Antonio Musio

List of Publications by Year
in descending order

56 papers	2,820 citations	218662 26 h-index	182417 51 g-index
61 all docs	61 docs citations	61 times ranked	3609 citing authors

#	Article	IF	Citations
1	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.	6.2	445
2	X-linked Cornelia de Lange syndrome owing to SMC1L1 mutations. Nature Genetics, 2006, 38, 528-530.	21.4	393
3	Physiologic Oxygen Enhances Human Embryonic Stem Cell Clonal Recovery and Reduces Chromosomal Abnormalities. Cloning and Stem Cells, 2006, 8, 16-23.	2.6	181
4	Mutation Spectrum and Genotype-Phenotype Correlation in Cornelia de Lange Syndrome. Human Mutation, 2013, 34, 1589-1596.	2.5	152
5	Loss-of-function HDAC8 mutations cause a phenotypic spectrum of Cornelia de Lange syndrome-like features, ocular hypertelorism, large fontanelle and X-linked inheritance. Human Molecular Genetics, 2014, 23, 2888-2900.	2.9	120
6	Spectrum and consequences ofSMC1Amutations: The unexpected involvement of a core component of cohesin in human disease. Human Mutation, 2010, 31, 5-10.	2.5	100
7	Cornelia de Lange syndrome mutations in SMC1A or SMC3 affect binding to DNA. Human Molecular Genetics, 2009, 18, 418-427.	2.9	92
8	SMC1 involvement in fragile site expression. Human Molecular Genetics, 2005, 14, 525-533.	2.9	86
9	The Coffinâ€“Siris syndrome: A proposed diagnostic approach and assessment of 15 overlapping cases. American Journal of Medical Genetics, Part A, 2012, 158A, 1865-1876.	1.2	69
10	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.	7.1	58
11	A conserved role for the mitochondrial citrate transporter Sea/SLC25A1 in the maintenance of chromosome integrity. Human Molecular Genetics, 2009, 18, 4180-4188.	2.9	58
12	The expanding universe of cohesin functions: a new genome stability caretaker involved in human disease and cancer. Human Mutation, 2010, 31, 623-630.	2.5	51
13	Separase prevents genomic instability by controlling replication fork speed. Nucleic Acids Research, 2018, 46, 267-278.	14.5	48
14	Inhibition of BUB1 results in genomic instability and anchorage-independent growth of normal human fibroblasts. Cancer Research, 2003, 63, 2855-63.	0.9	47
15	Cornelia de Lange syndrome: from molecular diagnosis to therapeutic approach. Journal of Medical Genetics, 2020, 57, 289-295.	3.2	45
16	Proteomic Profile Identifies Dysregulated Pathways in Cornelia de Lange Syndrome Cells with Distinct Mutations in SMC1A and SMC3 Genes. Journal of Proteome Research, 2012, 11, 6111-6123.	3.7	41
17	Evaluating Face2Gene as a Tool to Identify Cornelia de Lange Syndrome by Facial Phenotypes. International Journal of Molecular Sciences, 2020, 21, 1042.	4.1	40
18	AKTIP/Ft1, a New Shelterin-Interacting Factor Required for Telomere Maintenance. PLoS Genetics, 2015, 11, e1005167.	3.5	38

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19	Clinical utility gene card for: Cornelia de Lange syndrome. European Journal of Human Genetics, 2015, 23, 1431-1431.	2.8	37
20	Mutant cohesin affects RNA polymerase II regulation in Cornelia de Lange syndrome. Scientific Reports, 2015, 5, 16803.	3.3	35
21	Overexpression of the cohesin-core subunit SMC1A contributes to colorectal cancer development. Journal of Experimental and Clinical Cancer Research, 2019, 38, 108.	8.6	34
22	Pathogenic variants in <scp>EP300</scp> and <scp>ANKRD11</scp> in patients with phenotypes overlapping Cornelia de Lange syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1690-1696.	1.2	34
23	Longitudinal patterns similar to G-banding in untreated human chromosomes: evidence from atomic force microscopy. Chromosoma, 1994, 103, 225-229.	2.2	33
24	Atomic force microscope imaging of chromosome structure during G-banding treatments. Genome, 1997, 40, 127-131.	2.0	31
25	Damaging-agent sensitivity of Artemis-deficient cell lines. European Journal of Immunology, 2005, 35, 1250-1256.	2.9	30
26	Mutant cohesin drives chromosomal instability in early colorectal adenomas. Human Molecular Genetics, 2014, 23, 6773-6778.	2.9	30
27	Genome stability: What we have learned from cohesinopathies. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2016, 172, 171-178.	1.6	30
28	Enhanced expression of common fragile site with occupational exposure to pesticides. Cancer Genetics and Cytogenetics, 1995, 82, 123-127.	1.0	28
29	The dark side of cohesin: The carcinogenic point of view. Mutation Research - Reviews in Mutation Research, 2011, 728, 81-87.	5.5	28
30	Claspin inhibition leads to fragile site expression. Genes Chromosomes and Cancer, 2009, 48, 1083-1090.	2.8	26
31	p53 mitotic centrosome localization preserves centrosome integrity and works as sensor for the mitotic surveillance pathway. Cell Death and Disease, 2019, 10, 850.	6.3	26
32	Heterogeneous gene distribution reflects human genome complexity as detected at the cytogenetic level. Cancer Genetics and Cytogenetics, 2002, 134, 168-171.	1.0	25
33	SMC1B is present in mammalian somatic cells and interacts with mitotic cohesin proteins. Scientific Reports, 2015, 5, 18472.	3.3	24
34	Antioxidant treatment ameliorates phenotypic features of SMC1A-mutated Cornelia de Lange syndrome in vitro and in vivo. Human Molecular Genetics, 2018, 27, 3002-3011.	2.9	24
35	Cohesin mutations are synthetic lethal with stimulation of WNT signaling. ELife, 2020, 9, .	6.0	22
36	Aphidicolin-sensitive specific common fragile sites: A biomarker of exposure to pesticides. , 1997, 29, 250-255.		21

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37	The multiple facets of the SMC1A gene. Gene, 2020, 743, 144612.	2.2	21
38	A role for Separase in telomere protection. Nature Communications, 2016, 7, 10405.	12.8	20
39	Recapitulation of the Roberts syndrome cellular phenotype by inhibition of INCENP, ZWINT-1 and ZW10 genes. Gene, 2004, 331, 33-40.	2.2	19
40	SMC1A codon 496 mutations affect the cellular response to genotoxic treatments. American Journal of Medical Genetics, Part A, 2012, 158A, 224-228.	1.2	19
41	Claspin as a biomarker of human papillomavirus-related high grade lesions of uterine cervix. Journal of Translational Medicine, 2012, 10, 132.	4.4	18
42	Common fragile sites on human chromosomes represent transcriptionally active regions: evidence from camptothecin. Human Genetics, 1998, 102, 409-414.	3.8	17
43	CEP57 mutation in a girl with mosaic variegated aneuploidy syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 177-181.	1.2	17
44	Specific chromosomal aberrations correlated to transformation in Chinese hamster cells. Cancer Genetics and Cytogenetics, 1992, 62, 81-87.	1.0	12
45	Chromosome Missegregation in Single Human Oocytes Is Related to the Age and Gene Expression Profile. International Journal of Molecular Sciences, 2020, 21, 1934.	4.1	12
46	The multifaceted roles of cohesin in cancer. Journal of Experimental and Clinical Cancer Research, 2022, 41, 96.	8.6	11
47	Cycling-PRINS. Mutation Research - Genetic Toxicology and Environmental Mutagenesis, 1997, 390, 1-4.	1.7	9
48	Common and rare fragile sites on human chromosomes. Cancer Genetics and Cytogenetics, 1996, 88, 184-185.	1.0	8
49	Cytogenetic analysis of human cells reveals specific patterns of <scp>DNA</scp> damage in replicative and oncogeneâ€induced senescence. Aging Cell, 2013, 12, 312-315.	6.7	8
50	Proliferation of Multiple Cell Types in the Skeletal Muscle Tissue Elicited by Acute p21 Suppression. Molecular Therapy, 2015, 23, 885-895.	8.2	6
51	Disease-associated c-MYC downregulation in human disorders of transcriptional regulation. Human Molecular Genetics, 2022, 31, 1599-1609.	2.9	5
52	Chromosomes, genes, and cancer breakpoints. Cancer Genetics and Cytogenetics, 2002, 139, 141-142.	1.0	4
53	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.	1.0	4
54	Early senescence in heterozygous ABCA1 mutation skin fibroblasts: A gene dosage effect beyond HDL deficiency?. Biochemical and Biophysical Research Communications, 2014, 447, 231-236.	2.1	3

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55	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.	2.0	2
56	Cohesins. , 2016, , 1113-1116.		0