# Antonio Musio

List of Publications by Year
in descending order



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|---|---|---|---|
| 56 papers | 2,820 citations | 218592 26 h-index | 182361 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. | 2.6 | 445 |
| 2 | X-linked Cornelia de Lange syndrome owing to SMC1L1 mutations. Nature Genetics, 2006, 38, 528-530. | 9.4 | 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. | 1.1 | 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. | 1.4 | 120 |
| 6 | Spectrum and consequences ofSMC1Amutations: The unexpected involvement of a core component of cohesin in human disease. Human Mutation, 2010, 31, 5-10. | 1.1 | 100 |
| 7 | Cornelia de Lange syndrome mutations in SMC1A or SMC3 affect binding to DNA. Human Molecular Genetics, 2009, 18, 418-427. | 1.4 | 92 |
| 8 | SMC1 involvement in fragile site expression. Human Molecular Genetics, 2005, 14, 525-533. | 1.4 | 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. | 0.7 | 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. | 3.3 | 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. | 1.4 | 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. | 1.1 | 51 |
| 13 | Separase prevents genomic instability by controlling replication fork speed. Nucleic Acids Research, 2018, 46, 267-278. | 6.5 | 48 |
| 14 | Inhibition of BUB1 results in genomic instability and anchorage-independent growth of normal human fibroblasts. Cancer Research, 2003, 63, 2855-63. | 0.4 | 47 |
| 15 | Cornelia de Lange syndrome: from molecular diagnosis to therapeutic approach. Journal of Medical Genetics, 2020, 57, 289-295. | 1.5 | 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. | 1.8 | 41 |
| 17 | Evaluating Face2Gene as a Tool to Identify Cornelia de Lange Syndrome by Facial Phenotypes. International Journal of Molecular Sciences, 2020, 21, 1042. | 1.8 | 40 |
| 18 | AKTIP/Ft1, a New Shelterin-Interacting Factor Required for Telomere Maintenance. PLoS Genetics, 2015, 11, e1005167. | 1.5 | 38 |

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|---|---|---|---|
| 19 | Clinical utility gene card for: Cornelia de Lange syndrome. European Journal of Human Genetics, 2015, 23, 1431-1431. | 1.4 | 37 |
| 20 | Mutant cohesin affects RNA polymerase II regulation in Cornelia de Lange syndrome. Scientific Reports, 2015, 5, 16803. | 1.6 | 35 |
| 21 | Overexpression of the cohesin-core subunit SMC1A contributes to colorectal cancer development. Journal of Experimental and Clinical Cancer Research, 2019, 38, 108. | 3.5 | 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. | 0.7 | 34 |
| 23 | Longitudinal patterns similar to G-banding in untreated human chromosomes: evidence from atomic force microscopy. Chromosoma, 1994, 103, 225-229. | 1.0 | 33 |
| 24 | Atomic force microscope imaging of chromosome structure during G-banding treatments. Genome, 1997, 40, 127-131. | 0.9 | 31 |
| 25 | Damaging-agent sensitivity of Artemis-deficient cell lines. European Journal of Immunology, 2005, 35, 1250-1256. | 1.6 | 30 |
| 26 | Mutant cohesin drives chromosomal instability in early colorectal adenomas. Human Molecular Genetics, 2014, 23, 6773-6778. | 1.4 | 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. | 0.7 | 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. | 2.4 | 28 |
| 30 | Claspin inhibition leads to fragile site expression. Genes Chromosomes and Cancer, 2009, 48, 1083-1090. | 1.5 | 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. | 2.7 | 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. | 1.6 | 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. | 1.4 | 24 |
| 35 | Cohesin mutations are synthetic lethal with stimulation of WNT signaling. ELife, 2020, 9, . | 2.8 | 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. | 1.0 | 21 |
| 38 | A role for Separase in telomere protection. Nature Communications, 2016, 7, 10405. | 5.8 | 20 |
| 39 | Recapitulation of the Roberts syndrome cellular phenotype by inhibition of INCENP, ZWINT-1 and ZW10 genes. Gene, 2004, 331, 33-40. | 1.0 | 19 |
| 40 | SMC1A codon 496 mutations affect the cellular response to genotoxic treatments. American Journal of Medical Genetics, Part A, 2012, 158A, 224-228. | 0.7 | 19 |
| 41 | Claspin as a biomarker of human papillomavirus-related high grade lesions of uterine cervix. Journal of Translational Medicine, 2012, 10, 132. | 1.8 | 18 |
| 42 | Common fragile sites on human chromosomes represent transcriptionally active regions: evidence from camptothecin. Human Genetics, 1998, 102, 409-414. | 1.8 | 17 |
| 43 | CEP57 mutation in a girl with mosaic variegated aneuploidy syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 177-181. | 0.7 | 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. | 1.8 | 12 |
| 46 | The multifaceted roles of cohesin in cancer. Journal of Experimental and Clinical Cancer Research, 2022, 41, 96. | 3.5 | 11 |
| 47 | Cycling-PRINS. Mutation Research - Genetic Toxicology and Environmental Mutagenesis, 1997, 390, 1-4. | 0.9 | 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. | 3.0 | 8 |
| 50 | Proliferation of Multiple Cell Types in the Skeletal Muscle Tissue Elicited by Acute p21 Suppression. Molecular Therapy, 2015, 23, 885-895. | 3.7 | 6 |
| 51 | Disease-associated c-MYC downregulation in human disorders of transcriptional regulation. Human Molecular Genetics, 2022, 31, 1599-1609. | 1.4 | 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. | 0.4 | 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. | 1.0 | 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. | 0.9 | 2 |
| 56 | Cohesins. , 2016, , 1113-1116. | | 0 |