Bárbara Domingues Bitarello

Email: bbita@upenn.edu
Website: bbitarello.github.io
LinkedIn: barbara-bitarello
GitHub: github.com/bbitarello
Twitter: @dudutchy

ACADEMIC EMPLOYMENT

University of Pennsylvania

Philadelphia, USA

Postdoctoral Researcher in the Department of Genetics, Perelman School of Medicine

Advisor: Iain Mathieson

2018-Current

Max Planck Institute for Evolutionary Anthropology

Leipzig, Germany

Postdoctoral Researcher in the Department of Evolutionary Genetics

Advisor: Aida Andrés 2016–2017

EDUCATION

University of São Paulo

São Paulo, Brazil

Ph.D. in Biology (Genetics), Department of Genetics and Evolutionary Biology

Advisor: Diogo Meyer

2011-2016

- Thesis: "Balancing selection in the human genome: biological relevance and deleterious consequence"
- Visiting graduate student at Dr. Aida Andrés'group at the Max Planck Institute for Evolutionary Anthropology, 2013

University of São Paulo

São Paulo, Brazil

M.Sc. in Biology (Genetics), Department of Genetics and Evolutionary Biology

Advisor: Diogo Meyer

2009-2011

– Thesis: "Natural selection on HLA genes: a molecular investigation of the location and timing of selection events"

University of Campinas

Campinas, Brazil

B.Sc. and Licenciature in Biological Sciences

2004-2007

– Thesis: "Development of polymorphic microsatellite markers for the human botfly, Dermatobia hominis (Diptera: oestridae)"

KEY AREAS OF EXPERTISE

• Population Genetics

• Adaptive Evolution

• Comparative Genomics

• Human Genomics

• Balancing Selection

Immunogenetics

Bárbara D. Bitarello Page 1 of 5

^{*} In Brazil, a Licentiate degree in Biology includes contents in the areas of Chemistry, Physics and Health, to attend elementary and secondary education, as well as theoretical and supervised classroom experience in Pedagogical training. https://en.wikipedia.org/wiki/Licentiate(degree)#Brazil.

Teaching and Mentoring

- Teaching Assistant at R/Med

 Intro to R for clinicians (https://events.linuxfoundation.org/r-medicine/).
- Guest Lecture at University of São Paulo

 2015

 Population Genetics, graduate course (Lecture given: "Simulating neutral sequences").
- Co-supervisor undergraduate student at University of São Paulo

 The project was part of my Ph.D. project, namely analyzing the deleterious consequences of balancing selection in neighboring regions in the genome.
- Co-supervisor undergraduate student at University of São Paulo

 The project was part of my M.Sc. project, namely analyzing the evolutionary rates of HLA genes through phylogenetic methods. The student is now a graduate student at Rasmus Nielsen's lab at UC Berkeley.
- English Tutoring, self-employed

 I worked as an English tutor in Brazil, mostly focusing on students that needed to pass graduate program

 English examinations

Computational & Quantitative Skills

- Programming languages: R, Linux, Bash scripting, sed/awk, LATEX, markdown, Perl, Python (beginner)
- Software: ldpred, plink, Hail, MSMS, SLiM, PAML, Rstudio, bcftools, vcftools, bedtools
- Data Science: Machine Learning, Linear Modeling, Data Gathering, cleaning, exploration, visualization, reproducible research, documented code
- Statistics: Statistical Genetics, Biostatistics, Probability, MCMC
- Other: HPC, Cloud computing, parallel programming

LANGUAGES

- Portuguese: Native
- English: Native
- French: Intermediate Spanish: Intermediate

SCHOLARSHIPS AND AWARDS

- Best Graduate Student Paper on Genome Biology and Evolution [4], by the Society for Molecular Biology and Evolution, 2019
- Spotlight Trainee Paper [4], by the American Society of Human Genetics, 2019
- Ph.D. Student scholarship grant, São Paulo Research Foundation, Advisor: Diogo Meyer, Grant # 11/12500-2, 2011-2016
- Scholarship for visiting graduate student at the Max Planck Institute for Evolutionary Anthropology, São Paulo Research Foundation, Advisors: Diogo Meyer and Aida Andrés, Grant # 12/19563-2, 2013–2014

Bárbara D. Bitarello Page 2 of 5

- Scholarship to attend the Summer Institute for Statistical Genetics in Seattle, USA, 2012
- Master's Student Scholarship, São Paulo Research Foundation, Advisor: Diogo Meyer, Grant # 08/56502-6, 2009 -2011
- Entrance examination for the graduate program of the Department of Genetics and Evolutionary Biology of the University of São Paulo (1st place), 2009
- Undergraduate research scholarship from São Paulo Research Foundation, Advisor: Ana Maria Lima de Azeredo-Espin, Grant # 06/50793-3, 2006 –2008

SCIENTIFIC OUTREACH

• Guest speaker at the "Bioinformatics Chat" podcast Fall 2020 (upcoming)

I will talk about polygenic risk scores and why diversity matters for their accuracy, focusing on my most recent work. https://bioinformatics.chat

• My (mostly) science blog

I write personal essays, as well as tutorials and short articles about scientific concepts and publications.

https://bbitarello.github.io/post/

Ongoing Projects

See more on https://bbitarello.github.io/project as well as in my research statement

What drives reduced prediction accuracy of polygenic scores in non-European individuals?

Most genome-wide studies are carried out in Europeans. Several determinants are involved in calculating a meaningful polygenic risk score for other ancestries. The complex interplay of many factors that determine prediction accuracy across ancestries is the focus of this project.

Balancing Selection: methods, signatures, mechanisms

Balancing selection is an evolutionary force that maintains adaptive diversity in populations. It is an umbrella term that encompasses an array of mechanisms. Developing methods to detect its signature in genomes and well as understanding its impacts in humans and other species is the focus of this project.

SERVICE

• Brazilian Effort on Multifactorial Inheritance (BEMi)

2020-Current

This initiative aims at building collaborations between Brazilian researchers to gather data, methods, and analyses that address the genomics of multi-factorial diseases/conditions. The main goal is to develop population genomics projects in Brazilians.

Roles: Founding Member and Member of Steering Committee

• Member of SACNAS

2020-Current

SACNAS stands for Society for Advancement of Chicanos/Hispanics Native Americans in Science Roles: Review abstracts and scholarships and mentor younger scientists.

• Member of BPP Diversity Committee

2020-Current

Bárbara D. Bitarello Page 3 of 5

BPP stands for Biomedical Postdoctoral Council at the University of Pennsylvania

Roles: Advocating for diversity and inclusion for postdocs at the Biomedical Postdoctoral program at UPenn, and mentoring URM (underrepresented minority) graduate and undergraduate students.

• Ad hoc abstract reviewer

Biomedical Postdoctoral Council Research Symposium, University of Pennsylvania.

• Ad hoc manuscript reviewer

Genome Biology and Evolution, Human Immunology, Scientific Reports, Molecular Ecology Resources, Peer Community in Evolutionary Biology.

SELECTED CONFERENCE PRESENTATIONS

- Bitarello, BD. Ancestry Matters: why do polygenic risk scores remain limited to a few?, Annual Meeting of the Association of Genomic Diagnostics, Bonn, Germany, 2020 (upcoming) (Invited Talk)
- Bitarello, BD. What drives the reduced prediction accuracy of polygenic risk scores in non-European individuals?, Center for Theoretical and Evolutionary Genomics Seminar Series, Fall 2020 (Invited Talk)
- Bitarello, BD; Mathieson, I. Loss of predictive power of polygenic risk scores in admixed populations, The Allied Genetics Conference, Online, 2020 ([Interactive Poster]https://genetics-gsa.org/tagc-2020/
- Bitarello, BD; Mathieson, I. Low transferability of height polygenic risk scores in admixed ancestry populations, New York Area Population Genomics, New York, USA, 2020 (Contributed Talk)
- Bitarello, BD; Mathieson, I. Investigating the lack of transferability of polygenic risk scores in cohorts with admixed ancestry, Annual American Society of Human Genetics Meeting, Houston, TX, 2019 (Contributed Talk)
- Bitarello, BD; Mathieson, I. Polygenic risk scores perform poorly across populations, Annual American Society of Human Genetics Meeting, San Diego, USA, 2018 (Contributed Talk)
- Bitarello, BD; de Filippo, C; Andrés, A; Meyer, D. Balancing selection in humans: insights from a novel SFS-based method, Annual Meeting of the Society for Molecular Biology and Evolution, Vienna, Austria, 2015 (Poster)
- Bitarello, BD; de Filippo, C; Andrés, A; Meyer, D. NCV: A site frequency spectrum based method to detect balancing selection in humans, Annual Meeting of the Society for Molecular Biology and Evolution, San Juan, Puerto Rico, 2014 (Poster)
- Bitarello, BD; Meyer, D. Variation of dN/dS ratios at HLA genes over time and functional classes, Annual Meeting of the Society for Molecular Biology and Evolution, Chicago, USA, 2013 (Poster)
- Bitarello, BD; Meyer, D. Time dependence of dN/dS measures at HLA loci under balancing selection, Annual Meeting of the Society for Molecular Biology and Evolution, Lyon, France, 2010 (Poster)

Publications & Preprints

- [1] Mathieson, I., Day, F. R., Barban, N., Tropf, F. C., Brazel, D. M., Consortium, e., Consortium, B., Vaez, A., Zuydam, N., **Bitarello, B. D.**, Snieder, H., Hoed, M., Ong, K. K., Mills, M. C., Perry, J. R., "Genome-wide analysis identifies genetic effects on reproductive success and ongoing natural selection at the FADS locus", bioRxiv, 2020. DOI: 10.1101/2020.05.19.104455.
- [2] **Bitarello, B. D.**, Mathieson, I., "Polygenic scores for height in admixed populations", G3: Genes, Genomes, Genetics, g3.401658.2020, 2020. DOI: 10.1534/g3.120.401658.

Bárbara D. Bitarello Page 4 of 5

- [3] Giner-Delgado, C., Villatoro, S., Lerga-Jaso, J., Gayà-Vidal, M., Oliva, M., Castellano, D., Pantano, L., **Bitarello, B. D.**, Izquierdo, D., Noguera, I., Olalde, I., Delprat, A., Blancher, A., Lalueza-Fox, C., Esko, T., O'Reilly, P. F., Andrés, A. M., Ferretti, L., Puig, M., Cáceres, M., "Evolutionary and functional impact of common polymorphic inversions in the human genome", *Nature Communications*, vol. 10, no. 1, p. 4222, Dec. 2019. DOI: 10.1038/s41467-019-12173-x.
- [4] **Bitarello, B. D.**, Filippo, C., Teixeira, J. C., Schmidt, J. M., Kleinert, P., Meyer, D., Andrés, A. M., "Signatures of Long-Term Balancing Selection in Human Genomes", *Genome Biology and Evolution*, vol. 10, no. 3, pp. 939–955, Mar. 2018. DOI: 10.1093/gbe/evy054.
- [5] **Bitarello, B. D.**, Francisco, R. d. S., Meyer, D., "Heterogeneity of dN/dS Ratios at the Classical HLA Class I Genes over Divergence Time and Across the Allelic Phylogeny", *Journal of Molecular Evolution*, vol. 82, no. 1, pp. 38–50, Mar. 2016. DOI: 10.1007/s00239-015-9713-9.
- [6] Brandt, D. Y., Aguiar, V. R., **Bitarello, B. D.**, Nunes, K., Goudet, J., Meyer, D., "Mapping bias overestimates reference allele frequencies at the HLA genes in the 1000 genomes project phase I data", *G3: Genes, Genomes, Genetics*, vol. 5, no. 5, pp. 931–941, Mar. 2015. DOI: 10.1534/g3.114.015784.
- [7] dos Santos Francisco, R., Buhler, S., Nunes, J. M., **Bitarello, B. D.**, França, G. S., Meyer, D., Sanchez-Mazas, A., "HLA supertype variation across populations: new insights into the role of natural selection in the evolution of HLA-A and HLA-B polymorphisms", *Immunogenetics*, vol. 67, no. 11-12, pp. 651–663, Mar. 2015. DOI: 10.1007/s00251-015-0875-9.
- [8] Le Duc, D., Renaud, G., Krishnan, A., Almén, M. S., Huynen, L., Prohaska, S. J., Ongyerth, M., Bitarello, B. D., Schiöth, H. B., Hofreiter, M., Stadler, P. F., Prüfer, K., Lambert, D., Kelso, J., Schöneberg, T., "Kiwi genome provides insights into evolution of a nocturnal lifestyle", Genome Biology, vol. 16, no. 1, pp. 651–663, Mar. 2015. DOI: 10.1186/s13059-015-0711-4.
- [9] **Bitarello, B. D.**, Torres, T. T., Lyra, M. L., De Azeredo-Espin, A. M. L., "Development of polymorphic microsatellite markers for the human botfly, Dermatobia hominis (Diptera: Oestridae)", *Molecular Ecology Resources*, vol. 9, no. 1, pp. 409–411, Mar. 2009. DOI: 10.1111/j.1755-0998.2008.02454.x.

Review Articles

[10] Meyer, D., C. Aguiar, V., **Bitarello, B. D.**, C. Brandt, D., Nunes, K., "A genomic perspective on HLA evolution", *Immunogenetics*, 2017. DOI: 10.1007/s00251-017-1017-3.

Peer-reviewed Outreach Articles (in Portuguese)

- [11] **Bitarello, B. D.**, "Projeto Genográfico e as implicações da popularização dos estudos de genealogia gênica [The Genographic Project and the implications of the popularization of Gene Genealogy Studies]", Revista da Biologia, vol. 3, pp. 21–23, 2009.
- [12] **Bitarello, B. D.**, Meyer, D., "Intercruzmento de humanos modernos e neandertais: novas perspectivas a luz da genética [Interbreeding of modern humans with Neanderthals: new perspectives in the light of genetics]", Revista da Biologia, vol. 6, pp. 6–9, 2011.

Bárbara D. Bitarello Page 5 of 5