

BIOGRAPHICAL SKETCH

NAME: Mario Ventura

POSITION TITLE: Full Professor of Genetics

eRA COMMONS USERNAME: Not Applicable

EDUCATION/TRAINING:

Institution and Location	Degree	Completion Date	Field of Study
University of Bari “Aldo Moro”, Italy	Post Doc	2004	Genetics and Molecular Evolution
University of Bari “Aldo Moro”, Italy	Ph.D.	2003	Genetics and Molecular Evolution
University of Bari “Aldo Moro”, Italy	B.Sc.	1999	Biological Sciences

A. Personal Statement

I am a Full Professor of Genetics with over two decades of academic, research, and teaching experience in human and primate genomics. My scientific journey has been driven by a passion for understanding genome architecture, evolution, and their implications for human health. My research has focused on the structural and functional characterization of centromeres and neocentromeres, the evolutionary dynamics of segmental duplications, and the identification of genomic structural variations linked to diseases. These themes are unified by a central aim: to elucidate how genomic structural changes shape biological function and evolutionary trajectories.

I have successfully led national and international research projects and contributed to high-impact scientific publications, including landmark studies published in *Nature* and *Science*. I am actively involved in international collaborations, including work with the Genome Science Department at the University of Washington, where I serve as an Affiliate Professor. My laboratory applies state-of-the-art genomic technologies, including Oxford Nanopore and PacBio long-read sequencing, and advanced bioinformatics to decode genomic complexity across species.

Beyond research, I have played a key role in academic leadership, mentoring young researchers, coordinating teaching programs, and serving on evaluation panels and editorial boards. I am committed to fostering a collaborative and inclusive research environment, and to leveraging genomic science for both fundamental discovery and societal benefit. This proposal reflects my continued dedication to investigating the genomic basis of evolutionary innovation and human disease susceptibility. To date, I have published 135 full papers with peer review and six book chapters. I got an h-index of 51 and 11101 citations as reported in Scopus (<https://www.scopus.com>).

B. Positions, Scientific Appointments, and Honors

- 2021–present: Full Professor, Department of Biosciences, Biotechnology and Environment, University of Bari 'Aldo Moro', Italy
- 2013–2021: Associate Professor, Department of Biology, University of Bari 'Aldo Moro', Italy
- 2012–present: Affiliate Professor, Department of Genome Sciences, University of Washington, Seattle (WA)
- 2004–2013: Assistant Professor, University of Bari 'Aldo Moro', Italy
- Numerous Visiting Professorships at University of Washington (2007–2011)
- Honors include: Fulbright Fellowship (2007), Premio Ponzano for Genetics (2007), Cataldus d'Argento Award (2006), EMBO Short-Term Fellowship (2001)
- National Scientific Qualification (ASN) for Full Professor in Genetics (2017–2023)

C. Contributions to Science

My scientific contributions span multiple facets of human and primate genomics, with emphasis on structural variation, centromere biology, and the evolutionary dynamics of chromosomes. I have integrated cytogenetic and genomic approaches to generate reference-quality genome assemblies, discover lineage-specific genomic features, and investigate their implications in evolution and disease. Below, I outline selected key areas of contribution:

1. Centromere and Neocentromere Evolution in Primates

My research has led to the first high-resolution maps of complete centromere sequences in human and great apes. By applying cutting-edge long-read sequencing, FISH mapping, and computational analysis, we revealed evolutionary transitions of centromeres and the birth of neocentromeres. This work provides insights into chromosome identity, segregation mechanisms, and centromeric instability associated with disease.

Selected Publications:

- Logsdon GA, Rozanski AN, Ryabov F, Potapova T, Shepelev VA, Catacchio CR, Porubsky D, Mao Y, Yoo D, Rautiainen M, Koren S, Nurk S, Lucas JK, Hoekzema K, Munson KM, Gerton JL, Phillippy AM, **Ventura M**, Alexandrov IA, Eichler EE. The variation and evolution of complete human centromeres. *Nature*. 2024 Apr 3. doi: 10.1038/s41586-024-07278-3. Online ahead of print. PMID: 38570684.
- Mao, Y., Catacchio, C.R., Hillier, L.D.W., Porubsky, D., Li, R., Sulovari, A., Fernandes, J.D., Montinaro, F., Gordon, D.S., Storer, J.M., Haukness, M., Fiddes, I.T., Murali, S.C., Dishuck, P.C., Hsieh, P.H., Harvey, W.T., Audano, P.A., Mercuri, L., Piccolo, I., Antonacci, F., Munson, K.M., Lewis, A.P., Baker, C., Underwood, J.G., Hoekzema, K., Huang, T.-H., Sorensen, M., Walker, J.A., Hoffman, J., Thibaud-Nissen, F., Salama, S.R., Pang, A.W.C., Lee, J., Hastie, A.R., Paten, B., Batzer, M.A., Diekhans, M., **Ventura, M.** *, Eichler, E.E*. A high-quality bonobo genome refines the analysis of hominid evolution (2021) *Nature*, 594 (7861), pp. 77-81. *Corresponding authors
- Logsdon, G.A., Vollger, M.R., Hsieh, P.H., Mao, Y., Liskovych, M.A., Koren, S., Nurk, S., Mercuri, L., Dishuck, P.C., Rhie, A., de Lima, L.G., Dvorkina, T., Porubsky, D., Harvey, W.T., Mikheenko, A., Bzikadze, A.V., Kremitzki, M., Graves-Lindsay, T.A., Jain, C., Hoekzema, K., Murali, S.C., Munson, K.M., Baker, C., Sorensen, M., Lewis, A.M., Surti, U., Gerton, J.L., Larionov, V., **Ventura,**

M. , Miga, K.H., Phillippy, A.M., Eichler, E.E. The structure, function and evolution of a complete human chromosome 8 (2021) *Nature*, 593 (7857), pp. 101-107.

2. Structural Variation in Ape and Human Genomes

I have characterized numerous inversions, duplications, and deletions shaping primate genomes. My group pioneered the use of single-cell and long-read technologies to resolve complex structural variants. This work has shed light on hotspots of genome instability and has been instrumental in defining genome reference standards.

Selected Publications:

- Vollger MR, Guitart X, Dishuck PC, Mercuri L, Harvey WT, Gershman A, Diekhans M, Sulovari A, Munson KM, Lewis AP, Hoekzema K, Porubsky D, Li R, Nurk S, Koren S, Miga KH, Phillippy AM, Timp W, **Ventura M**, Eichler EE. Segmental duplications and their variation in a complete human genome. *Science* 2022 Apr;376(6588):eabj6965. doi: 10.1126/science.abj6965. Epub 2022 Apr 1.
- Mercuri L, Palmisano D, L'Abbate A, D'Addabbo P, Montinaro F, Catacchio CR, Hasenfeld P, **Ventura M**, Korbel JO, Sanders AD, Maggolini FAM, Antonacci F. A high-resolution map of small-scale inversions in the gibbon genome. *Genome Res.* 2022 Oct;32(10):1941-1951. doi: 10.1101/gr.276960.122. Epub 2022 Sep 30.
- Maggolini, F.A.M., Sanders, A.D., Shew, C.J., Sulovari, A., Mao, Y., Puig, M., Catacchio, C.R., Dellino, M., Palmisano, D., Mercuri, L., Bitonto, M., Porubský, D., Cáceres, M., Eichler, E.E., **Ventura, M.**, Dennis, M.Y., Korbel, J.O., Antonacci, F. Single-cell strand sequencing of a macaque genome reveals multiple nested inversions and breakpoint reuse during primate evolution (2020) *Genome research*, 30 (11), pp. 1680-1693.

3. Evolutionary Genomics and Disease Associations

My research uncovered links between structural variants and neurodevelopmental and psychiatric disorders. We explored how the evolution of disease-associated regions, such as 8p23.1 and 15q11-q13, may underlie susceptibility to genomic disorders. Our findings bridge evolutionary biology with medical genetics.

Selected Publications:

- Makova KD, Pickett BD, Harris RS, Hartley GA, Cechova M, Pal K, Nurk S, Yoo D, Li Q, Hebbar P, McGrath BC, Antonacci F, Aubel M, Biddanda A, Borchers M, Bomberg E, Bouffard GG, Brooks SY, Carbone L, Carrel L, Carroll A, Chang PC, Chin CS, Cook DE, Craig SJC, de Gennaro L, Diekhans M, Dutra A, Garcia GH, Grady PGS, Green RE, Haddad D, Hallast P, Harvey WT, Hickey G, Hillis DA, Hoyt SJ, Jeong H, Kamali K, Kosakovsky Pond SL, LaPolice TM, Lee C, Lewis AP, Loh YE, Masterson P, McCoy RC, Medvedev P, Miga KH, Munson KM, Pak E, Paten B, Pinto BJ, Potapova T, Rhie A, Rocha JL, Ryabov F, Ryder OA, Sacco S, Shafin K, Shepelev VA, Slon V, Solar SJ, Storer JM, Sudmant PH, Sweetalana, Sweeten A, Tassia MG, Thibaud-Nissen F, **Ventura M**, Wilson MA, Young AC, Zeng H, Zhang X, Szpiech ZA, Huber CD, Gerton JL, Yi SV, Schatz MC, Alexandrov IA, Koren S, O'Neill RJ, Eichler E, Phillippy AM. The Complete Sequence and Comparative Analysis
- Paparella A, L'Abbate A, Palmisano D, Chirico G, Porubsky D, Catacchio CR, **Ventura M**, Eichler EE, Maggolini FAM, Antonacci F. Structural Variation Evolution at the 15q11-q13 Disease-Associated Locus. *Int J Mol Sci.* 2023 Oct 31;24(21):15818. doi: 10.3390/ijms242115818

- Mohajeri, K., Cantsilieris, S., Huddleston, J., Nelson, B.J., Coe, B.P., Campbell, C.D., Baker, C., Harshman, L., Munson, K.M., Kronenberg, Z.N., Kremitzki, M., Raja, A., Catacchio, C.R., Graves, T.A., Wilson, R.K., **Ventura, M.**, Eichler, E.E. Interchromosomal core duplicons drive both evolutionary instability and disease susceptibility of the Chromosome 8p23.1 region (2016) *Genome Research*, 26 (11), pp. 1453-1467. DOI: 10.1101/gr.211284.116

4. Population Genomics and Human Diversity

Through collaborative studies, I have examined the genetic history and structure of human populations across Europe and the Americas. This includes work on genomic admixture, sex-biased inheritance, and selection patterns in response to environmental and cultural transitions.

Selected Publications:

- Raveane A, Molinaro L, Aneli S, Capodiferro MR, de Gennaro L, Ongaro L, Rambaldi Migliore N, Soffiati S, Scarano T, Torroni A, Achilli A, **Ventura M**, Pagani L, Capelli C, Olivieri A, Bertolini F, Semino O, Montinaro F. Assessing temporal and geographic contacts across the Adriatic Sea through the analysis of genome-wide data.
- Ongaro L, Mondal M, Flores R, Marnetto D, Molinaro L, Alarcón-Riquelme ME, Moreno-Estrada A, Mabunda N, **Ventura M**, Tambets K, Hellenthal G, Capelli C, Kivisild T, Metspalu M, Pagani L, Montinaro F. Continental-scale genomic analysis suggests shared post-admixture adaptation in the Americas. *Hum Mol Genet.* 2021 Nov 1;30(22):2123-2134. doi: 10.1093/hmg/ddab177
- De Gennaro L, Molinaro L, Raveane A, Santonastaso F, Saponetti SS, Massi MC, Pagani L, Metspalu M, Hellenthal G, Kivisild T, **Ventura M***, Montinaro F. * PANE: fast and reliable ancestral reconstruction on ancient genotype data with non-negative least square and principal component analysis. *Genome Biol.* 2025 Feb 11;26(1):29. doi: 10.1186/s13059-025-03491-z. *Co-corresponding.

D. Research Support

1. Ongoing – Telomere-to-telomere sequencing: the new era of Centromere and neocentromere eVolution (CenVolution)

Source: PRIN 2022, Ministry of University and Research (MIUR)

Role: Principal Investigator

Description: This project aims to apply ultra-long read sequencing to explore centromere and neocentromere evolution in primates and humans.

2. Completed – Omics explorations to discover substance use disorders: a journey from the brain to the gut

Source: Horizon Europe Seed Funding, University of Bari (2021)

Role: Principal Investigator

Description: This pilot initiative integrates genomics, transcriptomics, and microbiome data to investigate the genetic and epigenetic bases of substance use disorders. It aims to lay the groundwork for future EU-funded projects by identifying biomarkers and molecular pathways across brain-gut axes.

3. Completed – Structural genomic variation in primates and mammals: new perspectives for studying human evolution

Source: Future in Research 2010-RBFR103CE3

Role: Principal Investigator

Description: This project explored primate genome diversity with a focus on structural variants, using cytogenetic and sequencing approaches.

4. Completed – Promotion of ECO_sustainable Processes for the valorisation of Apulian Agri-food Productions (ECO_P4)

Source: PON02_00186_2866121

Role: Unit Leader

Description: This applied research project explored sustainable biotechnological innovations in agriculture using omics tools. My unit focused on transcriptomic profiling and structural genome analysis of table grape varieties.

5. Completed – Genomic Organisation and Sequence Characterization of Primate Pericentromeric Regions

Source: PRIN 2007Z5X4YA_001

Role: Principal Investigator

Description: A foundational project in my career, it aimed to map and characterize the sequence composition of pericentromeric regions in humans and non-human primates using FISH, BAC libraries, and early genomic tools.

E. Teaching Experience

Over the past two decades, I have developed and taught a broad portfolio of undergraduate and graduate (roughly 60) courses in genetics, molecular biology, and biotechnology. My teaching activity spans the Bachelor's, Master's, and Postgraduate Specialization School levels at the University of Bari 'Aldo Moro'. I have coordinated and delivered courses in Molecular Genetics, Genetic Engineering, Evolutionary Biology, and Cytogenetics, and contributed to advanced training in clinical cytogenomics through a dedicated Master's program. I am known for fostering student engagement and have received excellent evaluations from students over the years. Furthermore, I have supervised numerous undergraduate, Master's, and PhD theses, and coordinated teaching activities in the Medical and Pharmaceutical Biotechnology program.