# CATHERINE CORINNE ROBERTSON

Department of Computational Medicine and Bioinformatics
University of Michigan Medical School, Ann Arbor, MI

Molecular Genetics Section, National Human Genome Research Institute
National Institutes of Health, Bethesda, MD

919-812-2395 · ccrober@umich.edu · cassie.robertson@nih.gov

## EDUCATION

2016 - 2021 **PhD in Biochemistry and Molecular Genetics**
University of Virginia, Charlottesville, VA

Thesis Title: *Discovery, fine mapping, and functional characterization of genetic susceptibility loci in type 1 diabetes.*

Thesis Advisor: Stephen Rich, PhD

Thesis Committee: Stefan Bekiranov, PhD, Charles Farber, PhD, Michael Guertin, PhD, Coleen McNamara, MD, Suna Onengut-Gumuscu, PhD, and Stephen Rich, PhD

2012 - 2014 **MS in Biostatistics**
University of Michigan, Ann Arbor, MI
Advisors: Laura Scott, PhD and Michael Boehnke, PhD

2011 - 2012 **Postbaccalaureate Certificate in Applied Mathematics**
North Carolina State University, Raleigh, NC
Program Director: Steve Campbell, PhD

2005 - 2009 **BA, *cum laude***
Connecticut College, New London, CT
Majors (double): Mathematics & Dance
Advisor: Christopher Hammond, PhD

## RESEARCH EXPERIENCE

2022 - Present **Research Fellow**

University of Michigan (PI: Stephen Parker, PhD) and NIH/NHGRI (PI: Francis Collins, MD, PhD)
Research description: Analysis of single cell multi-omics data from pancreatic islets isolated from individuals with islet autoimmunity, type 1 diabetes, or no evidence of autoimmunity or diabetes. Analysis will explore effects of genetic variation and immunogenic exposures on chromatin state and transcriptional programs in pancreatic islets and investigate their causal role in the development of type 1 diabetes.

2017 - 2021 **Graduate Researcher**
University of Virginia (PI: Stephen Rich, PhD)

Functional genomics projects:
- Generating chromatin accessibility quantitative trait loci (caQTL) maps in $CD4^+$ T cells
- Integrating caQTLs with genetic association fine-mapping results and expression QTL (eQTL) data sets to prioritize causal variants and genes at type 1 diabetes-associated loci

Genetic association projects:
- Discovery and fine-mapping analysis of type 1 diabetes genetic associations in ~60,000 individuals genotyped with the Illumina ImmunoChip
- Imputation of HLA alleles in African American cohort using a multi-ethnic reference panel and SNP2HLA software
- Modeling association of genetic variation in HLA genes with risk for type 1 diabetes
- Developing a genetic risk score for type 1 diabetes using regularized regression techniques and comparing performance with traditional genetic risk prediction approaches
- Family-based genome-wide association analysis of type 1 diabetes in diverse ancestry groups

**Graduate Student Rotations,** University of Virginia

2018 Michael Guertin, Department of Biochemistry and Molecular Genetics
- Optimized immune cell stimulation protocol at scale feasible for high throughput nascent RNA transcription assays
- Learned nascent RNA transcription assay (PRO-seq)

2017 Kodi Ravichandran, Department of Microbiology, Immunology, and Cancer Biology
- Learned fundamental skills in cell culture and flow cytometry
- Performed assays to measure phagocytosis in human cell lines
- Optimized use of chloride indicator in phagocytosis assays

2016 Michael Brown, Carter Immunology Center
- Performed qPCR to measure viral DNA levels in mice
- Analyzed exome sequencing data from multiple mouse strains, including strains that have not been previously sequenced

2014 - 2016 **Statistical Analyst**
University of Michigan (PI: Matthew Sampson, MD)
- Integrated genetic, transcriptomic, and clinical data to study glomerular disease etiology and outcomes
- Led statistical analysis and contributed to manuscript preparation
- Performed preliminary data analysis for R01 (DK109905) to study molecular mechanisms of *APOL1*-associated glomerular disease

2012 - 2014 **Graduate Researcher**
University of Michigan (PI: Michael Boehnke, PhD)
- Performed genome-wide association analysis for 69 NMR-based lipid metabolites using Illumina Exome Chip, OmniExpress, and imputed genotyping data from large cohorts

## PUBLICATIONS

2024 **Robertson CC**†, Elgamal RM†, Henry-Kanarek BA†, Arvan P, Chen S, Dhawan S, Eizirik DL, Kaddis JS, Vahedi G, Parker SCJ, Gaulton KJ, Soleimanpour SA. Untangling the genetics of beta cell dysfunction and death in type 1 diabetes. *Mol Metab*. 2024 Jun 22;86:101973. doi: 10.1016/j.molmet.2024.101973. Epub ahead of print. PMID: 38914291.

2024 Grenko CM, Taylor HJ, Bonnycastle LL, Xue D, Lee BN, Weiss Z, Yan T, Swift AJ, Mansell EC, Lee A, **Robertson CC**, Narisu N, Erdos MR, Chen S, Collins FS, Taylor DL. Single-cell transcriptomic profiling of human pancreatic islets reveals genes responsive to glucose exposure over 24 h. *Diabetologia*. 2024 Jul 5. doi: 10.1007/s00125-024-06214-4. Epub ahead of print. PMID: 38967666.

2024 Bonnycastle LL, Swift AJ, Mansell EC, Lee A, Winnicki E, Li ES, **Robertson CC**, Parsons VA, Huynh T, Krilow C, Mohlke KL, Erdos MR, Narisu N, Collins FS. Generation of Human Isogenic Induced Pluripotent Stem Cell Lines with CRISPR Prime Editing. *CRISPR J*. 2024 Feb;7(1):53-67. doi: 10.1089/crispr.2023.0066. PMID: 38353623.

2024 Michalek DA, Tern C, Zhou W, **Robertson CC**, Farber E, Campolieto P, Chen WM, Onengut-Gumuscu S, Rich SS. A multi-ancestry genome-wide association study in type 1 diabetes. *Hum Mol Genet*. 2024 May 18;33(11):958-968. doi: 10.1093/hmg/ddae024. PMID: 38453145.

2022 Richardson TG, Crouch DJM, Power GM, Morales-Berstein F, Hazelwood E, Fang S, Cho Y, Inshaw JRJ, **Robertson CC**, Sidore C, Cucca F, Rich SS, Todd JA, Davey Smith G. Childhood body size directly increases type 1 diabetes risk based on a lifecourse

Mendelian randomization approach. *Nat Commun*. 2022 Apr 28;13(1):2337. doi: 10.1038/s41467-022-29932-y. PMID: 35484151.

2021 **Robertson CC**[†], Inshaw JRJ[†], Onengut-Gumuscu S, Chen WM, Santa Cruz DF, Yang H, Cutler AJ, Crouch DJM, Farber E, Bridges SL Jr, Edberg JC, Kimberly RP, Buckner JH, Deloukas P, Divers J, Dabelea D, Lawrence JM, Marcovina S, Shah AS, Greenbaum CJ, Atkinson MA, Gregersen PK, Oksenberg JR, Pociot F, Rewers MJ, Steck AK, Dunger DB; Type 1 Diabetes Genetics Consortium; Wicker LS, Concannon P, Todd JA, Rich SS. Fine-mapping, trans-ancestral and genomic analyses identify causal variants, cells, genes and drug targets for type 1 diabetes. *Nat Genet*. 2021 Jul;53(7):962-971. doi: 10.1038/s41588-021-00880-5. Epub 2021 Jun 14. PMID: 34127860.

2020 Sidore C, Orrù V, Cocco E, Steri M, Inshaw JR, Pitzalis M, Mulas A, McGurnaghan S, Frau J, Porcu E, Busonero F, Dei M, Lai S, Sole G, Virdis F, Serra V, Poddie F, Delitala A, Marongiu M, Deidda F, Pala M, Floris M, Masala M, Onengut-Gumuscu S, **Robertson CC**, Leoni L, Frongia A, Ricciardi MR, Chessa M, Olla N, Lovicu M, Loizedda A, Maschio A, Mereu L, Ferrigno P, Curreli N, Balaci L, Loi F, Ferreli LA, Pilia MG, Pani A, Marrosu MG, Abecasis GR, Rich SS, Colhoun H, Todd JA, Schlessinger D, Fiorillo E, Cucca F, Zoledziewska M. *PRF1* mutation alters immune system activation, inflammation, and risk of autoimmunity. *Mult Scler*. 2021 Aug;27(9):1332-1340. doi: 10.1177/1352458520963937. Epub 2020 Oct 14. PMID: 33566725.

2020 Brenner LN, Mercader JM, **Robertson CC**, Cole J, Chen L, Jacobs SBR, Rich SS, Florez JC. Analysis of glucocorticoid-related genes reveal *CCHCR1* as a new candidate gene for type 2 diabetes. *Journal of the Endocrine Society*. 2020 Aug; PMID: 33150273.

2019 Hippich M, Beyerlein A, Hagopian WA, Krischer JP, Vehik K, Winkler C, Toppari J, Lernmark A, Rewers MJ, Steck AK, She JX, Akolkar B, **Robertson CC**, Onengut-Gumuscu S, Rich SS, Bonifacio E, Ziegler A, TEDDY Study Group. Genetic contribution to the divergence in type 1 diabetes risk between children from the general population and children from affected families. *Diabetes*. 2019 Apr; PMID: 30655385.

2019 Onengut-Gumuscu S, Chen W, **Robertson CC**, Bonnie JK, Farber E, Oksenberg JR, Brant SR, Bridges SL, Edberg JC, Kimberly RP, Gregersen PK, Rewers MJ, Steck A, Black MH, Dabelea D, Pihoker C, Atkinson MA, Divers J, Bell AB, SEARCH for Diabetes in Youth, Type 1 Diabetes Genetics Consortium, Erlich HA, Concannon P, Rich SS. Type 1 diabetes risk in African-ancestry participation and utility of an ancestry-specific genetic risk score. *Diabetes Care*. 2019 Mar; PMID: 30659077.

2018 **Robertson CC** & Rich SS. Genetics of type 1 diabetes. *Current Opinions in Genetics and Development*. 2018 Feb 13; PMID: 29453110.

2017 Gillespie A, Lee H, **Robertson CC**, Cabot M, Brown MG. Genome-wide exome analysis of Cmv5-disparate mouse strains that differ in host resistance to murine cytomegalovirus infection. *G3: Genes, Genomes, Genetics*. 2017 Jun7; PMID: 28450376.

2016 Crawford BD, Gillies CE, **Robertson CC**, Kretzler M, Otto E, Vega-Wagner V, et al. Evaluating Mendelian nephrotic syndrome genes for evidence for risk alleles or oligogenicity that explain heritability. *Pediatric Nephrology*. 2016 Oct 20; PMID: 27766458.

2016 **Robertson CC**, Gillies CE, Putler RKB, Ng D, Reidy KJ, Crawford B, et al. An investigation of *APOL1* risk genotypes and preterm birth in African American population cohorts. *Nephrology Dialysis Transplantation*. 2016 Sep 16; PMID: 27638911.

2016 Sampson MG, Gillies CE, **Robertson CC**, Crawford B, Vega-Warner V, Otto EA, et al. Using Population Genetics to Interrogate the Monogenic Nephrotic Syndrome Diagnosis in a Case Cohort. *Journal of the American Society of Nephrology*. 2016 Jul 1; 27(7):1970–83. PMID: 26534921.

2016 Ng DK†, **Robertson CC**†, Woroniecki RP, Limou S, Gillies CE, Reidy KJ, et al. *APOL1* -associated glomerular disease among African-American children: a collaboration of the Chronic Kidney Disease in Children (CKiD) and Nephrotic Syndrome Study Network (NEPTUNE) cohorts. *Nephrology Dialysis Transplantation*. 2016 Apr 27; PMID: 27190333.

2016 *Sampson MG, **Robertson CC**, Martini S, Mariani LH, Lemley K V., Gillies CE, et al. Integrative Genomics Identifies Novel Associations with APOL1 Risk Genotypes in Black NEPTUNE Subjects. *Journal of the American Society of Nephrology*. 2016 Mar 1;27(3):814–23. PMID: 26150607.

*Featured in Nature Reviews Nephrology, Research Highlights (2015). doi:10.1038/nrneph.2015.123

2016 Gillies CE, Otto EA, Vega-Warner V, **Robertson CC**, Sanna-Cherchi S, Gharavi A, et al. tarSVM: Improving the accuracy of variant calls derived from microfluidic PCR-based targeted next generation sequencing using a support vector machine. *BMC Bioinformatics*. 2016 Dec 10;17(1):233. PMID: 27287006.

2015 Gillies CE, **Robertson CC**, Sampson MG, Kang HM. GeneVetter: a web tool for quantitative monogenic assessment of rare diseases. *Bioinformatics*. 2015 Jul 23; 31(22). PMID: 26209433.

† Equal contribution

## FUNDING AND AWARDS

2023 - 2024 Training Program in Endocrinology and Metabolism (T32 DK007245), University of Michigan

2022 - 2023 Training Program in Endocrinology and Metabolism (T32 DK007245), University of Michigan

2022 ASHG Epstein Award Finalist

2019 - 2020 Wagner Fellowship, University of Virginia

2019 Raven Society (Honor's society), University of Virginia

2018 - 2019 Biomedical Data Sciences Fellowship (T32 LM012416), University of Virginia

2017 - 2018 Biomedical Data Sciences Fellowship (T32 LM012416), University of Virginia

2016 - 2017 Peach Fellowship, University of Virginia

2013 - 2014 Genome Sciences Training Program (T32 HG000040), University of Michigan

2012 - 2013 Genome Sciences Training Program (T32 HG000040), University of Michigan

2012 Teaching Assistantship, North Carolina State University

2009 Distinction in Mathematics, Julia Wells Bower Award, Connecticut College

2009 Distinction in Dance, Connecticut College

2005 - 2009 Dean's Honors, Connecticut College

## ORAL PRESENTATIONS

### Abstract Presentations

2022 †*American Society of Human Genetics Annual Meeting*; Oct 26, Los Angeles, CA.
†Featured plenary presentation and 2022 Epstein Award Finalist

2022 *Human Islet Research Network Annual Investigator Meeting*; Sep 15, Washington, D.C.

2020 *American Diabetes Association 80th Scientific Sessions*; May 29, Virtual.

### Invited Presentations

2023 *American Diabetes Association 83rd Scientific Sessions*; Jun 26, San Diego, CA.

2022 Finland-United States Investigation of NIDDM Genetics (FUSION) study annual meeting; Apr 26, Ann Arbor, MI.

2021 Division of Biomedical Informatics and Personalized Medicine, University of Colorado Anschutz Medical Campus; Nov 3, Virtual.

2021 The Sugar Science, Ask the Expert Series; Oct 5, Virtual.

2021 Center for Combinatorial Gene Regulation, Duke University; Jul 28, Virtual.

2021 Laboratories of Drs. Stephen Parker (University of Michigan) and Francis Collins (NHGRI); Jul 16, Virtual.

2021 Laboratories of Drs. David Knowles and Tuuli Lappalainen, New York Genome Center; Jul 8, Virtual.

## POSTER PRESENTATIONS

2024 Robertson CC, Wang X, Orchard P, Chang ACN, Meng Z, Dolgachev V, Manickam N, Narisu N, Mike E, Collins FS, Chen S, Parker SCJ. Multiome profiling of human islets nominates beta cell-specific cytokine response profiles underlying type 1 diabetes. *American Diabetes Association 8th[d] Scientific Sessions*; Jun 22, Orlando, FL.

2021 Robertson CC, Onengut-Gumuscu S, Chen WM, Concannon P, Rich SS. Integrative analysis of chromatin accessibility and genetic risk in type 1 diabetes. *Cold Spring Harbor meeting on Systems Immunology*. Apr 20-23, Virtual.

2020 Robertson CC, Inshaw JRJ, Onengut-Gumuscu S, Chen WM, Flores DSC, Yang H, Cutler A, Crouch DM, Farber E, Wicker LS, Concannon P, Todd JA, Rich SS. Fine mapping and integrative genomic analyses identify potential mechanisms for type 1 diabetes. *American Society of Human Genetics Annual Meeting*; Oct 27-30, Virtual.

2019 Robertson CC, Family-based association analysis of type 1 diabetes replicates case-control associations and reveals sequence homology as a source of spurious association in exome chip analysis. *American Society of Human Genetics Annual Meeting*; Oct 16, Houston, TX.

2019 Robertson CC, Inshaw JR, Onengut-Gumuscu S, Chen W, Todd J, and Rich SS. Association analysis of type 1 diabetes in >60,000 ancestrally diverse participants identifies novel risk loci. *The Biology of Genomes*; May 10; Cold Spring Harbor, NY.

2018 Robertson CC, Inshaw JR, Onengut-Gumuscu S, Chen W, Todd J, and Rich SS. Association analysis of type 1 diabetes in >60,000 ancestrally diverse participants identifies novel risk loci. *American Society of Human Genetics Annual Meeting*; Oct 19, San Diego, CA.

2017 Robertson CC, Onengut-Gumuscu S, Chen W, and Rich SS. HLA imputation and association with type 1 diabetes in African Americans. *American Society of Human Genetics Annual Meeting*; Oct 18, Orlando, FL.

2014 Robertson, CC and Dharmarajan SH. A cohort study of changes in cognitive impairment trajectories over time. Poster presented at: Department of Biostatistics, University of Michigan; Apr 25, Ann Arbor, MI.

## DEPARTMENTAL TALKS

2021 Center for Public Health Genomics Research in Progress; Apr 12, Charlottesville, VA.

2019 Systems Biology Journal Club; Sep 11, Charlottesville, VA.

2019 Biochemistry and Molecular Genetics Department Research Club; Sep 10, Charlottesville, VA.

2019 Biochemistry and Molecular Genetics Department Research Club; Oct 2, Charlottesville, VA.

2018 Center for Public Health Genomics Research in Progress; Apr 9, Charlottesville, VA.

2017 Biochemistry and Molecular Genetics Department Research Club; Sep 12, Charlottesville, VA.

2014 University of Michigan Center for Statistical Genetics Seminar; Mar 21, Ann Arbor, MI.

2013 University of Michigan Genome Science Training Program student orientation; Aug 27, Ann Arbor, MI.

## PROFESSIONAL SERVICE

### Professional Societies

*American Diabetes Association*

2024 Abstract Reviewer and Session Chair
2023 Abstract Reviewer and Session Chair

*American Society of Human Genetics*

2024 Abstract Reviewer
2023 Abstract Reviewer
2022 Abstract Reviewer and Session Moderator
2019 – 2021 Program Committee Member

- Helped to shape scientific content of annual conference for leading society in human genetics
- Invited session programming: Reviewed proposals for invited sessions each year; Served as liaison between Program Committee and invited session proposer; provided constructive feedback for revising proposals
- Abstract session programming: Recruited three domain experts to my review section each year; Reviewed ~120 annual conference abstracts annually; designed and recruited moderators for approximately three abstract-driven sessions per year.
- Published advice for poster presenters leading up to 2019 meeting: Poster Session Tips for a Scientific Meeting. *ASHG News*. August 29, 2019; https://www.ashg.org/publications-news/ashg-news/poster-tips

2018 Session Moderator

### Journal Peer Reviewer

2023 Nature Genetics
2023 Nature Communications
2023 Frontiers in Molecular Medicine
2022 eLife

2022 BMC Medicine
2022 Diabetes
2022 NAR Genomics and Bioinformatics

## LEADERSHIP AND OUTREACH

2019 – 2020 **Women in Medical Sciences (WIMS)**, University of Virginia
Co-President

- Led organization promoting and supporting women in medical sciences at UVA
- Worked with team of five executive members to host social, academic, career development, and community outreach events
- Led "Seminar Diversity Initiative" to increase representation of underrepresented groups, including women and underrepresented minorities, in invited speaker seminars across the biomedical sciences at UVA

2018 - 2019 **Graduate Recruitment Initiative Team (GRIT)**, University of Virginia
Council Member

- Worked with council to organize full-day workshop to address challenges in recruitment and retention of Under-Represented Minorities (URM) in STEM graduate programs at UVA
- Analyzed data on URM representation among PhD applicants, admits, and matriculants

2018 - 2019 **Biomedical Data Sciences Training Grant (T32),** University of Virginia
Journal club founder and organizer

- Initiated and organized and monthly informal student-focused meeting dedicated to biomedical applications of data science

Hackathon participant, Apr 4-5, 2019

- Worked towards development of a Python package for the analysis of multi-omics single cell sequencing data type for a 36-hr period

Hackathon participant, Nov 8-9, 2017

- Participated in a team-driven analysis of single cell RNA-seq data over a 36-hr period
- Contributed to conceptualization and execution of data analysis plan
- Presented results at UVA Data Science Institute annual "Datapalooza" conference

2017 - 2019 **Graduate Biosciences Society**, University of Virginia
2018-19, Academic Committee Co-Chair

- Organized a full day symposium that included graduate student poster sessions, poster judges and prizes, student talks, and a keynote lecture from student-selected speaker, Dr. Erica Ollmann Saphire
- Organized poster session featuring faculty research to expose incoming graduate students to research rotation opportunities

2017-18, Treasurer

- Prepared budget and managed finances for society representing over 300 graduate students in biomedical sciences

July 2019 Selection Committee for Dean's Award for Excellence in Teaching, University of Virginia

Fall 2016 Planning Committee for Core Course in Integrated Biosciences Annual Lecture, University of Virginia

2013 - 2014 President of STATCOM (*pro bono* statistical consulting), University of Michigan

## TEACHING AND MENTORING EXPERIENCE

2024 - Present **Mentor for Nicholas Howell**, Postbaccalaureate Researcher at the National Human Genome Research Institute (NHGRI/NIH)

2021 - Present **Mentor for Alice Wang**, Candidate for PhD in Bioinformatics at the University of Michigan

2023 - Present **Mentor for Zoe Weiss**, Postbaccalaureate Researcher at the National Human Genome Research Institute (NHGRI/NIH)

2018 **Teaching Assistant for Core Course in Integrative Biosciences**, University of Virginia

- Led summer sessions on genetics and molecular biology for UVA graduate students

2012 **Graduate Teaching Assistant for Calculus I**, North Carolina State University

- Assistant for undergraduate NCSU calculcus course; Graded assignments and exams; held office hours and tutored students needing additional assistance.