Carmela Ca Fusco

List of Publications by Year
in descending order

45	6,181	304743	243625
papers	citations	22 h-index	44 g-index

47	47	47	16540
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). Autophagy, 2016, 12, 1-222.	9.1	4,701
2	Copy number variants at Williamsâ€“Beuren syndrome 7q11.23 region. Human Genetics, 2010, 128, 3-26.	3.8	134
3	7q11.23 dosage-dependent dysregulation in human pluripotent stem cells affects transcriptional programs in disease-relevant lineages. Nature Genetics, 2015, 47, 132-141.	21.4	108
4	Molecular Analysis, Pathogenic Mechanisms, and Readthrough Therapy on a Large Cohort of<scp>K</scp>abuki Syndrome Patients. Human Mutation, 2014, 35, 841-850.	2.5	87
5	Endothelial cell clonal expansion in the development of cerebral cavernous malformations. Nature Communications, 2019, 10, 2761.	12.8	87
6	Mutation spectrum of MLL2 in a cohort of kabuki syndrome patients. Orphanet Journal of Rare Diseases, 2011, 6, 38.	2.7	79
7	TRIM8 modulates p53 activity to dictate cell cycle arrest. Cell Cycle, 2012, 11, 511-523.	2.6	78
8	The E3-Ubiquitin Ligase TRIM50 Interacts with HDAC6 and p62, and Promotes the Sequestration and Clearance of Ubiquitinated Proteins into the Aggresome. PLoS ONE, 2012, 7, e40440.	2.5	76
9	Autophagy induction in atrophic muscle cells requires ULK1 activation by TRIM32 through unanchored K63-linked polyubiquitin chains. Science Advances, 2019, 5, eaau8857.	10.3	74
10	Smaller and larger deletions of the Williams Beuren syndrome region implicate genes involved in mild facial phenotype, epilepsy and autistic traits. European Journal of Human Genetics, 2014, 22, 64-70.	2.8	63
11	An atypical 7q11.23 deletion in a normal IQ Williamsâ€“Beuren syndrome patient. European Journal of Human Genetics, 2010, 18, 33-38.	2.8	62
12	TRIM8 downregulation in glioma affects cell proliferation and it is associated with patients survival. BMC Cancer, 2015, 15, 470.	2.6	61
13	Identification and characterization of seven novel mutations of elastin gene in a cohort of patients affected by supravalvular aortic stenosis. European Journal of Human Genetics, 2010, 18, 317-323.	2.8	51
14	The Tripartite Motif. Advances in Experimental Medicine and Biology, 2012, , 11-25.	1.6	49
15	Williamsâ€“Beuren syndrome TRIM50 encodes an E3 ubiquitin ligase. European Journal of Human Genetics, 2008, 16, 1038-1049.	2.8	43
16	TRIM50 regulates Beclin 1 proautophagic activity. Biochimica Et Biophysica Acta - Molecular Cell Research, 2018, 1865, 908-919.	4.1	39
17	Propranolol for familial cerebral cavernous malformation (Treat_CCM): study protocol for a randomized controlled pilot trial. Trials, 2020, 21, 401.	1.6	37
18	Using Transcription Modules to Identify Expression Clusters Perturbed in Williams-Beuren Syndrome. PLoS Computational Biology, 2011, 7, e1001054.	3.2	36

#	Article	IF	Citations
19	DPP6 gene disruption in a family with Gilles de la Tourette syndrome. Neurogenetics, 2014, 15, 237-242.	1.4	25
20	The tripartite motif: structure and function. Advances in Experimental Medicine and Biology, 2012, 770, 11-25.	1.6	25
21	Absence of deletion and duplication of MLL2 and KDM6A genes in a large cohort of patients with Kabuki syndrome. Molecular Genetics and Metabolism, 2012, 107, 627-629.	1.1	23
22	TRIM8-driven transcriptomic profile of neural stem cells identified glioma-related nodal genes and pathways. Biochimica Et Biophysica Acta - General Subjects, 2019, 1863, 491-501.	2.4	22
23	A Fish-Specific Transposable Element Shapes the Repertoire of p53 Target Genes in Zebrafish. PLoS ONE, 2012, 7, e46642.	2.5	17
24	HDAC6 mediates the acetylation of TRIM50. Cellular Signalling, 2014, 26, 363-369.	3.6	17
25	Characterization of Two Novel Intronic Variants Affecting Splicing in FBN1-Related Disorders. Genes, 2019, 10, 442.	2.4	17
26	A single-center study on 140 patients with cerebral cavernous malformations: 28 new pathogenic variants and functional characterization of a PDCD10 large deletion. Human Mutation, 2018, 39, 1885-1900.	2.5	16
27	TRIM8 interacts with KIF11 and KIFC1 and controls bipolar spindle formation and chromosomal stability. Cancer Letters, 2020, 473, 98-106.	7.2	16
28	A 1.3-Mb 7q11.23 Atypical Deletion Identified in a Cohort of Patients with Williams-Beuren Syndrome. Molecular Syndromology, 2013, 4, 143-147.	0.8	12
29	Mutational spectrum and clinical signatures in 114 families with hereditary multiple osteochondromas: insights into molecular properties of selected exostosin variants. Human Molecular Genetics, 2019, 28, 2133-2142.	2.9	12
30	Exon-Trapping Assay Improves Clinical Interpretation of COL11A1 and COL11A2 Intronic Variants in Stickler Syndrome Type 2 and Otospondylomegaepiphyseal Dysplasia. Genes, 2020, 11, 1513.	2.4	11
31	Pro-Fibrotic Phenotype in a Patient with Segmental Stiff Skin Syndrome via TGF-Î² Signaling Overactivation. International Journal of Molecular Sciences, 2020, 21, 5141.	4.1	9
32	Insights into the molecular pathogenesis of cardiospondylocarpofacial syndrome: MAP3K7 c.737-7AÂ>ÂG variant alters the TGFÎ²-mediated Î±-SMA cytoskeleton assembly and autophagy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2020, 1866, 165742.	3.8	7
33	Transcriptome Analysis Reveals Altered Expression of Genes Involved in Hypoxia, Inflammation and Immune Regulation in Pdcd10-Depleted Mouse Endothelial Cells. Genes, 2022, 13, 961.	2.4	6
34	Report of the First Clinical Case of a Moroccan Kabuki Patient with a Novel <i>MLL2</i> Mutation. Molecular Syndromology, 2013, 4, 152-156.	0.8	5
35	Uncommon functional properties of the first piscine 26S proteasome from the Antarctic notothenioid Trematomus bernacchii. Bioscience Reports, 2016, 36, .	2.4	5
36	TAB2 c.1398dup variant leads to haploinsufficiency and impairs extracellular matrix homeostasis. Human Mutation, 2019, 40, 1886-1898.	2.5	5

#	Article	IF	Citations
37	Molecular diagnostic workflow, clinical interpretation of sequence variants, and data repository procedures in 140 individuals with familial cerebral cavernous malformations. Human Mutation, 2019, 40, e24-e36.	2.5	3
38	Novel TONSL variants cause SPONASTRIME dysplasia and associate with spontaneous chromosome breaks, defective cell proliferation and apoptosis. Human Molecular Genetics, 2020, 29, 3122-3131.	2.9	3
39	Review of clinical and molecular variability in autosomal recessive cutis laxa 2A. American Journal of Medical Genetics, Part A, 2021, 185, 955-965.	1.2	2
40	GPR143 Mutational Analysis in Two Italian Families with X-Linked Ocular Albinism. Genetic Testing and Molecular Biomarkers, 2009, 13, 527-531.	0.7	1
41	Improving clinical interpretation of five <scp>KRIT1</scp> and <scp>PDCD10</scp> intronic variants. Clinical Genetics, 2021, 99, 829-835.	2.0	1
42	Genomic and Genetic Disorders Biobank. Open Journal of Bioresources, 2015, 2, .	1.5	1
43	Loss-of-function variants in exon 4 of TAB2 causeÂaÂrecognizable multisystem disorder with cardiovascular, facial, cutaneous, and musculoskeletalÂinvolvement. Genetics in Medicine, 2021, , .	2.4	1
44	Unusual Antioxidant Properties of 26S Proteasome Isolated from Cold-Adapted Organisms. International Journal of Molecular Sciences, 2017, 18, 1605.	4.1	0
45	Response to: Concern regarding classification of c.703G>A/p.Gly235Arg as a novel missense variant in KRIT1 gene. Human Mutation, 2020, 41, 1072-1074.	2.5	0