Stanford

Stephen B. Montgomery

Stanford Medicine Professor of Pathology, Professor of Genetics and of Biomedical Data Science and, by courtesy, of Computer Science

CONTACT INFORMATION

- **Alternate Contact**

  Char Armitage - Administrative Assistant

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Bio

BIO

Stephen Montgomery is an Endowed Professor of Pathology, Genetics, Biomedical Data Science and, by courtesy, Computer Science at Stanford University. He has trained in multiple countries including Canada, Germany, England, and Switzerland. He is best known for his work mapping the effects of genetic variation to gene expression and authored the first publications that compared whole genomes and transcriptome data within a human population and pioneered the use of molecular outliers to identify impactful rare variants (Montgomery et al, 2010, Montgomery et al, 2011).

Montgomery and his lab lead major genomics initiatives to understand the molecular mechanisms that underlie disease-associated variation. In 2017, they published analyses from the Genotype-Tissue-Expression (GTEx) Consortium which analyzed the impact of genetic variation on gene expression across tissues of the human body (GTEx Consortium, 2017). In 2024, his lab led major analyses in the NIH Common Fund MoTrPAC study identifying the molecular effects of exercise training across rat tissues (MoTrPAC, 2024). He is a Principal Investigator within multiple major NIH consortia including the GREGoR, MoTrPAC, TOPMed and Functional ADSP consortia and an Investigator in the Developmental GTEx, IGVF, SMaHT, AllOfUs, Undiagnosed Disease Network and ENCODE4 consortia demonstrating his labs ongoing impacts in multiple major genomics projects.

The Montgomery lab has a specific focus on mapping the molecular effects of rare and environment-responsive genetic variants. Work in his laboratory focuses on developing approaches for studying rare variants (such as Li et al, 2017; Ferraro et al, 2020) and seeing these approaches applied to understanding novel disease biology and providing diagnoses of individuals with genetic diseases (Fresard et al., 2017). As a PI of the GREGoR Stanford Site, his lab develops and applies these strategies to diagnose individuals with undiagnosed, rare diseases. The GREGoR Stanford site is currently recruiting 500 families with unsolved diagnoses in California to apply novel multi-omics and computational strategies to acheive diagnoses. His laboratory further has a specific focus on understanding the molecular consequences of structural variants and chromosomal copy number changes (Marderstein et al, 2024).

The Montgomery lab is also focused on advancing our understanding of common genetic variants and understudied RNAs. Examples of this work, his lab has demonstrated that multiple genetic variants contribute to genetic disease associations (Abell et al, 2022) and his lab has developed approaches to identify impactful long non-coding RNAs that contribute to complex disease (de Goede et al, 2021). Ongoing effort in his lab has focused on neurodegenerative and neurodevelopmental traits.

Montgomery is an active member of both the Stanford and broader research community. Among his contributions, he serves as a co-director of an NHGRI PhD T32 Training Grant, Faculty Director of Graduate Admissions for the Biomedical Data Science program and served for 4 years as a Stanford University Faculty Senator. He has/or is currently on the programming committee for major conferences such as ASHG, AGBT and WTSI Genomics of Rare Diseases. He is the incoming chair for the ASHG Awards committee. He is also a standing member of the NIH GHD Study Section.

In 2019, Montgomery was awarded the annual American Society of Human Genetics Early Career Award for his multi-faceted impacts on human genetics and genomics. In 2023, he was awarded the annual Stanford Prize in Population Genetics and Society. In 2024, he was awarded the Stanford Pathology Research Mentor Award.

ACADEMIC APPOINTMENTS

- Professor, Pathology
- Professor, Genetics
- Professor, Department of Biomedical Data Science
- Professor (By courtesy), Computer Science
- Member, Bio-X
- Member, Cardiovascular Institute
- Member, Wu Tsai Human Performance Alliance
- Member, Maternal & Child Health Research Institute (MCHRI)
- Member, Stanford Medicine Children’s Health Center for IBD and Celiac Disease
- Member, Wu Tsai Neurosciences Institute

ADMINISTRATIVE APPOINTMENTS

- Director of Genome Informatics, Department of Pathology, (2011- present)

PROFESSIONAL EDUCATION

- B.A.Sc., University of British Columbia , Engineering Physics (2002)
- Ph.D., University of British Columbia , Genetics (2006)

LINKS

- Montgomery Lab Official Website: http://montgomerylab.stanford.edu/
- GREGoR Stanford Site: https://gregor.stanford.edu/

Research & Scholarship

CURRENT RESEARCH AND SCHOLARLY INTERESTS

We focus on understanding the effects of genome variation on cellular phenotypes and cellular modeling of disease through genomic approaches such as next generation RNA sequencing in combination with developing and utilizing state-of-the-art bioinformatics and statistical genetics approaches. See our website at http://montgomerylab.stanford.edu/

Teaching

COURSES

2025-26

- Practical Application of AI/ML to Healthcare and Biotechnology: BMDS 283 (Spr)

2023-24

- Informatics in Industry: BIOMEDIN 206 (Spr)

2022-23

- Informatics in Industry: BIOMEDIN 206 (Spr)

STANFORD ADVISEES

Doctoral Dissertation Reader (AC)

Vidal Arroyo, Jon Bezney, Ziwei Chen, Tania Fabo, Karen Feng, Michael Hayes, Jodie Lunger, Aman Patel, Imani Porter, Taylor Pursell, Alp Tartici

Postdoctoral Faculty Sponsor

Iman Jaljuli, Evin Padhi, Yilin Xie

Doctoral Dissertation Advisor (AC)

Maggie Arriaga, Sohaib Hassan, Ronit Jain, Julie Lake, Kate Lawrence, Victoria Rosa, Sherry Yang

Orals Evaluator

Aman Patel

Doctoral Dissertation Co-Advisor (AC)

Jordan Cahoon, Haim Krupkin

Master's Program Advisor

Nate Demchak, Daniel Guo, Robert Igbokwe, Ananya Vasireddy, Patrick Walsh

Doctoral (Program)

Alex Belov, Sophia Kivelson, Esther Robb, Min Sun, Christine Yiwen Yeh

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Biomedical Data Science (Phd Program)
- Genetics (Phd Program)

Publications

PUBLICATIONS

- **Publisher Correction: The impact of exercise on gene regulation in association with complex trait genetics.** *Nature communications*
  Vetr, N. G., Gay, N. R., Montgomery, S. B.
  2026; 17 (1)

- **The Long Non-coding RNA Landscape of Endurance Exercise Training.** *Molecular metabolism*
  Bonilauri, B., Smith, G. R., Raja, A. N., Jimenez-Morales, D., Ahmed, A., Jin, C., Sparks, L. M., Walsh, M. J., Montgomery, S. B., Bodine, S. C., Ashley, E. A., Lindholm, M. E.
  2026: 102358

- **De Novo Variants Associated With Autosomal Recessive Conditions: Case Series and Implications for Genetic Testing and Counseling.** *American journal of medical genetics. Part A*
  Niehaus, A. D., Bonner, D. E., Carter, J., Avello, K., Jacob, N., Neu, M. B., Mendez, R., Qiao, W., Scott, S. A., Levy, R. J., Mattas, L., Schymick, J., Van Andel, et al
  2026

- **Multi-ancestry transcriptome prediction with functionally informed variants in TOPMed MESA improves performance of transcriptome-wide association studies.** *American journal of human genetics*

Hu, X., Araujo, D. S., Khunsriraksakul, C., Wang, L., Sun, Q., Wen, J., Zhou, L., Ekunwe, L., Lange, L. A., Lange, E. M., Montgomery, S. B., Reiner, A. P., Aguet, et al

2026; 113 (4): 828-841

- **Focus on single-gene effects limits discovery and interpretation of complex-trait-associated variants.** *American journal of human genetics*

  Lawrence, K. A., Gjorgjieva, T., Nachun, D., Montgomery, S. B.

  2026

- **Multi-omic identification of key transcriptional regulatory programs during endurance exercise training in rats.** *Nature communications*

  Smith, G. R., Zhao, B., Lindholm, M. E., Raja, A., Viggars, M., Pincas, H., Gay, N. R., Sun, Y., Vangeti, S., Ge, Y., Nair, V. D., Sanford, J. A., Amper, et al

  2026

- **Biallelic Variants in RNU6ATAC Result in a Minor Spliceopathy Characterized by Transcriptome-Wide Minor Intron Retention Events and Short Stature with Variable Multisystem Manifestations.** *HGG advances*

  Mendez, R., Arriaga, T. M., Ma, J., Bonner, D. E., Emami, S., Levy, R. J., Alsagheir, A., Alhaddad, B., Bakur, K., Ungar, R. A., Matalon, D. R., Miller, A. M., Nguyen, et al

  2026: 100588

- **Biallelic LAMP3 Variants in Five Families with Interstitial Lung Disease: Evidence of a Disease-Gene Association.** *Genetics in medicine : official journal of the American College of Medical Genetics*

  Keehan, L. A., Ono-Minagi, H., Hadhud, M., Rips, J., Hinds, D. M., Fischer, A. J., Bartlett, J. A., McCray, P. B., Qawasmi, N., Nathan, N., Louvrier, C., Desroziers, T., Damme, et al

  2026: 102531

- **Multi-omics analysis of endurance exercise reveals cardioprotective remodeling in rat heart**

  Brochet, P., Njoroge, J., Montalvo Hernandez, S., Lindholm, M., Smith, G., Amar, D., Gay, N., Zhao, B., Hung, C., Jin, C., Chavez, C., Nachun, D., Zaslavsky, et al

  LIPPINCOTT WILLIAMS & WILKINS.2025

- **Long-read, multi-omics resource uncovers structural variants driving molecular trait associations and neurodegenerative disease risk**

  Jensen, T., Le Guen, Y., Talozzi, L., Yang, S., Gorzynski, J., Tauber, A. P., Ashley, E. A., Montgomery, S., Greicius, M. D.

  SPRINGERNATURE.2025: 52

- **GREGoR: accelerating genomics for rare diseases.** *Nature*

  Dawood, M., Heavner, B., Wheeler, M. M., Ungar, R. A., LoTempio, J., Wiel, L., Berger, S., Bernstein, J. A., Chong, J. X., Délot, E. C., Eichler, E. E., Lupski, J. R., Shojaie, et al

  2025; 647 (8089): 331-342

- **Long-read genome sequencing and multi-omics in aging and neurodegeneration.** *medRxiv : the preprint server for health sciences*

  Jensen, T. D., Le Guen, Y., Talozzi, L., Yang, S., Gorzynski, J., Peña-Tauber, A., Stewart, I., Ferrasse, A., Nachun, D., Arriaga, M. T., Lee, J., Pulgrossi, R. C., Park, et al

  2025

- **The Long Non-coding RNA Landscape of Endurance Exercise Training.** *bioRxiv : the preprint server for biology*

  Bonilauri, B., Smith, G. R., Raja, A. N., Jimenez-Morales, D., Ahmed, A., Jin, C., Sparks, L. M., Walsh, M. J., Montgomery, S. B., Bodine, S. C., Ashley, E. A., Lindholm, M. E.

  2025

- **Mapping causal non-coding variants in coronary artery disease.** *Nature cardiovascular research*

  Montgomery, S. B.

  2025

- **Regulatory genomics at biobank scales.** *Nature reviews. Genetics*

  Montgomery, S. B.

  2025; 26 (10): 657-658

- **Transcriptome-wide outlier approach identifies individuals with minor spliceopathies.** *American journal of human genetics*

  Arriaga, T. M., Mendez, R., Ungar, R. A., Bonner, D. E., Matalon, D. R., Lemire, G., Goddard, P. C., Padhi, E. M., Miller, A. M., Nguyen, J. V., Ma, J., Smith, K. S., Scott, et al

  2025

- **Interactions Between Dietary Metabolites and Regulatory Risk Variants for Human Colon Cancer.** *bioRxiv : the preprint server for biology*
  Fabo, T. N., Meyers, R. M., Padhi, E., Kellman, L. N., Zhao, Y., Kundu, S., Reynolds, D. L., Chen, Z., Yang, X., Ko, L., Elfaki, I., Montgomery, S. B., Khavari, et al
  2025

- **Disruption of the cerebrospinal fluid-plasma protein balance in cognitive impairment and aging.** *Nature medicine*
  Farinas, A., Rutledge, J., Bot, V. A., Western, D., Ying, K., Lawrence, K. A., Oh, H. S., Yoon, S., Ding, D. Y., Tsai, A. P., Moran-Losada, P., Timsina, J., Le Guen, et al
  2025

- **The Somatic Mosaicism across Human Tissues Network.** *Nature*
  Coorens, T. H., Oh, J. W., Choi, Y. A., Lim, N. S., Zhao, B., Voshall, A., Abyzov, A., Antonacci-Fulton, L., Aparicio, S., Ardlie, K. G., Bell, T. J., Bennett, J. T., Bernstein, et al
  2025; 643 (8070): 47-59

- **Toward optimizing diversifying base editors for high-throughput mutational scanning studies.** *Nucleic acids research*
  Schwartz, C. I., Abell, N. S., Li, A., , Tycko, J., Truong, A., Montgomery, S. B., Hess, G. T.
  2025; 53 (12)

- **The evolutionarily conserved PRP4K-CHMP4B/vps32 splicing circuit regulates autophagy.** *Cell reports*
  Mathavarajah, S., Chipurupalli, S., Habib, E. B., Kim, W. D., Aoki, M. M., Corkery, D. P., Whelan, K. I., Lukacs, J., Erkan, M., Martinez, V. D., Smith, K. S., Montgomery, S. B., Salsman, et al
  2025; 44 (7): 115870

- **Focus on single gene effects limits discovery and interpretation of complex trait-associated variants.** *bioRxiv : the preprint server for biology*
  Lawrence, K., Gjorgjieva, T., Montgomery, S. B.
  2025

- **Transcriptomic signatures of rare variant impacts across sex and the X-chromosome.** *HGG advances*
  Ungar, R. A., Li, T., Vetr, N. G., Ersaro, N., Battle, A., Montgomery, S. B.
  2025: 100463

- **Predicting expression-altering promoter mutations with deep learning.** *Science (New York, N.Y.)*
  Jaganathan, K., Ersaro, N., Novakovsky, G., Wang, Y., James, T., Schwartzentruber, J., Fiziev, P., Kassam, I., Cao, F., Hawe, J., Cavanagh, H., Lim, A., Png, et al
  2025: eads7373

- **Integrated single-cell multiome analysis reveals muscle fiber-type gene regulatory circuitry modulated by endurance exercise.** *Genome research*
  Rubenstein, A. B., Smith, G. R., Zhang, Z., Chen, X., Chambers, T. L., Ruf-Zamojski, F., Mendelev, N., Cheng, W. S., Zamojski, M., Amper, M. A., Nair, V. D., Marderstein, A. R., Montgomery, et al
  2025

- **Integration of transcriptomics and long-read genomics prioritizes structural variants in rare disease.** *Genome research*
  Jensen, T. D., Ni, B., Reuter, C. M., Gorzynski, J. E., Fazal, S., Bonner, D., Ungar, R. A., Goddard, P. C., Raja, A., Ashley, E. A., Bernstein, J. A., Zuchner, S., Greicius, et al
  2025

- **DragonRNA: Generality of DNA-primed RNA-extension activities by DNA-directed RNA polymerases.** *Nucleic acids research*
  Greenwald, E., Galls, D., Park, J., Jain, N., Montgomery, S. B., Roy, B., Yin, Y. W., Fire, A. Z.
  2025; 53 (6)

- **Mapping the regulatory effects of common and rare non-coding variants across cellular and developmental contexts in the brain and heart.** *bioRxiv : the preprint server for biology*
  Marderstein, A. R., Kundu, S., Padhi, E. M., Deshpande, S., Wang, A., Robb, E., Sun, Y., Yun, C. M., Pomales-Matos, D., Xie, Y., Nachun, D., Jessa, S., Kundaje, et al
  2025

- **Functional analysis of cancer-associated germline risk variants.** *Nature genetics*
  Kellman, L. N., Neela, P. H., Srinivasan, S., Siprashvili, Z., Shanderson, R. L., Hong, A. W., Rao, D., Porter, D. F., Reynolds, D. L., Meyers, R. M., Guo, M. G., Yang, X., Zhao, et al

2025

- **Exercise intensity and training alter the innate immune cell type and chromosomal origins of circulating cell-free DNA in humans.** *Proceedings of the National Academy of Sciences of the United States of America*
  Rodrigues, K. B., Weng, Z., Graham, Z. A., Lavin, K., McAdam, J., Tuggle, S. C., Peoples, B., Seay, R., Yang, S., Bamman, M. M., Broderick, T. J., Montgomery, S. B.
  2025; 122 (3): e2406954122

- **The human and non-human primate developmental GTEx projects** *NATURE*
  Coorens, T. H. H., Guillaumet-Adkins, A., Kovner, R., Linn, R. L., Roberts, V. H. J., Sule, A., Van Hoose, P. M., the dGTEx Consortium, T.
  2025; 637 (8046): 557-564

- **regionalpcs improve discovery of DNA methylation associations with complex traits.** *Nature communications*
  Eulalio, T., Sun, M. W., Gevaert, O., Greicius, M. D., Montine, T. J., Nachun, D., Montgomery, S. B.
  2025; 16 (1): 368

- **Transcriptome-wide outlier approach identifies individuals with minor spliceopathies.** *medRxiv : the preprint server for health sciences*
  Arriaga, M. T., Mendez, R., Ungar, R. A., Bonner, D. E., Matalon, D. R., Lemire, G., Goddard, P. C., Padhi, E. M., Miller, A. M., Nguyen, J. V., Ma, J., Smith, K. S., Scott, et al
  2025

- **GREGoR: Accelerating Genomics for Rare Diseases.** *ArXiv*
  Dawood, M., Heavner, B., Wheeler, M. M., Ungar, R. A., LoTempio, J., Wiel, L., Berger, S., Bernstein, J. A., Chong, J. X., Délot, E. C., Eichler, E. E., Gibbs, R. A., Lupski, et al
  2024

- **High-coverage nanopore sequencing of samples from the 1000 Genomes Project to build a comprehensive catalog of human genetic variation.** *Genome research*
  Gustafson, J. A., Gibson, S. B., Damaraju, N., Zalusky, M. P., Hoekzema, K., Twesigomwe, D., Yang, L., Snead, A. A., Richmond, P. A., De Coster, W., Olson, N. D., Guarracino, A., Li, et al
  2024

- **Leaving no patient behind! Expert recommendation in the use of innovative technologies for diagnosing rare diseases.** *Orphanet journal of rare diseases*
  van Karnebeek, C. D., O'Donnell-Luria, A., Baynam, G., Baudot, A., Groza, T., Jans, J. J., Lassmann, T., Letinturier, M. C., Montgomery, S. B., Robinson, P. N., Sansen, S., Mehrian-Shai, R., Steward, et al
  2024; 19 (1): 357

- **Single-cell multi-omics map of human fetal blood in Down syndrome.** *Nature*
  Marderstein, A. R., De Zuani, M., Moeller, R., Bezney, J., Padhi, E. M., Wong, S., Coorens, T. H., Xie, Y., Xue, H., Montgomery, S. B., Cvejic, A.
  2024

- **A lymphocyte chemoaffinity axis for lung, non-intestinal mucosae and CNS.** *Nature*
  Ocón, B., Xiang, M., Bi, Y., Tan, S., Brulois, K., Ayesha, A., Kunte, M., Zhou, C., LaJevic, M., Lazarus, N., Mengoni, F., Sharma, T., Montgomery, et al
  2024

- **Deciphering the impact of genomic variation on function.** *Nature*
  2024; 633 (8028): 47-57

- **SINGLE-CELL MULTI-OMICS MAP OF HUMAN FOETAL BLOOD IN DOWN'S SYNDROME**
  Cvejic, A., Marderstein, A., Montgomery, S.
  ELSEVIER SCIENCE INC.2024

- **SINGLE-CELL MULTI-OMICS MAP OF HUMAN FOETAL BLOOD IN DOWN'S SYNDROME**
  Cvejic, A., Marderstein, A., Montgomery, S.
  ELSEVIER SCIENCE INC.2024

- **De novo variants in the RNU4-2 snRNA cause a frequent neurodevelopmental syndrome.** *Nature*
  Chen, Y., Dawes, R., Kim, H. C., Ljungdahl, A., Stenton, S. L., Walker, S., Lord, J., Lemire, G., Martin-Geary, A. C., Ganesh, V. S., Ma, J., Ellingford, J. M., Delage, et al
  2024

- **Impact of genome build on RNA-seq interpretation and diagnostics.** *American journal of human genetics*
  Ungar, R. A., Goddard, P. C., Jensen, T. D., Degalez, F., Smith, K. S., Jin, C. A., Bonner, D. E., Bernstein, J. A., Wheeler, M. T., Montgomery, S. B.
  2024

- **Loss of function of FAM177A1, a Golgi complex localized protein, causes a novel neurodevelopmental disorder.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Kohler, J. N., Legro, N. R., Baldridge, D., Shin, J., Bowman, A., Ugur, B., Jackstadt, M. M., Shriver, L. P., Patti, G. J., Zhang, B., Feng, W., McAdow, A. R., Goddard, et al
  2024: 101166

- **The impact of exercise on gene regulation in association with complex trait genetics.** *Nature communications*
  Vetr, N. G., Gay, N. R., MoTrPAC Study Group, Montgomery, S. B., Adkins, J. N., Albertson, B. G., Amar, D., Amper, M. A., Armenteros, J. J., Ashley, E., Avila-Pacheco, J., Bae, D., Balci, A. T., et al
  2024; 15 (1): 3346

- **Temporal dynamics of the multi-omic response to endurance exercise training.** *Nature*
  2024; 629 (8010): 174-183

- **regionalpcs: improved discovery of DNA methylation associations with complex traits.** *bioRxiv : the preprint server for biology*
  Eulalio, T., Sun, M. W., Gevaert, O., Greicius, M. D., Montine, T. J., Nachun, D., Montgomery, S. B.
  2024

- **Sexual dimorphism and the multi-omic response to exercise training in rat subcutaneous white adipose tissue.** *Nature metabolism*
  Many, G. M., Sanford, J. A., Sagendorf, T. J., Hou, Z., Nigro, P., Whytock, K. L., Amar, D., Caputo, T., Gay, N. R., Gaul, D. A., Hirshman, M. F., Jimenez-Morales, D., Lindholm, et al
  2024

- **Molecular adaptations in response to exercise training are associated with tissue-specific transcriptomic and epigenomic signatures.** *Cell genomics*
  Nair, V. D., Pincas, H., Smith, G. R., Zaslavsky, E., Ge, Y., Amper, M. A., Vasoya, M., Chikina, M., Sun, Y., Raja, A. N., Mao, W., Gay, N. R., Esser, et al
  2024: 100421

- **Molecular Transducers of Physical Activity Consortium (MoTrPAC): Human Studies Design and Protocol.** *Journal of applied physiology (Bethesda, Md. : 1985)*
  Group, M. R., Jakicic, J. M., Kohrt, W. M., Houmard, J. A., Miller, M. E., Radom-Aizik, S., Rasmussen, B. B., Ravussin, E., Serra, M., Stowe, C. L., Trappe, S., AbouAssi, H., Adkins, et al
  2024

- **The mitochondrial multi-omic response to exercise training across rat tissues.** *Cell metabolism*
  Amar, D., Gay, N. R., Jimenez-Morales, D., Jean Beltran, P. M., Ramaker, M. E., Raja, A. N., Zhao, B., Sun, Y., Marwaha, S., Gaul, D. A., Hershman, S. G., Ferrasse, A., Xia, et al
  2024

- **De novo variants in the non-coding spliceosomal snRNA gene RNU4-2 are a frequent cause of syndromic neurodevelopmental disorders.** *medRxiv : the preprint server for health sciences*
  Chen, Y., Dawes, R., Kim, H. C., Stenton, S. L., Walker, S., Ljungdahl, A., Lord, J., Ganesh, V. S., Ma, J., Martin-Geary, A. C., Lemire, G., D'Souza, E. N., Dong, et al
  2024

- **Integration of transcriptomics and long-read genomics prioritizes structural variants in rare disease.** *medRxiv : the preprint server for health sciences*
  Jensen, T. D., Ni, B., Reuter, C. M., Gorzynski, J. E., Fazal, S., Bonner, D., Ungar, R. A., Goddard, P. C., Raja, A., Ashley, E. A., Bernstein, J. A., Zuchner, S., Greicius, et al
  2024

- **Nanopore sequencing of 1000 Genomes Project samples to build a comprehensive catalog of human genetic variation.** *medRxiv : the preprint server for health sciences*
  Gustafson, J. A., Gibson, S. B., Damaraju, N., Zalusky, M. P., Hoekzema, K., Twesigomwe, D., Yang, L., Snead, A. A., Richmond, P. A., De Coster, W., Olson, N. D., Guarracino, A., Li, et al
  2024

- **RNA Sequencing in Disease Diagnosis.** *Annual review of genomics and human genetics*
  Smail, C., Montgomery, S. B.
  2024

- **Impact of genome build on RNA-seq interpretation and diagnostics.** *medRxiv : the preprint server for health sciences*
  Ungar, R. A., Goddard, P. C., Jensen, T. D., Degalez, F., Smith, K. S., Jin, C. A., Bonner, D. E., Bernstein, J. A., Wheeler, M. T., Montgomery, S. B.
  2024

- **Detection and analysis of complex structural variation in human genomes across populations and in brains of donors with psychiatric disorders** *Cell*
  Zhou, B., Arthur, J. G., Guo, H., et al
  2024; Published online September 30, 2024

- **Genetic architecture of cardiac dynamic flow volumes.** *Nature genetics*
  Gomes, B., Singh, A., O'Sullivan, J. W., Schnurr, T. M., Goddard, P. C., Loong, S., Amar, D., Hughes, J. W., Kostur, M., Haddad, F., Salerno, M., Foo, R., Montgomery, et al
  2023

- **Organ aging signatures in the plasma proteome track health and disease.** *Nature*
  Oh, H. S., Rutledge, J., Nachun, D., Pálovics, R., Abiose, O., Moran-Losada, P., Channappa, D., Urey, D. Y., Kim, K., Sung, Y. J., Wang, L., Timsina, J., Western, et al
  2023; 624 (7990): 164-172

- **Transcriptomics and chromatin accessibility in multiple African population samples.** *bioRxiv : the preprint server for biology*
  DeGorter, M. K., Goddard, P. C., Karakoc, E., Kundu, S., Yan, S. M., Nachun, D., Abell, N., Aguirre, M., Carstensen, T., Chen, Z., Durrant, M., Dwaracherla, V. R., Feng, et al
  2023

- **Multi- Omic Profiling of Macrophages Lacking Tet2 or Dnmt3a Reveals Mechanisms of Hyper-Inflammation in Clonal Hematopoiesis**
  Rodrigues, K. B., Gopakumar, J., Weng, Z., Mitchell, S., Maurer, M., Nachun, D., Eulalio, T., Estrada, D., Mazumder, T., Ma, L., Montgomery, S., Jaiswal, S.
  AMER SOC HEMATOLOGY.2023

- **Integrative analyses highlight functional regulatory variants associated with neuropsychiatric diseases.** *Nature genetics*
  Guo, M. G., Reynolds, D. L., Ang, C. E., Liu, Y., Zhao, Y., Donohue, L. K., Siprashvili, Z., Yang, X., Yoo, Y., Mondal, S., Hong, A., Kain, J., Meservey, et al
  2023

- **The functional impact of rare variation across the regulatory cascade.** *Cell genomics*
  Li, T., Ferraro, N., Strober, B. J., Aguet, F., Kasela, S., Arvanitis, M., Ni, B., Wiel, L., Hershberg, E., Ardlie, K., Arking, D. E., Beer, R. L., Brody, et al
  2023; 3 (10): 100401

- **Integrated single-cell multiome analysis reveals muscle fiber-type gene regulatory circuitry modulated by endurance exercise.** *bioRxiv : the preprint server for biology*
  Rubenstein, A. B., Smith, G. R., Zhang, Z., Chen, X., Chambers, T. L., Ruf-Zamojski, F., Mendelev, N., Cheng, W. S., Zamojski, M., Amper, M. A., Nair, V. D., Marderstein, A. R., Montgomery, et al
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