

Bárbara D. Bitarello

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EDUCATION

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|--|-----------------------------------|
| University of Pennsylvania
Postdoctoral Researcher in genetics, Advisor: Iain Mathieson | Philadelphia, USA
2018–Current |
| Max Planck Institute for Evolutionary Anthropology
Postdoctoral Researcher in genetics, Advisor: Aida Andrés | Leipzig, Germany
2016–2017 |
| University of São Paulo
Ph.D. in Biology(Genetics), Advisor: Diogo Meyer | São Paulo, Brazil
2011–2016 |
| – Thesis: “Balancing selection in the human genome: biological relevance and deleterious consequence” | |
| University of São Paulo
M.Sc. in Biology(Genetics), Advisor: Diogo Meyer | São Paulo, Brazil
2009–2011 |
| – Thesis: “Natural selection on HLA genes: a molecular investigation of the location and timing of selection events” | |
| University of Campinas
B.Sc. in Biological Sciences | Campinas, Brazil
2004–2007 |
| – Thesis: “Development of polymorphic microsatellite markers for the human botfly, <i>Dermatobia hominis</i> (Diptera: oestridae)” | |

RESEARCH GOALS

My main goal is to understand human evolution through the perspective of population and medical genetics by exploring the growing number genomes available, and developing better methods to make sense of these data. For this goal, it is important to understand the complex interplay of different evolutionary and demographic processes shaping human diversity, as well as the non-genetic factors that influence human phenotypes, both of which require a solid foundation in population and evolutionary genetics, computational biology, and productive collaborations with researchers from other fields. This research goal cannot truly be achieved unless human diversity at large is understood. A related research goal is to understand how ancestry affects inferences that have historic been linked to European ancestry only, in order to make the inevitable advances of genomic medicine applicable to people of all ancestries. Finally, on a pedagogical level, my goals are to teach evolutionary biology, statistical genetics, computational biology/bioinformatics, the ethics and history of the study of human genetics, and have a prolific scientific communication/outreach presence online as well as in my communities.

- [1] B. D. Bitarello and I. Mathieson, “Polygenic scores for height in admixed populations”, *bioRxiv*, p. 2020.04.08.030361, 2020. DOI: 10.1101/2020.04.08.030361.
- [2] I. Mathieson, F. R. Day, N. Barban, F. C. Troup, D. M. Brazel, E. Consortium, B. Consortium, A. Vaez, N. van Zuydam, B. D. Bitarello, H. Snieder, M. den Hoed, K. K. Ong, M. C. Mills, J. R. Perry, and o. b. o. t. H. R. B. Consortium, “Genome-wide analysis identifies genetic effects on reproductive success and ongoing natural selection at the FADS locus”, *bioRxiv*, p. 2020.05.19.104455, 2020. DOI: 10.1101/2020.05.19.104455.
- [3] C. Giner-Delgado, S. Villatoro, J. Lerga-Jaso, M. Gayà-Vidal, M. Oliva, D. Castellano, L. Pantano, B. D. Bitarello, D. Izquierdo, I. Noguera, I. Olalde, A. Delprat, A. Blancher, C. Lalueza-Fox, T. Esko, P. F. O’Reilly, A. M. Andrés, L. Ferretti, M. Puig, and M. Cáceres, “Evolutionary and functional impact of common polymorphic inversions in the human genome”, *Nature Communications*, vol. 10, no. 1, p. 4222, Dec. 2019, ISSN: 2041-1723. DOI: 10.1038/s41467-019-12173-x.
- [4] B. D. Bitarello, C. de Filippo, J. C. Teixeira, J. M. Schmidt, P. Kleinert, D. Meyer, and A. M. Andrés, “Signatures of Long-Term Balancing Selection in Human Genomes”, *Genome Biology and Evolution*, vol. 10, no. 3, P. Majumder, Ed., pp. 939–955, Mar. 2018, ISSN: 1759-6653. DOI: 10.1093/gbe/evy054.
- [5] D. Meyer, V. C. Aguiar, B. Bitarello, D. C. Brandt, and K. Nunes, “A genomic perspective on HLA evolution”, *Immunogenetics*, 2017, ISSN: 14321211. DOI: 10.1007/s00251-017-1017-3.
- [6] B. D. Bitarello, R. d. S. Francisco, and D. Meyer, “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, 2016, ISSN: 00222844. DOI: 10.1007/s00239-015-9713-9.
- [7] D. Y. Brandt, V. R. Aguiar, B. D. Bitarello, K. Nunes, J. Goudet, and D. Meyer, “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, 2015, ISSN: 21601836. DOI: 10.1534/g3.114.015784.
- [8] R. dos Santos Francisco, S. Buhler, J. M. Nunes, B. D. Bitarello, G. S. França, D. Meyer, and A. Sanchez-Mazas, “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, 2015, ISSN: 14321211. DOI: 10.1007/s00251-015-0875-9.
- [9] D. Le Duc, G. Renaud, A. Krishnan, M. S. Almén, L. Huynen, S. J. Prohaska, M. Ongyerth, B. D. Bitarello, H. B. Schiöth, M. Hofreiter, P. F. Stadler, K. Prüfer, D. Lambert, J. Kelso, and T. Schöneberg, “Kiwi genome provides insights into evolution of a nocturnal lifestyle”, *Genome Biology*, vol. 16, no. 1, 2015, ISSN: 1474760X. DOI: 10.1186/s13059-015-0711-4.
- [10] B. D. Bitarello, T. T. Torres, M. L. Lyra, and A. M. L. De Azeredo-Espin, “Development of polymorphic microsatellite markers for the human botfly, *Dermatobia hominis* (Diptera: Oestridae)”, *Molecular Ecology Resources*, vol. 9, no. 1, pp. 409–411, 2009, ISSN: 1755098X. DOI: 10.1111/j.1755-0998.2008.02454.x.

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;** 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**)

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, 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 grant 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 Grant, 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 scholarship research grant from São Paulo Research Foundation, Advisor: Ana Maria Lima de Azeredo-Espin, Grant # 06/50793-3, 2006 –2008

COMPUTATIONAL 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
- **Other:** HPC, Cloud computing, parallel programming

LANGUAGES

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

ONGOING PROJECTS

See full list of projects on bbitareлло.github.io/project/

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

Many different steps are involved in calculating a polygenic risk score that is meaningful. The interplay of the complex array of factors that determine prediction accuracy across ancestries is the focus of this project.

Balancing Selection: methods, signatures, mechanisms

I first started working with balancing selection in graduate school. Co-supervised by Aida Andrés and Diogo Meyer, and with several collaborators, we created a new method tailored to detect signatures of long-term balancing selection[4], which has since been implemented by researchers studying human and non-human organisms, and at least one modification of our original method has been developed.

TEACHING AND MENTORING

- **Volunteer Teaching Assistant** at R/Med Summer 2020
Intro to R for clinicians (<https://events.linuxfoundation.org/r-medicine/>)
- **Co-supervised undergraduate student** at University of São Paulo 2014
The project was part of my Ph.D project, namely analysing the deleterious consequences of balancing selection in neighboring regions in the genome.
- **Co-supervised undergraduate student** at University of São Paulo 2012
The project was part of my MS.C project, namely analysing the evolutionary rates of HLA genes through phylogenetic methods. The student is now a graduate student at Rasmus Nielsen's lab at UC Berkeley
- **Guest Lecture** at University of São Paulo 2015
Population Genetics, graduate course (Lecture given: "Simulating neutral sequences")

OUTREACH ACTIVITIES

- Guest speaker at the "Bioinformatics Chat" podcast Fall 2020 (upcomming)

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

- Member of Society for Advancement of Chicanos/Hispanics Native Americans in Science (SACNAS) 2020–Current
Volunteer reviewer of abstracts and travel scholarships for The National Diversity in STEM Virtual Conference, 2020
- My (mostly) science blog 2019–Current
I write personal essays, as well as tutorials and short articles about scientific concepts and publications. <https://bbitarello.github.io/post/>

OUTREACH PUBLICATIONS (IN PORTUGUESE)

- [11] B. Bitarello, “Projeto Genográfico e as implicações da popularização dos estudos de genealogia gênica”, *Revista da Biologia*, vol. 3, pp. 21–23, 2009.
- [12] B. Bitarello and D. Meyer, “Intercruzamento de humanos modernos e neandertais: novas perspectivas a luz da genética”, *Revista da Biologia*, vol. 6, pp. 6–9, 2011.