BIOGRAPHICAL SKETCH

NAME	POSITION TITLE
Antonio Musio	Researcher

EDUCATION/TRAINING

INSTITUTION AND LOCATION	DEGREE *(if applicable)*	YEAR(s)	FIELD OF STUDY
University of Pisa, Pisa, Italy	Master Degree	1991	Biological Sciences
University of Ferrara, Ferrara, Italy	Ph.D.	1995	Genetic Sciences
University of Pavia, Pavia, Italy	Specialty	1998	Human Cytogenetic

Positions and Employment
1991-1995 PhD student, University of Pisa
1995-1996 Postdoc, Institute of Mutagenesis, National Research Council (CNR), Pisa.
1996-1997 Postdoc, Joint Research Centre of European Commission
1997-1998 Postdoc, Desio Hospital (Mi)
1999-2000 Postdoc, Department of Man and Environment, University of Pisa, Pisa
2000-2002 Postdoc, Institute for Biomedical Technologies, CNR, Segrate (Mi)
2002 Visiting scientist, NIH/NCI, Bethesda (USA)
2005 Visiting scientist, Roslin Institute, Edinburgh (UK)
2002-2011 Researcher, Institute for Biomedical Technologies, CNR
2012-present Researcher, Institute for Genetics and Biomedical Research, CNR

Research Experience
Cell biology, molecular genetics, cytogenetics. Focus areas: Cornelia de Lange syndrome, Cohesin, Cohesinopathies, Cancer, DNA repair, Genome stability.

Ad Hoc reviewer: Science, Science Advances, Nature, Nature Cancer Review, Human Molecular Genetics, European Journal of Human Genetics, Human Mutation, PlosOne, PlosBiology, PlosGenetics, America Journal of Medical Genetics, Clinical Genetics, and many others; Padua University, Insubria University, Florence University, Ministry of Research; Agence Nationale de la Recerche, Israel Science Foundation, Agency for Health Quality and Assessment of Catalonia, German Federal Ministry for Education and Research.

Awards
1993. Travel award from the Società Italiana di Mutagenesi Ambientale
2002. Travel award from European Union
2006. Travel award from European Science Foundation
2006. Travel award from the Società Italiana di Biofisica e Biologia Molecolare

Editorial board member of:
2010-2012 World Journal of Medical Genetics
2009-2012 Recent Patents on DNA & Gene Sequences
2020-present Cells (IF 4.366)

2015-2019 Board member of the Italian Society of Environmental and Genomic Mutagenesis

2016-2020 Board member of the Institute for Genetics and Biomedical Research Committee

Organizer of four International meetings "Cohesin Biology and the Cohesinopathies":
2007 Villa Monastero, Varenna (Lecco)
2009 Certosa di Pontignano, Siena
2011 Castelvecchio Pascoli, Lucca
2013 Certosa di Pontignano, Siena

2014. Member of the organizing committee of the 13rd Life Science Congress, Pisa

Professional memberships
Italian Society of Environmental and Genomic Mutagenesis, member

Other activities
Teaching: Contract Professor, University of Pavia, and University of Pisa
Mentorship: Supervisor of graduate, postgraduate students, and postdoc fellows

Research support
2020-present Italian Association for Cancer Research (AIRC)
2017-present Fondazione Pisana per la Scienza
2016-2018 Italian Association for Cancer Research (AIRC)
2017-2018 Merck Serono
2015-2018 InterOmics Flagship Project
2012-2016 Istituto Toscano Tumori
2012-2014 Italian Association for Cancer Research (AIRC)
2010-2013 Regione Toscana
2008-2009 Istituto Toscano Tumori
2007-2009 Ministero della Salute
2007-2009 Fondazione Cariplo

Peer-reviewed publications

1. Simi S, **Musio A**, Vatteroni L, Piras A, Rainaldi G. Specific chromosome aberrations correlated to transformation in Chinese hamster cells. Cancer Genet Cytogenet 1992;62:81-87.

2. **Musio** A, Mariani T, Frediani C, Sbrana I, Ascoli C. Longitudinal patterns similar to G-banding in untreated human chromosomes: evidence from atomic force microscopy. Chromosoma 1994;103:225-229.

3. Mariani T, **Musio A**, Frediani C, Sbrana I, Ascoli C. An atomic force microscope for cytological and histological investigations. J Microscopy 1994;176:121-131.

4. Sbrana I, **Musio A**. Enhanced expression of common fragile site with occupational exposure to pesticides. Cancer Genet Cytogenet 1995;82:123-127.

5. Mariani T, **Musio A**, Simi S. No statistical association between fragile site and constitutional chromosome breakpoints. Cancer Genet Cytogenet 1995;85:78-81.

6. **Musio A**, Sbrana I. Common and rare fragile sites on human chromosomes: the cytogenetic expression of active and inactive genes? Cancer Genet Cytogenet 1996;88:184-185.

7. **Musio A**, Rainaldi G, Sbrana I. Spontaneous and aphidicolin-sensitive fragile site 3cen co-localizes with the telomeric sequence (TTAGGG)n in Chinese hamster cells. Cytogenet Cell Genet 1996;75:159-163.

8. **Musio A**, Mariani T, Frediani C, Ascoli C, Sbrana I. Atomic force microscope imaging of chromosome structure during G-banding treatments. Genome 1997;40:127-131.

9. **Musio A**, Sbrana I. Aphidicolin-sensitive specific common fragile sites expression: a biomarker of exposure to pesticides. Environ Mol Mutagen 1997;29:250-255.

10. **Musio A**, Rainaldi G. Cycling-PRINS. A method to improve the accuracy of telomeric detection in mammalian chromosomes. Mutation Res 1997;390:1-4.

11. Vatteroni L, **Musio A**, Meneveri R, Rainaldi G. Assignment of Chinese hamster p53 (TP53) to chromosome band 2p31, a region not involved in the karyotypic changes of a tumorigenic cell line. Cytogenet Cell Genet 1997;77:228-231.

12. Mariani T, **Musio A**, Simi S. Factors affecting the constitutional breakage of human chromosomes. J Environ Pathol Tox Onc 1997;16:263-272.

13. **Musio A**, Sbrana I. Detection of chromosome X in human sperm nuclei by direct- and indirect-primed in situ labeling (PRINS). Biochemica 1998;2:29-30.

14. Sbrana I, Zavattari P, **Musio A**. Common fragile sites on human chromosomes represent transcriptionally active regions: evidence from camptothecin. Hum Genet 1998;102:409-414.

15. **Musio A**, Baroli P, Sbrana I. Primed in situ labeling (PRINS) a method for rapid identification and quantification of human chromosomes in lymphocytes and sperm nuclei. Genome 1998;41:739-741.

16. **Musio A**, Mariani T. Distribution of interstitial telomere-related sequences in the human genome and their relationship with fragile sites. J Environ Pathol Tox Onc 1999;18:11-15.

17. Mariani T, Ascoli C, Bashieri P, Frediani C, **Musio A**. Scanning force images through the milliscope - a probe microscope with very wide scan range. J Microsc 2001;204:53-60.

18. Sobacchi C, Frattini A, Orchard P, Porras O, Tezcan I, Andolina M, **Musio A**, et al. The mutational spectrum of human malignant autosomal recessive osteopetrosis. Hum Mol Genet 2001;10:1767-1773.

19. **Musio A**, Mariani T, Vezzoni P, Frattini A. The heterogeneous gene distribution reflects human genome complexity as detected at the cytogenetic level. Cancer Genet Cytogenet 2002;134:168-171

20. **Musio A**, Zambroni D, Mariani T, Vezzoni P. Chromosomes, genes and cancer breakpoints. Cancer Genet Cytogenet 2002;139:141-142.

21. **Musio A**, Montagna C, Zambroni D, Indino E, Barbieri O, Citti L, Villa A, Ried T, Vezzoni P. Inhibition of BUB1 results in genomic instabilty and anchorage-independent growth of normal human fibroblasts. Cancer Res 2003;63:2855-2863

22. Sobacchi C, Vezzoni P, Ried MD, McGuigan FEA, Frattini A, Mirolo M, **Musio A**, Villa A, Ralston SH. Linkage disequilibrium between polymorphism in the human TCIRG1 gene and their association with bone mass in perimenopausal women. Calcific Tissue Int 2004;74:35-41.

23. **Musio A**, Marrella V, Sobacchi C, Rucci F, Fariselli L, Giliani S, Lanzi G, Notarangelo LD, Delia D, Colombo R, Vezzoni P, Villa A. Damaging-agent sensitivity of Artemis-deficient cell lines. Eur J Immun 2005;35:1250-1256.

24. **Musio A**, Montagna C, Mariani T, Tilenni M, Focarelli ML, Brait L, Indino E, Benedetti PA, Chessa L, Albertini A, Ried T, Vezzoni P. SMC1 involvement in fragile site expression. Hum Mol Genet 2005;14:525-533.

25. Frattini A, Blair HC, Sacco MG, Cerisoli F, Fagioli F, Mira Catò E, Pancrazio A, **Musio A**, Rucci F, Sobacchi C, Sharrow AC, Kalla SA, Buzzone MG, Magli MC, Vezzosi P, Villa A. Rescue of murine malignant osteopetrosis by bone marrow transplantation *in utero.* Proc Natl Acad Sci (USA) 2005; 102:14629-14634.

26. Forsyth NR, Musio **A**, Vezzoni P, Simpson AHRW, Noble BS, McWhir J. Physiologic oxygen enhances human embryonic stem cell clonal recovery and reduces chromosome abnormalities. Cloning Stem Cell 2006; 8:16-23.

27. **Musio A**, Selicorni A, Focarelli ML, Gervasini C, Donatella M, Russo S, Vezzoni P, Larizza L. X-linked Cornelia de Lange syndrome owing to SMC1L1 mutations. Nat Genet 2006; 38:520-530.

28. Deardorff MA, Kaur M, Yaeger D, Rampuria A, Korolev S, Pie J, Gil-Rodriguez C, Arnedo M, Loeys B, Kline AD, Wilson M, Lillquist K, Siu V, Ramos FJ, **Musio A**, Jackson LS, Dorsett D, Krantz ID. Mutations in cohesion complex members SMC3 and SMC1A cause a mild variant of Cornelia de Lange syndrome with predominat mental retardation. Am J Hum Genet 2007; 80:485-494.

29. Morciano P, Carrisi C, Capobianco L,Mannini L, Burgio G, Cestra G, De Benedetto GE, Corona DFV, **Musio A**, Cenci G. A conserved role for the mitochondrial citrate transporter Sea/SLC25A1 in the maintenance of chromosome integrity. Hum Mol Genet 2009; 8:4180-4188.

30. Focarelli ML, Souza S, Mannini L, Paulis M, Montecucco A, **Musio A**. Claspin inhibition leads to fragile site expression. Genes Chrom Cancer 2009; 48:1083-1090.

31. Revenkova E, Focarelli ML, Susani L, Paulis M, Bassi MT, Mannini L, Frattini A, Delia D, Krantz I, Vezzoni P, Jessberger R, **Musio A**. Cornelia de Lange syndrome mutations in SMC1A or SMC3 affect binding to DNA. Hum Mol Genet 2009; 18:418-427.

32. Mannini L, Menga S, **Musio A**. The expanding universe of cohesin functions: a new genome stability caretaker involved in human disease and cancer. Hum Mutat 2010; 31:623-630.

33. Mannini L, Liu J, Krantz ID, **Musio A**. Spectrum and consequences of SMC1A mutations: the unexpected involvement of a core component of cohesin in human disease. Hum Mutat 2010; 31:5-10.

34. Mannini L, **Musio** A. The dark side of cohesion: the carcinogenic point of view. Mutat Res 2011; 728:81-87.

35. Schrier SA, Bodurtha JN, Burton B, Chudley AE, Chiong MA, D'avanzo MG, Lynch SA, **Musio A**, Nyazov DM, Sanchez-Lara PA, Shalev SA, Deardorff MA. The Coffin-Siris syndrome: a proposed diagnostic approach and assessment of 15 overlapping cases. Am J Med Genet 2012; 158A:1865-1876.

36. Mannini L, Menga S, Tonelli A, Zanotti S, Bassi MT, Magnani C, **Musio A**. SMC1A codon 496 mutations affect the cellular response to genotoxic treatments. Am J Med Genet 2012; 158A:224-228.

37. Benevolo M, **Musio A**, Vocaturo A, Donà MG, Rollo F, Terrenato I, Carosi M, Pescarmona E, Vocaturo G, Mottolese M. Claspin as a biomarker of human papillomavirus-related high grade lesions of uterine cervix. J Transl Med 2012;10:132.

38. Gimigliano A, Mannini L, Bianchi L, Puglia M, Deardorff MA, Menga S, Krantz ID, **Musio A***, Bini L. Proteomic Profile Identifies Dysregulated Pathways in Cornelia de Lange Syndrome Cells with Distinct Mutations in SMC1A and SMC3 Genes. J Proteome Res 2012; 11:6111-6123. *corresponding author

39. Falcone G, Mazzola A, Michelini F, Bossi G, Censi F, Biferi MG, Minghetti L, Floridia G, Federico M, **Musio A**, Crescenzi M. Cytogenetic analysis of human cells reveals specific patterns of DNA damage in replicative and oncogene-induced senescence. Aging Cell 2013; 12:312-315.

40. Mannini L, Cucco F, Quarantotti V, Krantz ID, **Musio A**. Mutation spectrum and genotype-phenotype correlation in Cornelia de Lange syndrome. Hum Mutat 2013; 34:1589-1596.

41. Pinson L, Mannini L, Willems M, Cucco F, Sirvent N, Frebourg T, Quarantotti V, Collet C, Schneider A, Sarda P, Geneviève D, Puechberty J, Lefort G, **Musio A**. CEP57 mutation in a girl with mosaic variegated aneuploidy syndrome. Am J Med Genet A 2014; 164:177-181.

42. Cucco F, Servadio A, Gatti V, Bianchi P, Mannini L, Prodosmo A, De Vitis E, Basso G, Friuli A, Laghi L, Soddu S, Fontanini G, **Musio A**. Mutant cohesin drives chromosomal instability in early colorectal adenomas. Hum Mol Genet 2014; 23:6773-6778.

43. Baquero-Montoya C, Gil-Rodríguez MC, Braunholz D, Teresa-Rodrigo ME, Obieglo C, Gener B, Schwarzmayr T, Strom TM, Gómez-Puertas P, Puisac B, Gillessen-Kaesbach G, **Musio A**, Ramos FJ, Kaiser FJ, Pié J. Somatic mosaicism in a Cornelia de Lange syndrome patient with NIPBL mutation identified by different next generation sequencing approaches. Clin Genet 2014; 86:595-597.

44. Puntoni M, Bigazzi F, Sabatino L, Sbrana F, **Musio A**, Dal Pino B, Ragusa A, Corsano E, Sampietro T. Early senescence in heterozygous ABCA1 mutation skin fibroblasts: a gene dosage effect beyond HDL deficiency? Biochem Biophys Res Commun 2014; 447:231-236.

45. Ramos FJ, Puisac B, Baquero-Montoya C, Gil-Rodríguez MC, Bueno I, Deardorff MA, Hennekam RC, Kaiser FJ, Krantz ID, **Musio A**, Selicorni A, FitzPatrick DR, Pié J. Clinical utility gene card for: Cornelia de Lange syndrome. Eur J Hum Genet 2015; doi: 10.1038/ejhg.2014.270.

46. Burla R, Carcuro M, Raffa GD, Galati A, Raimondo D, Rizzo A, La Torre M, Micheli E, Ciapponi L, Cenci G, Cundari E, **Musio A**, Biroccio A, Cacchione S, Gatti M, Saggio I. AKTIP/Ft1, a New Shelterin-Interacting Factor Required for Telomere Maintenance. PLoS Genet 2015; 11(6):e1005167.

47. Biferi MG, Nicoletti C, Falcone G, Puggioni EM, Passaro N, Mazzola A, Pajalunga D, Zaccagnini G, Rizzuto E, Auricchio A, Zentilin L, De Luca G, Giacca M, Martelli F, **Musio A**, Musarò A, Crescenzi M. Proliferation of Multiple Cell Types in the Skeletal Muscle Tissue Elicited by Acute p21 Suppression. Mol Ther 2015; 23:885-895.

48. Kline AD, Calof AL, Lander AD, Gerton JL, Krantz ID, Dorsett D, Deardorff MA, Blagowidow N, Yokomori K, Shirahige K, Santos R, Woodman J, Megee PC, O'Connor JT, Egense A, Noon S, Belote M, Goodban MT, Hansen BD, Timmons JG, **Musio A**, Ishman SL, Bryan Y, Wu Y, Bettini LR, Mehta D, Zakari M, Mills JA, Srivastava S, Haaland RE. Clinical, developmental and molecular update on Cornelia de Lange syndrome and the cohesin complex: abstracts from the 2014 Scientific and Educational Symposium. Am J Med Genet A 2015; 167:1179-1192.

49. Mannini L, Cucco F, Quarantotti V, Amato C, Mara T, Tana L, Frattini A, Delia D, Krantz ID, Jessberger R, **Musio A**. SMC1B is present in mammalian somatic cells and interacts with mitotic cohesin proteins. Sci Rep 2015 5:18472. doi: 10.1038/srep18472.

50. Mannini L, Lamaze FC, Cucco F, Amato C, Quarantotti V, Rizzo IM, Krantz ID, Bilodeau S, **Musio A**. Mutant cohesin affects RNA polymerase II regulation in Cornelia de Lange syndrome. Sci Rep 2015; 5:16803. doi: 10.1038/srep16803.

51. Cucco F, **Musio A**. Genome instability: What we have learned from cohesinopathies. Am J Med Genet C Semin Med Genet 2016; 172:171-178.

52. Cipressa F, Morciano P, Bosso G, Mannini L, Galati A, Raffa GD, Cacchione S, **Musio A**, Cenci G. A role for Separase in telomere protection. Nat Commun 2016; 7:10405. doi: 10.1038/ncomms10405.

53. Kline AD, Krantz ID, Deardorff MA, Shirahige K, Dorsett D, Gerton JL, Wu M, Mehta D, Mills JA, Carrico CS, Noon S, Herrera PS, Horsfield JA, Bettale C, Morgan J, Huisman SA, Moss J, McCleery J, Grados M, Hansen BD, Srivastava S, Taylor-Snell E, Kerr LM, Katz O, Calof AL, **Musio A**, Egense A, Haaland RE. Cornelia de Lange syndrome and molecular implications of the cohesin complex: Abstracts from the 7th biennial scientific and educational symposium 2016. Am J Med Genet A 2017; 173:1172-1185.

54. Barone S, Parri C, Gazzi S, Rapalini E, **Musio A**. Aging and cohesin: a mini review. Curr Trends in Clin Embryol 2018; 5:30-36.

55. Cucco F, Palumbo E, Camerini S, D'Alessio B, Quarantotti V, Casella ML, Rizzo IM, Cukrov D, Delia D, Russo A, Crescenzi M, **Musio A**. Separase prevents genomic instability by controlling replication fork speed. Nucleic Acids Res 2018; 46:267-278.

56. Cukrov D, Newman T, Leask M, Leeke B, Sarogni P, Patimo A, Kline AD, Krantz ID, Horsfield J, **Musio A**. Antioxidant treatment ameliorates phenotypic features of SMC1A-mutated Cornelia de Lange syndrome in vitro and in vivo. Hum Mol Genet. 2018 May 30. doi: 10.1093/hmg/ddy203.

57. Sarogni P, Palumbo O, Servadio A, Astigiano S, D'Alessio B, Gatti V, Cukrov D, Baldari S, Pallotta MM, Aretini P, Dell'Orletta F, Soddu S, Carella M, Toietta G, Barbieri O, Fontanini G, **Musio A**. Overexpression of the cohesin-core subunit SMC1A contributes to colorectal cancer development. J Exp Clin Cancer Res. 2019; 38(1):108. doi: 10.1186/s13046-019-1116-0.

58. Kline AD, Krantz ID, Bando M, Shirahige K, Chea S, Sakata T, Rao S, Dorsett D, Singh VP, Gerton JL, Horsfield JA, Calof AL, Katz O, Grados M, Raible S, Barañano K, Lyon G, **Musio A**, Carrico CS, Clemens DK, Caudill P, Massa V, McGill BE, Dommestrup A, O'Connor J, Haaland RE. Cornelia de Lange syndrome, related disorders, and the Cohesin complex: Abstracts from the 8th biennial scientific and educational symposium 2018. Am J Med Genet A. 2019 179:1080-1090.

59. Contadini C, Monteonofrio L, Virdia I, Prodosmo A, Valente D, Chessa L, **Musio A**, Fava LL, Rinaldo C, Di Rocco G, Soddu S. p53 mitotic centrosome localization preserves centrosome integrity and works as sensor for the mitotic surveillance pathway. Cell Death Dis. 2019 10:850.

60. Latorre-Pellicer A, Ascaso Á, Trujillano L, Gil-Salvador M, Arnedo M, Lucia-Campos C, Antoñanzas-Pérez R, Marcos-Alcalde I, Parenti I, Bueno-Lozano G, **Musio A**, Puisac B, Kaiser FJ, Ramos FJ, Gómez-Puertas P, Pié J. Evaluating Face2Gene as a Tool to Identify Cornelia de Lange Syndrome by Facial Phenotypes. Int J Mol Sci. 2020 21(3).

61. Barone S, Sarogni P, Valli R, Pallotta MM, Silvia G, Frattini A, Khan AW, Rapalini E, Parri C, **Musio A**. Chromosome Missegregation in Single Human Oocytes Is Related to the Age and Gene Expression Profile. Int J Mol Sci. 2020 21(6).

62. Sarogni P, Pallotta MM, **Musio A**. Cornelia de Lange syndrome: from molecular diagnosis to therapeutic approach. J Med Genet. 2020 57:289-295.

63. Cucco F, Sarogni P, Rossato S, Alpa M, Patimo A, Latorre A, Magnani C, Puisac B, Ramos FJ, Pié J, **Musio A.** Pathogenic variants in EP300 and ANKRD11 in patients with phenotypes overlapping Cornelia de Lange syndrome. Am J Med Genet A. 2020; 182:1690-1696.

64. **Musio A**. The multiple facets of the SMC1A gene. Gene. 2020; 743:144612.

65. Chin CV, Antony J, Ketharnathan S, Labudina A, Gimenez G, Parsons KM, He J, George AJ, Pallota MM, **Musio A**, Braithwaite AW, Guilford P, Hannan RD, Horsfield JA. Cohesin mutations are synthetic lethal with stimulation of WNT signaling. Elife. 2020 Dec 7;9:e61405. doi: 10.7554/eLife.61405