

# Bárbara Domingues Bitarello

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## ACADEMIC EMPLOYMENT

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<b>University of Pennsylvania</b> Postdoctoral Researcher in the Department of Genetics, Perelman School of Medicine Advisor: Iain Mathieson	Philadelphia, USA 2018–Current
<b>Max Planck Institute for Evolutionary Anthropology</b> Postdoctoral Researcher in the Department of Evolutionary Genetics Advisor: Aida Andrés	Leipzig, Germany 2016–2017

## EDUCATION

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<b>University of São Paulo</b> Ph.D. in Biology (Genetics), Department of Genetics and Evolutionary Biology Advisor: Diogo Meyer	São Paulo, Brazil 2011–2016
<ul style="list-style-type: none"><li>– Thesis: “Balancing selection in the human genome: biological relevance and deleterious consequence”</li><li>– Visiting graduate student at Dr. Aida Andrés’group at the Max Planck Institute for Evolutionary Anthropology, 2013</li></ul>	
<b>University of São Paulo</b> M.Sc. in Biology (Genetics), Department of Genetics and Evolutionary Biology Advisor: Diogo Meyer	São Paulo, Brazil 2009–2011
<ul style="list-style-type: none"><li>– Thesis: “Natural selection on HLA genes: a molecular investigation of the location and timing of selection events”</li></ul>	
<b>University of Campinas</b> B.Sc. and Licenciature* in Biological Sciences	Campinas, Brazil 2004–2007
<ul style="list-style-type: none"><li>– Thesis: “Development of polymorphic microsatellite markers for the human botfly, <i>Dermatobia hominis</i> (Diptera: oestridae)”</li></ul>	

## KEY AREAS OF EXPERTISE

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- Population Genetics
- Comparative Genomics
- Balancing Selection
- Adaptive Evolution
- Human Genomics
- Immunogenetics

\* In Brazil, a Licenciature degree in Biology includes contents in the areas of Chemistry, Physics and Health, to attend elementary and secondary education, as well as pedagogical training, both theoretical (360 hours) and practical (**56 hours of supervised classroom experience**). [https://en.wikipedia.org/wiki/Licentiate\(degree\)#Brazil](https://en.wikipedia.org/wiki/Licentiate(degree)#Brazil).

## TEACHING AND MENTORING

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- **Teaching Assistant** at R/Med 2020  
*Intro to R for clinicians (<https://events.linuxfoundation.org/r-medicine/>). Length: 4 hours.*
- **Guest Lecturer** at University of São Paulo 2015  
*Population Genetics, graduate course (Lecture given: “Simulating neutral sequences”). Length: 4 hours.*
- **Co-supervisor of Caroline Simões (undergraduate student)** at University of São Paulo 2015  
*I supervised this student in getting some hands-on experience. We looked at data from my PhD project. Caroline is graduating in Physics and is applying to grad school in Physics. Length: one semester.*
- **Co-supervisor Débora Y.C Brandt (undergraduate student)** at University of São Paulo 2012  
*When Débora joined the lab towards the end of her undergraduate degree, I supervised her in getting to know genetics data, R programming and Linux/bash commands. She then pursued a Master’s in our lab and is now a graduate student at Rasmus Nielsen’s lab at UC Berkeley. Length: one semester*
- **English Tutoring**, self-employed 2008  
*I worked as an English tutor in Brazil, mostly focusing on students that needed to pass graduate program English examinations. Length: one semester*
- **Supervised classroom experience**, Elementary School in Campinas, Brazil 2007  
*I prepared and delivered Science classes to fifth-graders under the supervision of the Science teacher. Length: 56 hours (one school semester).*

## COMPUTATIONAL & QUANTITATIVE SKILLS

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- **Programming languages:** R, Linux, Bash scripting, sed/awk, L<sup>A</sup>T<sub>E</sub>X, 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

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

## SCHOLARSHIPS AND AWARDS

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- 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
- 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 (1<sup>st</sup> 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

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- 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 2019–Current  
*I write personal essays, as well as tutorials and short articles about scientific concepts and publications. <https://bbitarelo.github.io/post/>*

## ONGOING PROJECTS

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See more on <https://bbitarelo.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

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- **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 address under-representation of non-Europeans and ultimately reduce health disparities in precision medicine.*  
 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  
*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** 2019  
*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

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- **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**)
- **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**)

## PUBLICATIONS & PREPRINTS

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- [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.
- [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., “Intercruzamento 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.