

**Curriculum Vitae**  
**Joanne B. Cole, Ph.D.**

## 1. Personal History

### Primary Appointment:

Present Position: Assistant Professor  
Department of Biomedical Informatics  
University of Colorado School of Medicine  
Address: University of Colorado Anschutz Medical Campus  
1890 N Revere Ct, P12-7083  
Aurora, CO 80045  
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Email: <joanne.cole@cuanschutz.edu>

**Secondary Appointments:** Division of Renal Diseases and Hypertension, Department of Medicine, University of Colorado Anschutz Medical Campus; Colorado Center for Personalized Medicine, University of Colorado Anschutz Medical Campus

**Graduate School Appointments:** Training Faculty Member, Human Medical Genetics and Genomics Program, Biomedical Sciences Program, Biomedical Sciences and Biotechnology Program, University of Colorado Anschutz Medical Campus

## 2. Education

2006-2007 **Towson University, Towson, MD**  
2007-2010 **University of Maryland, College Park, MD**  
B.S. Cell Biology and Molecular Genetics

2011-2016 **University of Colorado Denver, Anschutz Medical Campus, Aurora, CO**  
Ph.D. Human Medical Genetics and Genomics

2016-2019 **The Broad Institute of MIT and Harvard, Massachusetts General Hospital, Boston Children's Hospital, Cambridge/Boston, MA**  
Postdoctoral Fellow, Medical and Population Genetics, Metabolism

### Additional Relevant Courses:

2002-2006 **Biotechnology Tech Magnet Program**, River Hill High School, Columbia, MD

2012 **NIGMS Short Course on Statistical Genetics & Genomics**, University of Alabama Birmingham School of Public Health, Birmingham, AL

2012 **Biostatistical Methods I**, University of Colorado School of Public Health, Aurora, CO

2013 **Biostatistical Methods II**, University of Colorado School of Public Health, Aurora, CO

2013 **Statistical Methods in Genetic Association**, University of Colorado School of Public Health, Aurora, CO

2015 **NIH BEST Learning How to Teach 8-Week Workshop**, University of Colorado Anschutz Medical Campus

- 2015                    **Topics in Probability & Statistics: Methods in Statistical Genetics**,  
University of Colorado Denver, Denver, CO
- 2015                    **Advanced Gene Mapping Course**, Rockefeller University, New York City, NY
- 2019                    **Nutritional Epidemiology I**, Harvard T.H. Chan School of Public Health,  
Boston, MA
- 2021                    **Bystander to Upstander Workshop**, Boston Children's Hospital, Virtual
- 2021                    **Nutritional Epidemiology II**, Harvard T.H. Chan School of Public Health,  
Boston, MA
- 2022                    **Mendelian Randomization Course**, University of Cambridge, Virtual
- 2022                    **Multicultural Mentoring**, University of Colorado Anschutz Medical Campus
- 2023                    **Mentorship Academy**, University of Colorado Anschutz Medical Campus

### 3. Academic Appointments

- 2016                    **Adjunct Faculty**, Regis University, Denver, CO
- 2019-2022            **Instructor in Investigation**, Department of Medicine, Massachusetts General  
Hospital/Harvard Medical School, Boston, MA
- 2022-present        **Assistant Professor**, Department of Biomedical Informatics, University of  
Colorado School of Medicine, Aurora, CO
- 2023-present        **Training Faculty Member**, Human Medical Genetics and Genomics Program,  
University of Colorado Anschutz Medical Campus
- 2023-present        **Training Faculty Member**, Biomedical Sciences Program, University of  
Colorado Anschutz Medical Campus
- 2023-present        **Training Faculty Member**, Biomedical Sciences and Biotechnology Program,  
University of Colorado Anschutz Medical Campus
- 2023-present        **Member**, Colorado Center for Personalized Medicine, University of Colorado  
Anschutz Medical Campus
- 2023-present        **Assistant Professor (Secondary Appointment)**, Division of Renal Diseases  
and Hypertension, Department of Medicine, University of Colorado Anschutz  
Medical Campus

### 4. Hospital, Government or Other Professional Positions

- 2005                    **Intern**, Brain Tumor Center, Johns Hopkins Hospital, Baltimore, MD
- 2010-2011            **R&D Associate I**, KPL, Kirkegaard & Perry Labs., Inc. (now SeraCare),  
Gaithersburg, MD

### 5. Honors and Awards

2008	Academic Honors, University of Maryland
2009-2010	Outstanding Student Award in the College of Chemical Life Sciences, University of Maryland
2010	Primannum Honor Society Mortar Board National Senior Honor Society
2010-2016	Golden Key International Honor Society
2012	Excellence in Science Program, AAAS Travel Award, NIGMS Short Course on Statistical Genetics & Genomics
2014	1 <sup>st</sup> place Trainee Oral Presentation Award & 2 <sup>nd</sup> place Trainee Poster Presentation, HMGGP Retreat
2015	Student Research Award, Society for Pediatric Research Travel Award, C. Werner and Kitty Hirs Research, University of Colorado Travel Award, Gordon Research Seminar Abstract 1 <sup>st</sup> place Poster Presentation Award, Methods in Statistical Genetics
2018	Reviewer's Choice Abstract, American Society of Human Genetics
2019	Peer Recognition Star Award, Boston Children's Hospital
2020	Outstanding Manuscript Award, Nutrition Obesity Research Center at Harvard
2023	Travel Award, CHARGE Consortium

## 6. Membership in Professional Organizations

2013-present	American Society of Human Genetics
2018, 2022-present	American Society for Nutrition
2018-2021	American Diabetes Association

## 7. Major Committee and Service Responsibilities

### Departmental & Program Committees:

2013-2015	Member, Admissions Committee, Human Medical Genetics and Genomics Program, University of Colorado Anschutz Medical Campus
2013-2015	Member, Curriculum Committee, Human Medical Genetics and Genomics Program, University of Colorado Anschutz Medical Campus
2020-2021	Chair, Communications Committee, Endocrinology Division, Boston Children's Hospital
2023-present	Chair, New PI Challenges Lunch Group, Department of Biomedical Informatics, University of Colorado Anschutz Medical Campus
2023-present	Faculty Member, Student Input and Feedback Forum, Human Medical Genetics and Genomics Program, University of Colorado Anschutz Medical Campus

### Institutional Committees:

2014-2015	Vice President, Graduate Student Council, University of Colorado Anschutz Medical Campus
2017-2019	Vice Chair, NextGen Trainee Association, Broad Institute of MIT and Harvard
2018-2019	Co-Founder and Executive Member, Sustainability Affinity Group, Broad Institute of MIT and Harvard

### National Committees: NA

### Expert Panels and Workshops: NA

### Community Service and Public Health Activities: NA

**Session Chair at Major Scientific Conferences and Workshops:**

2015                      Session Moderator, Gordon Research Seminar, Human Genetics and Genomics, Newport, RI

**8. Licensure and board certification**

NA

**9. Inventions, intellectual property and patents held or pending**

NA

**10. Review and Referee Work**

**Journal Editorial Service:**

2021-2022              Guest Associate Editor, Special Topic in Nutrigenomics, Frontiers in Genetics & Frontiers in Nutrition  
2023-present           Guest Associate Editor, Special Topic in Causal Inference and Mendelian Randomization, Frontiers in Nutrition

**Ad Hoc Journal Reviewer:**

BMJ Open  
Diabetes Research & Care  
Diabetes in America  
Food & Function  
Nature Genetics  
PLOS Genetics  
Scientific Reports  
Twin Research and Human Genetics  
Wellcome Open Research  
Gastrointestinal Disorders  
Journal of the American Society of Nephrology

**Grant Review and Study Section Membership:**

**Ad Hoc Grant Review:**

2022                      University of Colorado Diabetes Research Center Pilot and Feasibility Award

**11. Invited Extramural Lectures, Presentations and Visiting Professorships**

**Local**

2014                      Invited Speaker, Human Medical Genetics and Genomics Program Retreat, University of Colorado Anschutz Medical Campus, Colorado Springs, CO  
2017                      Invited Speaker, Diabetes Research Group Seminar, Broad Institute of MIT and Harvard, Cambridge, MA  
2018                      Invited Seminar, Division of Endocrinology, Boston Children's Hospital, Boston, MA  
2018                      Invited Speaker, Fusion-Oxford-Broad T2D Summit, Broad Institute of MIT and Harvard, Cambridge, MA  
2019                      Invited Speaker, 5<sup>th</sup> Annual Kidney-NExT5 Symposium, Broad Institute of MIT and Harvard, Cambridge, MA

- 2019 Invited Speaker (a), Medical and Population Genetics Seminar, Broad Institute of MIT and Harvard, Cambridge, MA
- 2019 Invited Speaker (b), Medical and Population Genetics Seminar, Broad Institute of MIT and Harvard, Cambridge, MA
- 2019 Invited Speaker, Diabetes Research Group Seminar, Broad Institute of MIT and Harvard, Cambridge, MA
- 2019 Oral Presentation, Trainee Retreat, Broad Institute of MIT and Harvard, Cambridge, MA
- 2022 Invited Speaker, Human Medical Genetics and Genomics Program Retreat, University of Colorado Anschutz Medical Campus, Colorado Springs, CO
- 2023 Invited Seminar, Section of Nutrition & Nutrition Obesity Research Center Seminar, University of Colorado Anschutz Medical Campus, Aurora, CO
- 2023 Invited Seminar, Human Medical Genetics and Genomics Program Seminar, University of Colorado Anschutz Medical Campus, Aurora, CO
- 2023 Invited Seminar, Division of Endocrinology, Metabolism and Diabetes Research Seminar, University of Colorado Anschutz Medical Campus, Aurora, CO
- 2023 Invited Seminar, Division of Renal Diseases and Hypertension Research Seminar, University of Colorado Anschutz Medical Campus, Aurora, CO

**Regional**

- 2019 Invited Seminar, Marcus Institute for Aging Research, Harvard Medical School, Boston, MA
- 2019 Invited Seminar, Mongan Institute and Clinical Translational Epidemiology Unit, Massachusetts General Hospital, Boston, MA
- 2020 Invited Seminar, Division of Preventive Medicine, Brigham and Women's Hospital
- 2022 Invited Seminar, AllRise Career Seminar Series, Division of Endocrinology, Boston Children's Hospital, Boston, MA

**National**

- 2015 Oral Presentation, Pediatric Academic Societies Meeting, San Diego, CA
- 2015 Oral Presentation, Gordon Research Conference: Human Genetics & Genomics, Newport, RI
- 2016 Invited Seminar, Framingham Heart Study Omics Conferences Series, Virtual
- 2018 Invited Seminar, Human Medical Genetics and Genomics Seminar Series, University of Colorado Anschutz Medical Campus, Aurora, CO
- 2019 Oral Presentation, American Diabetes Association Meeting, San Francisco, CA
- 2020 Invited Seminar, Department of Mathematical and Statistical Sciences, University of Colorado Denver, Denver, CO
- 2020 Invited Seminar, Institute for Behavioral Genetics, University of Colorado Boulder, Boulder, CO
- 2021 Invited Seminar, Department of Nutrition and Food Science, University of Maryland
- 2021 Invited Seminar, Center for Genetic Epidemiology, University of Southern California, Keck School of Medicine
- 2021 Invited Seminar, Bioinformatics & Integrative Biology Program, University of Massachusetts Chan Medical School
- 2022 Invited Seminar, Center for Quantitative Genetics, School of Biological Sciences, Georgia Institute of Technology
- 2022 Invited Seminar, Center for Genomic Health, Department of Genetics, Yale School of Medicine
- 2022 Invited Seminar, Department of Human Genetics, Emory School of Medicine

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| 2022 | Invited Seminar, Children's Nutrition Research Center, Baylor College of Medicine                     |
| 2022 | Invited Seminar, Department of Human Genetics, University of Chicago                                  |
| 2022 | Invited Seminar, Center for Health Artificial Intelligence, University of Colorado School of Medicine |
| 2023 | Oral Presentation, CHARGE Consortium Spring Meeting, Boston, MA                                       |
| 2023 | Oral Poster Presentation, American Society for Nutrition Meeting, Boston, MA                          |
| 2023 | Invited Seminar, Division of Human Genetics, Children's Hospital of Philadelphia, PA                  |

### International

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| 2023 | Invited Symposium Presentation, European Association for the Study of Diabetes, Hamburg, Germany |
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### 12. Teaching Record

**Program Director:** NA

**Course Director:** NA

#### Instructor of Record:

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|-----------|--|
| 2009-2010 | Instructor of Record Teaching Assistant, Principles of Molecular and Cellular Biology Laboratory, University of Maryland |
| 2014      | Instructor of Record Co-Teaching Assistant, General Genetics Recitation, University of Colorado Denver                   |
| 2016      | Adjunct Faculty, Molecular and Cellular Biology Laboratory, Regis University   |

#### Lectures in Courses/Teaching Seminars:

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|------|--|
| 2014 | General Genetics (BIOL3832) Lecture: Mutations, DNA Repair, and Genetic Diseases. University of Colorado Denver                              |
| 2023 | Foundations in Genetics (BSBT 6075) Lecture: Forward Genetics: GWAS. University of Colorado Anschutz Medical Campus                          |
| 2023 | Clinical Cytogenetics and Molecular Genetics (GENC 6120) Lecture: Genomics of Common Disease. University of Colorado Anschutz Medical Campus |

**Lectures to Lay Audiences:** NA

**High School/Undergraduate Mentor:** NA

**PhD Student Mentor:** NA

#### Postdoctoral/Medical Fellow Mentor:

- |              |   |  |
|--------------|---|--|
| 2018-2022    | Kenneth Westerman, PhD, Postdoctoral Fellow, Massachusetts General Hospital, The Broad Institute of MIT and Harvard<br>- NIDDK K01 Fellow<br>Current: Instructor, MGH | Primary Mentor: Manning<br>Role: Project and Career Mentor |
| 2022-present | Raymond Kreienkamp, MD/PhD, Pediatric Endocrine Fellow, Boston Children's Hospital  | Primary Mentors: Florez, Udler<br>Role: Project Mentor     |

- Pediatric Endocrine Society Rising Star Award
- 2023-present      Kristen James, PhD, Postdoctoral Fellow,      Role: Primary Mentor  
 University of Colorado
- NIH/NLM T15 Fellow in Bioinformatics
  - NIH/NLM T32 Fellow in Nutrition

**Research Committees: NA**

**Rotation Students:**

- 2023      Susana Lopez-Ignacio, BS, Graduate Student, Human Medical Genetics and Genomics PhD Program, University of Colorado Anschutz Medical Campus
- 2023      Shane Ridoux, BS, Graduate Student, PATHways in Genomic Data Science for Statistics & Applied Mathematics MS Programs, University of Colorado Denver

**Other Teaching Activities:**

- 2014-2016      High School STEM Club Leader, Green Mountain High School, Lakewood, CO, National Science Foundation STEM Club Program (PI: Michael Ferrara)

**13. Grant Support**

<u>Title</u>	<u>Period</u>	<u>Total Direct Funding</u>
<b>Active:</b> NIH-NIDDK: 4R00DK127196 <i>Genetically harmonized dietary intake and causal relationships with diabetes-related outcomes</i> Principal Investigator(s): Cole	01/10/2023- 11/30/2025	\$480,384
NIH-NIDDK: 1R01DK132299 <i>A functional genomics pipeline for genetic discovery in diabetic kidney disease</i> Principal Investigator: Jose Florez Role: Co-investigator	08/01/2022- 05/31/2027	\$84,935 (Sub-contract)
CU Ludeman Family Center for Women’s Health Early Career Faculty Research Development Award <i>Sex-specific genetic effects on lifestyle behavior and its causal role in obesity and diabetes risk</i> Principal Investigator(s): Cole	09/01/2023- 08/31/2024	\$25,000
Skaggs Scholar Program <i>Genetics of Drug-Induced Kidney Disease</i> Principal Investigator: Melanie Joy Role: Co-investigator	09/01/2023- 08/31/2025	\$150,000
<b>Completed:</b> NIH-NIDDK: 1K99DK127196	04/01/21-09/30/22	\$177,540

*Genetically harmonized dietary intake and causal relationships with diabetes-related outcomes*

Principal Investigator(s): Cole

American Diabetes Association Postdoctoral Fellowship: 01/01/19-3/31/21 \$161,650  
1-19-PDF-028

*Data-driven dietary habits: genetics, causality for diabetes and related traits, and transferability across studies*

Principal Investigator(s): Cole

NIH-NIDDK: 1T32DK110919 09/01/17-8/31/19 Salary support

*Harvard Training Program in Bioinformatics Applied to Diabetes, Obesity and Metabolism*

Principal Investigator(s): Florez, Patel  
Role: Trainee

NIH-NIDDK: 5T32DK007028 01/01/16-08/31/17 Salary support

*Massachusetts General Hospital Endocrinology Training Grant*

Principal Investigator(s): Avruch  
Role: Trainee

## 14. Bibliography

### Non-Competitive Abstracts (limited to first or senior author only):

1. **Cole JB**, Florez JC, Hirschhorn JN. Genetic architecture of data-driven dietary habits in UK Biobank highlights the importance of diverse phenotyping approaches. The Broad Institute of MIT and Harvard Annual Retreat, December 2018, Boston, MA. Poster Presentation.
2. **Cole JB**, Florez JC, Hirschhorn JN. Comprehensive genomic analysis of dietary intake identifies hundreds of genetic associations and supports causal relationships with lifestyle and human health. Massachusetts General Hospital Clinical Research Day, October 2020, Virtual. Poster Presentation.
3. Westerman KE, Udler MS, Florez JC, Manning AK, **Cole JB**. Systematic discovery of thousands of gene-environment interactions for cardiometabolic serum biomarkers. The Broad Institute of MIT and Harvard Annual Retreat, December 2020, Virtual. Poster Presentation.
4. **Cole JB**, Florez JC, Hirschhorn JN. Comprehensive genomic analysis of dietary intake elucidates relationships with metabolic disease. 9<sup>th</sup> Annual Obesity Research Incubator Session, May 2021, Virtual. Poster Presentation.
5. James K & **Cole JB**. Precise genetic instruments in Mendelian randomization improve causal estimates of complex traits. University of Colorado Department of Biomedical Informatics Annual Retreat, August 2023, Poster Presentation.

### Competitive Abstracts (limited to first or senior author only):

1. **Cole JB**, Manyama M, Larson J, Liberton DK, Ferrara TM, Riccardi SL, Li M, Mio W, Santorico SA, Hallgrimsson B, Spritz RA. Genetic Determinants of Normal Facial Variation. American Society of Human Genetics Annual Meeting, October 2014, San Diego, CA. Poster Presentation.
2. **Cole JB\***, Manyama M, Larson J, Liberton DK, Ferrara TM, Riccardi SL, Li M, Mio W, Santorico SA, Hallgrimsson B, Spritz RA. Genetic Determinants of Normal Facial Variation. Pediatric Academic Societies Annual Meeting, April 2015, San Diego, CA. Oral Presentation. \**Student Research Award*
3. **Cole JB**, Manyama M, Larson J, Liberton DK, Ferrara TM, Riccardi SL, Li M, Mio W, Santorico SA, Hallgrimsson B, Spritz RA. Genetic Determinants and Heritability of Normal Facial Variation. Gordon



- Research Conference: Human Genetics & Genomics, July 2015, Newport, RI. Oral and Poster Presentation.
4. **Cole JB**, Manyama. Mio W, Santorico SA, Hallgrímsson B, Spritz RA. Heritability of Normal Human Facial Shape. American Society of Human Genetics Annual Meeting, October 2015, Baltimore, MD. Poster Presentation.
  5. **Cole JB**, Florez JC, Hirschhorn JN. Data-driven approach to dietary phenotypes for nutrigenomics in UK Biobank. American Society of Human Genetics Annual Meeting, October 2017, Orlando, FL. Poster Presentation.
  6. **Cole JB**, Florez JC, Hirschhorn JN. Uncovering the genetic architecture of data-driven dietary habits in UK Biobank. American Society of Human Genetics Annual Meeting, October 2018, San Diego, CA. Poster Presentation. *\*Reviewer's Choice Award.*
  7. **Cole JB**, Florez JC, Hirschhorn JN. Genetic architecture of data-driven dietary habits in UK Biobank. Conference of the Program in Quantitative Genomics, November 2018, Boston, MA. Poster Presentation.
  8. **Cole JB**, Leong A, Florez JC, Hirschhorn JN. Genome-wide association study of food frequency questionnaire single nutrients and dietary patterns uncover hundreds of genetic associations. American Diabetes Association Annual Meeting, June 2019, San Francisco, CA. Oral Presentation.
  9. **Cole JB**, Florez JC, Hirschhorn JN. Comprehensive genomic analysis of data-driven dietary patterns in UK Biobank identifies novel loci and reveals causal associations. American Society of Human Genetics Annual Meeting, October 2019, Houston, TX. Poster Presentation.
  10. **Cole JB**, Florez JC, Hirschhorn JN. Comprehensive genomic analysis of data-driven dietary habits in UK Biobank identifies hundreds of novel loci and reveals causal associations. Keystone Symposium Conference: Beyond a Million Genomes: From Discovery to Precision Health, January 2020, Breckenridge, CO. Poster Presentation.
  11. **Cole JB**, Florez JC, Hirschhorn JN. Genomic Analysis of Dietary Intake identifies hundreds of novel associations and reveals causal relationships. UK Biobank Annual Scientific Conference, June 2020, Virtual. Poster Talk.
  12. Westerman KE, Chen Y, Chen H, Florez JC, **Cole JB**, Manning AK. Large-scale, genome-wide gene-diet interaction testing for HA1c using derived dietary patterns in the UK Biobank. American Society for Nutrition Annual Meeting, June 2020, Virtual. Poster presentation.
  13. **Cole JB**, Florez JC, Hirschhorn JN. Understanding how the genetics of dietary patterns in Europeans translates across ancestries. American Society of Human Genetics Annual Meeting, October 2020, Virtual. Poster Presentation.
  14. Westerman KE, Udler MS, Florez JC, Manning AK, **Cole JB**. Systematic discovery of thousands of gene-environment interactions for cardiometabolic serum biomarkers. CHARGE Consortium Meeting, May 2021, Virtual. Oral Presentation.
  15. Westerman KE, Udler MS, Florez JC, Manning AK, **Cole JB**. Systematic discovery of thousands of gene-environment interactions for cardiometabolic serum biomarkers. American Heart Association EpiLifestyle Meeting, June 2021, Virtual. Poster Talk.
  16. **Cole JB**, Florez JC, Hirschhorn JN. Novel phenotyping approaches: Empirical Bayes methods improve heritability of dietary phenotypes from repeat diet questionnaire data. American Society of Human Genetics Annual Meeting, October 2021, Virtual. Poster Presentation.
  17. Westerman KE, Giulianini F, Florez JC, Chen H, Chasman DI, Udler MS, Manning AK, **Cole JB**. Systematic discovery of gene-environment interactions for metabolic serum biomarkers. American Society of Human Genetics Annual Meeting, October 2021, Virtual. Oral Presentation. *\*KEW: Semifinalist for the Charles J. Epstein Trainee Award.*
  18. **Cole JB**, Westerman KE, Manning AK. Genetic heritability as a tool to optimize phenotyping decisions: An application in nutritional epidemiology. The Cohorts for Heart and Aging Research in Genomic Epidemiology (CHARGE) Consortium Spring Meeting, May 2023, Boston, MA. Oral Presentation. *\*CHARGE Travel Award.*
  19. **Cole JB**. Identifying genes with direct biological effects on dietary intake to improve personalized medicine. American Society for Nutrition Meeting, July 2023, Boston, MA. Oral Poster Presentation. *\*ASN Research & Media Highlight*

20. James K & **Cole JB**. Precise genetic instruments in Mendelian randomization improve causal estimates of complex traits. American Society of Human Genetics Annual Meeting, November 2023, Poster Presentation.

### Peer-Reviewed Publications (37)

1. Jin Y, Birlea SA, Fain PR, Ferrara TM, Ben S, Riccardi SL, **Cole JB**, Gowan K, Holland PJ, Bennett DC, Luiten RM, Wolkerstorfer A, van der Veen JP, Hartmann A, Eichner S, Schuler G, van Geel N, Lambert J, Kemp EH, Gawkrödger DJ, Weetman AP, Taïeb A, Jouary T, Ezzedine K, Wallace MR, McCormack WT, Picardo M, Leone G, Overbeck A, Silverberg NB, & Spritz RA. (2012). Genome-wide association analyses identify 13 new susceptibility loci for generalized vitiligo. *Nature Genetics*, 44(6), 676-680. doi:10.1038/ng.2272
2. **Cole JB**, Manyama M, Kimwaga E, Mathayo J, Larson JR, Liberton DK, Lukowiak K, Ferrara TM, Riccardi SL, Li M, Mio W, Prochazkova M, Williams T, Li H, Jones KL, Klein OD, Santorico SA, Hallgrímsson B, & Spritz RA. (2016). Genomewide Association Study of African Children Identifies Association of SCHIP1 and PDE8A with Facial Size and Shape. *PLOS Genetics*, 12(8), e1006174. doi:10.1371/journal.pgen.1006174
3. Shaffer JR, Orlova E, Lee MK, Leslie EJ, Raffensperger ZD, Heike CL, Cunningham ML, Hecht JT, Kau CH, Nidey NL, Moreno LM, Wehby GL, Murray JC, Laurie CA, Laurie CC, **Cole J**, Ferrara T, Santorico S, Klein O, Mio W, Feingold E, Hallgrímsson B, Spritz RA, Marazita ML, & Weinberg SM. (2016). Genome-Wide Association Study Reveals Multiple Loci Influencing Normal Human Facial Morphology. *PLOS Genetics*, 12(8), e1006149. doi:10.1371/journal.pgen.1006149
4. **Cole JB**, Manyama M, Larson JR, Liberton DK, Ferrara TM, Riccardi SL, Li M, Mio W, Klein OD, Santorico SA, Hallgrímsson B, & Spritz RA. (2017). Human Facial Shape and Size Heritability and Genetic Correlations. *Genetics*, 205(2), 967-978. doi:10.1534/genetics.116.193185
5. **Cole JB**, & Spritz RA. (2017). The genetics of facial morphology. *eLS*, 1-9. doi:10.1002/9780470015902.a0027240
6. Li M, **Cole JB**, Manyama M, Larson JR, Liberton DK, Riccardi SL, Ferrara TM, Santorico SA, Bannister JJ, Forkert ND, Spritz RA, Mio W, & Hallgrímsson B. (2017). Rapid automated landmarking for morphometric analysis of three-dimensional facial scans. *Journal of Anatomy*, 230(4), 607-618. doi:10.1111/joa.12576
7. **Cole JB**, Manyama MF, Nikitovic D, Gonzalez PN, Liberton DK, Wilson WM, Rolian C, Larson JR, Kimwaga E, Mathayo J, Roseman CC, Santorico SA, Lukowiak K, Spritz RA, & Hallgrímsson B. (2018). Facial shape manifestations of growth faltering in Tanzanian children. *Journal of Anatomy*, 232(2), 250-262. doi:10.1111/joa.12748
8. Larson JR, Manyama MF, **Cole JB**, Gonzalez PN, Percival CJ, Liberton DK, Ferrara TM, Riccardi SL, Kimwaga EA, Mathayo J, Spitzmacher JA, Rolian C, Jamniczky HA, Weinberg SM, Roseman CC, Klein O, Lukowiak K, Spritz RA, & Hallgrímsson B. (2018). Body size and allometric variation in facial shape in children. *American Journal of Physical Anthropology*, 165(2), 327-342. doi:10.1002/ajpa.23356
9. Udler MS, Kim J, von Grotthuss M, Bonàs-Guarch S, **Cole JB**, Chiou J, Christopher DAoboM, the I, Boehnke M, Laakso M, Atzmon G, Glaser B, Mercader JM, Gaulton K, Flannick J, Getz G, & 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. doi:10.1371/journal.pmed.1002654
10. Salem RM\*, Todd JN\*, Sandholm N\*, **Cole JB\***, Chen W-M, Andrews D, Pezzolesi MG, McKeigue PM, Hiraki LT, Qiu C, Nair V, Di Liao C, Cao JJ, Valo E, Onengut-Gumuscu S, Smiles AM, McGurnaghan SJ, Haukka JK, Harjutsalo V, Brennan EP, van Zuydam N, Ahlqvist E, Doyle R, Ahluwalia TS, Lajer M, Hughes MF, Park J, Skupien J, Spiliopoulou A, Liu A, Menon R, Boustany-Kari CM, Kang HM, Nelson RG, Klein R, Klein BE, Lee KE, Gao X, Mauer M, Maestroni S, Caramori ML, de Boer IH, Miller RG, Guo J, Boright AP, Tregouet D, Gyorgy B, Snell-Bergeon JK, Maahs DM, Bull SB, Canty AJ, Palmer CNA, Stechemesser L, Paulweber B, Weitgasser R, Sokolovska J, Rovite V, Pīrāgs V, Prakapiene E, Radzeviciene L, Verkauskiene R, Panduru NM, Groop LC, McCarthy MI, Gu HF, Möllsten A, Falhammar H, Brismar K, Martin F, Rossing P, Costacou T, Zerbini G, Marre M,

- Hadjadj S, McKnight AJ, Forsblom C, McKay G, Godson C, Maxwell AP, Kretzler M, Susztak K, Colhoun HM, Krolewski A, Paterson AD, Groop P-H, Rich SS, Hirschhorn JN, & Florez JC. (2019). Genome-Wide Association Study of Diabetic Kidney Disease Highlights Biology Involved in Glomerular Basement Membrane Collagen. *Journal of the American Society of Nephrology*, ASN.2019030218. doi:10.1681/asn.2019030218
11. Brenner LN, Mercader JM, Robertson CC, **Cole J**, Chen L, Jacobs SB, Rich SS, & Florez JC. (2020). Analysis of glucocorticoid-related genes reveal CCHCR1 as a new candidate gene for type 2 diabetes. *Journal of the Endocrine Society*.
  12. **Cole JB**, & Florez JC. (2020). Genetics of diabetes mellitus and diabetes complications. *Nature Reviews Nephrology*. doi:10.1038/s41581-020-0278-5
  13. **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), 1467. doi:10.1038/s41467-020-15193-0 \*Outstanding Manuscript Award, Nutrition Obesity Research Center at Harvard.
  14. Hsu Y-HH, Astley CM, **Cole JB**, Vedantam S, Mercader JM, Metspalu A, Fischer K, Fortney K, Morgen EK, Gonzalez C, Gonzalez ME, Esko T, & 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. doi:10.1038/s41366-020-0603-x
  15. Whiffin N, Armean IM, Kleinman A, Marshall JL, Minikel EV, Goodrich JK, Quaipe NM, **Cole JB**, Wang Q, Karczewski KJ, Cummings BB, Francioli L, Laricchia K, Guan A, Alipanahi B, Morrison P, Baptista MAS, Merchant KM, Ware JS, Havulinna AS, Iliadou B, Lee JJ, Nadkarni GN, Whiteman C, Daly M, Esko T, Hultman C, Loos RJJ, Milani L, Palotie A, Pato C, Pato M, Saleheen D, Sullivan PF, Alfoldi J, Cannon P, & MacArthur DG. (2020). The effect of LRRK2 loss-of-function variants in humans. *Nature Medicine*, 26(6), 869-877. doi:10.1038/s41591-020-0893-5
  16. Goodrich JK, Singer-Berk M, Son R, Sveden A, Wood J, England E, **Cole JB**, Weisburd B, Watts N, Caulkins L, Dornbos P, Koesterer R, Zappala Z, Zhang H, Maloney KA, Dahl A, Aguilar-Salinas CA, Atzmon G, Barajas-Olmos F, Barzilai N, Blangero J, Boerwinkle E, Bonnycastle LL, Bottinger E, Bowden DW, Centeno-Cruz F, Chambers JC, Chami N, Chan E, Chan J, Cheng CY, Cho YS, Contreras-Cubas C, Córdova E, Correa A, DeFronzo RA, Duggirala R, Dupuis J, Garay-Sevilla ME, García-Ortiz H, Gieger C, Glaser B, González-Villalpando C, Gonzalez ME, Grarup N, Groop L, Gross M, Haiman C, Han S, Hanis CL, Hansen T, Heard-Costa NL, Henderson BE, Hernandez JMM, Hwang MY, Islas-Andrade S, Jørgensen ME, Kang HM, Kim BJ, Kim YJ, Koistinen HA, Kooner JS, Kuusisto J, Kwak SH, Laakso M, Lange L, Lee JY, Lee J, Lehman DM, Linneberg A, Liu J, Loos RJJ, Lyssenko V, Ma RCW, Martínez-Hernández A, Meigs JB, Meitinger T, Mendoza-Caamal E, Mohlke KL, Morris AD, Morrison AC, Ng MCY, Nilsson PM, O'Donnell CJ, Orozco L, Palmer CNA, Park KS, Post WS, Pedersen O, Preuss M, Psaty BM, Reiner AP, Revilla-Monsalve C, Rich SS, Rotter JI, Saleheen D, Schurmann C, Sim X, Sladek R, Small KS, So WY, Spector TD, Strauch K, Strom TM, Tai ES, Tam CHT, Teo YY, Thameem F, Tomlinson B, Tracy RP, Tuomi T, Tuomilehto J, Tusié-Luna T, van Dam RM, Vasani RS, Wilson JG, Witte DR, Wong TY, Burt NP, Zaitlen N, McCarthy MI, Boehnke M, Pollin TI, Flannick J, Mercader JM, O'Donnell-Luria A, Baxter S, Florez JC, MacArthur DG, & Udrler MS. (2021). Determinants of penetrance and variable expressivity in monogenic metabolic conditions across 77,184 exomes. *Nature Communications*, 12(1), 3505. doi:10.1038/s41467-021-23556-4
  17. Guindo-Martínez M, Amela R, Bonàs-Guarch S, Puiggròs M, Salvoro C, Miguel-Escalada I, Carey CE, **Cole JB**, Rüeger S, Atkinson E, Leong A, Sanchez F, Ramon-Cortes C, Ejarque J, Palmer DS, Kurki M, Aragam K, Florez JC, Badia RM, Mercader JM, & Torrents D. (2021). The impact of non-additive genetic associations on age-related complex diseases. *Nature Communications*, 12(1), 2436. doi:10.1038/s41467-021-21952-4
  18. Haukka J, Sandholm N, Valo E, Forsblom C, Harjutsalo V, **Cole JB**, McGurnaghan SJ, Colhoun HM, & Groop P-H. (2021). Novel linkage peaks discovered for diabetic nephropathy in individuals with type 1 diabetes. *Diabetes*, 70(4), 986-995.
  19. Leong A, **Cole JB**, Brenner LN, Meigs JB, Florez JC, & Mercader JM. (2021). Cardiometabolic risk factors for COVID-19 susceptibility and severity: A Mendelian randomization analysis. *PLoS Medicine*, 18(3), e1003553. doi:10.1371/journal.pmed.1003553

20. Smyth LJ, Kilner J, Nair V, Liu H, Brennan E, Kerr K, Sandholm N, **Cole J**, Dahlström E, Syreeni A, Salem RM, Nelson RG, Looker HC, Wooster C, Anderson K, McKay GJ, Kee F, Young I, Andrews D, Forsblom C, Hirschhorn JN, Godson C, Groop PH, Maxwell AP, Susztak K, Kretzler M, Florez JC, & 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. doi:10.1186/s13148-021-01081-x
21. Topless RKG, Major TJ, Florez JC, Hirschhorn JN, Cadzow M, Dalbeth N, Stamp LK, Wilcox PL, Reynolds RJ, **Cole JB\***, & Merriman TR\*. (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), 75. doi:10.1186/s13075-021-02444-8
22. Westerman KE, Miao J, Chasman DI, Florez JC, Chen H, Manning AK, & **Cole JB**. (2021). Genome-wide gene–diet interaction analysis in the UK Biobank identifies novel effects on hemoglobin A1c. *Human Molecular Genetics*. doi:10.1093/hmg/ddab109
23. Yilmaz F, Null M, Astling D, Yu HC, Cole J, Santorico SA, Hallgrímsson B, Manyama M, Spritz RA, Hendricks AE, & Shaikh TH. (2021). Genome-wide copy number variations in a large cohort of bantu African children. *BMC Medical Genomics*, 14(1), 129. doi:10.1186/s12920-021-00978-z
24. **Cole JB**, & Gabbianelli R. (2022). Editorial: Recent advances in nutrigenomics: Making strides towards precision nutrition. *Front Genet*, 13, 997266. doi:10.3389/fgene.2022.997266
25. DiCorpo D\*, LeClair J\*, **Cole JB\***, Sarnowski C, Ahmadizar F, Bielak LF, Blokstra A, Bottinger EP, Chaker L, Chen YI, Chen Y, de Vries PS, Faquih T, Ghanbari M, Gudmundsdottir V, Guo X, Hasbani NR, Ibi D, Ikram MA, Kavousi M, Leonard HL, Leong A, Mercader JM, Morrison AC, Nadkarni GN, Nalls MA, Noordam R, Preuss M, Smith JA, Trompet S, Vissink P, Yao J, Zhao W, Boerwinkle E, Goodarzi MO, Gudnason V, Jukema JW, Kardia SLR, Loos RJF, Liu CT, Manning AK, Mook-Kanamori D, Pankow JS, Picavet HSJ, Sattar N, Simonsick EM, Verschuren WMM, Willems van Dijk K, Florez JC, Rotter JI, Meigs JB, Dupuis J, & Udlér MS. (2022). Type 2 Diabetes Partitioned Polygenic Scores Associate With Disease Outcomes in 454,193 Individuals Across 13 Cohorts. *Diabetes Care*, 45(3), 674-683. doi:10.2337/dc21-1395
26. Dornbos P, Koesterer R, Rutténburg A, Nguyen T, **Cole JB**, AMP-T2D Consortium, Leong A, Meigs JB, Florez JC, Rotter JI, Udlér MS, & Flannick J. (2022). A combined polygenic score of 21,293 rare and 22 common variants improves diabetes diagnosis based on hemoglobin A1C levels. *Nature Genetics*. doi:10.1038/s41588-022-01200-1
27. Kim H, Westerman KE, Smith K, Chiou J, **Cole JB**, Majarian T, von Grotthuss M, Kwak SH, Kim J, Mercader JM, Florez JC, Gaulton K, Manning AK, & Udlér MS. (2022). High-throughput genetic clustering of type 2 diabetes loci reveals heterogeneous mechanistic pathways of metabolic disease. *Diabetologia*. doi:10.1007/s00125-022-05848-6
28. Null M, Yilmaz F, Astling D, Yu H-C, **Cole JB**, Hallgrímsson B, Santorico SA, Spritz RA, Shaikh TH, & Hendricks AE. (2022). Genome-wide analysis of copy number variants and normal facial variation in a large cohort of Bantu Africans. *Human Genetics and Genomics Advances*, 3(1), 100082. doi:10.1016/j.xhgg.2021.100082
29. O'Connor MJ, Schroeder P, Huerta-Chagoya A, Cortés-Sánchez P, Bonàs-Guarch S, Guindo-Martínez M, **Cole JB**, Kaur V, Torrents D, Veerapen K, Grarup N, Kurki M, Rundsten CF, Pedersen O, Brandslund I, Linneberg A, Hansen T, Leong A, Florez JC, & Mercader JM. (2022). Recessive Genome-Wide Meta-analysis Illuminates Genetic Architecture of Type 2 Diabetes. *Diabetes*, 71(3), 554-565. doi:10.2337/db21-0545
30. Sandholm N\*, **Cole JB\***, Nair V, Sheng X, Liu H, Ahlqvist E, van Zuydam N, Dahlström EH, Fermin D, Smyth LJ, Salem RM, Forsblom C, Valo E, Harjutsalo V, Brennan EP, McKay GJ, Andrews D, Doyle R, Looker HC, Nelson RG, Palmer C, McKnight AJ, Godson C, Maxwell AP, Groop L, McCarthy MI, Kretzler M, Susztak K, Hirschhorn JN, Florez JC, & Groop PH. (2022). Genome-wide meta-analysis and omics integration identifies novel genes associated with diabetic kidney disease. *Diabetologia*, 65(9), 1495-1509. doi:10.1007/s00125-022-05735-0
31. Shah RV, Steffen LM, Naylor M, Reis JP, Jacobs DR, Jr., Allen NB, Lloyd-Jones D, Meyer K, **Cole J**, Piaggi P, Vasán RS, Clish CB, & Murthy VL. (2022). Dietary metabolic signatures and cardiometabolic risk. *European Heart Journal*. doi:10.1093/eurheartj/ehac446

32. Smyth LJ, Dahlström EH, Syreeni A, Kerr K, Kilner J, Doyle R, Brennan E, Nair V, Fermin D, Nelson RG, Looker HC, Wooster C, Andrews D, Anderson K, McKay GJ, **Cole JB**, Salem RM, Conlon PJ, Kretzler M, Hirschhorn JN, Sadlier D, Godson C, Florez JC, Forsblom C, Maxwell AP, Groop PH, Sandholm N, & McKnight AJ. (2022). Epigenome-wide meta-analysis identifies DNA methylation biomarkers associated with diabetic kidney disease. *Nature Communications*, 13(1), 7891. doi:10.1038/s41467-022-34963-6
33. Wang Q, Li H, Tajima K, Verkerke ARP, Taxin ZH, Hou Z, **Cole JB**, Li F, Wong J, Abe I, Pradhan RN, Yamamuro T, Yoneshiro T, Hirschhorn JN, & Kajimura S. (2022). Post-translational control of beige fat biogenesis by PRDM16 stabilization. *Nature*, 609(7925), 151-158. doi:10.1038/s41586-022-05067-4
34. Westerman KE, Majarian TD, Giulianini F, Jang D-K, Miao J, Florez JC, Chen H, Chasman DI, Udler MS, Manning AK, & **Cole JB**. (2022). Variance-quantitative trait loci enable systematic discovery of gene-environment interactions for cardiometabolic serum biomarkers. *Nature Communications*, 13(1), 3993. doi:10.1038/s41467-022-31625-5
35. **Cole JB**, Westerman KE, Manning AK, Florez JC, & Hirschhorn JN. (2023). Genetic heritability as a tool to evaluate the precision of 24-hour recall dietary questionnaire variables in UK Biobank. *Frontiers in Genetics*, 13. doi:10.3389/fgene.2022.1070511
36. Cromer SJ, Lakhani CM, Mercader JM, Majarian TD, Schroeder P, **Cole JB**, Florez JC, Patel CJ, Manning AK, & Burnett-Bowie S-AM. (2023). Association and Interaction of Genetics and Area-Level Socioeconomic Factors on the Prevalence of Type 2 Diabetes and Obesity. *Diabetes Care*, dc221954.
37. Huerta-Chagoya A, Schroeder P, Mandla R, Deutsch AJ, Zhu W, Petty L, Yi X, **Cole JB**, Udler MS, Dornbos P, Porneala B, DiCorpo D, Liu CT, Li JH, Szczerbiński L, Kaur V, Kim J, Lu Y, Martin A, Eizirik DL, Marchetti P, Marselli L, Chen L, Srinivasan S, Todd J, Flannick J, Gubitosi-Klug R, Levitsky L, Shah R, Kelsey M, Burke B, Dabelea DM, Divers J, Marcovina S, Stalbow L, Loos RJJ, Darst BF, Kooperberg C, Raffield LM, Haiman C, Sun Q, McCormick JB, Fisher-Hoch SP, Ordoñez ML, Meigs J, Baier LJ, González-Villalpando C, González-Villalpando ME, Orozco L, García-García L, Moreno-Estrada A, Aguilar-Salinas CA, Tusié T, Dupuis J, Ng MCY, Manning A, Highland HM, Cnop M, Hanson R, Below J, Florez JC, Leong A, & Mercader JM. (2023). The power of TOPMed imputation for the discovery of Latino-enriched rare variants associated with type 2 diabetes. *Diabetologia*. doi:10.1007/s00125-023-05912-9