Joanne B. Cole, Ph.D.

Instructor, Harvard Medical School
The Broad Institute of MIT and Harvard,
Medical and Population Genetics & Metabolism Programs
Massachusetts General Hospital,
Center for Genomic Medicine
Boston Children's Hospital,
Center for Basic and Translational Obesity Research

Education

Ph.D. Human Medical Genetics and Genomics University of Colorado Denver, Anschutz Medical Campus	Aurora, CO	September 2016
B.S. Cellular Biology and Molecular Genetics University of Maryland	College Park, MD	December 2010

Research Experience

Postdoctoral Research Fellow — Cambridge, MA — 2016-Present
The Broad Institute of MIT and Harvard
Advisors: Jose C. Florez, M.D./Ph.D. & Joel N. Hirschhorn, M.D./Ph.D.

Elucidate the underlying relationships between dietary intake and overall human health and lifestyle with the use of computational and statistical genetic approaches in large biobanks and cohorts. Additional genetic investigation focuses on cardiometabolic disease and related traits including diabetic kidney disease, diabetes, and anthropometric traits.

K99/R00 NIDDK Pathway to Independence Award 2021-2026
American Diabetes Association Postdoctoral Fellowship 2019-2021
Harvard Training Fellowship: Bioinformatics Applied to Diabetes, Obesity and Metabolism 2017-2019
Massachusetts General Hospital Training Fellowship: Endocrinology 2016-2017

Graduate Research Assistant — Aurora, CO — 2011-2016
University of Colorado Denver, Anschutz Medical Campus
Advisor: Richard Spritz, M.D.

Thesis: *Heritability and Genetic Determinants of Normal Human Facial Shape and Body Size*
Applied quantitative genetics to estimate heritability and identify genetic determinants of normal human facial shape and body size from complementary multivariate phenotypes derived from 3D facial scan images and anthropometric measurements in over 6,000 African Bantu children from Tanzania. Graduate work resulted in nine publications (four first author) and two oral presentations (one student research award) at national conferences.

R&D Research Associate — Gaithersburg, MD — 2010-2011
KPL, Kirkegaard & Perry Labs., Inc.

- Developed and brought to market two new western blot products for mass production and a custom-ordered purified and fluorescently labeled antibody.
- Wrote manufacturing procedures, product information documents, and technical white notes for customers on ELISA and western blot techniques, trouble-shooting, and advanced applications.

Biotechnology Magnet Practicum Student — Clarksville, MD — 2002-2006
River Hill High School

- Intensive four-year high school technology magnet program with coursework and research at the Howard County Applications and Research Laboratory for 50% of my junior and senior years.
- Independent research project: sequence the unannotated *Rudbeckia hirta* NADH dehydrogenase F subunit gene.

Brain Tumor Center Intern — Baltimore, MD — 2005
Johns Hopkins Hospital

- Designed and instructed on a Microsoft Access patient database for personalized treatment plans.

Grants & Awards

NIDDK K99/R00 Pathway to Independence Award	2021-2026
Nutrition Obesity Research Center at Harvard Outstanding Manuscript Award	2020
American Diabetes Association Postdoctoral Fellowship	2019-2021
Peer Recognition Star Award, Boston Children's Hospital	2019
American Society of Human Genetics Reviewer's Choice Abstract	2018
Harvard/MGH Diabetes and Bioinformatics Training Fellow	2017-2019
Harvard/MGH Endocrine Training Fellow	2016-2017
Methods in Statistical Genetics Best Poster Presentation	2015
Gordon Research Seminar Abstract Travel Award	2015
Society for Pediatric Research Student Research Award at PAS 2015	2015
C. Werner and Kitty Hirs Research Travel Award	2015
Rockefeller University Gene Mapping Course Travel Award	2015
Human Medical Genetics Program Retreat Best Trainee Presentation	2014
Human Medical Genetics Program Retreat Poster Presentation – 2nd place	2014
AAAS member, Excellence in Science Program	2012
UAB Short Course on Statistical Genetics Travel Award	2012
Outstanding Student Award in the College of Chemical Life Sciences	2009-2010
Academic Honors, University of Maryland	2008-2010

Publications

Original Research Articles

1. Sandholm N*, **Cole JB***, Nair V, Sheng X, …, GENIE Consortium. (2021). Genome-wide meta-analysis and omics integration identifies novel genes associated with diabetic kidney disease. *Submitted.*
2. Wang Q, Li H, Tajima K, …, **Cole JB**, …, Kajimura S. (2021). Post-translational control of beige fat biogenesis and energy metabolism via PRDM16 protein stabilization. *Submitted.*
3. Westerman KE, Majarian TD, Giulianini F, Jang DK, …, **Cole JB** (2021). Variance-quantitative trait loci enable systematic discovery of gene-environment interactions for cardiometabolic serum biomarkers. *Nature Communications. Under review.*
4. Dornbos P, Koesterer K, Ruttenburg A, **Cole JB**, …, Flannick JA (2021). A combined polygenic score of 21,293 rare and 22 common variants significantly improves diabetes diagnosis based on hemoglobin A1C levels. *Nature Genetics. Under review.*
5. Null M, Yilmaz F, Astling D, …, **Cole JB**, …, Hendricks AE. (2021). Genome-wide analysis of copy number variants and normal facial variation in a large cohort of Bantu Africans. *Human Genetics and Genomics Advances*. *Accepted.*
6. DiCorpo D*, LeClair J*, **Cole JB***, Sarnowski C, …, Udler MS. (2021). Type 2 diabetes partitioned polygenic scores associate with disease outcomes in 454,193 individuals across 13 cohorts. *Diabetes Care*. *Accepted.*
7. O'Connor MJ, Heurta-Chagoya A, Cortes-Sanchez P, …, **Cole JB**, …, Mercader JM (2021). Genome-wide association meta-analysis using a recessive model illuminates genetic architecture of type 2 diabetes. *Diabetes* db210545.
8. Goodrich JK, Singer-Berk M, Son R, …, **Cole JB**, …, Udler MS (2021). Determinants of penetrance and variable expressivity in monogenic metabolic conditions across 77,184 exomes. *Nature Communications* 12(1):3505.
9. Yilmaz F, Null M, Astling D, …, **Cole JB**, …, Shaikh TH (2021). Genome-wide copy number variations in a large cohort of bantu African children. *BMC Medical Genomics* 14(1):129.
10. Smyth LJ, Kilner J, Nair V, …, **Cole JB**, …, McKnight AJ (2021). Assessment of differentially methylated loci in individuals with end-stage kidney disease attributed to diabetic kidney disease: an exploratory study. *Clinical Epigenetics* 13(1):99.
11. Guindo-Martinez M, Amela R, Bonas-Guarch S, …, **Cole JB**, …, Torrents D (2021). The impact of non-additive genetic associations on age-related complex diseases. *Nature Communications* 12(1):2436.
12. Westerman KE, Miao J, Chasman DI, Florez JC, …, **Cole JB** (2021). Genome-wide gene-diet interaction analysis in the UK Biobank identifies novel effects on Hemoglobin A1c. *Human Molecular Genetics* ddab109.
13. Haukka J, Sandholm N, Valo E, …, **Cole JB**, …, Groop PH (2021). Novel linkage peaks discovered for diabetic nephropathy in individuals with type 1 diabetes. *Diabetes* 70(4):986-995.

14. Topless RKG, Major TJ, Florez JC, …, **Cole JB***, Merriman T**** (2021). The comparative effect of exposure to various risk factors on the risk of hyperuricaemia: Diet has a weak causal effect. *Arthritis Research & Therapy* 23(1):1-12.
15. Leong A, **Cole JB**, Brenner LN, Meigs JB, …, Mercader JM (2021). Cardiometabolic risk factors for COVID-19 susceptibility and severity: A Mendelian randomization analysis. *PLOS Medicine* 183(3):e1003553.
16. Brenner LN, Mercader JM, Robertson CC, **Cole JB**, …, Florez JC (2020). Analysis of glucocorticoid-related genes reveal *CCHCR1* as a new candidate gene for type 2 diabetes. *Journal of the Endocrine Society* 4(11):bvaa121.
17. Hsu YH, Astley CM, **Cole JB**, Vedantam S, …, Hirschhorn JN (2020). Integrating untargeted metabolomics, genetically informed causal inference, and pathway enrichment to define the obesity metabolome. *International Journal of Obesity* 44(7):1596-1606.
18. Whiffin N, Armean IM, Kleinman A, …, **Cole JB**, …, MacArthur DG (2020). The effect of *LRRK2* loss-of-function variants in humans. *Nature Medicine* 26(6): 869-877.
19. **Cole JB**, Florez JC, Hirschhorn JN (2020). Comprehensive genomic analysis of dietary habits in UK Biobank identifies hundreds of genetic associations. *Nature Communications* 11(1):1-11.
    Nutrition Obesity Research Center at Harvard 2020 Outstanding Manuscript Award
20. Salem RM*, Todd JN*, Sandholm N*, **Cole JB***, …, GENIE Consortium (2019). Genome-wide association study of diabetic kidney disease highlights biology involved in glomerular basement membrane collagen. *Journal of the American Society of Nephrology* 30(10):2000-2016.
21. Udler MS, Kim J, Grotthus M, **Cole JB**, …, Florez JC (2018). Type 2 diabetes genetic loci informed by multi-trait associations point to disease mechanisms and subtypes: A soft clustering analysis. *PLoS Medicine* 15(9):e1002654.
22. **Cole JB**, Manyama MF, Nikitovic, D, Gonzalez PN, …, Hallgrimsson B (2018). Facial shape manifestations of growth faltering in Tanzanian children. *Journal of Anatomy* 232(2):250-262.
23. Larson JR, Manyama MF, **Cole JB**, Gonzalez PN, …, Hallgrimsson B (2018). Body size and allometric variation in facial shape in children. *American Journal of Physical Anthropology* 165(2):327-342.
24. **Cole JB**, Manyama MF, Larson JR, Liberton DK, …, Spritz RA (2017). Human facial shape and size heritability and genetic correlations. *Genetics* 205(2):967-978.
25. Li M, **Cole JB**, Manyama MF, Larson JR, …, Hallgrimsson B (2017). Rapid automated landmarking for morphometric analysis of three-dimensional facial scans. *Journal of Anatomy 230*(4):607-618.
26. **Cole JB**, Manyama MF, Kimwaga E, Mathayo J, …, Spritz RA (2016). Genomewide association study in African children identifies association of *SCHIP1* and *PDE8A* with facial size. *PLoS Genetics* 12(8):e1006174.
27. Shaffer JR, Orlova E, Lee MK, …, **Cole JB**, …, Weinberg SM (2016). Genome-wide association study reveals multiple loci influencing normal human facial morphology. *PLoS Genetics* 12(8):e1006149.
28. Jin Y, Birlea SA, Fain PR, …, **Cole JB**, …, Spritz RA (2012). Genome-wide association analyses identify 13 new susceptibility loci for generalized vitiligo. *Nature Genetics* 44(6):676-680.

Peer-reviewed Review Articles

29. **Cole JB** and Florez JC (2020). Genetics of diabetes mellitus and diabetes complications. *Nature Reviews Nephrology* 16(7):377-390.
30. **Cole JB** and Spritz RA (2017). The genetics of facial morphology. *Encyclopedia of Life Sciences* 2017:1-9.

*equal author contribution

Presentations

Genetics of Dietary Intake

Division of Preventive Medicine Faculty Seminar, Brigham and Women's Hospital. Invited Seminar	2021
American Society of Human Genetics Annual Meeting. Poster Presentation. **2018 Reviewer's Choice*	2017-2021
UK Biobank Scientific Conference, Virtual. Oral Poster Presentation.	2020
Institute for Behavioral Genetics, University of Colorado Boulder, Boulder, CO. Invited Seminar.	2020
Department of Mathematical and Statistical Sciences, University of Colorado Denver, Denver, CO. Invited Seminar.	2020
Mongan Institute Clinical and Translational Epidemiology Unit, Massachusetts General Hospital, Boston, MA. Invited Seminar.	2019
Trainee Retreat, The Broad Institute of MIT and Harvard, Boston, MA. Oral Presentation.	2019
Marcus Institute for Aging Research, Harvard Medical School, Boston, MA. Invited Seminar.	2019
American Diabetes Association Annual Meeting, San Francisco, CA. Oral Presentation.	2019

Human Medical Genetics and Genomics Program, University of Colorado Denver Anschutz Medical Campus, Aurora, CO. Invited Seminar. 2018

Genetics of Diabetic Kidney Disease
5th Annual Kidney-NExT5 Symposium, Cambridge, MA. Invited Presentation. 2019

Genetics of Normal Human Facial Shape
Framingham Heart Study Omics Conference Series, Boston, MA. Invited Seminar. 2016
American Society of Human Genetics Annual Meeting, Poster Presentation. 2014-2015
Gordon Research Conference Human Genetics & Genomics, Newport, RI. Oral Presentation. 2015
Pediatric Academic Societies Meeting, San Diego, CA. Oral Presentation. **Society for Pediatric Research Student Research Award* 2015
Methods in Statistical Genetics, University of Colorado Denver, Denver CO. Poster Presentation. **Best Poster Presentation* 2015
Human Medical Genetics and Genomics Program Retreat, Colorado Springs, CO. Oral Presentation. **Best Trainee Oral Presentation* 2014

Related Professional Experience

Positions
Frontiers in Genetics Journal Guest Editor (Special Topic in Nutrigenomics) 2021-Present
BCH Endocrinology Communications Committee Chair 2020-2021
Broad Institute Sustainability Group Co-founder and Executive Member 2018-2019
Broad Institute NextGen Trainee Association Vice-Chair 2017-2019
Broad Institute Mentoring Program Mentor 2017-2018
UCD Graduate School Dean Search Committee Member 2015
UCD Graduate Student Council Vice President 2014-2015
UCD Graduate Peer Mentor 2014-2015
UCD Graduate Program Curriculum Committee Member 2013-2015
UCD Graduate Program Admissions Committee Member 2013-2015

Professional Affiliations
American Diabetes Association member 2018-Present
American Society of Human Genetics member 2013-Present
Golden Key International Honor Society 2010-2016
Mortar Board National Senior Honor Society 2010
Primannum Honor Society 2010

Teaching Experience

Adjunct Professor, Molecular & Cellular Biology Lab — Denver, CO — 2016
Regis University

- Instructed 25 undergraduates through biology topics including enzymatic digestion, fermentation, photosynthesis, and bacterial transformation and ending with an independent research project.
- Managed and mentored an undergraduate teaching assistant.

Certificate, Learning How to Teach Workshop — Aurora, CO — 2015
NIH Broadening Experiences in Scientific Training

- Eight- week workshop covering How People Learn, Enhancing the Intercultural Classroom, Learning Outcomes and Objectives, Assessment and Grading, and Teaching Philosophies.

STEM Club Instructor — Lakewood, CO — 2014-2015
National Science Foundation STEM Club Program

- Prepared, budgeted, and taught biweekly STEM lessons to Green Mountain High School students. Activities included DNA extractions, forensics, chemical reactions, robotics, and career planning.

Independent Teaching Assistant, Genetics Discussion Denver, CO 2014
University of Colorado Denver

- Independent weekly instruction of 25 undergraduates for two semesters through introductory genetics topics.
- Mentored undergraduate and graduate teaching assistants.
- Guest lectured 100+ students on mutations, DNA repair mechanisms, and genetic diseases.

Independent Teaching Assistant, Molecular Biology Lab College Park, MD 2009-2010
University of Maryland

- Independent weekly instruction and course management of 25 undergraduates for three semesters through molecular biology laboratory experiments and poster presentations based on a recent molecular biology journal article.