

# SARA A. CARIOSCIA

*Computational biologist with expertise in statistical genetics and data science*

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## SUMMARY

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- 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.

## EDUCATION

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### Johns Hopkins University

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

2025

Baltimore, MD

### Georgetown University

B.S. in Biology & Classical Studies

2017

Washington, DC

## TECHNICAL SKILLS

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**Statistical genetics:** Genome-, transcriptome, and phenome-wide association studies (GWAS, TWAS, PheWAS), phenotyping, colocalization, RNA-seq, scRNA-seq

**Genomics:** Imputation, WES/WGS/array QC, algorithm development, low-coverage DNA sequencing analysis

**Public datasets:** gnomAD, TOPMed, 1000 Genomes

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

**Bioinformatics tools:** bcftools, plink, bedtools, ldsc, IGV, SLiM

## AWARDS & HONORS

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- **Oppenheimer Thesis Award;** Johns Hopkins University Department of Biology
- **National Science Foundation Graduate Research Fellowship (NSF GRFP)** (\$138,000)
- **Outstanding Speaker Award;** Mutations in Time & Space 2025, Cambridge, MA

## PROFESSIONAL EXPERIENCE

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### Data Scientist | Valo Health

2025 – pres.

- Build statistical models to support engineering of human tissue models.
- Contribute to shared data science infrastructure using Python, SQL, and cloud-based data platforms, enabling reproducible analysis across computational and experimental teams.

### Computational Biologist | Tempus AI

Summer 2023

- Collaborated with the Translational Research team to scope, design, and implement an interactive Shiny dashboard to visualize integration test status.
- Used GCP infrastructure and Docker to deploy reproducible pipelines using R and SQL.
- Established robust unit testing frameworks for team software tools, streamlining development and deployment.

### Statistical modeling of mosaic aneuploidy in development | Graduate Researcher, Johns Hopkins

2023 – 2025

- 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.

### Statistical genetics of aneuploidy in human embryos | Graduate Researcher, Johns Hopkins

2022 – 2025

- 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 and transcriptomic evidence, linking GWAS hits for aneuploidy to meiosis genes.

- Integrated functional genomics data (including PheWAS and TWAS analyses) to demonstrate that aneuploidy, recombination, and female reproductive aging share a genetic basis.
- Used Git for branch-based development, remote collaboration, protected branches, and merge request workflows.

## **Quantitative genetics of allele transmission in human sperm** | *Graduate Researcher, Johns Hopkins* 2019 – 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**

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### **SELECTED PUBLICATIONS**

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

Yang A, **Carioscia SA,** *et al.* (2025). Approximate Bayesian computation supports a high incidence of chromosomal mosaicism in blastocyst-stage human embryos. *Genetics*. doi: <https://doi.org/10.1093/genetics/iyaf149>.

**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.

### **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**

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### **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**

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### **Board Member** | *Friends of the Mount Vernon Trail* 2021 – pres.

- 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.