

BIOGRAPHICAL SKETCH

NAME: Claudia Rita Catacchio

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POSITION TITLE: Associate Professor

EDUCATION/TRAINING

INSTITUTION AND LOCATION	DEGREE (if applicable)	Completion Date MM/YYYY	FIELD OF STUDY
European Molecular Biology Laboratory (EMBL) (Heidelberg)	Visiting Scientist	10/2017	Human and Primate Genomic Structural Variations
Department of Biosciences, biotechnology, and environment (University of Bari, Italy)	Post-Doc	10/2015	Mammalian Genome Evolution
Department of Biology (University of Bari, Italy)	PhD	03/2012	Primate Genome Evolution and Structural Variations
Department of Genome Sciences (University of Washington)	Visiting Scholar	09/2011	Primate Genome Evolution
Edinburgh Cancer Research Center (University of Edinburgh)	Visiting Scientist	10/2008	Neocentromeres in Human
University of Bari, Italy	Msc (2 years)	10/2007	Medical Biotechnology and Molecular Medicine
University of Bari, Italy	BA (3 years)	07/2005	Medical and Pharmaceutical Biotechnology

A. Personal Statement

My research focuses on understanding genome evolution in primates, with particular emphasis on structural variation, repetitive DNA, and centromere organization. Since establishing my scientific independence in 2018, I have pursued longstanding questions on how genome architecture contributes to species diversity, drawing on an integrated approach that combines cytogenetics, sequencing, molecular biology and computational genomics.

My training includes international experience, notably more than one year in Evan Eichler's laboratory, where I contributed to the first bonobo reference genome published in *Nature*. I later led the generation of the second bonobo reference genome as first author, coordinating a large international collaboration. These experiences shaped my expertise in reference genome construction and comparative primate genomics.

I am currently involved in the Primate T2T Centromere Consortium, where I analyze centromeres from gapless great ape genomes. In parallel, I developed a bioinformatic tool to detect and characterize centromeric satellite DNA and higher-order repeats, now submitted to *Genome Biology* (<https://www.biorxiv.org/content/10.1101/2025.01.07.631657v1>). This tool is central to my research program and directly underpins the proposed project.

Following a research visit to Ian Korbel's laboratory at EMBL, I gained hands-on experience with Strand-seq technology, which contributed to its adoption within our research environment in Bari. This expertise enables the integration of long-read sequencing, Strand-seq, and cytogenetic approaches for high-resolution analysis of genome structure.

I have demonstrated the ability to attract competitive funding: I am Principal Investigator of a PRIN 2022 project focused on gibbon genome evolution, which directly supports the scientific framework of the present proposal. In addition, I have participated as a team member in multiple nationally funded projects (PRIN 2020, PRIN 2022 PNRR, and PNRR flagship initiatives), reflecting sustained involvement in large-scale, collaborative research efforts.

In addition to research, I am actively involved in doctoral education, teaching genetics and genomics at undergraduate, master's, and PhD levels, and I hold several institutional responsibilities at the University of Bari (such as University of Bari Delegate to the European University Association (EUA) network). The proposed project represents a natural extension of my research trajectory and leverages my expertise to address fundamental questions in genome evolution using gibbons as a model system.

B. Positions, Scientific Appointments, and Honors

Since 2021: Associate Professor in Genetics (BIOS-14/A). Department of Biosciences, Biotechnology and Environment, University of Bari "Aldo Moro", Italy.

2018 – 2021: Tenure track Researcher (rtd-B) in Genetics. Department of Biology, University of Bari "Aldo Moro", Italy.

2015 – 2018: Junior Researcher (rtd-A) in Genetics. Department of Biology, University of Bari "Aldo Moro", Italy.

C. Contributions to Science

My research has made significant contributions to the field of genome evolution, particularly in primate genomics, structural variation, and centromere biology.

1. Reference genome generation and primate comparative genomics

I contributed to the generation of the first bonobo reference genome, published in Nature, and subsequently led the production of the second bonobo reference genome as first author. These resources have become foundational for comparative genomics in great apes and have enabled detailed analyses of structural variation, segmental duplications, and evolutionary divergence.

2. Structural variation and Strand-seq applications

Through training at EMBL and collaboration with pioneers of Strand-seq, I introduced this technology into my laboratory, making it one of the few worldwide capable of mapping balanced chromosomal rearrangements at high resolution. I have applied Strand-seq to evolutionary studies and to clinically relevant contexts, including the investigation of inversions associated with infertility and neurodevelopmental disorders.

3. Centromere biology and repeat evolution

I am an active member of the Primate T2T Centromere Consortium, contributing to the analysis of centromeres from gapless ape genomes. To address the lack of dedicated tools for centromeric repeat analysis, I developed a novel bioinformatic pipeline for the detection and characterization of satellite DNA and higher-order repeats. This tool, currently under review at Genome Biology, enables comparative analyses of centromere organization across species and represents a key methodological advance in the field.

4. Scientific productivity and leadership

Since 2018, I have coauthored 45 peer-reviewed publications, including 11 as first or corresponding author. My work integrates genomics, cytogenetics, and bioinformatics, and is characterized by strong international collaborations and methodological innovation.

Together, these contributions have advanced our understanding of primate genome evolution and laid the groundwork for the proposed project, which extends these approaches to gibbons to address fundamental questions on structural variation, centromere evolution, and genomic diversity.