Richard Border   border.richard@gmail.com
Assistant Professor   0000-0002-6293-2968
Department of Computational Biology   github.com/rborder
School of Computer Science   user=z11Dt0UAAAAJ
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Research statement

I study problems 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.

I am moving to CMU to start my lab in January 2025. In the meantime, I'm working with Noah Zaitlen in the UCLA David Geffen School of Medicine Neurology Department and Sriram Sankararaman in the UCLA Computer Science Department.

Peer-reviewed publications

19.   Sadowski, M., ..., **Border**, **R**., ..., Zaitlen, N. (2024). "Genetics modulates the impact of widely-used drugs on outcomes and side effects." *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

Manuscripts in preparation

- **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." *Out for review* at *Nature Genetics.* doi.org/10.1101/2024.10.16.618755

- 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

- 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

- 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." *Out for review* at the *American Journal of Human Genetics.*

- Medford, J., ..., **Border**, **R**., ..., Zaitlen, N.. "Beyond predictive R-squared: Complex relationships between traits and their corresponding polygenic scores are revealed using non-equivalence tests of quantile-specific linear models." *Invited for resubmission* at the *American Journal of Human Genetics.*

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

- 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

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

Conference presentations

- **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* annual meeting of the Behavior Genetics Association in San Diego, CA.

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*

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*

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

Dissertation and master's thesis

- **Border**, **R**.. "Topics in the quantitative analysis of complex trait genetic architectures". Psychology and Neuroscience Graduate Theses & Dissertations. scholar.colorado.edu/concern/articles/vd66w090s

- **Border**, **R**.. "Stochastic Lanczos likelihood estimation of genomic variance components". Applied Mathematics Graduate Theses & Dissertations. 120. scholar.colorado.edu/appm_gradetds/120

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*
- *IEEE Transactions on Computational Biology and Bioinformatics*
- *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)*

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.