

SARA A. CARIOSCIA

Statistical geneticist with expertise in human reproductive genetics

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SUMMARY

- Experienced statistical geneticist with 5+ years of expertise in GWAS, PheWAS, and integrative analysis of large-scale genomic & transcriptomic datasets.
- Skilled computational biologist proficient in R, Python, and bioinformatics tools, with a track record of developing reproducible computational methods and statistical models.
- Deep subject matter expertise in human reproductive genetics, including embryonic aneuploidy, preimplantation genetic testing, and genetic transmission in sperm.
- Proven collaborator and communicator possessing cross-functional experience in both academic and industry settings.

EDUCATION

Johns Hopkins University

Ph.D. in Cell, Molecular, Developmental Biology and Biophysics

2019 – Present

Baltimore, MD

Georgetown University

B.S. in Biology & Classical Studies

2013 – 2017

Washington, DC

TECHNICAL SKILLS

Statistical genetics: Genome- and phenome-wide association studies (GWAS, PheWAS), phenotyping, colocalization

Genomics: Imputation, SNP array genotyping, algorithm development, low-coverage DNA sequencing analysis

Data science: Statistics, regression models, power analysis, simulation, data visualization

Programming: R, Python, Bash, Git, Snakemake, R package development, R Shiny, Quarto, GCP, Docker

Bioinformatics tools: bcftools, plink, bedtools, Idsc, IGV, SLiM

AWARDS & HONORS

- **National Science Foundation Graduate Research Fellowship (NSF GRFP)** (\$138,000)
- **Outstanding Speaker Award;** Mutations in Time & Space 2025, Cambridge, MA
- **Margolies Travel Award;** Johns Hopkins University Department of Biology (\$2,000)
- **Instructional Enhancement Grant;** Johns Hopkins University Center for Teaching Excellence & Innovation (\$5,500)

PROFESSIONAL EXPERIENCE

Statistical genetics of aneuploidy in human embryos | *PhD Student, Johns Hopkins University*

2022 – pres.

- Led analyses for GWAS, phenotyping, and imputation across a dataset of 139,416 *in vitro* fertilized embryos and 22,850 parents to identify genetic risk factors for embryonic aneuploidy.
- Performed colocalization of genetic, transcriptomic, and epigenetic evidence, linking GWAS hits for aneuploidy to meiosis-related genes.
- Designed PheWAS and TWAS analyses to demonstrate that aneuploidy, recombination, and female reproductive aging share a genetic basis.

Statistical modeling of mosaic aneuploidy in development | *PhD Student, Johns Hopkins University*

2023 – pres.

- Sole supervisor of one undergraduate and one PhD rotation student modeling genetic errors and three-dimensional development of early human embryos.
- Leveraged approximate Bayesian computation to estimate the true frequencies of embryonic aneuploidy and mosaicism.
- Led project to a \$4,000 Provost's Undergraduate Research Award and publication as a preprint on *bioRxiv*.

Computational Biologist | *Computational Biologist, Tempus Labs*

Summer 2023

- Developed an R package to automate integration testing across large-scale internal datasets and analysis pipelines.
- Collaborated closely with the Translational Research team to scope, design, and implement an interactive Shiny dashboard to visualize integration test status.
- Established robust unit testing frameworks for team software tools, streamlining development and deployment.

Quantitative genetics of allele transmission in human sperm | *PhD Student, Johns Hopkins University* 2021 – 2022

- Developed a statistical method to perform imputation, phasing, and mapping of recombination events for low-coverage, single-cell gamete sequencing data.
- Applied method to a dataset of 41,189 human sperm to show balanced allele transmission across the genome, demonstrating well-powered evidence for consistent Mendelian inheritance in humans.

Science Policy Fellow | *Science Policy Fellow, IDA Science & Technology Policy Institute* 2017 – 2019

- Provided rapid-response policy analysis for federal agencies, including the White House Office of Science and Technology Policy, NIH, NASA, and the Department of Energy, focusing on research strategy and national science initiatives.
- Authored 12 reports and gave 6 client-facing presentations on dual-use research of concern, genomic data governance, and emerging biotechnologies to inform federal decision-making.

PUBLICATIONS & PRESENTATIONS

SELECTED PUBLICATIONS

Carioscia SA,* Biddanda A,* *et al.* (2025). Common variation in meiosis genes shapes human recombination phenotypes and aneuploidy risk. *medRxiv*. *Co-first author DOI: 10.1101/2025.04.02.25325097.

Yang A, **Carioscia SA**, *et al.* (2024). Approximate Bayesian computation supports a high incidence of chromosomal mosaicism in blastocyst-stage human embryos. *bioRxiv*. DOI: 10.1101/2024.11.26.625484.

Carioscia SA,* Weaver KJ,* *et al.* (2022). A method for low-coverage single-gamete sequence analysis demonstrates adherence to Mendel's first law across a large sample of human sperm. *eLife*. *Co-first author DOI: 10.7554/eLife.76383.

Rydzewski W, **Carioscia SA**, *et al.* (2016). Sexual antagonism and meiotic drive cause stable linkage disequilibrium and favor reduced recombination on the X chromosome. *Journal of Evolutionary Biology*. DOI: 10.1111/jeb.12866.

SELECTED TALKS

2025 Mutations in Time and Space, Broad Institute, Cambridge, MA

2024 Johns Hopkins Chromatin and Chromosomes Workshop, Baltimore, MD

2024 Biology of Genomes, Cold Spring Harbor (CSHL), NY

SCIENTIFIC COMMUNICATION

Technology Fellow | *Johns Hopkins University Center for Teaching Excellence and Innovation* 2021 – 2023

- Awarded a \$5,500 grant to develop open-source teaching materials in Python, intended to introduce undergraduates with no coding experience to computational design of CRISPR reagents.
- Designed open-source modules intended for use as a five-week component of a developmental genetics lab course.

Course Developer & Instructor | *Johns Hopkins University* 2022 – 2023

- Developed and taught a 10-student course on population genetics simulation and visualization, as well as a 12-student course on modeling biological populations, taught in R, Python, and SLiM.
- Created teaching materials for a two-day workshop on hidden Markov models for undergraduate biology majors.

LEADERSHIP & SERVICE

Board Member | *Friends of the Mount Vernon Trail* 2021 – Present

- Board member of a National Park Service partner organization dedicated to enhancing the safety and natural beauty of the 18-mile Mount Vernon Trail.
- Lead volunteer events focused on invasive species removal, trail maintenance, and habitat restoration.
- Engaged over 1,400 volunteers and addressed 864 issues on the trail in 2024, resulting in a Community Impact Award from the Rosslyn Business Improvement District.

Organizing Committee Member | *symBIOsis, Johns Hopkins University* 2020 – 2022

- Founding organizing committee member of symBIOsis, a student organization dedicated to the mentorship and support of 120+ graduate students in the Department of Biology.
- Organized advising events for first- and second-year students on topics including grant applications, lab joining, and thesis proposal preparation.