, 101547.	0.7	2
14	Schilbachâ€“Rott syndrome associated with 9q22.32q22.33 duplication, involving the PTCH1 gene. European Journal of Human Genetics, 2019, 27, 1260-1266.	2.8	1
15	MYC-containing amplicons in acute myeloid leukemia: genomic structures, evolution, and transcriptional consequences. Leukemia, 2018, 32, 2152-2166.	7.2	70
16	TRIM50 regulates Beclin 1 proautophagic activity. Biochimica Et Biophysica Acta - Molecular Cell Research, 2018, 1865, 908-919.	4.1	39
17	A NGS-Targeted Autism/ID Panel Reveals Compound Heterozygous GNB5 Variants in a Novel Patient. Frontiers in Genetics, 2018, 9, 626.	2.3	9
18	Dissecting KMT2D missense mutations in Kabuki syndrome patients. Human Molecular Genetics, 2018, 27, 3651-3668.	2.9	49

#	Article	IF	Citations
19	Clinical and Neurobehavioral Features of Three Novel Kabuki Syndrome Patients with Mosaic KMT2D Mutations and a Review of Literature. International Journal of Molecular Sciences, 2018, 19, 82.	4.1	19