

# Antonio Musio

## List of Publications by Year in descending order

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Version: 2024-02-01

56  
papers

2,820  
citations

249298

26  
h-index

206121

51  
g-index

61  
all docs

61  
docs citations

61  
times ranked

3928  
citing authors

#	ARTICLE	IF	CITATIONS
1	Disease-associated <i>c-MYC</i> downregulation in human disorders of transcriptional regulation. <i>Human Molecular Genetics</i> , 2022, 31, 1599-1609.	1.4	5
2	The multifaceted roles of cohesin in cancer. <i>Journal of Experimental and Clinical Cancer Research</i> , 2022, 41, 96.	3.5	11
3	Pathogenic variants in <i>EP300</i> and <i>ANKRD11</i> in patients with phenotypes overlapping Cornelia de Lange syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1690-1696.	0.7	34
4	Chromosome Missegregation in Single Human Oocytes Is Related to the Age and Gene Expression Profile. <i>International Journal of Molecular Sciences</i> , 2020, 21, 1934.	1.8	12
5	Evaluating Face2Gene as a Tool to Identify Cornelia de Lange Syndrome by Facial Phenotypes. <i>International Journal of Molecular Sciences</i> , 2020, 21, 1042.	1.8	40
6	Cornelia de Lange syndrome: from molecular diagnosis to therapeutic approach. <i>Journal of Medical Genetics</i> , 2020, 57, 289-295.	1.5	45
7	The multiple facets of the <i>SMC1A</i> gene. <i>Gene</i> , 2020, 743, 144612.	1.0	21
8	Cohesin mutations are synthetic lethal with stimulation of WNT signaling. <i>ELife</i> , 2020, 9, .	2.8	22
9	p53 mitotic centrosome localization preserves centrosome integrity and works as sensor for the mitotic surveillance pathway. <i>Cell Death and Disease</i> , 2019, 10, 850.	2.7	26
10	Overexpression of the cohesin-core subunit <i>SMC1A</i> contributes to colorectal cancer development. <i>Journal of Experimental and Clinical Cancer Research</i> , 2019, 38, 108.	3.5	34
11	Separase prevents genomic instability by controlling replication fork speed. <i>Nucleic Acids Research</i> , 2018, 46, 267-278.	6.5	48
12	Antioxidant treatment ameliorates phenotypic features of <i>SMC1A</i> -mutated Cornelia de Lange syndrome in vitro and in vivo. <i>Human Molecular Genetics</i> , 2018, 27, 3002-3011.	1.4	24
13	A role for Separase in telomere protection. <i>Nature Communications</i> , 2016, 7, 10405.	5.8	20
14	Genome stability: What we have learned from cohesinopathies. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2016, 172, 171-178.	0.7	30
15	Cohesins. , 2016, , 1113-1116.		0
16	Mutant cohesin affects RNA polymerase II regulation in Cornelia de Lange syndrome. <i>Scientific Reports</i> , 2015, 5, 16803.	1.6	35
17	<i>SMC1B</i> is present in mammalian somatic cells and interacts with mitotic cohesin proteins. <i>Scientific Reports</i> , 2015, 5, 18472.	1.6	24
18	Clinical utility gene card for: Cornelia de Lange syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 1431-1431.	1.4	37

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19	Proliferation of Multiple Cell Types in the Skeletal Muscle Tissue Elicited by Acute p21 Suppression. <i>Molecular Therapy</i> , 2015, 23, 885-895.	3.7	6
20	AKTIP/Ft1, a New Shelterin-Interacting Factor Required for Telomere Maintenance. <i>PLoS Genetics</i> , 2015, 11, e1005167.	1.5	38
21	CEP57 mutation in a girl with mosaic variegated aneuploidy syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 177-181.	0.7	17
22	Mutant cohesin drives chromosomal instability in early colorectal adenomas. <i>Human Molecular Genetics</i> , 2014, 23, 6773-6778.	1.4	30
23	Loss-of-function HDAC8 mutations cause a phenotypic spectrum of Cornelia de Lange syndrome-like features, ocular hypertelorism, large fontanelle and X-linked inheritance. <i>Human Molecular Genetics</i> , 2014, 23, 2888-2900.	1.4	120
24	Early senescence in heterozygous ABCA1 mutation skin fibroblasts: A gene dosage effect beyond HDL deficiency?. <i>Biochemical and Biophysical Research Communications</i> , 2014, 447, 231-236.	1.0	3
25	Mutation Spectrum and Genotype-Phenotype Correlation in Cornelia de Lange Syndrome. <i>Human Mutation</i> , 2013, 34, 1589-1596.	1.1	152
26	Cytogenetic analysis of human cells reveals specific patterns of DNA damage in replicative and oncogene-induced senescence. <i>Aging Cell</i> , 2013, 12, 312-315.	3.0	8
27	Proteomic Profile Identifies Dysregulated Pathways in Cornelia de Lange Syndrome Cells with Distinct Mutations in <i>SMC1A</i> and <i>SMC3</i> Genes. <i>Journal of Proteome Research</i> , 2012, 11, 6111-6123.	1.8	41
28	Claspin as a biomarker of human papillomavirus-related high grade lesions of uterine cervix. <i>Journal of Translational Medicine</i> , 2012, 10, 132.	1.8	18
29	The Coffin-Siris syndrome: A proposed diagnostic approach and assessment of 15 overlapping cases. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 1865-1876.	0.7	69
30	<i>SMC1A</i> codon 496 mutations affect the cellular response to genotoxic treatments. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 224-228.	0.7	19
31	The dark side of cohesin: The carcinogenic point of view. <i>Mutation Research - Reviews in Mutation Research</i> , 2011, 728, 81-87.	2.4	28
32	Spectrum and consequences of <i>SMC1A</i> mutations: The unexpected involvement of a core component of cohesin in human disease. <i>Human Mutation</i> , 2010, 31, 5-10.	1.1	100
33	The expanding universe of cohesin functions: a new genome stability caretaker involved in human disease and cancer. <i>Human Mutation</i> , 2010, 31, 623-630.	1.1	51
34	A conserved role for the mitochondrial citrate transporter Sea/SLC25A1 in the maintenance of chromosome integrity. <i>Human Molecular Genetics</i> , 2009, 18, 4180-4188.	1.4	58
35	Claspin inhibition leads to fragile site expression. <i>Genes Chromosomes and Cancer</i> , 2009, 48, 1083-1090.	1.5	26
36	Cornelia de Lange syndrome mutations in <i>SMC1A</i> or <i>SMC3</i> affect binding to DNA. <i>Human Molecular Genetics</i> , 2009, 18, 418-427.	1.4	92

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37	Mutations in Cohesin Complex Members SMC3 and SMC1A Cause a Mild Variant of Cornelia de Lange Syndrome with Predominant Mental Retardation. American Journal of Human Genetics, 2007, 80, 485-494.	2.6	445
38	Physiologic Oxygen Enhances Human Embryonic Stem Cell Clonal Recovery and Reduces Chromosomal Abnormalities. Cloning and Stem Cells, 2006, 8, 16-23.	2.6	181
39	X-linked Cornelia de Lange syndrome owing to SMC1L1 mutations. Nature Genetics, 2006, 38, 528-530.	9.4	393
40	SMC1 inhibition results in FRA3B expression but has no effect on its delayed replication. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2006, 595, 23-28.	0.4	4
41	Damaging-agent sensitivity of Artemis-deficient cell lines. European Journal of Immunology, 2005, 35, 1250-1256.	1.6	30
42	SMC1 involvement in fragile site expression. Human Molecular Genetics, 2005, 14, 525-533.	1.4	86
43	Rescue of ATPa3-deficient murine malignant osteopetrosis by hematopoietic stem cell transplantation in utero. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 14629-14634.	3.3	58
44	Recapitulation of the Roberts syndrome cellular phenotype by inhibition of INCENP, ZWINT-1 and ZW10 genes. Gene, 2004, 331, 33-40.	1.0	19
45	Inhibition of BUB1 results in genomic instability and anchorage-independent growth of normal human fibroblasts. Cancer Research, 2003, 63, 2855-63.	0.4	47
46	Heterogeneous gene distribution reflects human genome complexity as detected at the cytogenetic level. Cancer Genetics and Cytogenetics, 2002, 134, 168-171.	1.0	25
47	Chromosomes, genes, and cancer breakpoints. Cancer Genetics and Cytogenetics, 2002, 139, 141-142.	1.0	4
48	Common fragile sites on human chromosomes represent transcriptionally active regions: evidence from camptothecin. Human Genetics, 1998, 102, 409-414.	1.8	17
49	Primed in situ labeling (PRINS): a method for rapid identification and quantification of human chromosomes in both lymphocytes and sperm nuclei. Genome, 1998, 41, 739-741.	0.9	2
50	Atomic force microscope imaging of chromosome structure during G-banding treatments. Genome, 1997, 40, 127-131.	0.9	31
51	Cycling-PRINS. Mutation Research - Genetic Toxicology and Environmental Mutagenesis, 1997, 390, 1-4.	0.9	9
52	Aphidicolin-sensitive specific common fragile sites: A biomarker of exposure to pesticides. , 1997, 29, 250-255.		21
53	Common and rare fragile sites on human chromosomes. Cancer Genetics and Cytogenetics, 1996, 88, 184-185.	1.0	8
54	Enhanced expression of common fragile site with occupational exposure to pesticides. Cancer Genetics and Cytogenetics, 1995, 82, 123-127.	1.0	28

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55	Longitudinal patterns similar to G-banding in untreated human chromosomes: evidence from atomic force microscopy. <i>Chromosoma</i> , 1994, 103, 225-229.	1.0	33
56	Specific chromosomal aberrations correlated to transformation in Chinese hamster cells. <i>Cancer Genetics and Cytogenetics</i> , 1992, 62, 81-87.	1.0	12