Bárbara Domingues Bitarello, Ph.D

Assistant Professor | Bryn Mawr College

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APPOINTMENTS

Department of Biology, Bryn Mawr College

07/2021-Present

Assistant Professor

Perelman School of Medicine, University of Pennsylvania

03/2018 - 06/2021

Postdoctoral Researcher, Department of Genetics

Advisor: Iain Mathieson

Main project: Loss of accuracy in polygenic risk score in admixed populations

Max Planck Institute for Evolutionary Anthropology

10/2016 - 12/2017

Postdoctoral Researcher, Department of Evolutionary Genetics

Advisor: Aida Andrés

Main project: Adaptive evolution in the human lineage

EDUCATION

Ph.D. in Biology (Genetics)

08/2016

Department of Genetics and Evolutionary Biology, University of São Paulo, São Paulo, Brazil

Advisor: Diogo Meyer

M.Sc. in Biology (Genetics)

08/2011

Department of Genetics and Evolutionary Biology, University of São Paulo, São Paulo, Brazil

B.Sc. in Biological Sciences

12/2007

University of Campinas, Campinas, Brazil

Licentiate Degree in Biological Sciences: teaching training for Elementary school

RESEARCH & TEACHING INTERESTS

Research interests

Human genetics, population genetics, population genomics, genomic medicine, predictive modeling, statistical genetics, genetic epidemiology, immunogenetics, evolutionary genetics, neuropsychiatric genetics, adaptive evolution, comparative genomics.

Teaching interests

Evolutionary biology, comparative genomics, phylogenetics, statistical genetics, history of human genetics, computational biology, population genetics, biostatistics, introduction to genetics, introduction to evolutionary biology, machine learning, programming with R.

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Publications & Preprints

1. Predicting skeletal stature using ancient DNA

Cox, SL; Moots, H; Stock, JT; Shbat, A; **Bitarello, BD**; Haak, W; Rosenstock, E; Ruff, CB; Mathieson, I bioRxiv, 2021

2. Genome-wide analysis identifies genetic effects on reproductive success and ongoing natural selection at the FADS locus.

Mathieson, I; Day, FR; Barban, N; Tropf, FC; Brazel, DM; eQTLGen Consortium; BIOS Consortium; Vaez, A; Zuydam, N; **Bitarello, BD**; Snieder, H; Hoed, M; Ong, KK; Mills, MC; Perry, JR bioRxiv, 2020

3. Polygenic scores for height in admixed populations.

Bitarello, BD; Mathieson, I G3:Genes, Genomes, Genetics, 2020

4. Evolutionary and functional impact of common polymorphic inversions in the human genome.

Giner-Delgado, C; Villatoro, S; Lerga-Jaso, J; Gayà-Vidal, M; Oliva, M; Castellano, D; Pantano, L; Bitarello, BD et al. (12 additional authors)

Nature Communications, 2019

5. Signatures of Long-Term Balancing Selection in Human Genomes. Bitarello, BD; Filippo, C; Teixeira, JC; Schmidt, JM; Kleinert, P; Meyer, D; Andrés, AM

Genome Biology and Evolution, 2018

6. Heterogeneity of dN/dS Ratios at the Classical HLA Class I Genes over Divergence Time and Across the Allelic Phylogeny.

Bitarello, BD; Francisco, RS; Meyer, D Journal of Molecular Evolution, 2016

7. Mapping bias overestimates reference allele frequencies at the HLA genes in the 1000 genomes project phase I data.

Brandt, DY; Aguiar, VR; **Bitarello, BD**; Nunes, K; Goudet, J; Meyer, DG3: Genes, Genomes, Genetics, 2015

8. HLA supertype variation across populations: new insights into the role of natural selection in the evolution of HLA-A and HLA-B polymorphisms.

Francisco, RDS; Buhler, S; Nunes, JM; **Bitarello, BD**; França, GS; Meyer, D; Sanchez-Mazas, A Immunogenetics, 2015

9. Kiwi genome provides insights into evolution of a nocturnal lifestyle.

Le Duc, D; Renaud, G; Krishnan, A; Almén, MS; Huynen, L; Prohaska, SJ; Ongyerth, M; **Bitarello, BD** et al. 7 additional authors Genome Biology, 2015

10. Development of polymorphic microsatellite markers for the human botfly, Dermatobia hominis (Diptera: Oestridae).

Bitarello, BD; Torres, TT; Lyra, ML; Azeredo-Espin, AML

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Review & Opinion Articles

- A genomic perspective on HLA evolution.
 Meyer, D, Aguiar, VRC, Bitarello, BD, Brandt, D, Nunes, K Immunogenetics, 2017.
- 11. 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]

 Bitarello, BD; Meyer, D

 Revista da Biologia, 2011.
- 12. 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]

 Bitarello, BD

 Revista da Biologia, 2009.

OTHER RESEARCH EXPERIENCE

University of São Paulo, São Paulo, Brazil

08/2011-09/2016

Graduate Researcher, Department of Genetics and Evolutionary Biology

Advisor: Diogo Meyer

Ph.D. thesis: "Balancing selection in the human genome: biological relevance and deleterious consequence", 2016

Visiting graduate, Planck Institute for Evolutionary Anthropology, 2013

Master's Thesis: "Natural Selection on HLA genes: a molecular investigation of the location and timing of selection events", 2011

University of Campinas, Campinas, Brazil

09/2005-12/2007

Research Assistant, Department of Genetics

Advisor: Ana Maria Lima de Azeredo-Espin

Main project: "Development of polymorphic microsatellite markers for the human botfly, Dermatobia hominis (Diptera: Oestridae)"

Teaching & Mentoring

University of Pennsylvania

Mentoring of undergraduate researcher

Nina Tansey, 03/2021–06/2021

Children's Hospital of Philadelphia

Teaching Assistant, 08/2020

Mini course: Intro to R for clinicians. Length: 4 hours.

University of São Paulo

Mentoring of undergraduate and Master's students, 2012-2017

Caroline Simões: We looked at data from my Ph.D. project. Caroline majored in Physics and is applying for grad school. 11/2014–04/2016

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Débora Brandt: We explored HLA gene diversity in public databases. She pursued a Master's degree in our lab and is now a graduate student at UC Berkeley. 09/2011–05/2015

Luiz Carlos Machado de Oliveira Junior: We explored the hitchiking of deleterious variants in regions under positive selection in human genomes as his Master's project in our lab. He is now applying for grad school in the US. 06/2014 –Present

Guest lecturer, 04/2015

Graduate course in Evolutionary Genomics. I taught neutral coalescent simulations. Length: 4 hours

Elementary School in Campinas, Brazil

Supervised classroom experience, 2007

I prepared and delivered Science classes to fifth-graders under supervision. Length: 56 hours.

Invited talks & Contributed conference presentations

"Increased local genetic load as a result of selective sweeps in human populations", Society for Molecular Biology and Evolution, Online, **Poster**, 07/2021

"Investigating the lack of transferability of polygenic risk scores in cohorts with admixed ancestry", Department of Genetics and Evolutionary Biology, University of São Paulo, **Invited Talk**, 06/2021

"Investigating the lack of transferability of PRS in cohorts with mixed ancestry", Annual Meeting of the Association of Genomic Diagnostics, Bonn, Germany, Invited Talk, 12/2020

What drives the reduced prediction accuracy of polygenic risk scores in non-European individuals?, Center for Theoretical and Evolutionary Genomics Seminar Series, Berkeley, USA, **Invited Talk**, 09/2020

Loss of predictive power of polygenic risk scores in admixed populations, The Allied Genetics Conference, Online, Poster, 05/2020

Low transferability of height polygenic risk scores in admixed ancestry populations, New York Area Population Genomics, New York, USA, Contributed Talk, 01/2020

Investigating the lack of transferability of polygenic risk scores in cohorts with admixed ancestry, Annual American Society of Human Genetics Meeting, Houston, USA, Contributed Talk, 10/2019

Polygenic risk scores perform poorly across populations, Annual American Society of Human Genetics Meeting, San Diego, USA Contributed Talk, 10/2018

Balancing selection in humans: insights from a novel SFS-based method, Annual Meeting of the Society for Molecular Biology and Evolution, Vienna, Austria, **Poster**, 07/2015

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, **Poster**, 06/2014

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, Poster, 07/013

Time dependence of dN/dS measures at HLA loci under balancing selection, Annual Meeting of the Society for Molecular Biology and Evolution, Lyon, France, **Poster**, 07/2010

SCHOLARSHIPS & AWARDS

Best Graduate Student Paper, Genome Biology and Evolution [4], Society for Molecular Biology and Evolution, \$2,000, 07/2019.

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Spotlight Trainee Paper [4], American Society of Human Genetics, 05/2019.

Ph.D. scholarship grant, São Paulo Research Foundation, Grant # 11/12500-2, 10/2011-04/2016

Visiting graduate student scholarship grant, São Paulo Research Foundation, Grant # 12/19563-2, 02/2013-01/2014

Scholarship to attend the Summer Institute for Statistical Genetics in Seattle, USA, 07/2012

Master's Student Scholarship, São Paulo Research Foundation, Grant # 08/56502-6, 03/2009-02/2011.

 $1^{\rm st}$ place in entrance examination for the graduate program, Department of Genetics and Evolutionary Biology, University of São Paulo, 12/2009

Undergraduate research scholarship from São Paulo Research Foundation, Grant # 06/50793-3, 09/2006-12/2007

SCIENTIFIC OUTREACH

Guest speaker 03/2021

"Bioinformatics Chat" podcast

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

Public seminar 06/2021

Department of Genetics and Evolutionary Biology, University of São Paulo
I presented my work on polygenic risk scores to graduate students and professors. The lecture (in Portuguese) was recorded and made public here: https://youtu.be/Tdiqs1p40cM

Opinion peer-reviewed articles in Portuguese

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] Bitarello, BD; Meyer, D; Revista da Biologia, 2011.

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] **Bitarello, BD**; Revista da Biologia, 2009.

ACADEMIC, PROFESSIONAL & COMMUNITY SERVICE

LatinGenomes: Latin American Alliance for Genomic Diversity 09/2020–Present Goal: to design and implement a network of cohorts and biobanks focused on and led my Latin American scientists.

Roles: Founding member and member of the Imputation Resources and Community Engagement workgroups.

https://twitter.com/LatinGenomes.

Biomedical Postdoctoral Council Diversity Committee, 07/2020–Present

Roles: Mentoring Circles - co-lead a small mentoring circle for undergraduate and graduate students; advocate for diversity and inclusion for postdocs

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SACNAS, 08/2020–Present

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

Brazilian Effort on Multifactorial Inheritance (BEMi), 08/2020-Present

Goal: address under-representation of non-Europeans in genomic studies.

Roles: Founding and Steering Committee member

Ad hoc Reviewer, 01/2015

Abstracts: Biomedical Postdoctoral Council Research Symposium, University of Pennsylvania. Manuscripts: Genome Biology and Evolution, Human Immunology, Scientific Reports, Molecular Ecology Resources, Peer Community in Evolutionary Biology.

Consultant, 03/2015

Consultant on ancient DNA for identification of forensic skeletal remains from victims of the military dictatorship in Brazil.

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