

# Carmela Ca Fusco

## List of Publications by Year in descending order

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45  
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#	ARTICLE	IF	CITATIONS
1	Transcriptome Analysis Reveals Altered Expression of Genes Involved in Hypoxia, Inflammation and Immune Regulation in Pcd10-Depleted Mouse Endothelial Cells. <i>Genes</i> , 2022, 13, 961.	2.4	6
2	Improving clinical interpretation of five <i>KRIT1</i> and <i>PDCD10</i> intronic variants. <i>Clinical Genetics</i> , 2021, 99, 829-835.	2.0	1
3	Review of clinical and molecular variability in autosomal recessive cutis laxa 2A. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 955-965.	1.2	2
4	Loss-of-function variants in exon 4 of <i>TAB2</i> cause a recognizable multisystem disorder with cardiovascular, facial, cutaneous, and musculoskeletal involvement. <i>Genetics in Medicine</i> , 2021, , .	2.4	1
5	<i>TRIM8</i> interacts with <i>KIF11</i> and <i>KIFC1</i> and controls bipolar spindle formation and chromosomal stability. <i>Cancer Letters</i> , 2020, 473, 98-106.	7.2	16
6	Novel <i>TONSL</i> variants cause <i>SPONASTRIME</i> dysplasia and associate with spontaneous chromosome breaks, defective cell proliferation and apoptosis. <i>Human Molecular Genetics</i> , 2020, 29, 3122-3131.	2.9	3
7	Pro-Fibrotic Phenotype in a Patient with Segmental Stiff Skin Syndrome via $TGF-\beta$ Signaling Overactivation. <i>International Journal of Molecular Sciences</i> , 2020, 21, 5141.	4.1	9
8	Exon-Trapping Assay Improves Clinical Interpretation of <i>COL11A1</i> and <i>COL11A2</i> Intronic Variants in Stickler Syndrome Type 2 and Otospondylomegalepiphyseal Dysplasia. <i>Genes</i> , 2020, 11, 1513.	2.4	11
9	Propranolol for familial cerebral cavernous malformation (Treat_CCM): study protocol for a randomized controlled pilot trial. <i>Trials</i> , 2020, 21, 401.	1.6	37
10	Insights into the molecular pathogenesis of cardio-spondylocarpofacial syndrome: <i>MAP3K7</i> c.737-7A>G variant alters the $TGF-\beta$ -mediated $\beta$ -SMA cytoskeleton assembly and autophagy. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2020, 1866, 165742.	3.8	7
11	Response to: Concern regarding classification of c.703G>A/p.Gly235Arg as a novel missense variant in <i>KRIT1</i> gene. <i>Human Mutation</i> , 2020, 41, 1072-1074.	2.5	0
12	<i>TAB2</i> c.1398dup variant leads to haploinsufficiency and impairs extracellular matrix homeostasis. <i>Human Mutation</i> , 2019, 40, 1886-1898.	2.5	5
13	Molecular diagnostic workflow, clinical interpretation of sequence variants, and data repository procedures in 140 individuals with familial cerebral cavernous malformations. <i>Human Mutation</i> , 2019, 40, e24-e36.	2.5	3
14	Endothelial cell clonal expansion in the development of cerebral cavernous malformations. <i>Nature Communications</i> , 2019, 10, 2761.	12.8	87
15	Autophagy induction in atrophic muscle cells requires <i>ULK1</i> activation by <i>TRIM32</i> through unanchored K63-linked polyubiquitin chains. <i>Science Advances</i> , 2019, 5, eaau8857.	10.3	74
16	Characterization of Two Novel Intronic Variants Affecting Splicing in <i>FBN1</i> -Related Disorders. <i>Genes</i> , 2019, 10, 442.	2.4	17
17	Mutational spectrum and clinical signatures in 114 families with hereditary multiple osteochondromas: insights into molecular properties of selected exostosin variants. <i>Human Molecular Genetics</i> , 2019, 28, 2133-2142.	2.9	12
18	<i>TRIM8</i> -driven transcriptomic profile of neural stem cells identified glioma-related nodal genes and pathways. <i>Biochimica Et Biophysica Acta - General Subjects</i> , 2019, 1863, 491-501.	2.4	22

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19	TRIM50 regulates Beclin 1 proautophagic activity. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , 2018, 1865, 908-919.	4.1	39
20	A single-center study on 140 patients with cerebral cavernous malformations: 28 new pathogenic variants and functional characterization of a <i>PDCD10</i> large deletion. <i>Human Mutation</i> , 2018, 39, 1885-1900.	2.5	16
21	Unusual Antioxidant Properties of 26S Proteasome Isolated from Cold-Adapted Organisms. <i>International Journal of Molecular Sciences</i> , 2017, 18, 1605.	4.1	0
22	Uncommon functional properties of the first piscine 26S proteasome from the Antarctic nototheniid <i>Trematomus bernacchii</i> . <i>Bioscience Reports</i> , 2016, 36, .	2.4	5
23	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). <i>Autophagy</i> , 2016, 12, 1-222.	9.1	4,701
24	TRIM8 downregulation in glioma affects cell proliferation and it is associated with patients survival. <i>BMC Cancer</i> , 2015, 15, 470.	2.6	61
25	7q11.23 dosage-dependent dysregulation in human pluripotent stem cells affects transcriptional programs in disease-relevant lineages. <i>Nature Genetics</i> , 2015, 47, 132-141.	21.4	108
26	Genomic and Genetic Disorders Biobank. <i>Open Journal of Bioresources</i> , 2015, 2, .	1.5	1
27	Smaller and larger deletions of the Williams Beuren syndrome region implicate genes involved in mild facial phenotype, epilepsy and autistic traits. <i>European Journal of Human Genetics</i> , 2014, 22, 64-70.	2.8	63
28	DPP6 gene disruption in a family with Gilles de la Tourette syndrome. <i>Neurogenetics</i> , 2014, 15, 237-242.	1.4	25
29	Molecular Analysis, Pathogenic Mechanisms, and Readthrough Therapy on a Large Cohort of <i>Kanabuki Syndrome</i> Patients. <i>Human Mutation</i> , 2014, 35, 841-850.	2.5	87
30	HDAC6 mediates the acetylation of TRIM50. <i>Cellular Signalling</i> , 2014, 26, 363-369.	3.6	17
31	Report of the First Clinical Case of a Moroccan Kabuki Patient with a Novel <i>MLL2</i> Mutation. <i>Molecular Syndromology</i> , 2013, 4, 152-156.	0.8	5
32	A 1.3-Mb 7q11.23 Atypical Deletion Identified in a Cohort of Patients with Williams-Beuren Syndrome. <i>Molecular Syndromology</i> , 2013, 4, 143-147.	0.8	12
33	Absence of deletion and duplication of <i>MLL2</i> and <i>KDM6A</i> genes in a large cohort of patients with Kabuki syndrome. <i>Molecular Genetics and Metabolism</i> , 2012, 107, 627-629.	1.1	23
34	The E3-Ubiquitin Ligase TRIM50 Interacts with HDAC6 and p62, and Promotes the Sequestration and Clearance of Ubiquitinated Proteins into the Aggresome. <i>PLoS ONE</i> , 2012, 7, e40440.	2.5	76
35	A Fish-Specific Transposable Element Shapes the Repertoire of p53 Target Genes in Zebrafish. <i>PLoS ONE</i> , 2012, 7, e46642.	2.5	17
36	TRIM8 modulates p53 activity to dictate cell cycle arrest. <i>Cell Cycle</i> , 2012, 11, 511-523.	2.6	78

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37	The Tripartite Motif. <i>Advances in Experimental Medicine and Biology</i> , 2012, , 11-25.	1.6	49
38	The tripartite motif: structure and function. <i>Advances in Experimental Medicine and Biology</i> , 2012, 770, 11-25.	1.6	25
39	Mutation spectrum of MLL2 in a cohort of kabuki syndrome patients. <i>Orphanet Journal of Rare Diseases</i> , 2011, 6, 38.	2.7	79
40	Using Transcription Modules to Identify Expression Clusters Perturbed in Williams-Beuren Syndrome. <i>PLoS Computational Biology</i> , 2011, 7, e1001054.	3.2	36
41	Copy number variants at Williams-Beuren syndrome 7q11.23 region. <i>Human Genetics</i> , 2010, 128, 3-26.	3.8	134
42	An atypical 7q11.23 deletion in a normal IQ Williams-Beuren syndrome patient. <i>European Journal of Human Genetics</i> , 2010, 18, 33-38.	2.8	62
43	Identification and characterization of seven novel mutations of elastin gene in a cohort of patients affected by supraaortic stenosis. <i>European Journal of Human Genetics</i> , 2010, 18, 317-323.	2.8	51
44	<i>GPR143</i> Mutational Analysis in Two Italian Families with X-Linked Ocular Albinism. <i>Genetic Testing and Molecular Biomarkers</i> , 2009, 13, 527-531.	0.7	1
45	Williams-Beuren syndrome TRIM50 encodes an E3 ubiquitin ligase. <i>European Journal of Human Genetics</i> , 2008, 16, 1038-1049.	2.8	43