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## BIOGRAPHICAL SKETCH

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NAME: Francesca Antonacci

eRA COMMONS USER NAME: ANTONACCI\_F

POSITION TITLE: Full Professor of Genetics (BIOS-14/A), University of Bari Aldo Moro

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### EDUCATION/TRAINING

| INSTITUTION AND LOCATION               | DEGREE        | Completion Date | FIELD OF STUDY                   |
|----------------------------------------|---------------|-----------------|----------------------------------|
| University of Washington, Seattle, USA | Postdoc       | 05/2012         | Genome Sciences                  |
| University of Bari, Bari, Italy        | Ph.D.         | 03/2008         | Genetics and Molecular Evolution |
| University of Bari, Bari, Italy        | Master Degree | 07/2004         | Biological Sciences              |

### A. Personal Statement

I am a Full Professor of Genetics (BIOS-14/A) at the University of Bari Aldo Moro with extensive experience in the study of genome structural variation and its implications in human diseases and evolution. After completing a PhD in Molecular Genetics and Evolution, I conducted postdoctoral research at the University of Washington under Prof. Evan Eichler supervision, focusing on human and primate genome variation. I currently coordinate several research projects investigating idiopathic infertility, human genome structural variation and genome evolution. My expertise spans from cytogenetics technologies to single-cell strand sequencing. I have authored over 50 peer-reviewed publications with an h-index of 31 and over 5,798 citations.

### B. Positions, Scientific Appointments, and Honors

2023–Present Full Professor, University of Bari Aldo Moro, Italy  
2020–2023 Associate Professor, University of Bari Aldo Moro, Italy  
2012–2020 Assistant Professor, University of Bari Aldo Moro, Italy  
2007–2012 Postdoctoral Fellow, University of Washington, Seattle, USA

#### Honors:

2018 Awarded FFABR Research Funding, ANVUR  
2012 Finalist, ISSNAF Young Investigator Award  
2006 EMBO Short-Term Fellowship

### C. Contributions to Science

#### 1. Structural Variants and Human Disease

My research has uncovered key mechanisms by which structural variations—such as inversions and segmental duplications—contribute to human genetic diseases. I have focused on idiopathic infertility and neurodevelopmental disorders, applying cytogenomics and long-range sequencing to characterize complex chromosomal rearrangements. A core aspect of my work involves analyzing inversion polymorphisms and recurrent rearrangement hotspots, as well as identifying structural haplotypes that predispose to disease by promoting genomic instability.

- Paparella A et al., Int J Mol Sci. 2023; doi: 10.3390/ijms242115818.
- Porubsky D et al. Cell. 2022; doi: 10.1016/j.cell.2022.04.017.
- Maggiolini FAM et al., PLoS Genet. 2019; doi: 10.1371/journal.pgen.1008075.
- Antonacci F et al., Nat Genet. 2014; doi: 10.1038/ng.3120.
- Mefford HC et al., Am J Med Genet A. 2010; doi: 10.1002/ajmg.a.33557.
- Antonacci F et al., Nat Genet. 2010; doi: 10.1038/ng.643.
- Girirajan S et al., Nat Genet. 2010; doi: 10.1038/ng.534.
- Antonacci F, Hum Mol Genet. 2009; doi: 10.1093/hmg/ddp187.

## 2. Comparative Genomics and Primate Genome Evolution

I contributed to the sequencing and analysis of great ape genomes, providing insights into primate genome evolution and human-specific genes. My work has focused on identifying structural variation patterns across primate lineages, elucidating the mechanisms that drive genomic rearrangements, and understanding their evolutionary consequences. This includes the study of inversion polymorphisms, segmental duplications, and the emergence of novel gene families.

- Yoo D et al., Nature 2025; doi: 10.1038/s41586-025-08816-3.
- Zhang S et al., Nature 2025; doi: 10.1038/s41586-025-08596-w.
- Makova KD et al., Nature 2024; doi: 10.1038/s41586-024-07473-2.
- Mercuri L et al., Genome Res. 2022; doi:10.1101/gr.276960.122.
- Mao Y et al., Nature 2021; 10.1038/s41586-021-03519-x.
- Warren WC et al., Science 2020; doi: 10.1126/science.abc6617.
- Porubsky D et al., Nat Genet. 2020; doi: 10.1038/s41588-020-0646-x.
- Catacchio CR et al., Genome Res. 2018; doi: 10.1101/gr.234831.118.
- Dennis MY et al., Genome Biol. 2017; doi: 10.1186/s13059-017-1163-9.
- Nettle X et al., Nature. 2016; doi: 10.1038/nature19075
- Giannuzzi G et al., Genome Res. 2013; doi: 10.1101/gr.156240.113.
- Nettle X et al., Nat Methods. 2013; doi: 10.1038/nmeth.2572.
- Sudmant PH et al., Genome Res. 2013; doi: 10.1101/gr.158543.113.
- Steinberg KM\*, Antonacci F\* et al., Nat Genet. 2012; doi: 10.1038/ng.2335.
- Dennis MY et al., Cell. 2012; doi: 10.1016/j.cell.2012.03.033.
- Cellamare A et al., Mol Biol Evol. 2009; doi: 10.1093/molbev/msp101.
- Bekpen C et al., PLoS Genet. 2009; doi: 10.1371/journal.pgen.1000403.
- Zody MC et al., Nat Genet. 2008; doi: 10.1038/ng.193.
- Ventura M et al., Science. 2007; doi: 10.1126/science.1197005.

## 3. Genomic Technologies and Method Development

I have applied and optimized novel sequencing technologies to resolve complex genomic rearrangements in human and primate genomes.

- Maggolini FAM et al., Genome Res. 2020; doi: 10.1101/gr.265322.120.
- Eslami Rasekh M et al., BMC Genomics 2017; doi: 10.1186/s12864-016-3444-1.
- Chaisson MJP et al. Nature 2015. doi: 10.1038/nature13907.
- Huddleston J et al. Genome Res. 2014; doi: 10.1101/gr.168450.113.
- Sudmant PH et al., Science. 2010; doi:10.1126/science.1197005.
- Kidd JM et al., Nat Methods. 2010; doi: 10.1038/nmeth.2572.
- Alkan C et al., Nat Genet. 2009; doi: 10.1038/ng.437.
- Kidd JM et al., Nature. 2008; doi: 10.1038/nature06862.

## 4. Cytogenetics and Genomic Diagnostics

As coordinator of a Master's program in Clinical Cytogenomics, I integrated teaching and research in diagnostic cytogenetics, supervising clinical case studies and developing workflows for chromosomal disorder diagnosis.

- Montanari A et al., Mol Cytogenet. 2024; doi: 10.1186/s13039-024-00700-5.
- Riviello FN et al., Genes 2023. doi.org/10.3390/genes14122194.
- Cellamare A et al. Genes. 2021; doi: 10.3390/genes12060877.
- Buysse K\*, Antonacci F\* et al., Eur J Med Genet. 2009; doi: 10.1016/j.ejmg.2008.10.007.
- Buysse K et al., J Med Genet. 2008; doi: 10.1136/jmg.2008.058883.