

# BÁRBARA BITARELLO, PHD

Computational Biologist

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## SUMMARY

- Computational biologist with expertise in large-scale data, genomics, population genetics, statistical genetics, immunogenetics
- Strong record of collaborative work in academia and management of research projects, with 4 successful grant proposals
- Excellent communication skills and record of written communication and scientific writing, including 10 scientific papers
- Self-motivated, problem-solving scientist, with deep understanding of next-generation sequencing, genotype data, and biobanks
- Experienced speaker, with 3 talks and several poster presentations delivered to international scientific and stakeholder audiences
- Driven to use bioinformatics to gather and analyze human genetic data and advance genomic medicine and R&D

## TECHNICAL SKILLS

- **Programming:** R programming, Python, Bash/shell scripting, sed/awk
- **Genomics:** GWAS, linear modeling, polygenic risk score implementation, DNA sequence simulations, ancestry inference, gene ontology analyses, functional annotation
- **Genomic Data analysis:** Biobank data analyses, frequentist statistics, biostatistics
- **Software** Plink, Ldpred, MSMS, SLiM, Microsoft Word, Overleaf, RStudio, VSCode, bcftools, Hail, Apache Spark (beginner)
- **Reproducible Research:** GitHub, R Notebooks
- **Data Visualization:** ggplot2, flexdashboard, htmlwidgets
- **Languages:** English and Portuguese: native. French and Spanish: conversational.
- **Task and time management:** Quire, Slack

## RESEARCH EXPERIENCE

### University of Pennsylvania, Perelman School of Medicine

Postdoctoral Researcher, Department of Genetics

March 2018-present

- Led project to understand the drivers of polygenic risk prediction accuracy in admixed populations, resulting in 1 first author publication<sup>2</sup>
- Used big data from UK Biobank, 1000 Genomes, Women's Health Initiative, Health and Retirement Study and gnomAD
- Used findings to influence scientific and stakeholders audiences on the need for diversity in human genetics
- Performed ancestry inference, leveraged individual genetic ancestry into polygenic risk predictions
- Performed GWAS and linear modelling of polygenic traits
- Developed tools to correct for biases, showing the determinants of loss of prediction power across different ancestries
- Delivered 3 conference talks, 1 local talk, and 1 interactive poster
- Collaborated on a project looking for the genetic determinant of reproductive success performing tests to detect balancing selection signatures, resulting in one co-authorship<sup>1</sup>
- Developed a bioinformatics pipeline to derive polygenic risk scores using different databases and methods

### Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany

Postdoctoral Researcher, Department of Evolutionary Genetics

Oct 2016-Dec 2017

- Led and collaborated on projects to investigate signatures of balancing selection in human genomes, resulting in 2 co-authorship publications<sup>3,5</sup>
- Used 1000 Genomes data, ancient genomes, R data analysis, population genetics methods

### University of São Paulo, São Paulo, Brazil

Doctoral Researcher, Genetics and Evolutionary Biology Department

Aug 2011-Aug 2016

- Quantified the pervasiveness of balancing selection throughout human evolutionary history and its effects in neighboring regions in the genome, resulting in 1 first author paper<sup>4</sup>, 2 international conference posters, and 2 local talks
- Developed an innovative statistical method that has since been widely used<sup>4</sup>, leading to the discovery of new targets for functional validation
- Analyzed regulatory and evolutionary properties of genomic targets of selection
- MSMS software for balancing selection simulations, R programming and bash/linux for data analysis and visualization, ENSEMBL for genomic annotations, bcftools to analyze 1000 Genomes dataset, parallel computing in R
- Mentored 1 undergraduate student and guest lectured for 1 graduate course in genomics
- Performed phylogenetic analyses in PAML for Collaborative international research effort to assemble the genome of the kiwi<sup>7</sup>
- Performed data analysis on two projects on HLA genes' diversity and mapping bias, resulting in two co-authorships<sup>8,9</sup>

### University of São Paulo, São Paulo, Brazil

MSc Researcher, Genetics and Evolutionary Biology Department

March 2009 to Aug 2011

- Investigated the timescale of natural selection in human HLA genes, resulting in 1 first author paper<sup>6</sup> and 2 international conference poster presentations
- Phylogenetic analyses of HLA genes using PAML software, DNA sequence simulations, R programming and bash/linux to analyze and visualize the data
- Mentored one masters student working on HLA genes

- Engaged in scientific outreach resulting in 2 peer-reviewed publications in Portuguese

## State University of Campinas, Campinas, Brazil

Undergraduate Researcher, Department of Genetics

Aug 2005 to Dec 2007

- Developed genetic markers for the human botfly in order to understand its diversity in Brazil, resulting in one first author publication<sup>10</sup> and 2 conference posters
- Extracted DNA, designed primers, performed sanger sequencing, analyzed polyacrylamide gels, used statistical approaches to assess and compare genetic diversity in 2 populations

## EDUCATION

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- PhD, Biology (Genetics), University of São Paulo, Brazil, 2016
- MSc, Biology (Genetics), University of São Paulo, Brazil, 2011
- BS, Biological Sciences, State University of Campinas, Brazil, 2007

## AWARDS

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### Grants

- PhD Research Grant from São Paulo Research Foundation, 2011-2016
- PhD Research Grant for visiting period at the Max Planck Institute from São Paulo Research Foundation, 2013-2013
- Master's Research Grant from São Paulo Research Foundation, 2009-2011
- Undergraduate Research Grant from São Paulo Research Foundation, 2005-2007

### Prizes

- Best graduate student paper award for Genome Biology and Evolution (SMBE, 2019)
- Spotlight Trainee Paper, by the American Society of Human Genetics (ASHG, 2019)
- 1<sup>st</sup> place in for the graduate program, University of São Paulo, Department of Genetics and Evolutionary Biology (2009)

## CONFERENCE PRESENTATIONS (4 OUT OF 15)

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- **Bitarello, BD** Mathieson, I (2020) *Low transferability of height polygenic risk scores in admixed ancestry populations*. Contributed talk, NY Area Population Genomics, New York, NY.
- **Bitarello, BD** Mathieson, I (2019) *Investigating the lack of transferability of polygenic risk scores in cohorts with admixed ancestry*, Contributed talk at the American Society of Human Genetics Annual Meeting, Houston, TX.
- **Bitarello, BD** Mathieson, I (2019) *Polygenic risk scores perform poorly across populations*, Contributed talk at the American Society of Human Genetics Annual Meeting, San Diego, CA.
- **Bitarello, BD**; de Filippo, C; Andrés, A; Meyer, D (2015) *Balancing selection in humans: insights from a novel SFS-based method*. Poster session presented at the Annual Meeting of the Society for Molecular Biology and Evolution, Vienna, Austria.

## PUBLICATIONS & PREPRINTS

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- [1] Mathieson, I, Day, F, Barban, N, Tropf, F, Brazel, D, Vaez, A, Zuydam, N, **Bitarello, BD** et al. Genome-wide analysis identifies genetic effects on reproductive success and ongoing natural selection at the FADS locus. *BiorXiv*, 2020
- [2] **Bitarello, BD** Mathieson, I. Polygenic scores for height in admixed populations. *G3: Genes, Genomes, Genetics*, in press, 2020
- [3] Giner-Delgado, Villatoro, CS, Lerga-Jaso, J, Gayà-Vidal, M, Oliva, M, Castellano, D, Pantano, L, **Bitarello, BD** et al. Evolutionary and functional impact of common polymorphic inversions in the human genome. *Nature Communications*, 2019.
- [4] **Bitarello, BD** et. al. Signatures of long-term balancing selection in human genomes. *Genome Biology and Evolution*, 2018.
- [5] Meyer, D, Aguiar, V, **Bitarello, BD** et al. A genomic perspective on HLA evolution. *Immunogenetics*, 2017.
- [6] **Bitarello, BD** et al. Heterogeneity of dN/dS ratios at the classical HLA class I genes over divergence time and across the allelic phylogeny. *Journal of Molecular Evolution*, 2016
- [7] Le Duc, D, Renaud, G, Krishnan, A, Almén, MS, Huynen, L, Prohaska, SJ, Ongyerth, M **Bitarello, BD** et al. Kiwi genome provides insights into evolution of a nocturnal lifestyle. *Genome Biology*, 2015.
- [8] Brandt, D, Aguiar, V, **Bitarello, BD** et al. Mapping bias overestimates reference allele frequencies at the HLA genes in the 1000 genomes project phase I data. *G3: Genes, Genomes, Genetics*, 2015.
- [9] Francisco, RDS, Buhler, S, Nunes, JM, **Bitarello, BD** et al. HLA supertype variation across populations: new insights into the role of natural selection in the evolution of HLA-A and HLA-B polymorphisms. *Immunogenetics*, 2015.
- [10] **Bitarello, BD**, Torres, TT, Lyra, ML, Azeredo-Espin, AML. Development of polymorphic microsatellite markers for the human botfly, *Dermatobia hominis* (Diptera: Oestridae). *Molecular Ecology Resources*, 2009.

## VOLUNTEER EXPERIENCE

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- Abstract and grant reviewer for The National Diversity in STEM Conference, Society for the Advancement of Chicanos/Hispanics and Native Americans in Science, 2020
- Member of Diversity Committee of Biomedical Postdoctoral Council, University of Pennsylvania, 2020
- Reviewer of scientific manuscripts for peer-reviewed journals, 2016-present
- Consultant on ancient DNA for identification of forensic skeletal remains, 2015