Richard Border   rborder@cs.cmu.edu
Assistant Professor   ⓘ 0000-0002-6293-2968
Department of Computational Biology   ◈ github.com/rborder
School of Computer Science   ⑧ [user=z11Dt0UAAAAJ](https://scholar.google.com/citations?user=z11Dt0UAAAAJ)
Carnegie Mellon University   ∂ borderlab.org

Research statement

I study problems in computational biology at the intersections of human genetics, statistics, quantitative psychology, and applied mathematics. My primary research interests include:

1. Nonrandom mating and other forms of population structure, particularly how they impact inference about genetic architecture

2. Scalable Bayesian inference and other methods for the efficient and flexible analysis and simulation of genome-wide data.

3. Metascience, measurement, model misspecification, and sensitivity analysis.

Preprints and manuscripts under review

- **Border**, **R**., , Wang, J., Caggiano, C., Sankararaman, S., Schork, A.J., Turley, P., Young, A.S., Benjamin, D.J., Dahl, A.W., Zaitlen, N.. Simple models of non-random mating and environmental transmission bias standard human genetics statistical methods." *Invited for resubmission* at *Nature Genetics*. doi.org/10.1101/2024.10.16.618755

- Ercelen, D., Caggiano, C., **Border, R**., Sankararaman, S., Mangul, S., Zaitlen, N., Thompson, M.. "Identifying novel genetic and phenotypic associations to genomic features by leveraging off-target reads in exome sequencing data." doi.org/10.1101/2024.11.30.625754

- Fan, C. C., Dehkordi, S. R., **Border, R.,** Shao, L., Xu, B., Loughnan, R., Thompson, W. K., Hsu, L., Lin, M., Cheng, C., Lai, R., Su, M.-H., Kao, W., Werge, T., Wu, C., Schork, A. J., Zaitlen, N., Demur, A. B., Wang, S. Assortative mating across nine psychiatric disorders is consistent and persistent over cultures and generations. *Invited for resubmission* at *Nature Human Behavior*. doi.org/10.1101/2024.09.19.24314024

- Fu, B., ..., **Border**, **R**., ..., Sankararaman, S.. "A biobank-scale test of marginal epistasis reveals genome-wide signals of polygenic epistasis." Under review at *Nature Communications*. doi.org/10.1101/2023.09.10.557084

- Krebs, K.D., ..., **Border**, **R**., ..., Schork, A.J.. "The relationship between genotype- and phenotype-based estimates of genetic liability to psychiatric disorders, in practice and in theory." *Invited for resubmission* at the *American Journal of Human Genetics*.

- Petter, E., LoPiccolo, J., Groha, S., **Border, R.**, Pasaniuc, B., Kehl, K., Zaitlen, N., Gusev, A. "Leveraging tumor dynamics to discover mutations influencing progression and treatment response for precision oncology." *Under review* at *Science Translational Medicine*.

- Sadowski M., Dahl, A.W., Zaitlen, N., **Border, R.**. "The geometry of G × E: how scaling and endogenous treatment effects shape interaction direction." *Under review* at *PLOS Genetics*. doi.org/10.1101/2025.07.15.664999

- Tan, T., ..., **Border**, **R**., ..., Young, S.. "Family-GWAS reveals effects of environment and mating on genetic associations." *Invited for resubmission* at *Nature.* doi.org/10.1101/2024.10.01.24314703

- Van der Laan, C.M., ..., **Border, R.**, ..., Boomsma, D.I.. Meta-analysis of genome wide association studies on childhood ADHD symptoms and diagnosis reveals 17 novel loci and 22 potential effector genes." doi.org/10.1101/2024.04.17.24305817

- Wainschtein, P., Zhang, Y., Schwartzentruber, J., Fiziev, P. P., Sidorenko, J., Wang, H., Kassam, I., McRae, J., **Border, R.**, Zaitlen, N., Sankararaman, S., Goddard, M. E., Zeng, J., Visscher, P. M., Farh, K. K.-H., Yengo, L. "Estimation and mapping of the missing heritability of human phenotypes". *Invited for resubmission* at *Nature.*

- Wei, A., **Border**, **R**., ..., Arboleda, V.. "The genetic underpinnings of variable penetrance and expressivity of pathogenic mutations in cardiometabolic traits." *Invited for resubmission* at *Nature Communications.* doi.org/10.1101/2023.09.14.23295564

Peer-reviewed publications

20. Medford, J., ..., **Border**, **R**., ..., Zaitlen, N.. "Beyond predictive R2: Quantile regression and non-equivalence tests reveal complex relationships of traits and polygenic scores." *The American Journal of Human Genetics.* doi.org/10.1016/j.ajhg.2025.04.013

19. Sadowski, M., ..., **Border**, **R**., ..., Zaitlen, N. (2024). "Characterizing the genetic architecture of drug response using gene-context interaction methods." *Cell Genomics.* doi.org/10.1016/j.xgen.2024.100722

18. Ma, S., Wang, F., **Border, R.**, Buxbaum, J., Zaitlen, N., Ionita-Laza, I. (2024). Local genetic correlation via knockoffs reduces confounding due to cross-trait assortative mating." *The American Journal of Human Genetics.* doi.org/10.1016/j.ajhg.2024.10.012

17. Krebs, K.D., ..., **Border**, **R**., ..., Schork, A.J. (2024). "PA-FGRS is a novel estimator of pedigree-based genetic liability that complements genotype-based inferences into the genetic architecture of major depressive disorder." *The American Journal of Human Genetics.* doi.org/10.1016/j.ajhg.2024.09.009

16. Dahl, A., ..., **Border**, **R**., ..., Cai, N. (2023). "Phenotype integration improves power and preserves specificity in biobank-based genetic studies of major depressive disorder." *Nature Genetics.* doi.org/10.1038/s41588-023-01559-9

15. **Border**, **R**. and Malik, O.A. (2023). "rBahadur: efficient simulation of structured high-dimensional genotype data with applications to assortative mating." *BMC Bioinformatics.* 10.1186/s12859-023-05442-6

14. Romero Villela, P.N., ..., **Border**, **R**., ..., Ehringer, M.A. (2023). "Loci on chromosome 20 interact with rs16969968 to influence cigarettes per day in European ancestry individuals." *Drug and Alcohol Dependence.* 10.1016/j.drugalcdep.2024.111126

13. **Border**, **R**., Athanasiadis, G., Build, A., Schork, A., Cai, N., Young, A., Werge, T., Flint, J., Kendler, K., Sankararaman, S., Dahl, A., Zaitlen, N. (2022). "Cross-trait assortative mating is widespread and inflates genetic correlation estimates." *Science.* doi.org/10.1126/science.abo2059

12. Jami, J.S., ..., **Border**, **R**., ..., Middeldorp, C.M. (2022). "Genome-wide association meta-analysis of childhood and adolescent internalising symptoms." *Journal of the American Academy of Child and Adolescent Psychiatry.* doi.org/10.1016/j.jaac.2021.11.035

11. **Border**, **R**., O'Rourke, S., de Candia, T., Goddard, M. E., Visscher, P. M., Yengo, L., Jones, M., Keller, M. C. (2022). "Assortative mating biases marker-based heritability estimates." *Nature Communications.* doi.org/10.1038/s41467-022-28294-9

10. Ip, H.F., ..., **Border**, **R**., ..., Boomsma, D. (2021). "Genetic association study of childhood aggression across raters, instruments and age." *Translational Psychiatry.* doi.org/10.1038/s41398-021-01480-x

9. Adjangba, C., **Border, R**., Romero, Villela P.N., Ehringer, M.A., Evans, L.M. (2021). "Little evidence of modified genetic effect of rs16969968 on heavy smoking based on age of onset of smoking." *Nicotine and Tobacco Research.* doi.org/10.1093/ntr/ntaa229

8. **Border**, **R**. and Becker, S. (2019). Stochastic Lanczos estimation of genomic variance components for linear mixed-effects models. *BMC Bioinformatics.* doi.org/10.1186/s12859-019-2978-z

7. **Border**, **R**., Johnson, E.C., Evans, L.M., Berley, N., Sullvan, P.F., Keller, M.C. (2019). "No support for historic candidate gene or candidate gene-by-interaction hypotheses for major depression across multiple large samples." *American Journal of Psychiatry.* doi.org/10.1176/appi.ajp.2018.18070881

6. **Border**, **R**., Johnson, E.C., Evans, L.M., Keller, M.C. (2019). "Measurement error cannot account for failed replications of historic candidate gene-by-environment hypotheses: response to Vrshek-Schallhorn et al.." *American Journal of Psychiatry.* doi.org/10.1176/appi.ajp.2019.19040374r

5. **Border, R.**, Smolen, A., Corley, R., Stallings, M., Brown, S., Conger, R., Derringer, J., Donnellan, B., Haberstick, B., Hewitt, J., Hopfer, C., Krauter, K., McQueen, M., Wall, T., Keller, M., Evans, L. (2019). "Imputation of behavioral candidate gene repeat polymorphisms in 486,551 publicly-available UK Biobank individuals." *European Journal of Human Genetics.* doi.org/10.1038/s41431-019-0349-x

4. **Border**, **R**., Corley, R.C., Brown, S.A., Hewitt, J.K., Hopfer, C.J., Williams, S.K., Rhea, S., Shriver, C.L., Stallings, M.C., Wall, T.L., Woodward, K.E., Rhee, S.H. (2018). "Independent predictors of mortality in adolescents ascertained for conduct disorder and substance use problems, their siblings, and community controls." *Addiction.* doi.org/10.1111/add.14366

3. **Border**, **R**., Corley, R.C., Brown, S.A., Hewitt, J.K., Hopfer, C.J., Stallings, M.C., Wall, T.L., Young, S.E., Rhee, S.H. (2018). "Predictors of adult outcomes in clinically- and legally-referred youth with antisocial behavior." *PLOS ONE.* doi.org/10.1371/journal.pone.0206442

2. Johnson, E.C., **Border**, **R**., Melroy-Greif, W.E., de Leeuw, C., Ehringer, M.A., Keller, M.C. (2017). "No evidence that schizophrenia candidate genes are more associated with schizophrenia than non-candidate genes." *Biological Psychiatry.* doi.org/10.1016/j.biopsych.2017.06.033

1. **Border**, **R**. and Keller, M.C. (2017). "Fundamental problems with candidate gene-by-environment interaction studies." *Journal of Child Psychology and Psychiatry.* doi.org/10.1111/jcpp.12669

Conference presentations

- Sadowski, M., Zaitlen, N., **Border, R**.. Towards resolving scaling and endogeneity induced confounding in gene-environment interaction tests. Presented at the 2025 *Probabilistic Modeling in Genomics* meeting.

- **Border, R.**, Sankararaman, S., Benjamin, D. J., Young, A., Dahl, A. W., Zaitlen, N. "(less un)Realistic simulation of complex trait architectures for open benchmarking of statistical genetic estimators." Poster presented at the 2024 annual meeting of the *American Society of Human Genetics*.

- Kar, A., Fu, B., Sukhatme, M. G., Lee, S. T., **Border, R.**, Garske, K. M., Chen, Z., Kaminska, D., Männistö, V., Pihlajamäki, J., Sankararaman, S., Pajukanta, P.. "Epistatic SNPs land in regulatory regions and possess a similar risk as GWAS SNPs in a common liver disease, MASLD." Poster presented at the 2024 annual meeting of the *American Society of Human Genetics*.

- Fu, B., Pazokitoroudi, A., Xue, A., Anand, A., Anand, P., **Border, R.**, Zaitlen, N., Sankararaman, S.. "A biobank-scale test of marginal epistasis reveals genome-wide signals of polygenic epistasis." Poster presented at the 2024 annual meeting of the *American Society of Human Genetics*.

- **Border**, **R**.. "Simulation of complex phenogenetic architectures incorporating multivariate assortative mating, vertical transmission, cross-trait causal dependencies, and fine-scale population structure." Talk presented at the 2023 *Probabilistic Modeling in Genomics* meeting.

- Wu, Y. and **Border**, **R**.. "Emerging topics in biobank-scale association analysis." Co-moderator of platform-talk at the 2022 annual meeting of the *American Society of Human Genetics*.

- **Border**, **R**., Athanasiadis, G., Buil Demur, A., Schork, A., Cai, N., Young., A., Werge, T., Kendler, K., Flint, J., Dahl, A., Price, A., Zaitlen, N. (2022). "Quantifying the effects of high-dimensional cross-trait assortative mating on complex trait genetic architectures." Reviewers' Choice Abstract poster presented at the 2022 annual meeting of the *American Society of Human Genetics*.

- **Border**, **R**., Athanasiadis, G., Buil Demur, A., Schork, A., Werge, T., Kendler, K., Flint, J., Dahl, A., Zaitlen, N. (2021). "Widespread evidence of systematic bias in estimates of genetic correlation due to cross-trait assortative mating." Plenary talk presented at the 2021 annual meeting of the *American Society of Human Genetics*.

- **Border**, **R**., Becker, S. (2019). "Randomized algorithms for genomic variance components estimation in mixed models." Poster presented at the 2019 *International Workshop on Statistical Genetic Methods for Human Complex Traits*, Boulder, CO.

- **Border**, **R**., Johnson, E.C., Evans, L.M., Berley, N., Sullvan, P.F., Keller, M.C. (2018). "Quantitative reconcilliation of GWAS and candidate gene findings: measurement error, nonlinearity, and artifactual results." Paper presentation *accepted* for the 48*th* meeting of the Behavior Genetics Association in Boston, MA. (Talk delivered by M.C. Keller due to illness)

- **Border**, **R**., Johnson, E.C., Berley, N., Medland, S.E., Sullvan, P.F., Keller, M.C. (2018). "Examining the relevance of canonical candidate genes for major depression." Poster *accepted* for the 48*th* meeting of the Behavior Genetics Association in Boston, MA.

- Evans, L.M., **Border, R.**, du Pont, A., Friedman, N.P., Johnson, E., Yang, J., Visscher, P., Keller, M.C. (2018). "Exploring the genetic architecture of psychiatric disorders using partitioned heritability approaches." Symposium presented by Luke Evans at the *World Congress of Psychiatric Genetics*, October 2018, Glasgow, Scotland.

- **Border**, **R**., Johnson, E.C., Berley, N., Sullvan, P.F., Keller, M.C. (2017). "Discrepancies between candidate gene and genome-wide studies of complex traits and endophenotypes." Poster presented at the 25*th* annual meeting of the World Congress of Psychiatric Genetics, Orlando, Florida, October 13-17, 2017

- Park, A. L., Tsai, K. H., Guan, K., **Border, R.**, and Chorpita, B. F. (2017). "Unintended consequences of evidence-based treatment policy reform." In *Use of Evidence in Mental Health Treatment and Clinical Decision-Making.* Symposium held at the 4*th* Biennial Society for Implementation Research Collaboration Conference, Seattle, WA.

- Johnson, E.C., Melroy-Greif, W.E., **Border**, **R**., Keller, M.C., Ehringer, M.A. (2016). "Examining 25 classic schizophrenia candidate genes in the context of GWAS data: evidence for relevance?" Poster presented at the 2016 meeting of the American Society of Human Genetics in Vancouver, British Colombia.

- **Border**, **R**., Sawaya, S., Huggett, S., Brown, S.A., Wall, T.L., and Stallings, M.C. (2015). "Sensitivity of random forests algorithm to population stratification in GWAS data." Postter presented at the 45*th* meeting of the Behavior Genetics Association in San Diego, CA.

Invited talks

- (less un)Realistic models of the genetic architecture of complex traits.
  Center for Computational Molecular Biology, Brown University. February, 2025.

- dynamic modeling / modeling dynamics.
  Computational Genetics Summer Institute, University of California, Los Angeles. July, 2024.

- Investigating genetic architectures in the context of complex intergenerational dynamics.
  Institute for Human Genetics, University of California, San Francisco. April, 2024.
  Department of Computational Biology, Carnegie Mellon University. February, 2024.
  Department of Human Genetics, University of Chicago. February, 2024.
  Department of Psychiatry, Virginia Commonwealth University. February, 2024.

- Forward time simulation of complex phenogenetic architectures.
  Institute for Behavioral Genetics, University of Colorado Boulder. May, 2023.

- Non-random mating, vertical transmission, and causal dependencies distort our understanding of complex trait genetics.
  Psychiatric Genetics Consortium Substance Use Disorders Group. April, 2023.

- Understanding genetic correlation estimates in the context of multivariate assortative mating.
  Program in Genetic Epidemiology and Statistical Genetics, Harvard T.H. Chan School of Public Health. October, 2021.

- Genetic correlation doesn't imply pleiotropy: understanding the effects of cross-trait assortative mating and errors in diagnosis.
  Genome Sequencing Program Methods Group, Rutgers University. June, 2021.

- Assortative mating and marker-based heritability estimation.
  Program in Genetic Epidemiology and Statistical Genetics, Harvard T.H. Chan School of Public Health. March, 2020.

- Stochastic Lanczos REML algorithms for linear mixed models in genetic association studies.
  Computational Medicine Seminar, University of California, Los Angeles. March, 2020.

Software

- **Border, R.** (2023). "xftsim: eXtensible Forward Time SIMulator." Python 3 module.
  pypi.org/project/xftsim, github.com/rborder/xftsim

- **Border, R**. and Malik O.A. (2023). "rBahadur: Efficient simulation of genotype-phenotype data under assortative mating by generating Bahadur order-2 multivariate Bernoulli distributed random variates." R library. cran.r-project.org/package=rBahadur, github.com/rborder/rBahadur

- **Border, R.** (2019). "SL_REML: Stochastic Lanczos REML algorithms for linear mixed models." Python 3 module. github.com/rborder/SL_REML

Education

2019	Doctor of Philosophy *Behavioral, Psychiatric, and Statistical Genetics* *Advised by Matthew C. Keller* *Department of Psychology and Neuroscience, University of Colorado Boulder*
2018	Master of Science *Applied Mathematics* *Advised by Stephen Becker* *Department of Applied Mathematics, University of Colorado Boulder* Master of Arts *Behavioral, Psychiatric, and Statistical Genetics* *Advised by Soo Hyun Rhee* *Department of Psychology and Neuroscience, University of Colorado Boulder*
2011	Bachelor of Arts *Japanese Language and Literature* *Advised by Terry Kawashima* *Department of East Asian Studies, Wesleyan University*

Academic Positions

2024–	Assistant Professor, *Department of Computational Biology, School of Computer Science, Carnegie Mellon University*
2020–2024	Postdoctoral Fellow, *mentored by Noah Zaitlen and Sriram Sankararaman, Departments of Neurology and Computer Science, University of California Los Angeles*

2021–2023   Visiting Scientist, *mentored by Alkes Price and Sriram Sankararaman, Department of Epidemiology, Harvard T.H. Chan School of Public Health*

2015–2019   Predoctoral Fellow, *advised by Matthew Keller and Soo Rhee, Institute for Behavioral Genetics, University of Colorado Boulder*

2018–2019   Course Assistant, *Department of Applied Mathematics, University of Colorado Boulder*

2014–2016   Teaching Assistant, *Department of Psychology and Neuroscience, University of Colorado Boulder*

2012–2014   Research Assistant, *Departments of Psychiatry and Psychology, University of California Los Angeles*

2012–2014   Clinical Trainee, *Child OCD, Anxiety, and Tic Disorders Clinic, University of California Los Angeles*

Selected honors and awards

2023   Outstanding Mentorship Award
Bruins in Genomics Summer Research Program

2022   Reviewers' Choice Abstract
American Society for Human Genetics

      Random Matrix Theory and Numerical Linear Algebra Conference Travel Award
National Science Foundation

2021   Charles J. Epstein Research Semifinalist Award
American Society for Human Genetics

      National Institutes of Health Postdoctoral Trainee   *T32NS048004*
Semel Institute for Neuroscience and Human Behavior, University of California, Los Angeles

2020   Dosier Muenzinger Award for Outstanding Contribution to Basic Research
Department of Psychology and Neuroscience, University of Colorado Boulder

2016–2019   National Institutes of Health Predoctoral Trainee   *T32MH016880*
Selected by faculty training committee thrice consecutively (maximum number of times awarded to any graduate student), Institute for Behavioral Genetics, University of Colorado Boulder

2018   Behavior Genetics Association Travel Award
Behavior Genetics Association

      Departmental Travel Grant
Department of Psychology and Neuroscience, University of Colorado Boulder

2017   United Government of Graduate Students Individual Travel Award
University of Colorado Boulder Graduate School

2015–2019   Predoctoral Fellowship
Institute for Behavioral Genetics, University of Colorado Boulder

Peer review

I have served as a special topic guest editor for *Frontiers in Genetics.* Additionally, I have refereed for the following journals / conferences:

- Alcoholism: Clinical and Experimental Research
- American Journal of Medical Genetics
- American Journal of Psychiatry
- Bioscience Reports
- BMC Bioinformatics
- BMC Psychiatry
- eLife
- Frontiers in Public Health
- Genes, Brain and Behavior
- Genome Research
- IEEE Transactions on Computational Biology and Bioinformatics
- Intelligent Systems for Molecular Biology (ISMB)

- JAMA Psychiatry
- Journal of Clinical Medicine
- Journal of Psychiatric Research
- Molecular Psychiatry
- Nature Human Behavior
- Nature Communications
- Nature Genetics
- Psychiatry Research
- Psychological Bulletin
- Psychoneuroendocrinology
- Research in Computational Molecular Biology (RECOMB)
- Social Science and Medicine

Teaching positions

2019   Statistical Methods and Applications II (Combined Undergraduate and Graduate Sections)
Course Assistant, Department of Applied Mathematics.

2018   Mathematical Statistics (Combined Undergraduate and Graduate Sections)
Course Assistant, Department of Applied Mathematics.

Statistical Methods (Combined Undergraduate and Graduate Sections)
Course Assistant, Department of Applied Mathematics.

2016   Statistical Programming with R (Graduate)
Teaching Assistant, Department of Psychology and Neuroscience.

Statistics II (Graduate)
Teaching Assistant, Department of Psychology and Neuroscience.

2015	Introduction to Statistics (Undergraduate)
	Teaching Assistant, Department of Psychology and Neuroscience.
	Statistical Programming with R (Graduate)
	Teaching Assistant, Department of Psychology and Neuroscience.

Guest lectures

- Randomized algorithms for genomic variance components analyses (2019). *Randomized Algorithms, Department of Applied Mathematics, University of Colorado Boulder.*

- Mapping genes for complex traits (2019). *Physiological Genetics and Genomics, Department of Integrative Physiology, University of Colorado Boulder.*

- Statistical power (2016). *Introduction to Statistics, Department of Psychology and Neuroscience, University of Colorado Boulder.*

- Functional programming concepts in R (2015). *Statistical Programming with R, Department of Psychology and Neuroscienc, University of Colorado Boulder.*

- Methods for reproducible research in R (2015). *Statistical Programming with R. Department of Psychology and Neuroscience, University of Colorado Boulder.*

Departmental service

2024-2025	Admissions Committee member for Joint CMU-Pitt Ph.D. Program in Computational Biology
	Faculty host for Computational Biology Department faculty candidate
	Awards Committee for Newell Undergraduate Research Award