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	MYC-containing amplicons in acute myeloid leukemia: genomic structures, evolution, and transcriptional consequences. Leukemia, 2018, 32, 2152-2166.	7.2	70
2	Dissecting KMT2D missense mutations in Kabuki syndrome patients. Human Molecular Genetics, 2018, 27, 3651-3668.	2.9	49
3	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
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	TRIM50 regulates Beclin 1 proautophagic activity. Biochimica Et Biophysica Acta - Molecular Cell Research, 2018, 1865, 908-919.	4.1	39
6	DNA Methylation in the Diagnosis of Monogenic Diseases. Genes, 2020, 11, 355.	2.4	28
7	DNA methylation episignature testing improves molecular diagnosis of Mendelian chromatinopathies. Genetics in Medicine, 2022, 24, 51-60.	2.4	24
8	Common, low-frequency, rare, and ultra-rare coding variants contribute to COVID-19 severity. Human Genetics, 2022, 141, 147-173.	3.8	22
9	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
10	TRIM8 interacts with KIF11 and KIFC1 and controls bipolar spindle formation and chromosomal stability. Cancer Letters, 2020, 473, 98-106.	7.2	16
11	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
12	Identification of a DNA Methylation Episignature in the 22q11.2 Deletion Syndrome. International Journal of Molecular Sciences, 2021, 22, 8611.	4.1	15
13	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
14	Loss of Function of the Gene Encoding the Histone Methyltransferase KMT2D Leads to Deregulation of Mitochondrial Respiration. Cells, 2020, 9, 1685.	4.1	10
15	A NGS-Targeted Autism/ID Panel Reveals Compound Heterozygous GNB5 Variants in a Novel Patient. Frontiers in Genetics, 2018, 9, 626.	2.3	9
16	A small 7q11.23 microduplication involving <scp>GTF2I</scp> in a family with intellectual disability. Clinical Genetics, 2020, 97, 940-942.	2.0	4
17	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
18	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

#	Article	IF	Citations
19	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